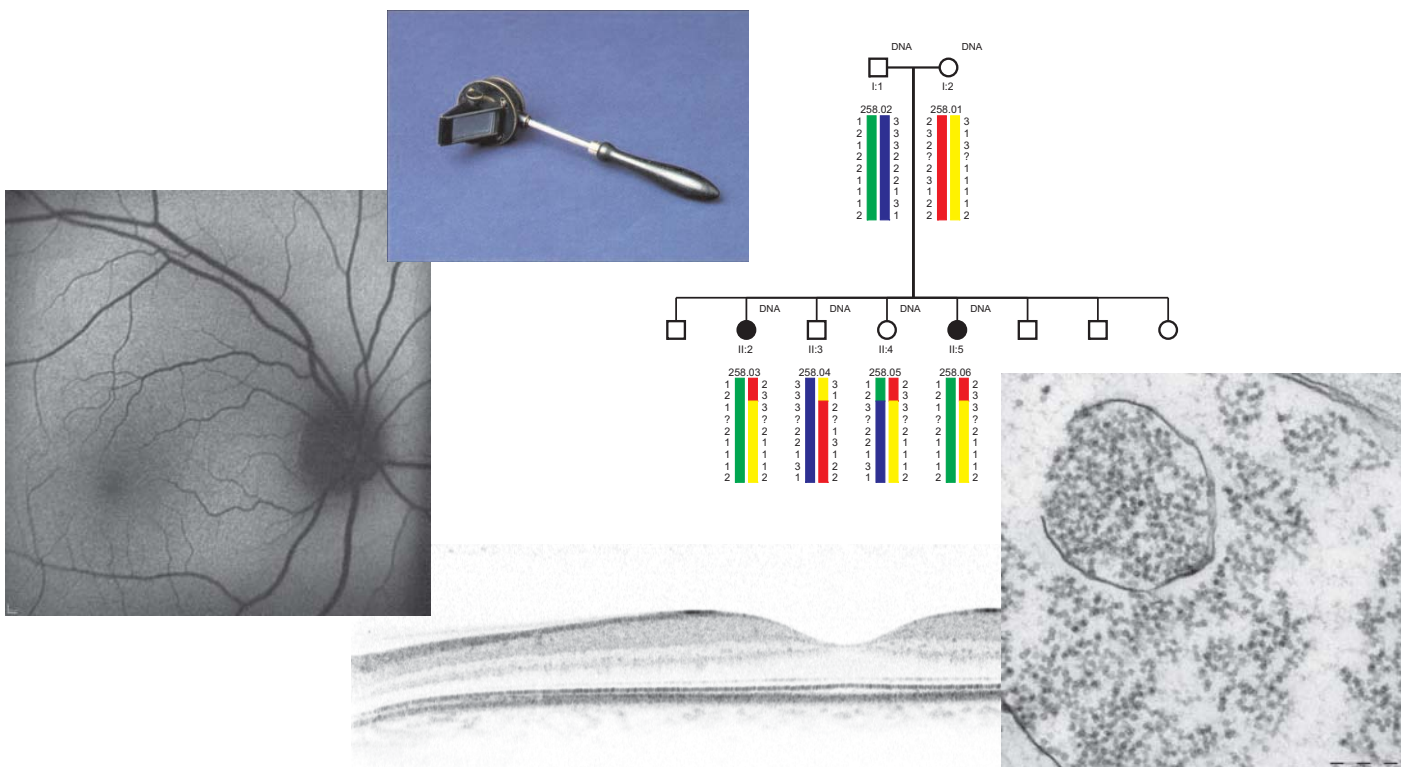


36<sup>th</sup> Annual Meeting of the  
European Paediatric Ophthalmological Society  
Hotel Dolce, Bad Nauheim, Germany  
30.09. - 02.10.2010

EPOS 2010

# New Challenges in Paediatric Ophthalmology

Local Hosts: Birgit Lorenz, Markus Preising



Proceedings of the 36<sup>th</sup> Annual Meeting of the European Paediatric Ophthalmological Society (EPOS) 2010

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## Welcome by the President of EPOS

Dear Friends, dear Members, dear Colleagues,

It is our great pleasure to welcome you to the 36<sup>th</sup> Annual Meeting of the European Paediatric Ophthalmological Society EPOS here in Bad Nauheim which is very close to our home university in Giessen. It is a very special congress for me, as it will be the last under my presidency.

The Scientific Program Committee consisted of the board members of EPOS Ingele Casteels, Pascal Dureau, Albert Francescetti, Nicoline Schalijs-Delfos, Branka Stirn Kranjc, Gabriela Wirth-Barben, Markus Preising and myself. Markus and I would like to express our sincere thanks to all board members who helped to put together again a very interesting scientific program.

This year, the main topic is "New Challenges in Paediatric Ophthalmology and as always, free papers and posters from other fields of genetic and paediatric ophthalmology are also scheduled. Almost one third of the 69 free contributions will be presented by young researchers who will be evaluated during the meeting to receive one of the Best Presentation Awards in the two categories poster and paper.

Outstanding speakers have accepted our invitation to present at the meeting: Under the title "Go with the in-crowd" Jean Bennett will give a key lecture on her latest results related to human RPE65 gene therapy. Challenges in genotyping will be addressed by Frans Cremers and Andreas Gal. As an expanding society mirroring an expanding EU we have selected to have a session on "Developing countries: Epidemiologic, technical and financial challenges". We are expecting outstanding data presented by Richard Bowman, Constanta Nascutzy and Tatjana Ciomartan. Exciting data come from new imaging methods. In the session "I spy with my little eye: Challenges in imaging and evaluation" David Wallace will report on his exciting research. In two sessions an "Invitation to dance" reports on the challenges in interdisciplinary research that will be highlighted by Hans-Peter Hammes, Silke Haverkamp, Michael Hofmann, and Klaus Preissner.

One key mission of EPOS is to promote clinical and scientific paediatric ophthalmology throughout Europe and beyond, and in particular to attract young clinicians and researchers in the field. In this spirit, this meeting continues to have, in addition to keynote lectures, papers with sufficient time for discussion and ample poster sessions that encourage vivid exchange of ideas. This year, we will have 12 keynote lectures, 27 free papers, and 43 posters. Researchers from as many as 26 countries will present their results, and we are happy to welcome an increasing number of colleagues from Europe's neighbour states in North Africa and the Middle East.

The General Assembly of the EPOS will be held on Saturday, from 11.15 h to 12.30 h. The agenda has been circulated to all members. Attendance is extremely important since all members have the opportunity to design the aims of the society and propagate paediatric ophthalmology throughout Europe. The President, Treasurer, and Secretary are completing their current terms this year and



## Welcome by the President of EPOS

new officers need to be elected to these posts. Our statutes have served us well for the past ten years but now the first amendments proposed need to be discussed and implemented.

As in previous years, we would like to thank all exhibitors and sponsors for their significant efforts in supporting the meeting, and encourage all participants to visit the technical exhibit during coffee breaks and the scientific lunch symposium by Théa on Friday.

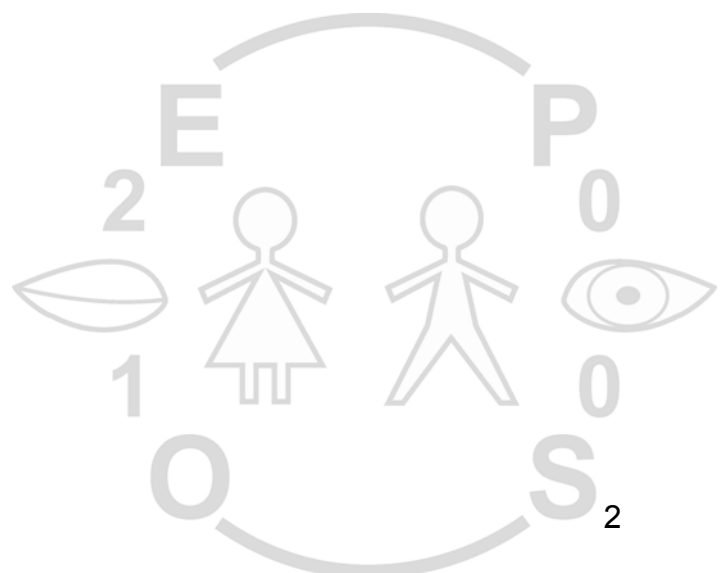
Further, we wish to express our sincere thanks to Christine Mais, the thesis students and the staff of the Laboratory for Molecular Ophthalmology of the Department of Ophthalmology of the University of Giessen who helped with the arrangements to make this meeting go smoothly and agreeably for all of us.

Since the foundation of EPOS, formerly EPOG, numerous fruitful relations have been built up among EPOS members and continue to grow by including new clinicians and researchers in the very dynamic field of Paediatric and Genetic Ophthalmology.

Markus and I hope that this year's meeting will again trigger new fruitful collaborations and personal contacts between participants to make the Society grow even further as well as strengthening old friendships.

Birgit Lorenz, MD  
Congress President  
President of EPOS

Markus Preising, PhD  
Congress President





## Welcome by the Dean of the Medical Faculty of the Justus Liebig University

Dear Prof. Lorenz, Distinguished Guests and Speakers, Ladies and Gentlemen,

Let me, on behalf of the University of Giessen and its Faculty of Medicine, extend a very warm welcome to the delegates and participants of the European Paediatric Ophthalmological Society here at Bad Nauheim.

Vision is one of our most important senses and it is estimated that about one-third of the brain function is involved in visual processing. Visual impairment in childhood has a major impact on mental processes and personal development of a child with important social consequences. Therefore identification and prevention of visual processing defects is a key to progress in disease alleviation.

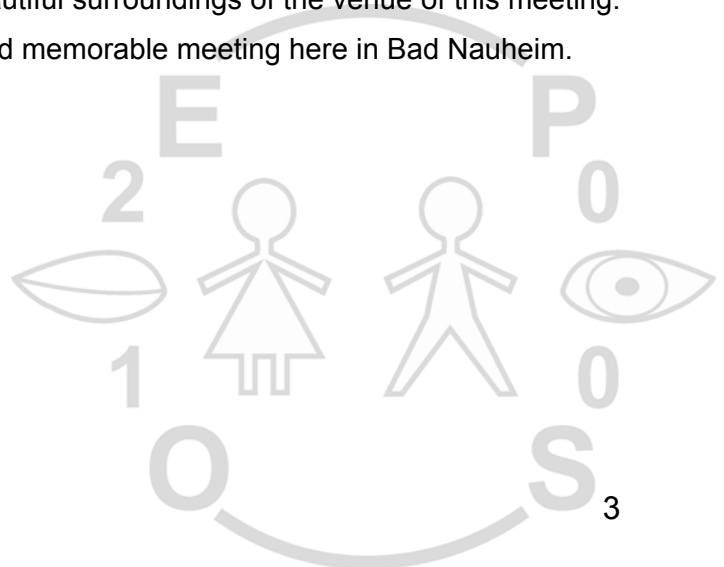
The paediatric ophthalmologist holds a key position in the interaction with other specialist fields of medicine such as in paediatrics, neuropaediatrics, ear-nose and throat, craniofacial surgery, endocrinology, neuroradiology, immunology and genetics. Indeed, the paediatric ophthalmologist is a team player translating diagnosis into therapeutic options and rehabilitation.

This year's meeting focuses on current challenges in paediatric ophthalmology and has a broad agenda. This is mirrored in many of the talks and posters to be presented here over the next two days that will feature developments in diagnostics and gene therapy, and discuss problems in implementing state-of-the art techniques in developing countries where needs are pressing

As with many clinical vocations there is a need to attract and support young clinicians and researchers for future tasks and challenges in paediatric ophthalmology. I am delighted to learn that EPOS has therefore implemented travel- and research- grants for young ophthalmologists and researchers.

The organizers of this meeting are to be congratulated on putting together an excellent program with distinguished local, national and international speakers. Weather permitting; I would encourage you to use your vision to take in the beautiful surroundings of the venue of this meeting. Finally, I wish you all an eye-opening, enjoyable and memorable meeting here in Bad Nauheim.

Trinad Chakraborty  
Dean, Faculty of Medicine





## Information on Bad Nauheim

Ladies and Gentlemen,

I am very pleased by your decision to choose the health city Bad Nauheim to host the 36<sup>th</sup> Annual Meeting of the European Paediatric Ophthalmological Society. Further more the venue, Congress Hotel Dolce, provides a superior choice of an ambience for your ambitious programme.

Exchange among colleagues and the opportunity to experience lectures on up to date research by world wide renowned scientists are mandatory in times of quickly emerging medical knowledge especially in hereditary and paediatric eye disorders. You get the opportunity to discuss topics related to your profession and to enlarge your knowledge since high quality service depends on recent and comprehensive information.

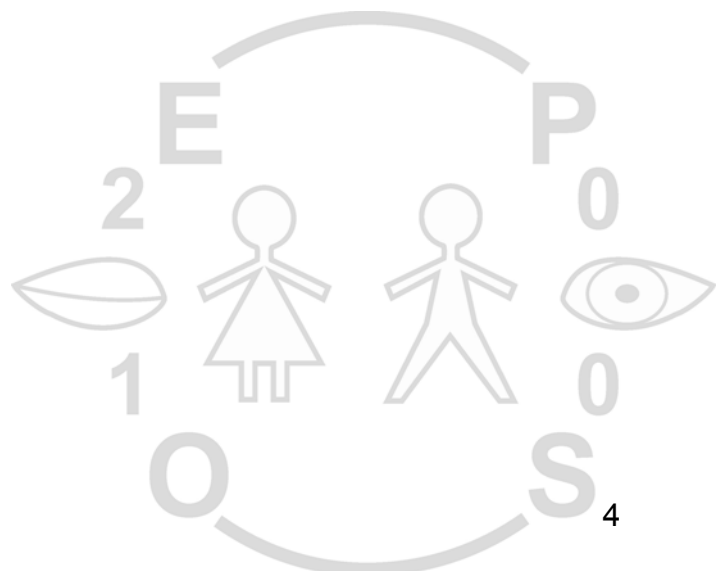
The evening social with charity concert and charity dinner promises to become an extraordinary event. All those who contribute to the evening social may feel as goodwill ambassador. Your participation supports precious research projects and shows your appreciation of the efforts taken on these topics.

I like to express my gratitude to the organizers and all those involved in the success of the meeting. Take the chance to extend your views and knowledge with your colleagues from almost 30 countries.

I wish you highly informative presentations, active discussions, and a convenient stay in our beautiful art nouveau and health city Bad Nauheim. Take your chance to visit the terrain of the Landesgartenschau 2010 in our town. You will find many inspirations for a timely garden design based on a fortunate and successful alliance of utility space and recreation area!

Yours

Bernd Witzel  
Mayor





## **Art Nouveau spa with unique options of well-being**

Over many years tradition and an innovative research made Bad Nauheim a leading medical location. The city of health is not only centre of famous hospitals but also a place for well-being. In front of a gorgeous Art Nouveau heritage different kinds of spa-water provide an ideal opportunity for regeneration and healing. The surname “Three empresses’ spa” indicates Bad Nauheim used to be a place for the European nobility. Crowned heads like the Austrian empress Elisabeth, the German empress Auguste Viktoria or the Russian tsarina Alexandra came to Bad Nauheim as well as politicians, intellectuals and other notables. The curative effect of the salted and carbonated water especially for heart cases spread among doctors and patients even far beyond the European borders.

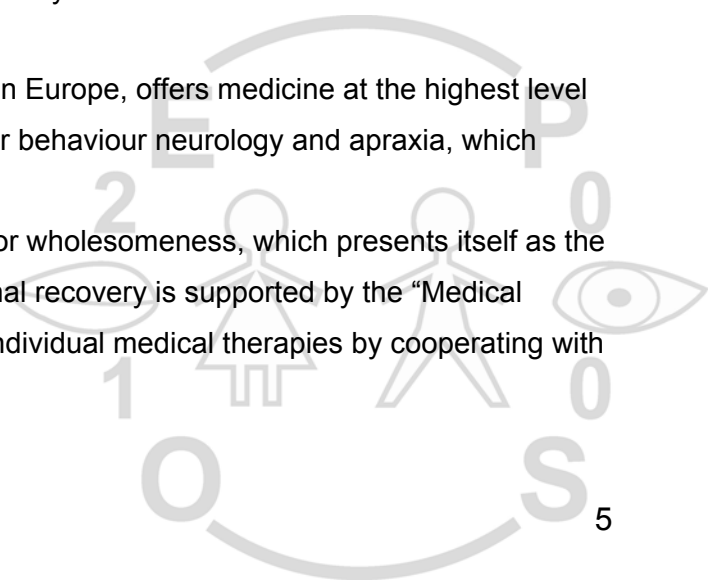
## **Leading medicine for the good of the patients**

Still today Bad Nauheim has an excellent reputation as a centre of excellence for heart, and cardiovascular diseases. Specialized hospitals of international reputation take care of the well-being of their guests. The Max-Planck-Institute for Heart and Lung Research, which is closely linked to the Kerckhoff campus, is very important for the city as a centre of science. The Kerckhoff campus consists of the newly erected Kerckhoff Klinik, the Kerckhoff rehabilitation centre, as well as the Diabetes-Klinik of the Pitzer group and the HELIOS William Harvey Klinik for vascular diseases. Patients benefit from this unique centre for heart and vascular diseases in Germany by the treatment of a broad spectrum of disease. The close cooperation between the hospitals results in an ideal attendance for the patients, in short distances and in fast diagnoses as well as in closely coordinated therapies.

World-famous athletes appreciate the professional competence and the flair of the spa. Many professional athletes like the Formel 1 champion Michael Schumacher visit the “Sportklinik” in Bad Nauheim regularly to gain fitness at the highest level. During the football world championship in 2006 Bad Nauheim was the accommodation for the team of Saudi Arabia, half century after King Ibn Saud already visited Bad Nauheim with his whole royal household to take a medical rehab for four weeks.

The hospital for Parkinsonism, which is the biggest in Europe, offers medicine at the highest level and collaborates with the Soemmerring institution for behaviour neurology and apraxia, which linked to the universities of Gießen and Marburg.

Bad Nauheim is a place of international reputation for wholesomeness, which presents itself as the first place to go also for foreign patients. The personal recovery is supported by the “Medical Coordinator” who is responsible for organising the individual medical therapies by cooperating with all hospitals and leading doctors.





## **Unique Art Nouveau ensemble**

The salted water which has its source between the Taunus and the Wetterau region since 500.000 years and already provided wealth and power to the Celts, constituted Bad Nauheim's rise to a world spa of empresses and kings. The change to a spa took place in the first half of the 19<sup>th</sup> century when the healing power of the brine spring was discovered. To fulfil the the rising demands of the bathers, Wilhelm Jost got an order from the art loving emperor Ernst Ludwig von Hessen and bei Rhein to build a spacious bathing complex. Between 1905 and 1912 arose the Sprudelhof with its arcaded framing, six bath houses including approximately 267 cubicles, the Trinkkuranlage, a hall for concerts and theatres as well as buildings for administration and engines. Only the best material was used in an integrated work of art consisting of many small details. In bath house 3 you can experience the bathing from the past with all its gorgeous decorated waiting and recreation rooms and the Australian wooden bath tubs. The quiet inner courtyards and gardens of the bath houses, which are probably the most important resort experiences because of its amenity and its beauty, are still used as an area of relaxation and quietness after a bath. Thanks to the biggest complete Art Nouveau building of Europe, Bad Nauheim became the first German member of the EU-promoted "Reseau Art Nouveau Network" which is a cooperation between 19 European cities like Barcelona, Brussels and Budapest.

The brilliant height of the Art Nouveau Spa can be experienced closely in September during the Art Nouveau festival. Music, literature, movie, culinary art, architecture and fashion – the Art Nouveau festival emblazes a variety of facets of the art genre like guided tours through the bath houses, concerts or expositions.

## **Regenerating with an exhilarant sea breeze**

Located idyllically on the brink of the Taunus, Bad Nauheim offers a varied programme all about health and "Medical Wellness". The spa treatments go from Ayurveda to yoga. Treatments are also available in the "Therme am Park", which invites its guests to a time full of pleasant bathing and relaxing in the 36° hot thermal pools. Due to the wave machine, flumes and Jacuzzis, the "Usa-Wellenbad" (swimming pool) inspires the young and the old.

Another way to perceive the healing effect of the water is by inhalation. Five salt works (graduation houses) with a length of 650 m as a whole arrange for the trickling of the brine. In this process the finest salted droplets are set free and ease the mucous membrane of the respiratory system. In the inhalation room you can enjoy the healthy sea breeze very intensively. Near the salt work II the health garden with its 15 stations provides new experiences for all senses.

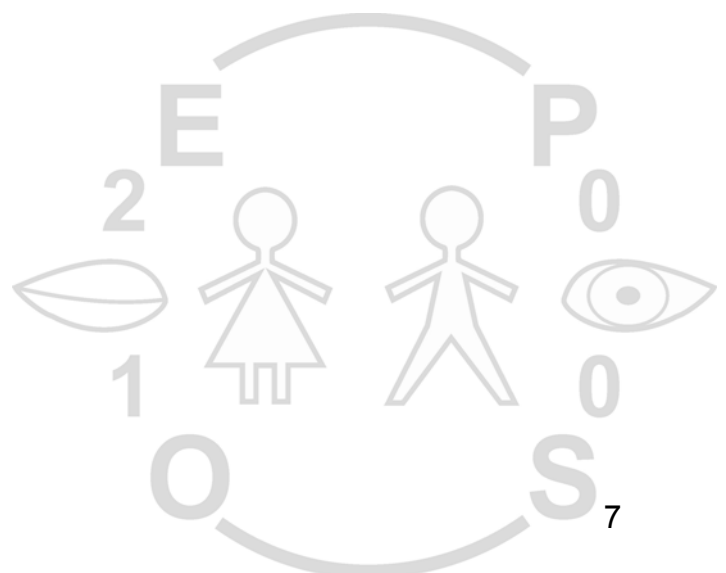


## **On the traces of Elvis**

Even Elvis Presley stocked up on the bracing sea breeze in his “European home” Bad Nauheim. Who wants to follow the traces of the “King of Rock ‘n’ Roll” can join a guided tour which includes the dwelling houses where the legendary superstar used to live in his period of military service from 1958 to 1960. The European Elvis Festival in August attracts Elvis-Fans from all over the world to watch concerts from well-known interpreters or parades of Cadillacs and Harleys.

## **Fascination roses**

A visit to Bad Nauheim is especially worth in summer when all rose gardens flourish. The district Steinfurth has a very long tradition back to 1868 when Heinrich Schultheis founded the first school of roses. Until today the roses from Steinfurth have an excellent international reputation: due to their robust quality and the high duration of blossoming, they are shipped to many places throughout the world. The museum of roses is unique in the world and expresses the beauty of the flower in an artistic and literate way. Every second year the gardeners of Steinfurth celebrate the queen of flowers with the big rose festival. The highlight of the festival is a gorgeous rose-parade led by the rose queen.



**Thursday 30.09.2010**

**17.30 h** **Congress Bag Counter opens for pre-registered participants**

**18.00 – 20.00 h** **Onsite Registration**

**18.00 – 22.00 h** **Opening Reception**

**20.00 h** **Board Meeting**

**Friday 01.10.2010**

**8.30 h Birgit Lorenz** **Welcome by the President and Local Host**

**8.40 h Trinad Chakraborty** **Welcome by the Dean of the Medical Faculty of the Justus-Liebig University Giessen**

**8.50 – 10.00 h I. Scientific Session** **Developing Countries – Epidemiologic, Technical and Financial Challenges**

**Chairperson David Wallace –**  
**Nadiya Bobrova**

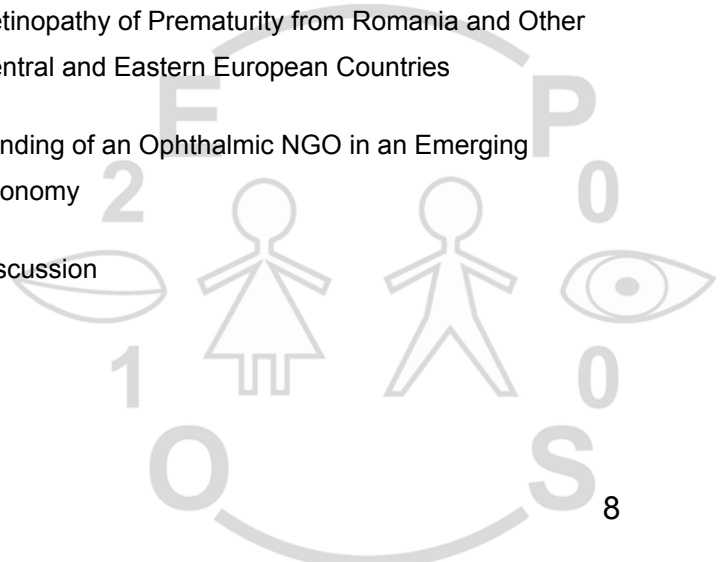
**L1** 8.50 h **Richard Bowman** **Challenges of Managing Paediatric Cataract in Sub Saharan Africa**

**L2** 9.10 h **Constanta Nascutz** **Challenges in the Screening and Treatment of Retinopathy of Prematurity in Romania and Other Central and Eastern European Countries in Europe**

**L3** 9.30 h **Tatiana Ciomartan** **Challenges in the Care of Preterm Babies with Retinopathy of Prematurity from Romania and Other Central and Eastern European Countries**

**T1** 9.40 h **Raymond Brown** **Funding of an Ophthalmic NGO in an Emerging Economy**

9.50 h **Discussion**



**Topic Related Posters in Friday Poster Session**

- |           |                 |  |
|-----------|-----------------|--|
| <b>P1</b> | Nicolaos Ziakas | Preschool Visual Screening in the Municipality of Thessaloniki |
| <b>P2</b> | Patricia Domsa  | Place of Web-Based Visual Screening in Pediatric Eye Care      |

10.00 – 10.30 h

**Coffee Break**

10.30 - 11.45 h

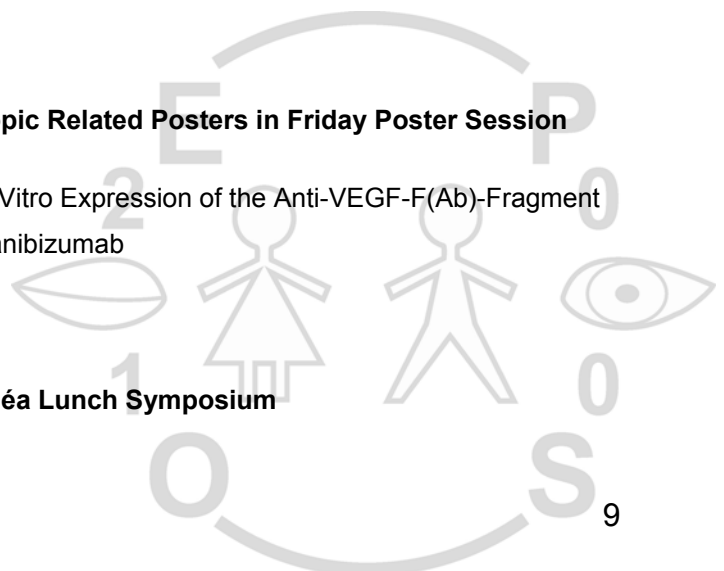
**II. Scientific Session****Invitation to Dance –  
The Challenge of Interdisciplinary Research I  
Neovascularization and the Eye****Chairperson****Klaus T. Preissner –  
Pascal Dureau**

- |           |         |                    |   |
|-----------|---------|--------------------|---|
| <b>L4</b> | 10.30 h | Hans-Peter Hammes  | Neovascularization in the Eye – Lessons from Translational Blood Vessel Research                                |
| <b>L5</b> | 10.50 h | Klaus T. Preissner | Challenges in the Treatment of Pathological Neovascularization: New Players and Mechanisms                      |
| <b>T2</b> | 11.10 h | Birgit Lorenz      | The Giessen Experience with Anti-VEGF Therapy for APROP   |
| <b>T3</b> | 11.20 h | Phanthipha Wongwai | Outcome of Adjunctive Intravitreal Bevacizumab Combined with Laser Indirect Ophthalmoscopy for Treatment of ROP |
|           | 11.30 h |                    | Discussion  |

**Topic Related Posters in Friday Poster Session**

- |           |               |   |
|-----------|---------------|---|
| <b>P3</b> | Tobias Wimmer | In Vitro Expression of the Anti-VEGF-F(Ab)-Fragment Ranibizumab |
|-----------|---------------|---|

11.45 – 12.15 h

**Théa Lunch Symposium**



12.15 – 13.30 h

**Théa Luncheon**

13.30 – 14.30 h

**Friday Poster Session****Posters unrelated to Paper Sessions**

<b>P4</b>	Nikolaos Kozeis	Keratitis in Reiter's Syndrome in Childhood
<b>P5</b>	Aneta Skupin	Oculoglandular Syndrom Parinaud
<b>P6</b>	Nicolaos Ziakas	Bilateral Traumatic Optic Neuropathy in Child - Case Report
<b>P7</b>	Victoria Balasanyan	And where is Pathology at Oculomotor Pareses? New Approach to Old Challenge
<b>P8</b>	Nikolaos Kozeis	Visual Skills and Gross Motor Function in Spastic Diplegic Children
<b>P9</b>	Judit Körtvélyes	Impaired Foveal and Peripheral Face Processing in Amblyopia
<b>P10</b>	Giorgio Porro	Visual Fields Defects by Sturge Weber Syndrome
<b>P11</b>	Nikolaos Kozeis	Visual Function vs. Psychomotor Development of Premature Infants with No Severe Adverse Effects

14.30 – 16.00 h

**III. Scientific Session****Go with the In-Crowd –****Challenged by Up to Date Science I****Phenotypes Between New Genes and Modifiers**

**Chairperson: Andreas Gal –  
Matthias Seeliger**

<b>L6</b>	14.30 h	Frans Cremers	Homozygosity Mapping in Nonconsanguineous Families Facilitates the Identification of Novel Retinal Dystrophy Genes
<b>L7</b>	14.50 h	Andreas Gal	Genetic Modifiers of Disease Phenotype
<b>T4</b>	15.10 h	Hanno J. Bolz	PDZD7 is a Modifier of Retinal Disease and a Contributor to Digenic Usher Syndrome



<b>T5</b>	15.20 h	Hélène Dollfus	How Many Genes for Bardet-Biedl Syndrome?
	15.30 h		Discussion

**Topic Related Posters in Friday Poster Session**

<b>P12</b>		Konstantinos Aliferis	Overlapping of Alström and Bardet-Biedl Syndrome Early Phenotype Confirmed by Systematic High Throughput Ciliopathy Genes Sequencing
<b>P13</b>		Jutta Hosch	Development of a Humanized Mouse-Model for X-Linked Retinitis Pigmentosa Caused by a Point Mutation in the <i>RPGR</i> Gene

16.00 – 16.30 h

**Coffee Break**

16.30 – 17.50 h    **IV. Scientific Session    Go with the In-Crowd –  
Challenged by Up to Date Science II  
Gene Therapy**

**Chairperson:    Frans Cremers –  
Knut Stieger**

<b>L8</b>	16.30 h	Jean Bennett	Restoration of Cortical Vision After Gene Therapy for Congenital Blindness
<b>L9</b>	16.50 h	Mathias Seeliger	Molecular Therapy in Retinal Degenerations – What's in the Pipeline?
<b>T6</b>	17.10 h	Knut Stieger	Evaluation of AAV Mediated Gene Therapy for <i>RPE65</i> Patients by Highly Sensitive Psychophysical Techniques
<b>T7</b>	17.20 h	Mariya Moosajee	Novel S/MAR Vectors Provide a Non-Viral Gene Therapy for Choroideraemia
	17.30 h		Discussion

**Topic Related Posters in Friday Poster Session**

**P14** Bert Constantin Giers Immunohistochemical Characterization of AAV Transduced Retinae Following Subretinal Injection in Rats

**19.00 h** Limburger  
Domsingknaben and  
Alison Browner

**Charity Concert at the Theater Dolce**

**20.30 h**

**Charity Dinner at the Hotel Dolce**

**Patron: Prof. Dr. phil. Joachim-Felix Leonhard**

*Saturday*

**02.10.2010**

**8.00 – 9.20 h V. Scientific Session Challenges in Treatment**

**Chairperson: Nicoline Schalijs-Delfos - Ingele Casteels**

**T8** 8.00 h Alicia Serra Our Experience with Paediatric Aphakic Glaucoma

**T9** 8.10 h Marije Sminia Corneal Endothelial Cell Density after Artisan Aphakia IOL Implantation for Crystalline Lens Subluxation in Marfan Syndrome.

**T10** 8.20 h Nadiya Bobrova New Treatment Modality of Combined (Local and Systemic) Retinoblastoma Chemotherapy. (First Results).

**T11** 8.30 h Marie-Claire Gaillard Ranibizumab (Lucentis®) in the Management of Late-Stage Coats' Disease

**T12** 8.40 h Tomoko Maeda-Chubachi Phase 3, Prospective, 12-Week, Double-Masked, Multicentre Study of Latanoprost (LAT) and Timolol (TIM) in Paediatric Glaucomas: Age and Diagnosis Subgroup Analysis



**T13** 8.50 h Francis L. Munier Preliminary Results of Superselective Ophthalmic Artery Chemotherapy (SOAC) in 14 Patients with Advanced Retinoblastoma

**T14** 9.00 h Anton Gerinec Orbital Tumors in Children

9.10 h Discussion

### **Topic Related Posters in Saturday Poster Session**

**P15** Nadiya Bobrova Liquid Implant in Congenital Glaucoma Surgical Treatment.

**P16** Nadiya Bobrova Infants Implantation Surgery – New Approaches

**P17** Birgit Lorenz Secondary Infantile Cataract Associated with Presumed Intrauterine Infection

**P18** Werner Schmidt Cataract Surgery and Postoperative Outcome in a Child with Hallermann-Streiff Syndrome with Dysproportional Microphthalmia

**P19** Marta Morales Uveal Effusion Syndrome in Hallermann-Streiff Syndrome

**P20** Gianfranco Bellizzi Treatment of Severe Vernal Keratoconjunctivitis with 1% Topical Cyclosporine in Children

**P21** Anne-Claudia Stefanut The Management of the Bilateral Palpebral Necrotizing Fasciitis in a Newborn with Agammaglobulinemia Bruton

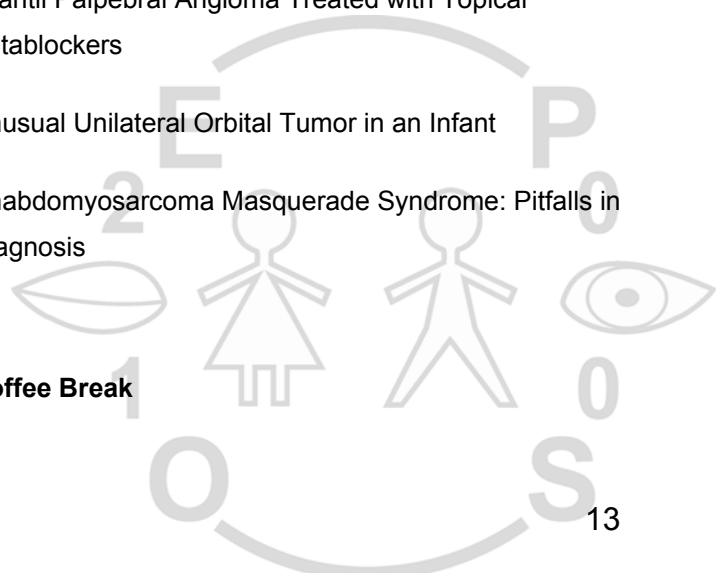
**P22** Julia Escudero Infantil Palpebral Angioma Treated with Topical Betablockers

**P23** Melanie Jäger Unusual Unilateral Orbital Tumor in an Infant

**P24** Reshma Thampy Rhabdomyosarcoma Masquerade Syndrome: Pitfalls in Diagnosis

**9.20 – 9.50 h**

**Coffee Break**



**9.50 – 10.50 h VI. Scientific Session Retinopathy of Prematurity****Chairperson: Constanta Nascutzy –  
Gerd Holmström**

- |            |         |                          |  |
|------------|---------|--------------------------|--|
| <b>T15</b> | 9.50 h  | Dordi Austeng            | Screening for Retinopathy of Prematurity in Infants Born Before 27 Weeks of Gestation in Sweden  |
| <b>T16</b> | 10.00 h | Nicoline Schalijs-Delfos | The Incidence of Visual Impairment Due to ROP and Their Concomitant Disabilities in the Netherlands. A Thirty Year Overview.                               |
| <b>T17</b> | 10.10 h | Arlette van Sorge        | Preliminary Results of the NEDROP-Study: a National Inventory on Screening for Retinopathy of Prematurity.   |
| <b>T18</b> | 10.20 h | J Kate Barnes            | Five Year Visual Outcome of Children, Following Treatment with Diode Laser for Retinopathy of Prematurity Under Sub-Tenon's Local Anaesthesia.             |
| <b>T19</b> | 10.30 h | Catherine Cassiman       | Functional and Structural Ophthalmological Outcome in Cryo- or Laser Treated Premature Babies with Retinopathy of Prematurity (ROP) Between 1989 and 2008. |
|            | 10.40 h |                          | Discussion   |

**Topic Related Posters in Saturday Poster Session**

- |            |  |                                  |   |
|------------|--|----------------------------------|---|
| <b>P25</b> |  | Nicolaos Ziakas                  | Screening of Retinopathy of Prematurity Using Sucrose As Analgesia                                  |
| <b>P26</b> |  | Isabel George or Karel Allegaert | Early Neonatal Creatinaemia is an Indicator of the Subsequent Risk to Develop Threshold Retinopathy |
| <b>P27</b> |  | Anne-Claudia Stefanut            | Oxidative Stress - a Biomarker in Retinopathy of Prematurity  |
| <b>P28</b> |  | Anne-Claudia Stefanut            | Oxidative Stress Parameters in a Model of Oxygen Induced Retinopathy                                |
| <b>P29</b> |  | Nadiya Bobrova                   | Efficacy of Green Laser Photocoagulation for  |



		Aggressive Posterior Retinopathy of Prematurity
<b>P30</b>	Dana Tomcikova	Benefit of Paint Diode Laser Coagulation in Treatment of ROP.
<b>P31</b>	Natalya Fomina	The Challenge of the Relations Between the Parents of the Premature Babies and Paediatric Ophthalmologists.

**11.00 – 12.15 h**

**EPOS General Assembly**

**12.15 – 13.45 h**

**Lunch Break**

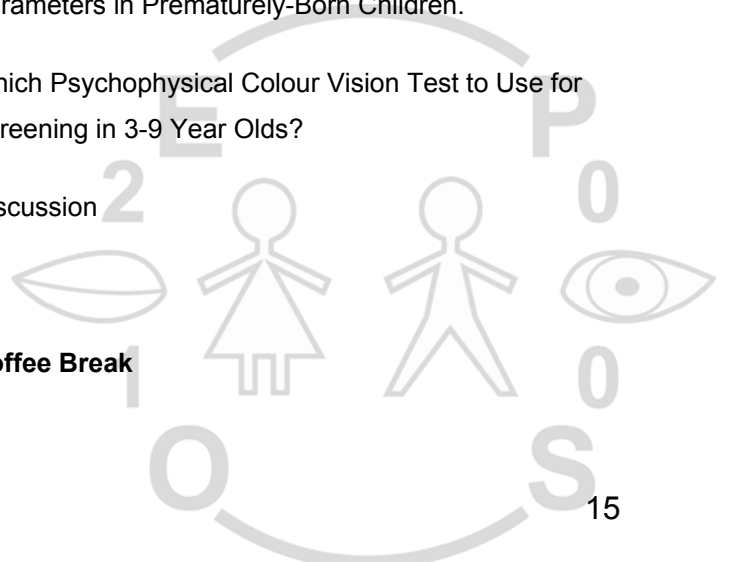
**13.45 – 14.30 h**

**Saturday Poster Session**

**14.30 – 15.30 h VII. Scientific Session Invitation to Dance – The Challenge of Interdisciplinary Research II Retinal Development**

**Chairperson: Nikos Kozeis - Helene Dollfus**

<b>L10</b>	14.30 h	Michael Hoffmann	Misrouting of the Optic Nerves – the Visual Cortex Keeps Track of Abnormal Tracks!
<b>L11</b>	14.50 h	Silke Haverkamp	Ectopic Ribbon Synapse Formation in the Outer Retina of Mutant Mice Lacking Functional Rods and Cones
<b>T20</b>	15.00 h	Hanna Akerblom	Retinal Nerve Fibre Layer and Optic Nerve Head Parameters in Prematurely-Born Children.
<b>T21</b>	15.10 h	Manca Tekavcic Pompe	Which Psychophysical Colour Vision Test to Use for Screening in 3-9 Year Olds?
	15.20 h		Discussion
<b>15.30 – 16.00 h</b>			<b>Coffee Break</b>



**16.00 – 17.00 h VIII. Scientific Session I Spy with My Little Eye -****Challenges in Imaging and Evaluation****Chairperson: Michael Hofmann -  
Birgit Lorenz**

<b>L12</b>	16.00 h	David Wallace	Pediatric Retinal Imaging: The Future is Here!
<b>T22</b>	16.20 h	Ramiro S. Maldonado	Evaluation of the Neonatal Macula Using Spectral Domain Optical Coherence Tomography (SD OCT)
<b>T23</b>	16.30 h	Jaume Català-Mora	OCT and Clinical Features of Chorioretinal Colobomas in Children
<b>T24</b>	16.40 h	Christoph Friedburg	Foveal Ultrastructure and Fundus Autofluorescence in Achromatopsia.
	16.50 h		Discussion

**Topic Related Posters in Saturday Poster Session**

<b>P32</b>		Christina Pieh-Beisse	Retinal Nerve Fiber Layer Thickness in Children Measured by Spectral Domain OCT
<b>P33</b>		Reshma Thampy	Fluorescein Angiography-Guided Management of Retinopathy in Incontinentia Pigmenti: A Case Series.
<b>P34</b>		Lígia Ribeiro	Macula and Nerve Fiber Layer Thickness in Amblyopia: an Optical Coherence Tomography Study
<b>P35</b>		Alexander Ehnes	Optical Coherence Tomography – Device Independent Automatic Segmentation of Intraretinal Layers
<b>P36</b>		Matthäus Pilch	Optical Coherence Tomography - Automatic Segmentation of Locally Limited Structures
<b>P37</b>		Steffen Zahn	Introducing an Application for the Analysis, Segmentation and Interpretation of OCT Investigations Applicable for Multiple Devices

**17.00 – 17.40 h IX. Scientific Session Challenges in Genetics II****Phenotypes****Chairperson: Hanno Bolz**

<b>T25</b>	17.00 h	Pascal Dureau	Ocular Manifestations of Incontinentia Pigmenti
<b>T26</b>	17.10 h	Markus Preising	<i>RD3</i> mutation in a consanguineous LCA family
<b>T27</b>	17.20 h	Francesco Testa	New Insight in Retinal Phenotype of Patient with <i>AIPL1</i> Mutations
	17.30 h		Discussion

**Topic Related Posters in Saturday Poster Session**

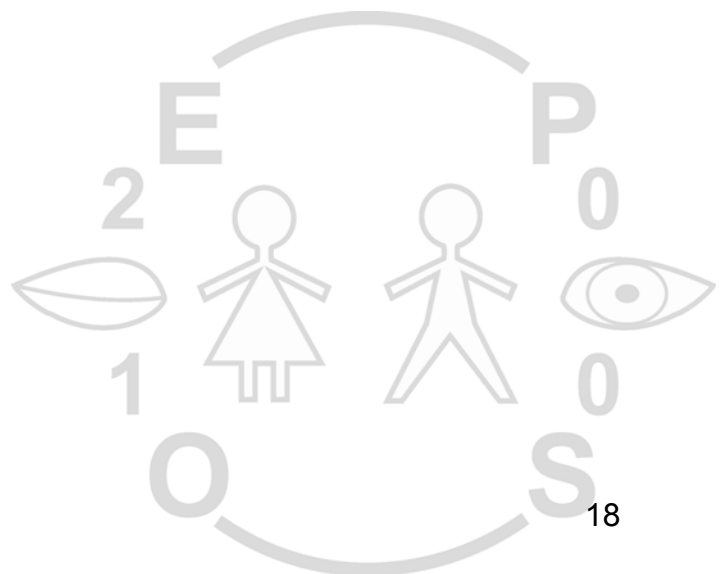
<b>P38</b>		Elzbieta Markowska	Best Disease - A Case Report
<b>P39</b>		Yaumara Perdomo-Trujillo	Severe Optic Neuropathy Mimicking Leber Hereditary Optic Atrophy in a Friedreich's Ataxia Patient.
<b>P40</b>		Francoise Roulez	Retinal Neovascularisation, Blepharokeratoconjunctivitis and Progressive Entropion in a Young Girl with Autosomal Dominant Severe Dyskeratosis Congenita (DC) Due to <i>TINF2</i> Gen
<b>P41</b>		Eduardo Marinho Saraiva	Nanophthalmos: A Family Case Report
<b>P42</b>		Nikolaos Kozeis	Increased C/D Ratio in Noonan Syndrome
<b>P43</b>		Francoise Meire	Non-syndromic bilateral and unilateral optic nerve aplasia associated with microdeletion of 10q23.33q23.33: first familial case and potential role of <i>CYP26A1</i> and <i>CYP26C1</i> genes in optic nerve development
<b>17.40 h</b>		<b>Birgit Lorenz</b>	<b>Scientific Awards</b>
<b>17.50 h</b>		<b>Birgit Lorenz</b>	<b>Closing Remarks</b>
<b>18.00 h</b>			<b>End of Meeting</b>



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Abstracts  
**Abstracts**

Lectures  
**Lectures**



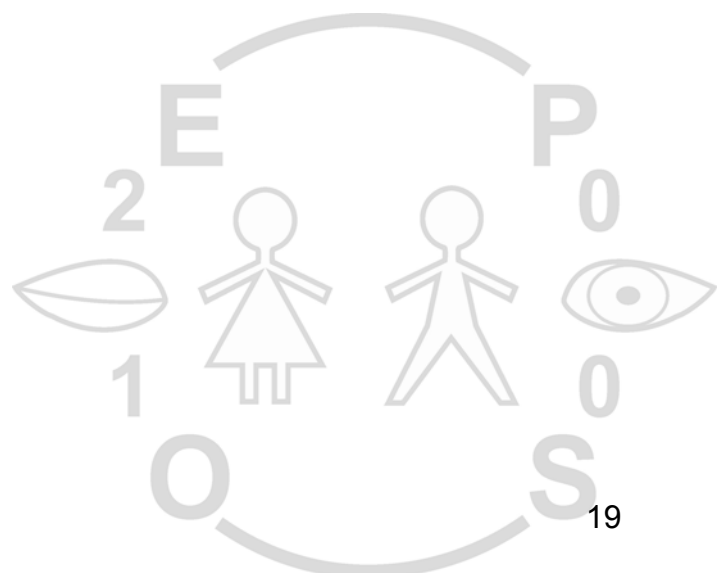
**L01: Challenges of Managing Paediatric Cataract in Sub Saharan Africa**

Richard Bowman

CCBRT Hospital, Dar es Salaam, Tanzania

*Presented by Richard Bowman*

A number of studies have suggested that cataract has become the leading cause of blindness in children in sub Saharan Africa, having overtaken corneal blindness from vitamin A deficiency and measles. This is probably due to successful mass vitamin A supplementation and measles vaccination programs rather than any change in cataract incidence. Little is known about the causes of cataract in children in Africa. Our studies of cataract surgery for children in east Africa have shown that some of the challenges involved in managing this condition include late presentation, poor accessibility, and poor follow up. Additional studies have tried to investigate these barriers further in order to try to devise strategies to address them. Despite these barriers our outcome study from CCBRT showed that two thirds of children who came back for follow up had post-operative visual acuities of 6/18 or better in their better eye which should allow mainstream education. The WHO has recommended that paediatric cataract surgery be confined to specialist childrens eye centres or Child Eye Health Tertiary Facilities (CEHTFs). We have conducted a situation analysis on CEHTFs for the Africa and have established a training program in CCBRT including a 6 month paediatric ophthalmology fellowship and shorter training for other members of the team such as nurses, refractionists and vision therapists.

**Conflict of Interest: None**



## L02: Challenges in the Screening and Treatment of Retinopathy of Prematurity in Romania and Other Central and Eastern European Countries in Europe

Constanta Nascutzky

*Institute for Mother and Child Care "A. Rusescu"*

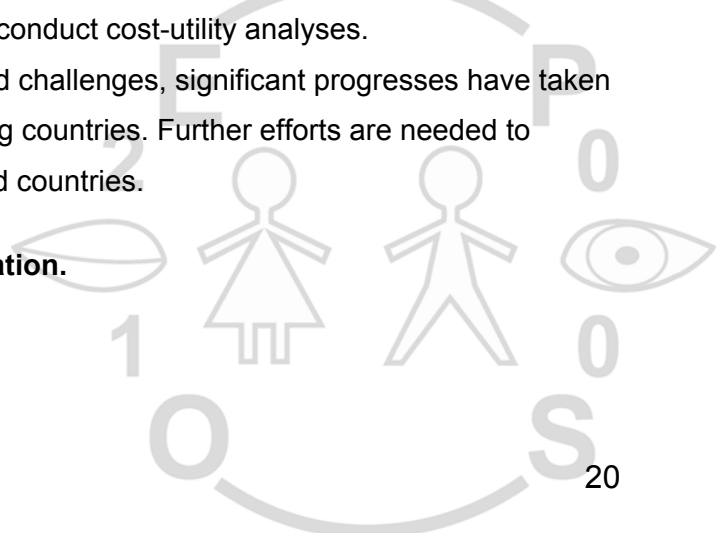
Presented by Constanța Nascutzky

Objective: to provide an overview of major challenges in the screening and treatment of preterm babies with retinopathy of prematurity (ROP) in Romania and other developing countries in Europe. ROP is currently the major avoidable cause of blindness in these countries, with a steady increase in incidence, including that of severe stages in relatively more mature and larger babies. This is described as “the third ROP epidemic”. Significant regional variations in ROP incidence, usually correlated with neonatal practices, are reported in various developing countries. Few have national population-based data on ROP incidence. Romania is no exception, incidence varies from one region to another (20% in the north, 55% in the south; severe ROP from 7% to 28%, 20% of the severe ROP cases would have been missed if we would have used birthweight less than 1500g as a screening guideline). Setting-up national ROP programmes, including registries, is still a challenge. Developing countries face the same, if not more severe shortage in experienced paediatric ophthalmologists than developed countries. Telemedicine is useful for screening and follow-up, as well as training of ophthalmologists, neonatologists, paediatricians, nurses etc. Gradual improvements are occurring in equipment available for screening and treatment. Logistical challenges are represented by problems in universal patient access to screening centres or that of ROP specialists to patients; these could be overcome by telemedicine. ROP screening and laser treatment are performed following international protocols, adapted to the needs and resources of the local population. Tertiary level child eye-care centres are still few in developing countries and vitreoretinal surgeons even more so. Limited financial resources are allocated by national governments and international agencies (WHO, UNICEF), in some developing countries health is not a major political priority. Cost estimates for ROP screening and treatment are difficult to obtain in developing countries due to lack of resources to conduct cost-utility analyses.

**Conclusions:** In spite of all of the above-mentioned challenges, significant progresses have taken place in ROP screening and treatment in developing countries. Further efforts are needed to achieve results comparable with those of developed countries.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





### **L03: Challenges in the Care of Preterm Babies with Retinopathy of Prematurity from Romania and Other Central and Eastern European Countries**

Tatiana Ciomartan

Institute for Mother and Child Care "A. Rusescu"

*Presented by Tatiana Ciomartan*

Objective: to provide an overview of major challenges in the care of preterm babies with retinopathy of prematurity (ROP) in Central and Eastern Europe. In the past 20 years considerable progress has been achieved in the quality of neonatal services, increasingly more immature and smaller babies survive in this region. Nevertheless, some countries have less resources to save extremely low birthweight infants. National ROP programmes exist in many of these countries, but often they do not manage to cover the entire target population. Registries that would allow proper evaluation of incidence of ROP are difficult to establish in some countries, and thus there is a risk of multiple reporting, as well as of missing some preemies. No unique patient record is used at national level. In spite of saving more and more immature babies, ROP has been diagnosed in significantly more mature and larger babies in Romania and other countries in the region, which prompted the adaptation of ROP screening guidelines used in the developed countries to the local situation in each Central and Eastern European country. We still see babies that go blind because they reach a referral centre when it is too late (in a large Romanian sample 30 children presented at their first examination with stage bilateral 5 ROP, numbers are now decreasing). Long-term follow-up of babies with ROP is hindered by logistical difficulties – children are referred from second level maternities, screening is often done by transporting the baby from the maternity to the reference centre; after discharge low-income families have a hard time coming for the follow-up visits. Severe shortage in trained personnel for ROP screening and treatment is a major issue impeding on the success of ROP programmes. Telemedicine, a possible solution to this problem, is available in very few centres. Expenses for ROP screening and treatment are covered exclusively from governmental sources – these are often insufficient and delayed. In many countries health is not a political priority.

**Conclusions:** Central and Eastern European countries have made considerable progress in the care of preterm babies with ROP. In this region more mature and larger babies have ROP, possibly because of variations in neonatal care (O<sub>2</sub> monitoring). Increased public and governmental awareness of ROP will have these countries reach the level of care achieved in Western Europe.

**Conflict of Interest: None**

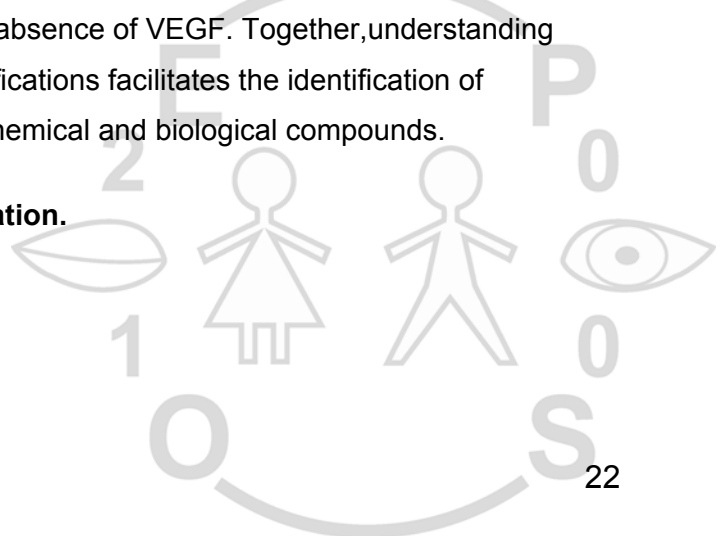
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**L04: Neovascularization in the Eye – Lessons from Translational Blood Vessel Research**

Hans-Peter Hammes

5<sup>th</sup> Medical Department, Universitätsmedizin Mannheim, University of Heidelberg*Presented by Hans-Peter Hammes*

Diabetic retinopathy is the leading cause of blindness in western countries, a fact that has not been substantially altered by the implementation of intensified insulin therapy or polypharmacy. Even if the translation of knowledge gathered through large network program sponsored by the EU and the BMBF would lead to a significant prevention of diabetes, the burden of vascular complications would not drop during the next 10-20 years. Diabetic retinopathy is not a proliferative disease, as often thought, contrasting with the general vasoregressive pattern of other target tissues of diabetic complications, it is primarily also a vasoregressive disease, however with a neovascular response to injury. Animal models cannot fully represent the complexity of hyperglycemia-induced cell and matrix alterations so that the clinically meaningful transition from vasoregressive to vasoproliferative predominance cannot be modelled in animals. Still, some important aspects of angiogenesis can be investigated in its complexity in vivo, and the mouse retina has evolved as a suitable model of angiogenesis. Applying state of the art histological techniques and novel genetic and pharmaceutical tool in genetically modified mice, important aspects contributing to the relevant stages of angiogenesis (permeability, migration, proliferation, lumen formation, stabilization) have found some molecular targets which may prove useful in translation to medical therapy. In this line, concepts such as the tip cell – stalk cell – phalanx cell have found their limitations, and pericytes, previously conceived as cellular mediators of angiogenesis inhibition, have conceptually converted into tour guides of angiogenesis. It has also become clear that the plethora of growth factors have specific roles in the angiogenic retina, and their relative contributions to physiological and pathological angiogenesis is now becoming clear. For example, the interaction of vascular endothelial growth factor and the angiotensin family has been considered important in angiogenesis. However, recent evidence suggests that proliferative retinopathy occurs in the absence of Ang-2, and angiogenesis occurs in the absence of VEGF. Together, understanding general concepts and possible distinct tissue specifications facilitates the identification of therapeutic targets, given the myriad of available chemical and biological compounds.

**Conflict of Interest: None****Author does not allow recording of the presentation.**



## **L05: Challenges in the Treatment of Pathological Neovascularization: New Players and Mechanisms**

Klaus T. Preissner

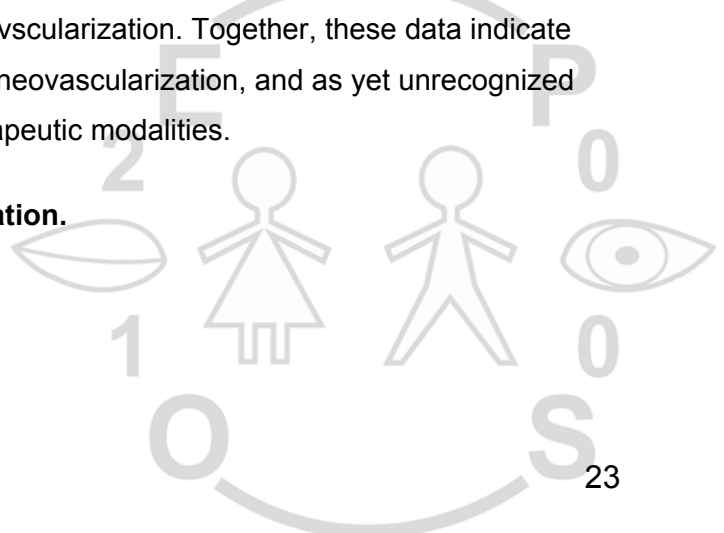
Depart. Biochemistry, Medical School, Justus-Liebig-Universität, Giessen

### **Presented by Klaus T. Preissner**

The process of neovascularization greatly depends on the induction of the angiogenic phenotype of endothelial cells that is strictly controlled by humoral factors as well as by cellular communications in the vascular system. Based on the kinetics of gene expression of cytokines such as "vascular endothelial growth factor" (VEGF-A) or adhesion receptors (such as integrin  $\alpha v \beta 3$ ) in the mouse model of "retinopathy of prematurity" (ROP) respective therapeutic approaches with anti-VEGF modalities or cyclic RGD-peptides were successful. Moreover, platelets were found to be a major contributor to retinal neovascularization, since in the ROP-model a 35-45% reduction in angiogenesis was found in the situation of thrombocytopenia as well as by inhibiting platelet aggregation. Besides platelet-derived VEGF, lipid mediators such as sphingosine-1-phosphate were identified as predominant activators of sprouting angiogenesis. Moreover, tissue remodeling related to neovascularization as well as cell injury may lead to exposure of intracellular material and is associated with increased permeability of blood vessels in the vicinity of the respective process or damage. Previous work from our laboratory demonstrated that natural cellular RNA as well as artificial RNA (poly: IC) significantly increased the permeability across brain microvascular endothelial cells in vitro and in vivo. Extracellular RNA-/poly: IC-induced hyperpermeability of tight monolayers of endothelial cells was mediated through the VEGF-A/VEGF-receptor 2/neuropilin system and was accompanied by topographic alterations of tight junction proteins and VE-cadherin. Both, antisense oligonucleotides against VEGF-receptor 2 as well as treatment with RNase1 prevented the permeability-inducing activity of extracellular RNA completely. Finally, in a mouse embryoid body model of stem cell differentiation, extracellular RNA was found to induce vasculogenesis and sprouting of newly formed endothelial cells, whereas administration of RNase1 in the ROP-model resulted in 50% reduction of neovascularization. Together, these data indicate that several "new players" appear to dictate retinal neovascularization, and as yet unrecognized antagonistic regimens may be useful as novel therapeutic modalities.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





## L06: Homozygosity Mapping in Nonconsanguineous Families Facilitates the Identification of Novel Retinal Dystrophy Genes

Frans Cremers<sup>1</sup>, Rob Collin<sup>1</sup>, Tamar Ben-Yosef<sup>2</sup>, Dikla Bandah-Rozenfeld<sup>3</sup>, Dror Sharon<sup>3</sup>, Anneke Den Hollander<sup>4</sup>

<sup>1</sup>Department of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands, <sup>2</sup>Genetics Department, Technion, Haifa, Israel, <sup>3</sup>Department of Ophthalmology, Hadassah-Hebrew University Medical Center, Jerusalem, Israel, <sup>4</sup>Department of Ophthalmology, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands

*Presented by Frans Cremers*

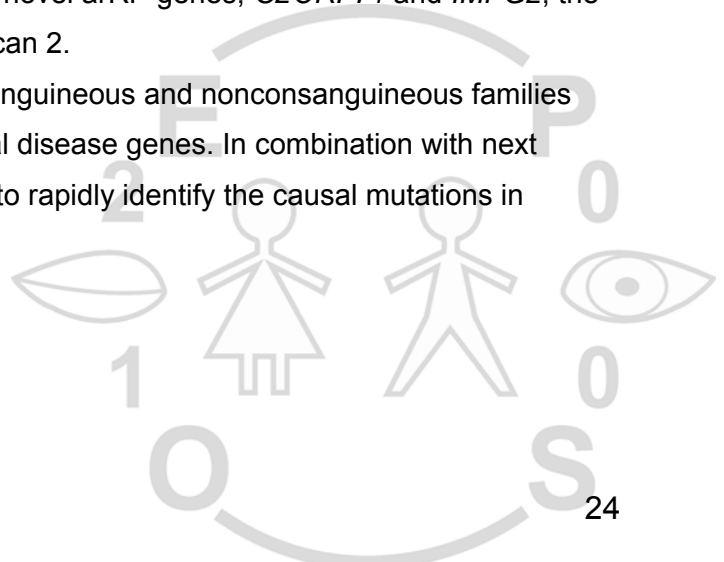
**Introduction:** Inherited retinal diseases display a high degree of genetic heterogeneity, which renders the identification of the underlying disease genes very challenging. We develop novel genomics strategies to identify these genes.

**Methods:** The generally low prevalence of retinal disease-associated mutations can be used to our advantage as these mutations often occur homozygously in patients. By using genome-wide single nucleotide polymorphism (SNP) arrays, we can identify the regions encompassing these mutations, both in consanguineous and nonconsanguineous families.

**Results:** In the Dutch population, ~1/3 of patients with inherited retinal diseases carry homozygous mutations. This suggests that the Dutch population contains subpopulations that until two generations ago, were relatively isolated. We performed genome-wide homozygosity mapping using high-density SNP arrays in ~400 patients with autosomal recessive retinitis pigmentosa (RP), Leber congenital amaurosis (LCA), cone- or cone-rod dystrophy (CD, CRD), or achromatopsia (ACHM). Patients were ascertained predominantly in the Netherlands, Canada, Israel, and Germany. These studies revealed many patients from nonconsanguineous families with conspicuous homozygous regions. By employing bioinformatic tools, we subsequently identified the LCA gene *LCA5*, the elusive arRP gene *EYS* located at the RP25 locus, and the CD/ACHM gene *PDE6C*. Very recently, by combining homozygosity data of patients from consanguineous and nonconsanguineous families, we identified two novel arRP genes, *C2ORF71* and *IMPG2*, the latter encoding interphotoreceptor matrix proteoglycan 2.

**Conclusion:** Homozygosity mapping in both consanguineous and nonconsanguineous families has proven to be effective in identifying novel retinal disease genes. In combination with next generation sequencing, this approach will allow us to rapidly identify the causal mutations in families with retinal dystrophies.

**Conflict of Interest:** None



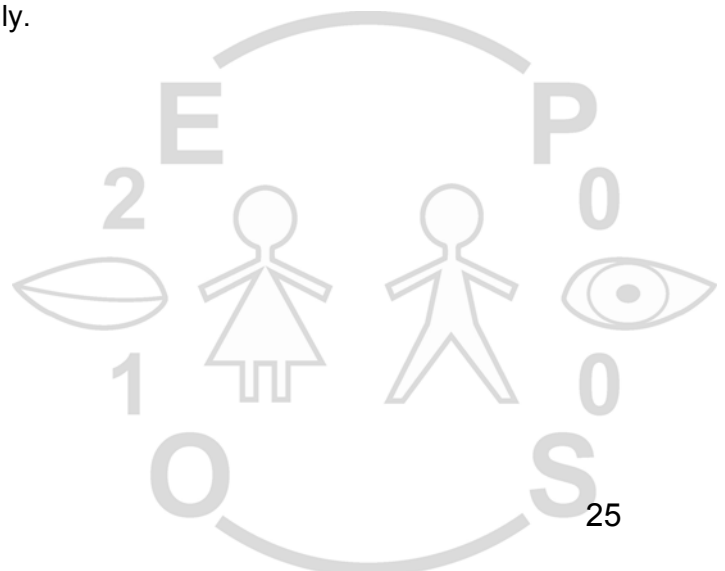
**L07: Genetic Modifiers of Disease Phenotype**

Andreas Gal

Institut für Humangenetik, UKHE, Hamburg

*Presented by Andreas Gal*

Heterogeneity is a common feature of human genetic diseases. Genetic heterogeneity consists of allelic heterogeneity in which different mutations in one gene result in the same trait, and locus heterogeneity in which mutations in two or more genes produce the same phenotype. Clinical heterogeneity refers to largely variable phenotypes that can be observed in patients with the same disease. This phenotypic variability, both among patients from the same family and in unrelated patients affected by the same disease, seems to be, at least in part, due to genetic modifiers. Modifier genes may qualitatively or quantitatively alter the phenotype produced by another gene, due to effects on penetrance, dominance, age of onset, progression, expressivity, or pleiotropy. Usually there is a single allele that is not highly evolutionarily conserved and has a modest effect. Modifier genes may alter the phenotype in various ways, and their effect can be observed if they are present or absent. In the case of penetrance, the modifier may change the frequency of affecteds among mutant homozygotes or heterozygotes by moving the threshold for expressing the trait, i.e. a greater or smaller proportion will be affected. Variable expressivity, the extent/severity of the trait in affecteds, can be explained by a modifier that shifts the distribution for mutant homozygotes (or heterozygotes) relative to the phenotypic threshold. Dominance of an allele can be altered by a modifier through moving the threshold for expressing the trait, i.e. heterozygotes are (or are no longer) affected. In my presentation, I will review examples from the literature on genetic modifiers affecting the various genetic features and the molecular mechanisms of their action in various retinal diseases. Modifiers may act in both directions and as such may represent susceptibility alleles that increase disease risk, or protective alleles that reduce it. The latter may be promising targets to study in order to better understand 'natural genetic resistance to disease' and develop strategies to exploit it pharmacologically.

**Conflict of Interest: None**



### L08: Restoration of Cortical Vision After Gene Therapy for Congenital Blindness

Bennett Jean<sup>1</sup>, Laura Cyckowski<sup>2</sup>, Justin Monroe<sup>2</sup>, Albert Maguire<sup>1</sup>, Kenneth Shindler<sup>1</sup>, Manzar Ashtari<sup>2</sup>

<sup>1</sup>University of Pennsylvania, <sup>2</sup>The Children's Hospital of Philadelphia

*Presented by Jean Bennett*

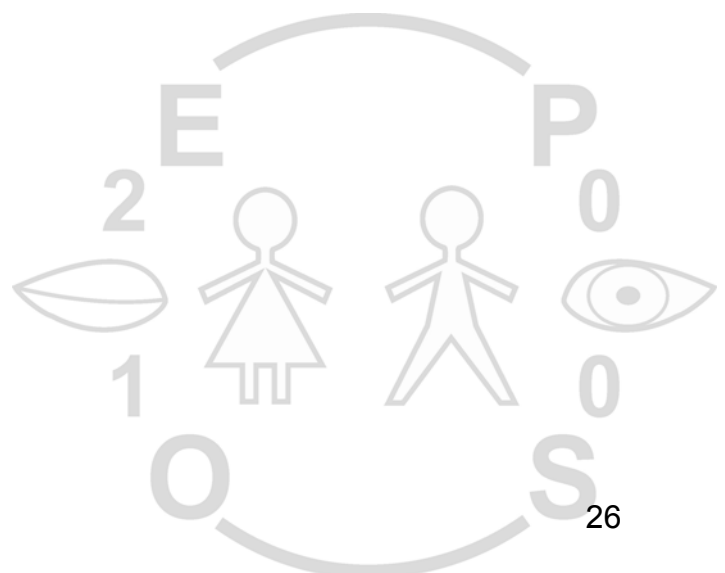
**Introduction:** Gene therapy improved vision in all 12 subjects who received unilateral subretinal injection of an adeno-associated virus (AAV) carrying the normal hRPE65 cDNA (AAV2-hRPE65v2) in a gene therapy clinical trial for Leber's congenital amaurosis (LCA). An unanswered question is how the visual cortex responds to recovery of retinal function after prolonged sensory deprivation.

**Methods:** Functional magnetic resonance imaging (fMRI) was carried out on 3 children (ages 8-10yo) and one adult (35yo) in the study, long after injection of AAV2-hRPE65v2. Full-field flickering stimuli were presented at 3 levels of contrast to each eye individually.

**Results:** fMRI results showed increased cortical activations after stimulus presentation to the treated eye only in all subjects. There was close correlation between the predicted field maps from the injection site and distribution of treated eye cortical activations.

**Conclusion:** This study shows that severe, long term (up to 35 years) visual impairment in LCA-RPE65 does not alter the integrity or responsiveness of neurons in the visual cortex. Gene therapy results in sustained, improved visual ability as reflected by function of the visual cortex. The challenges and issues surrounding extrapolation of the results of LCA-RPE65 gene therapy to other pediatric retinal diseases will be discussed. Additional collaborators: Bart LeRoy, Alberto Auricchio, Francesca Simonelli

**Conflict of Interest:** Yes, Co-author on a pending patent, "Method of treating or retarding the development of blindness"; However, waived any potential financial interest in 2002.



**L09: Molecular Therapy in Retinal Degenerations – What's in the Pipeline?**

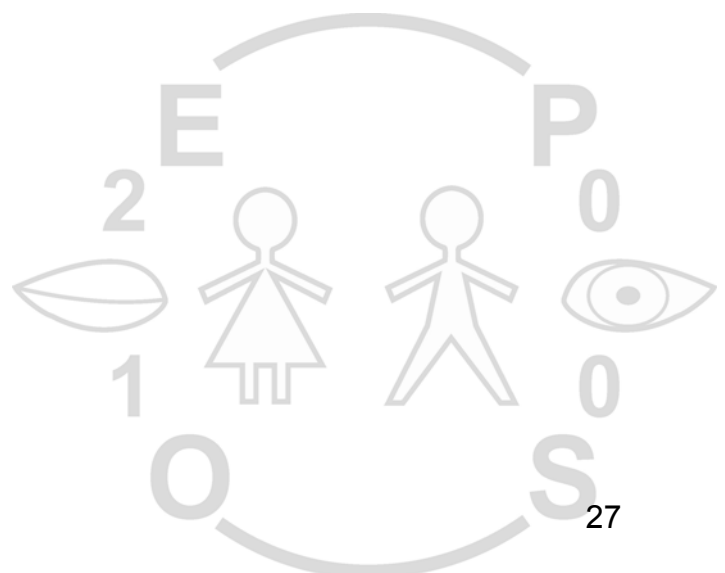
Mathias W. Seeliger

Division of Ocular Neurodegeneration, Institute for Ophthalmic Research, Department of Ophthalmology, Eberhard-Karls-University of Tuebingen, Germany

*Presented by Mathias Seeliger*

Several lines of promising therapeutic strategies for retinal degenerations have been developed in the immediate past years. So far, none has progressed as far as gene therapy. Nevertheless, substantial advances have been made in other areas, and some examples will also be highlighted in this presentation. Gene therapy, the main representative of curative approaches, has led to the successful restoration of function using recombinant adeno-associated viral vectors (rAAV) but also lentiviral vectors (LV), and has in the case of *RPE65* culminated in human clinical trials. Based on proof-of-principle studies in animal models, some other candidates for human gene transfer like X-linked retinoschisis and achromatopsia now come to the fore. Some symptomatic approaches aim at more central structures of the visual system that degenerate late in the course of a disease, like the use of halorhodopsin to make bipolar and ganglion cells intrinsically light sensitive. Other attempts try to retard the disease process by slow release of neuroprotective factors. The rescue of vision requires however an adequate functional and morphological assessment at different sites along the visual pathway. For retinal degenerations, optical coherence tomography (OCT) and electroretinography (ERG) appear best suited for the clinical and experimental follow-up. However, as the application of treatment commonly leads to local differences in functionality, novel concepts and/or techniques to take care of the topographic differences may be required.

**Conflict of Interest: None**



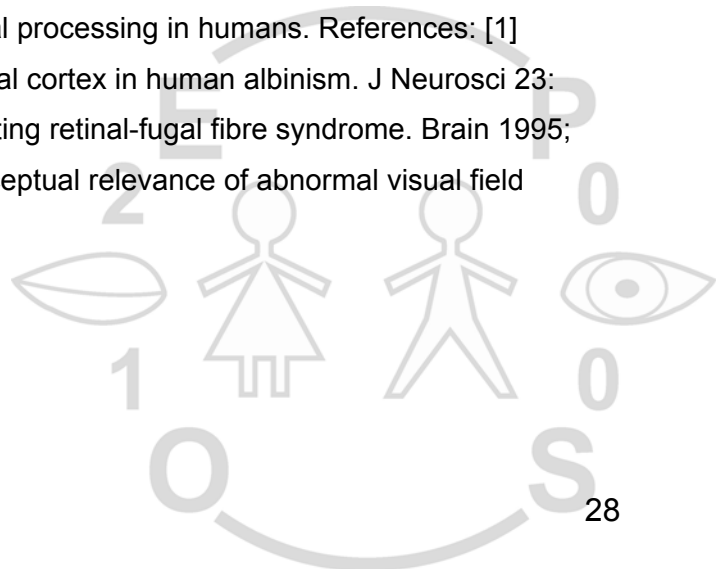
**L10: Misrouting of the Optic Nerves – the Visual Cortex Keeps Track of Abnormal Tracks!**

Michael Hoffmann

Visual Processing Lab, Ophthalmic Department, Otto-von-Guericke-University Magdeburg, Germany

*Presented by Michael Hoffmann*

The human visual cortex of each hemisphere receives binocular input from the respective contralateral visual hemifield. The partial decussation of the optic nerves at the optic chiasm is an indispensable prerequisite for this cortical visual field representation. In patients with albinism and achiasmia this projection pattern is disturbed such that the visual cortex receives an additional input of the ipsilateral hemifield [1,2]. The actual projection error of the optic nerves is entirely different in the two patient groups. In albinism it is a part of the temporal, in achiasmia it is the nasal retina that is abnormally represented. Investigating the integration of these abnormal visual field representations for visual processing using functional magnetic resonance imaging offers unique insights into fundamental principles and mechanisms in self- and reorganisation of the human visual cortex. Here, the retinotopic organisation of the visual cortex is compared between patients with albinism or achiasmia and the following features are common for both conditions of misrouted optic nerves: (1) Large scale congenitally abnormal input is represented as a retinotopic map. (2) The abnormal map is superimposed onto the normal retinotopic map, such that mirror-symmetrical visual field positions along the central vertical meridian are represented in close vicinity. (3) Visual field tests demonstrate the relevance of the abnormal visual field representation for visual perception. This indicates the plasticity not only of early, but also of higher tier visual areas [3]. These findings demonstrate that in humans with albinism and with achiasmia the abnormal input to the visual cortex is accommodated in a similar way. Thus, the mechanisms governing the accommodation of the abnormal input to the visual cortex appear to be independent of the nature and the origin of the abnormal input. In conclusion, the observed specific mapping of large-scale congenitally abnormal input to the visual cortex appears to reflect a general principle to make the abnormal visual input available for cortical processing in humans. References: [1] Hoffmann MB et al. (2003) Organisation of the visual cortex in human albinism. *J Neurosci* 23: 8921-8930 [2] Apkarian et al. (1995). Non-decussating retinal-fugal fibre syndrome. *Brain* 1995; 118: 1195–216. [3] Hoffmann MB et al. (2007) Perceptual relevance of abnormal visual field representations. *Brit J Ophthalmol* 91: 509-513

**Conflict of Interest: None**



## L11: Ectopic Ribbon Synapse Formation in the Outer Retina of Mutant Mice Lacking Functional Rods and Cones

Silke Haverkamp<sup>1</sup>, Isabella Spiwoks-Becker<sup>2</sup>, Karin Schäferhoff<sup>3</sup>, Michael Bonin<sup>3</sup>, Martin Biel<sup>4</sup>, Stylianos Michalakis<sup>4</sup>

<sup>1</sup>Max-Planck-Institute for Brain Research, Frankfurt/M, <sup>2</sup>Johannes Gutenberg University, Mainz, <sup>3</sup>Microarray Facility, Tuebingen, <sup>4</sup>LMU Muenchen

*Presented by Silke Haverkamp*

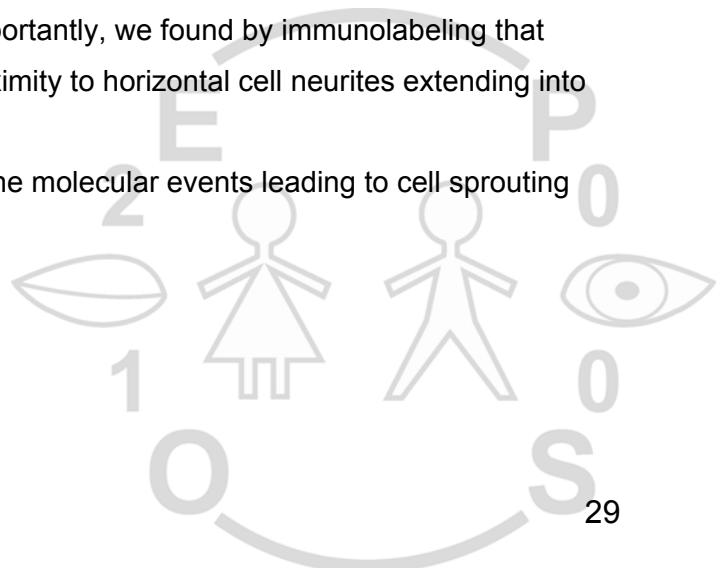
**Introduction:** At the first chemical synapse of the mammalian retina, rod photoreceptors transfer light signals to rod bipolar cells and horizontal cells. After loss of rod photoreceptor function, postsynaptic partners respond by extending neurites into the outer nuclear layer (ONL). In order to study this plasticity in detail and to evaluate how the cone pathway influences this response, we analyzed horizontal cell sprouting in mouse lines with targeted deletions of *CNGA3* and/or *CNGB1*. These genes encode essential subunits of the cone or rod cyclic nucleotide-gated channels, and are indispensable for normal function of the respective photoreceptor class.

**Methods:** The developmental time course of the outgrowth of horizontal cell processes and the formation of ectopic synapses was studied by immunocytochemistry and confocal laser scanning microscopy. Microarray-based gene expression analysis was performed using Affymetrix Mouse Genome 430 2.0 Arrays.

**Results:** In *CNGA3/B1* double knockouts, horizontal cell sprouting into the ONL was first observed around postnatal day 12. At first, the horizontal cell outgrowths seemed to have no apparent target. Two weeks later, the number of sprouting processes decreased, but the remaining sprouts developed synapse-like contacts in the ONL. The ultrastructural appearance of ectopic ribbon synapses at the base of rod somata within the ONL strongly suggests the formation of new synapses due to neuronal growth. To identify molecules involved in the induction of horizontal cell sprouting, we compared global retinal gene expression in *CNGB1* single and *CNGA3/B1* double knockouts at P12. We found differential expression of several genes implicated in formation, outgrowth and guidance of axons and neurites. Importantly, we found by immunolabeling that upregulated candidates localized to or in close proximity to horizontal cell neurites extending into the outer nuclear layer.

**Conclusion:** These results may help to elucidate the molecular events leading to cell sprouting and synaptic plasticity in the diseased retina.

**Conflict of Interest: None**



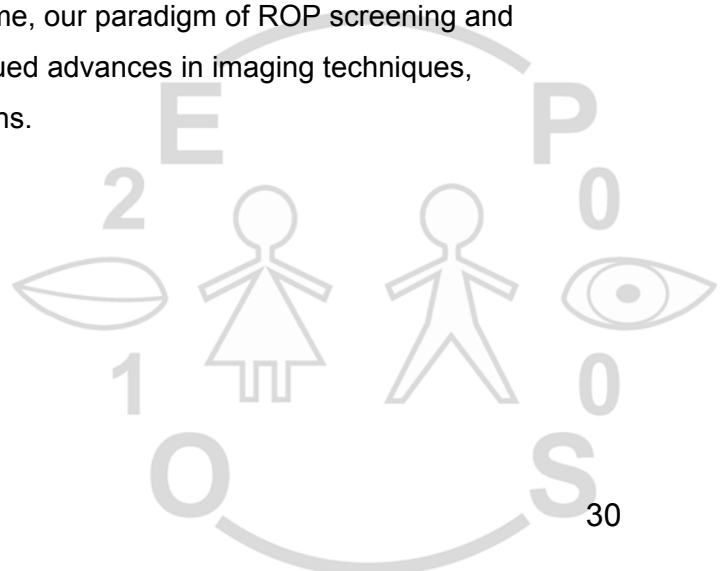
**L12: Pediatric Retinal Imaging: The Future is Here!**

David Wallace, Ramiro Maldonado, Sharon Freedman, Sina Farsiu, Cynthia Toth

Duke Eye Center

*Presented by David Wallace*

In the past decade, there have been a number of important technological advancements that have improved our ability to image and analyze the pediatric retina, including video indirect ophthalmoscopy, high-quality retinal photography, computer-assisted analysis of plus disease, and the application of spectral-domain ocular coherence tomography (SD-OCT) to infants and children. Improvements in video indirect ophthalmoscopy have allowed acquisition of very good quality still images during routine bedside screening examinations. Newly developed image processing methods have created panoramic montages using multiple video indirect images from a single eye. Images that have lower quality can be processed to improve their quality sufficiently to allow automated analysis of retinal features. The RetCam camera, which has for years allowed acquisition of still images of very high quality, now has improved portability with the development of the RetCam Shuttle. High-quality retinal images from multiple sources can be analyzed using computer software such as ROPtool to quantify the degree of vascular dilation and tortuosity. ROPtool now has a clinically meaningful overall measure of “plusness” that combines values for dilation and tortuosity. In addition, its application as a real-time second opinion for plus disease diagnosis is being piloted at the bedside. Finally, application of spectral-domain optical coherence tomography (SD-OCT) has revealed new insights into pediatric retinal development and pathology. Human foveal development has for the first time been observed in vivo using SD-OCT. In infants with retinopathy of prematurity (ROP), SD-OCT has revealed a myriad of retinal changes that are not evident by indirect ophthalmoscopy, including macular edema and cysts, epiretinal tissue and membranes, and retinoschisis. This imaging modality is also useful to visualize the extent of retinal detachment and presence of epiretinal membranes, providing retinal surgeons with key information that influences surgical intervention. In years to come, our paradigm of ROP screening and treatment will evolve considerably based on continued advances in imaging techniques, preventative strategies, and therapeutic interventions.

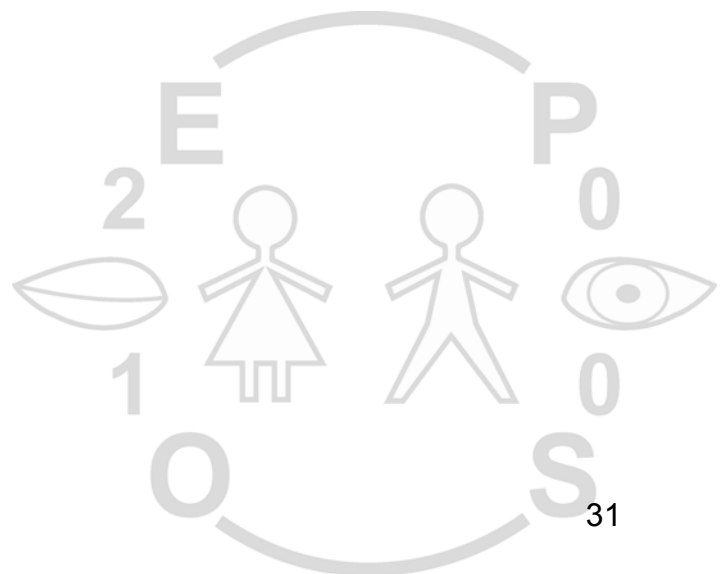
**Conflict of Interest: None**



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# Paper Presentations

## Paper Presentations





**T01: Funding of an Ophthalmic NGO in an Emerging Economy**

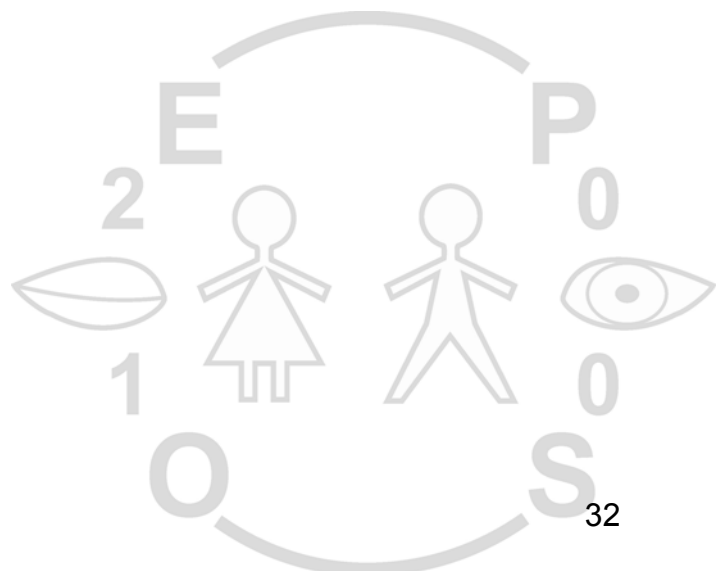
Raymond Brown

University Hospital of North Staffordshire

*Presented by Raymond Brown*

**Introduction:** Funding of an Ophthalmic NGO in an emerging economy; a personal experience. Paraguay, in South America is a country where the health care is, in some ways, half way between the devastating need seen in sub-saharan Africa and the relatively comprehensive healthcare offered in Europe and the USA. It has a stable political base in which to develop a paediatric ophthalmology training, while at the same time has a large population without easy access to health care. I have been fortunate to have worked with Fundación Visión in Asunción, for a few weeks in each of the past 8 years and have seen its development both in infrastructure, the training offered and in the scope of treatment. I have been impressed by the innovative ways that have been found to fund the programme and have put together my impression of the various methods by which the NGO funds itself from local sources. In addition to local income, an NGO in a developing country can access a number of different international sources of funding. There are ways of doing this which increase the NGO's chances of obtaining these funds. The third arm of funding work in a developing country is, what funds can an ophthalmologist in Europe provide and how? I discuss my experiences of this and how my own small NGO in the UK raises money to support Fundación Visión and the, sometimes difficult, decisions to be made as how to best spend this money.

**Conflict of Interest: None**



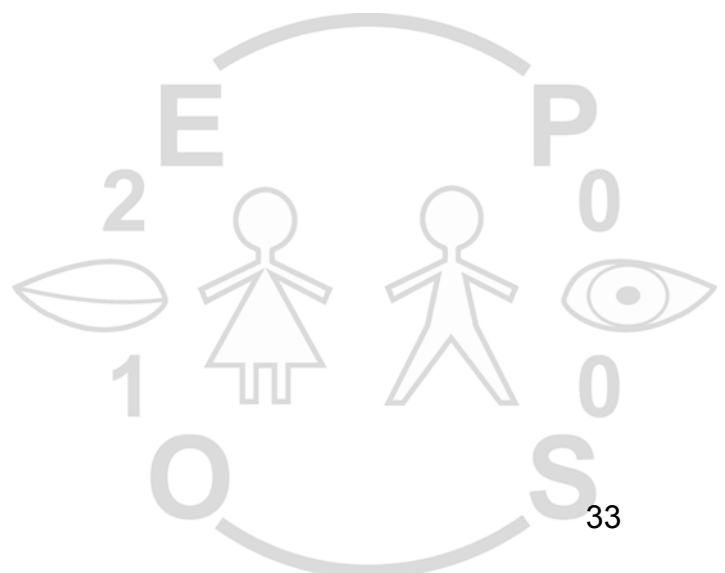
**T02: The Giessen Experience with Anti-VEGF Therapy for APROP**

Birgit Lorenz

Department of Ophthalmology, Universitaetsklinikum Giessen and Marburg GmbH, Giessen, Germany

*Presented by Birgit Lorenz*

**Background and Purpose** Aggressive posterior retinopathy of prematurity APROP carries the worst prognosis as to structural and functional outcome. Laser therapy has limited success. Intravitreal anti-VEGF therapy that is currently evaluated in several multicenter studies could be a better alternative. **Methods** Analysis of the actual literature on anti-VEGF therapy in acute ROP. Wide-field-digital imaging of anterior and posterior segment and serial fluorescein angiography (FLA) with a RetCAM II (Clarity, Pleasanton, USA) in infants with APROP. **Results** Three extreme prematures (GA 22 to 24+5 wks, BW 380 - 770g) with APROP received a single dose of bevacizumab intravitreally (Avastin®, two 0.312 mg/eye; one 0.625 mg/eye) 1.5 mm posterior to the limbus at PMA 32 - 34 wks. Regression of hyperaemic and persistent tunica vasculosa lentis anterior occurred within days, regression of APROP with further vascularisation of the peripheral retina within several weeks. FLA visualised continuing physiological vascularisation to the peripheral retina and absence of pathological exudation. **Conclusions** The positive Giessen experience corroborates the positive results from larger multicenter studies. Single doses of intravitreal bevacizumab alone are capable not only to induce regression of APROP but also results in further maturation of the normal retinal vascularisation. However, because of the lack of long-term results the authors recommend to limit the use of anti-VEGF to APROP as less aggressive forms of acute ROP have a success rate of > 95% with conventional laser therapy.

**Conflict of Interest: None**



**T03: Outcome of Adjunctive Intravitreal Bevacizumab Combined with Laser Indirect Ophthalmoscopy for Treatment of ROP**

Phanthipha Wongwai

Khon Kaen University

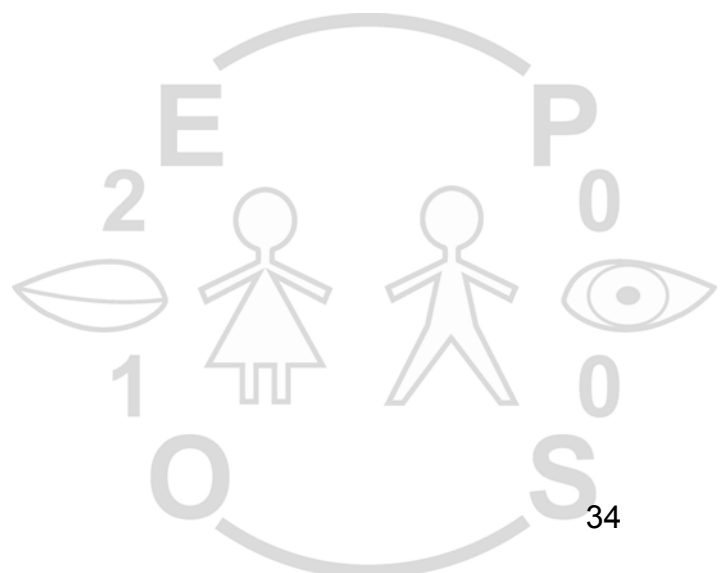
*Presented by Phanthipha Wongwai*

**Objective:** To evaluate outcome of combine intravitreal Bevacizumab with laser indirect ophthalmoscopy for treatment of ROP Design: Historical cohort study Method: Data was collected at 1 year followed-up after treatment of ROP with combine laser and intravitreal Bevacizumab.

**Results:** 40 newborns (80 eyes) diagnosed ROP were treated with combine intravitreal Bevacizumab and laser. Mean gestational age was 30.36 (25-34 week), mean birth-weight was 1254.64 (770-1900g), mean age of diagnosed ROP was 7.46 (2-16 wk), the majority of the patient was diagnosed stage 3 zone II ROP with plus disease. Regression was found in 73 eyes (91%), progression to Stage IV 5 eyes (6%) and 1 patient was died due to RDS. In regressed ROP 73 eyes, myopia within -2.00 D was found in majority of the patient, no strabismus and all of them were good fixed and followed. No unanticipated adverse event from the treatment.

**Conclusion:** Outcome of combine intravitreal Bevacizumab and laser in ROP was preferable, however long term followed up and randomized control trial are necessary to prove the effectiveness

**Conflict of Interest: None**



**T04: PDZD7 is a Modifier of Retinal Disease and a Contributor to Digenic Usher Syndrome**

Hanno Bolz<sup>1</sup>, Jennifer Phillips<sup>2</sup>, Max Liebau<sup>3</sup>, Inga Ebermann<sup>4</sup>, Monte Westerfield<sup>2</sup>, Thomas Benzing<sup>3</sup>

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*Presented by Hanno J. Bolz*

**Introduction:** Usher syndrome is a genetically heterogeneous recessive disease with hearing loss and retinitis pigmentosa (RP). It frequently presents with unexplained, often intrafamilial, variability of the visual phenotype.

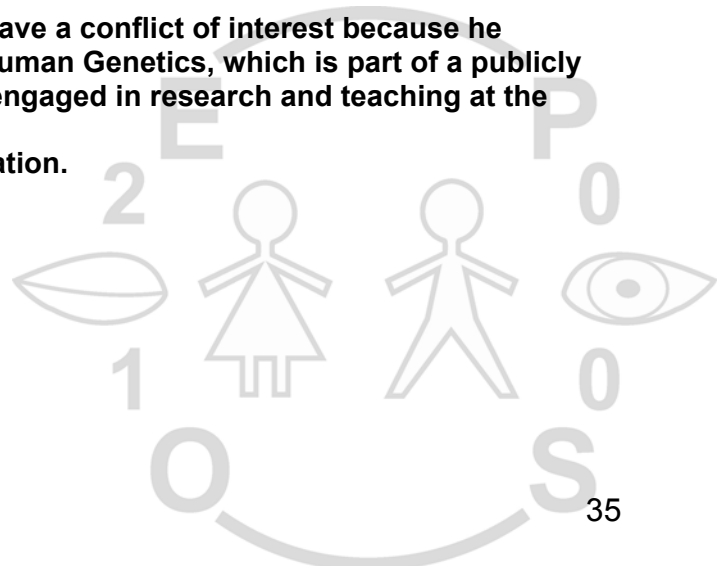
**Methods:** We cloned human and zebrafish *PDZD7* and characterized the proteins by immunohistochemistry and interaction studies. Human genotypes were validated using zebrafish.

**Results:** We demonstrate interaction between *PDZD7* and *GPR98* (*USH2C*) and *USH2A*, and their colocalization in the photoreceptor's connecting cilium region. On a homozygous *USH2A* mutation background, *PDZD7* aggravates RP. Heterozygous *PDZD7* mutations were present with truncating mutations in *USH2A*, *GPR98*, and an unidentified locus. Knockdown studies in zebrafish were consistent with digenic inheritance of *PDZD7* and *GPR98*, and with *PDZD7* as a retinal disease modifier in *USH2A* patients. *Pdzd7* knockdown produced an Usher-like phenotype in zebrafish, exacerbated retinal cell death in combination with *ush2a* or *gpr98*, and significantly reduced *Gpr98* localization in the region of the photoreceptor connecting cilium.

**Conclusion:** Our data challenge the view of Usher syndrome as a traditional Mendelian disorder. As in Bardet-Biedl syndrome, reclassification of Usher syndrome as an oligogenic disease permits a better understanding of its phenotypic variability. With the advance of new DNA technologies such as next-generation sequencing, similar constellations may be discovered for a number of other recessive diseases.

**Conflict of Interest:** Yes, H.J.B. may appear to have a conflict of interest because he currently works for the Bioscientia Center for Human Genetics, which is part of a publicly traded diagnostic company. He is still actively engaged in research and teaching at the University Hospital

**Author does not allow recording of the presentation.**





**T05: How Many Genes for Bardet-Biedl Syndrome?**

Dollfus H el ene<sup>1</sup>, Schaefer Elise<sup>1</sup>, Vincent Marie-Claire<sup>2</sup>, Muller Jean<sup>2</sup>, Mandel Jean-Louis<sup>3</sup>, Stoetzel Corinne<sup>1</sup>

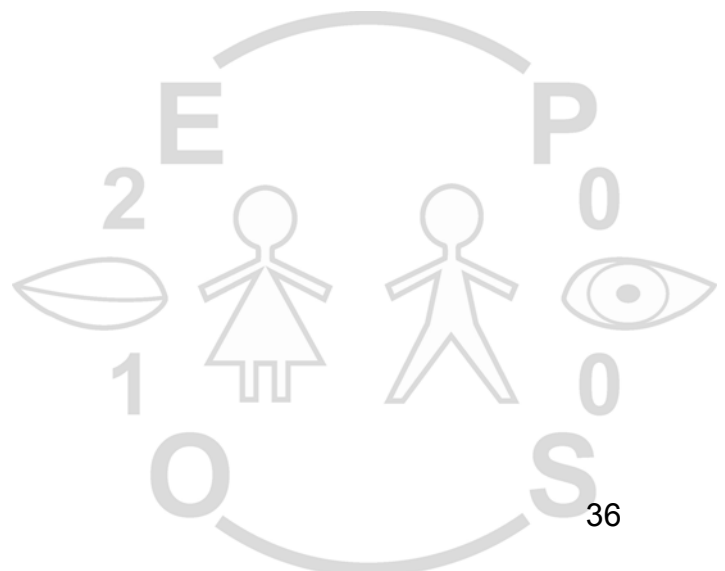
<sup>1</sup>Laboratoire Avenir Inserm EA 3949, Facult e de M edecine, H opitaux Universitaires de Strasbourg, Strasbourg, France., <sup>2</sup>Laboratoire de Diagnostic g en tique, H opitaux Universitaires de Strasbourg, Strasbourg France, <sup>3</sup>IGBMC, Illkirch-Graffenstaden, France

*Presented by H el ene Dollfus*

Bardet-Biedl syndrome (BBS) is a syndromic early-onset retinitis pigmentosa associated with obesity, polydactyly, cognitive impairment and kidney dysfunction related to primary cilia dysfunction. This autosomal recessive condition is characterized by genetic heterogeneity that challenges rapid molecular confirmation of the clinical diagnosis as well as prenatal diagnosis. To date, fifteen genes have been reported to be related to the BBS ciliopathy phenotype, the last of which is the very recent result of an international collaborative work that reveals a switch from classical homozygosity mapping to exome capture approaches. We will summarize the overall mutational load found in the French BBS study group series compared to other groups and question whether all the BBS genes have been identified or not?

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**



**T06: Evaluation of AAV Mediated Gene Therapy for RPE65 Patients by Highly Sensitive Psychophysical Techniques**

Knut Stieger, Markus Preising, Christoph Friedburg, Elisabeth Strohmayer, Steffen Zahn, Birgit Lorenz

Department of Ophthalmology, Justus Liebig University, Giessen, Germany

*Presented by Knut Stieger*

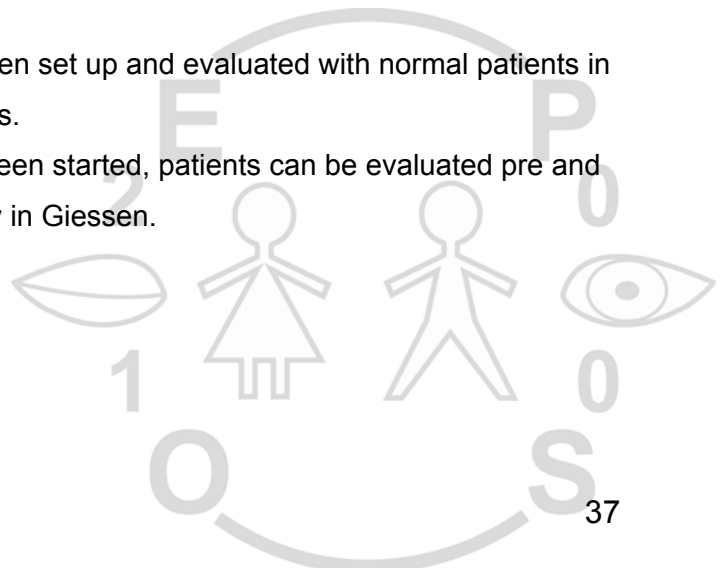
**Introduction:** The University Hospital of the University of Nantes is currently preparing a phase I clinical trial for AAV mediated gene therapy for patients with early onset severe retinal dystrophy (EOSRD) due to mutations in the *RPE65* gene ([www.clinical-trials.gov](http://www.clinical-trials.gov); patient screening: NCT00422721). Therapeutic efficacy of the treatment has been difficult to demonstrate in published clinical trials from the US and United Kingdom, as electroretinogram recordings and other objective measurements continued to be below threshold values. Highly sensitive psychophysical techniques represent an improvement to reliably quantify the lowest levels of visual perception, but are only of limited availability. In this study, the ten patients that are going to be enrolled in the initial phase I clinical trial in Nantes, will be evaluated by two-color threshold perimetry, full field stimulus testing, chromatic pupillometry and spectral domain OCT at the Department of Ophthalmology in Giessen before and after treatment by an AAV2/4.hRPE65.RPE65 gene therapy.

**Methods:** The Department of Ophthalmology in Giessen possesses profound experience using a two-color threshold perimeter (modified Humphrey field analyzer (HFA), which is not available at the Department of Ophthalmology in Nantes. Full field stimulus testing is going to be performed with a diagnosis espion E2 apparatus (Diagnosys LLC) modified for red, blue and white light stimuli. In addition, a newly developed binocular chromatic pupillometer (AMTech, and Diagnosys Colordome Ganzfeld stimulator) will be used to assess changes in the diameter of the pupil in response to light. A Heidelberg Spectralis OCT will be used to visualize the morphology of the retina.

**Results:** The four examination techniques have been set up and evaluated with normal patients in order to generate normal ranges for human subjects.

**Conclusion:** Once the clinical trial in Nantes has been started, patients can be evaluated pre and post operation at the Department of Ophthalmology in Giessen.

**Conflict of Interest: None**





**T07: Novel S/MAR Vectors Provide a Non-Viral Gene Therapy for Choroideraemia**

Mariya Moosajee, Elham Ostad-Saffari, Dhani Tracey-White, Miguel Seabra, Richard Harbottle

Molecular Medicine, Imperial College London, United Kingdom

*Presented by Mariya Moosajee*

**Introduction:** Non-viral gene therapy vectors are attractive alternatives to viral-based delivery systems due to their low toxicity and reduced immunogenicity. Recently, a novel plasmid vector was developed employing a human scaffold/matrix attachment region (S/MAR) to provide functional episomal persistence within cells. Choroideraemia is an X-linked recessive disorder caused by mutations in the *REP1* (Rab escort protein 1) gene. This progressive chorioretinal dystrophy has no effective treatment. We investigate whether non-viral S/MAR vectors can provide persistent *REP1* expression in the retina, and thus develop a potential gene therapy for choroideraemia.

**Methods:** Generation of S-MAR vectors containing either *REP1* cDNA, or reporter eGFP and luciferase genes, driven by the human elongation factor 1 short (EFS) or ubiquitin-C promoter. DNA was formulated with cationic polymer linear polyethylenimine (DNA: PEI) and 2 µl was injected subretinally into one eye of each MF-1 mouse (4-6 weeks old), the contralateral eye served as a control (n=50). The temporal expression of luciferase was followed utilising in vivo bioluminescent imaging. Histological and immunofluorescence analysis was used to evaluate integration of the vector within the retina.

**Results:** DNA: PEI complexes were successfully delivered to the eye via subretinal injection. Once-only vector administration demonstrated persistent episomal transgene expression for up to 6 months. Expression of the transgene was localised to the RPE, confirmed by immunofluorescence of retinal histology.

**Conclusion:** The novel S-MAR vector shows effective gene delivery to the RPE with long-term expression of *REP1*. This non-viral gene therapy may provide a potential treatment for choroideraemia, with application to other genetic retinal diseases.

**Conflict of Interest: None**



**T08: Our Experience with Paediatric Aphakic Glaucoma**Alicia Serra<sup>1</sup>, Marta Morales<sup>1</sup>, Marta Garcia<sup>1</sup><sup>1</sup>Hospital de Sant Joan de Deu, Barcelona, <sup>2</sup>Hospital de Sant Pau, Barcelona*Presented by Alicia Serra*

**Purpose:** To evaluate the clinical and therapeutic characteristics of Paediatric Aphakic Glaucoma (PAG)

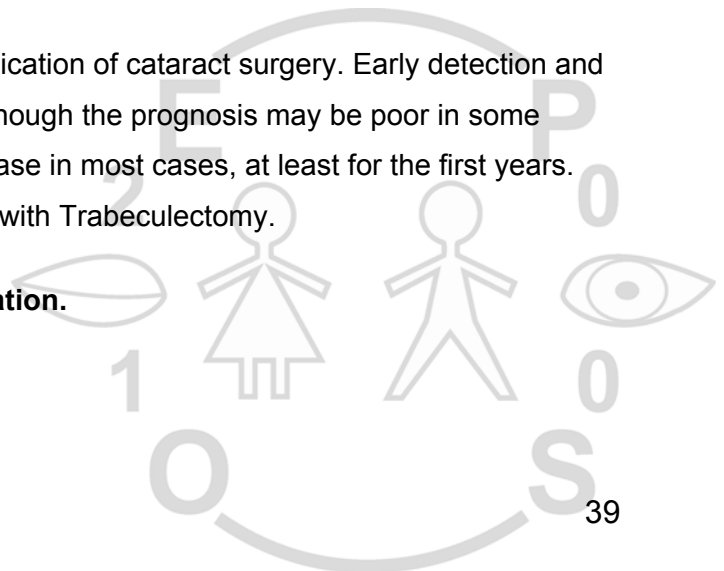
**Methods:** Retrospective study in 26 patients (37 eyes) diagnosed of PAG from 1988. We analyse the following characteristics: age at surgery of cataracts, type and etiology of cataracts, time from surgery and the beginning of glaucoma, response to medical and surgical treatment, and VA at the end of follow-up. In patients operated of cataracts after 1995 (13 patients, 19 eyes) we calculate the incidence of glaucoma for each etiology of cataract

**Results:** The etiology of cataracts was idiopathic in 13 patients, familial in 7, associated to Persistence of Fetal Vasculature in 3, to neurologic retardation in 2 and evolutive in 1 patient. 32/37 were bilateral cataracts, 12/37 associated to microftalmia. The age at surgery of cataracts ranged from 1 to 56 months, mean 7.5m, with 78% of the eyes operated before 6m of age and only 13% operated after 1 year. Time from surgery and the beginning of glaucoma ranged from 1 month to 12 years, 9/37 of the eyes before 1 year. All the eyes received medical treatment after diagnosis of PAG. In 16 eyes it has been enough to control the disease for a period ranged from 1 to 15 years (mean 6,8y). But 21/37 of the eyes needed surgical treatment 5m to 15y after diagnosis (mean 5y). 6 Trabeculectomies were performed in 5 eyes, in 4/6 failed during the first year. Valves were implanted in 14 eyes: 10 are working well without medical treatment from 1 to 6 years after implantacion, 3 needed medical treatment after 1 to 3 years of postop, and in one case a vitreous haemorrhage and ptisis bulbi occurred in the first weeks post op. Laser diode cyclophotocoagulation was performed in 7 eyes, in all of them the hypotensive effect was transitory (< 6m). Visual acuity at the end of follow-up ranged from no LP to 8/10, with 9 eyes being legally blind.

**Conclusions:** PAG is a frequent and severe complication of cataract surgery. Early detection and treatment is necessary to prevent loss of vision, although the prognosis may be poor in some cases. Medical treatment is able to control the disease in most cases, at least for the first years. Surgery of PAG has better results with valves than with Trabeculectomy.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**



**T09: Corneal Endothelial Cell Density after Artisan Aphakia IOL Implantation for Crystalline Lens Subluxation in Marfan Syndrome.**Marije Sminia<sup>1</sup>, Liesbeth Prick<sup>1</sup>, Monica Odenthal<sup>2</sup><sup>1</sup>Academic Medical Centre, Amsterdam, The Netherlands, <sup>2</sup>Diaconessenhuis, Leiden, The Netherlands*Presented by Marije Sminia*

**Introduction:** In the absence of capsular support the Artisan® (Ophtec, Groningen, The Netherlands) aphakia intraocular lens (IOL) can be used. For example in aphakia after surgery for the (sub)luxation of the crystalline lens (i.a. Marfan syndrome) or traumatic cataract. In these cases the Artisan aphakia IOL is preferred above implantation of angle supported or scleral sutured IOLs in some European countries and there are reports about safe implantation of these lenses in children. A point of concern with Artisan IOL implantation is corneal endothelial cell loss. Safety with regard to the corneal endothelium is of great importance in the paediatric age group, because of their long life expectancy

**Purpose:** To report on the long term clinical outcome of 2 young female patients with Marfan syndrome after Artisan aphakia IOL implantation for crystalline lens subluxation. And to compare the corneal endothelial cell density (ECD) of these two patients with the ECD of an age matched control group of patients with Marfan syndrome, without a history of intraocular surgery. Setting: Academic Medical Centre, Amsterdam, The Netherlands

**Methods:** A retrospective study was performed, evaluating the charts and endothelial photographs of the two study patients (4 eyes), and 8 control patients (16 eyes). BSCVA, corneal diameter, keratometry, axial eye length, anterior chamber depth, iris details, complications and intraocular surgical procedures were reported. The main outcome measure was the endothelial cell density at the last follow-up visit.

**Results:** The youngest of the two study patients (case 1) was 4.4/ 5.3 years old at the time of Artisan aphakia IOL implantation and 15.7 years at the last follow-up visit. The oldest of the two study patients (case 2) was 9.2/11.8 years at the time of the IOL implant and 25 years old at the last visit. The mean follow up of the 4 study eyes was 12.7 years (range 10.3 to 15.8 years). In all 4 study eyes a prophylactic or therapeutic cerclage was performed. The mean corneal endothelial cell density in the four study eyes was 3045 cells/ mm<sup>2</sup> at the last follow up visit. The mean ECD in the 16 control eyes was 2678 cells/mm<sup>2</sup>. The mean coefficient of variation of cell size was 36.6 in the study eyes and 27.1 in the control eyes. Details on the clinical outcome, ECD and CV of cell size will be discussed during the presentation.

**Conflict of Interest: None**

**T10: New Treatment Modality of Combined (Local and Systemic) Retinoblastoma Chemotherapy. (First Results).**

Nadiya Bobrova

The V.P. Filatov Institute of Eye Diseases and Tissue Therapy

*Presented by Nadiya Bobrova*

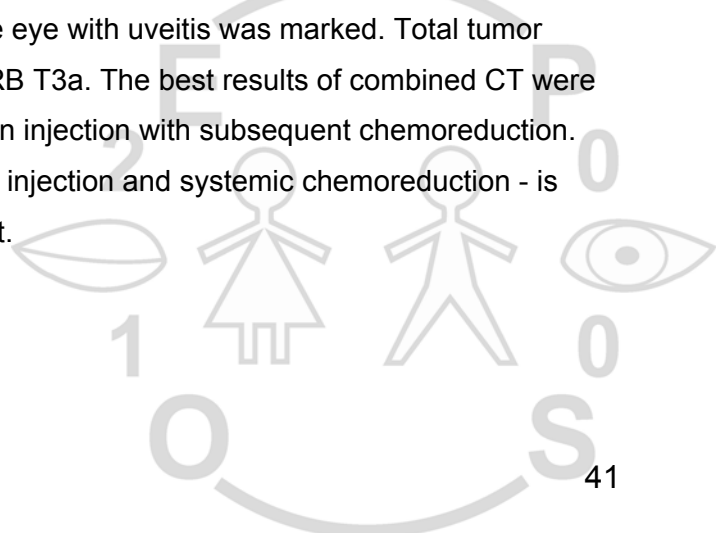
**Introduction:** To work out the new treatment modality of combined local and systemic retinoblastoma (RB) chemotherapy (CT).

**Methods:** Combined CT: systemic chemoreduction (Carboplatin, Etoposide, Vincristin) [Shields C et al, 1996] and local intravitreal Melphalan injection [Kaneko A, Suzuki S, 2003; 2004] was conducted to 26 children (30 eyes) with RB at age 5mo/o - 6 y/o (mean age 27±12mo). According TNM WHO classification T3 stage RB was on majority of eyes (19), T2 was on 7 eyes and T1 - only on 4. Tumor invasion to the vitreous and vitreal seeds were found on 17 eyes with a various T-stage. Unilateral RB was at 13 children, bilateral - at 13 (17 eyes). Additionally local tumour destruction (cryo-, lasertherapy, plaque St-90 radiotherapy) - 11 eyes; external beam radiotherapy – 4 eyes, combination of different destructive methods (4) were used. Follow up 2 - 10 months (mean 4.6±2.1mo) has seen at 15 children (16 eyes).

**Results:** No complications during intravitreal Melphalan injection and in postop were observed. Positive effect after combined local and systemic CH was marked on 20 from 30 eyes already in 2-4 weeks and consisted in reduction of size and prominence, fragmentation, calcination or scarring of the big tumors, resorbition of the small retinal focuses and vitreal seeds, provided repeated local Melphalan chemotherapy on 9 eyes. 14 children continued systemic chemoreduction, 2 patients with multifocal tumor growth have received EBRT, 1 has continued supervision. In follow up partial and total tumor regress was marked on 13 from 16 eyes. 3eyes with T3 RB were enucleated caused by the progressive tumor growth (1), hemophthalmos (1), uveitis with vitreous opacification and retinal detachment (1). Pathohistological examination in all cases verified RB with expressed tumor necrosis, no tumour cells spreading through the injections channels and in eye coats was found, total tumour necrosis and calcinations in one eye with uveitis was marked. Total tumor regress and calcination diagnosed on 3 eyes with RB T3a. The best results of combined CT were occurred in 5 cases of primary intravitreal Melphalan injection with subsequent chemoreduction.

**Conclusion:** Combined CT - intravitreal Melphalan injection and systemic chemoreduction - is perspective modality of RB salvage globe treatment.

**Conflict of Interest:** None



**T11: Ranibizumab (Lucentis®) in the Management of Late-Stage Coats' Disease**

Marie-Claire Gaillard, Aubin Balmer, Francis L. Munier

Hôpital Ophtalmique Jules Gonin, Lausanne, Switzerland

*Presented by Marie-Claire Gaillard*

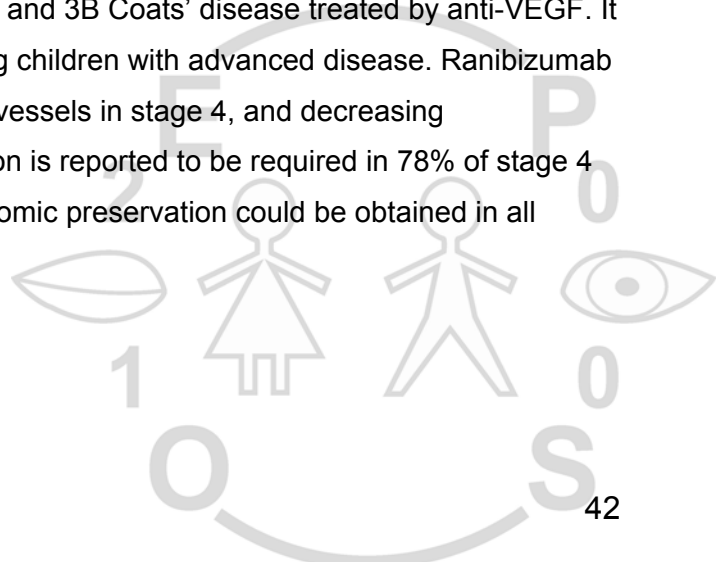
**Introduction:** Coats' disease is characterized by idiopathic telangiectasia with lipid exudates, and by retinal ischemia. Advanced cases develop total retinal detachment (stage 3B) and iris neovascularization with glaucoma (stage 4). Globe salvage depends on the destruction of the telangiectasia by laser photocoagulation and cryoablation of the ischemic retina, inapplicable at late-stage disease because of retinal detachment or poor dilatation due to rubeosis iridis.

**Methods:** Between 2006 and 2010, 8 consecutive cases of Coats' disease stage 3B (4 cases) and 4 (4 cases) received 1 or more intravitreal injections of 0.5 ml ranibizumab after parental and Swissmedic consent to off-label use. In 3 cases with bullous detachment filling the vitreous cavity, subretinal fluid drainage by sclerotomy was performed. The mean age at diagnosis was 17.9 months (range 1-43 months). After treatment, ERG was recorded and amblyopia treatment attempted in a subset of patients (n=4 and 6 respectively). The mean follow-up was 25.5 months.

**Results:** There were no ocular or systemic side effects. Rubeosis iridis disappeared during the first week and retinal reattachment occurred within 4 months post-injection in all patients, rendering the pathological vessels and ischemic retina accessible to conventional therapy. Ten months after complete reattachment of the retina, one patient developed severe fibrous vitreo-retinopathy evolving to tractional retinal detachment. Amblyopia treatment restored measurable visual acuity to 0.25 in 1 stage 4 patient. Scotopic ERG displayed flat recording in 3 patients, and reduced amplitudes in 1. Dysfunction of photopic ERG was severe in the 3 cases with flat scotopic ERG and mild in the last child.

**Conclusion:** Anti-VEGF therapy in management of Coats' disease has recently been reported in 9 distinct publications totalizing 13 patients including only 3 with advanced disease (2 stage 3B, 1stage 4). Our study is the largest series of stage 4 and 3B Coats' disease treated by anti-VEGF. It suggests that ranibizumab is well-tolerated in young children with advanced disease. Ranibizumab facilitates the management by suppressing iris neovessels in stage 4, and decreasing telangiectasia exudation. In the literature, enucleation is reported to be required in 78% of stage 4 and 7% of stage 3A and B cases. In our study anatomic preservation could be obtained in all patients, with a good visual outcome in 1 case.

**Conflict of Interest: None**



**T12: Phase 3, Prospective, 12-Week, Double-Masked, Multicentre Study of Latanoprost (LAT) and Timolol (TIM) in Paediatric Glaucomas: Age and Diagnosis Subgroup Analysis**

Tomoko Maeda-Chubachi<sup>1</sup>, Katherine S Chi-Burris<sup>1</sup>, Barbara Wirostko<sup>2</sup>, Dominique Brémond-Gignac<sup>3</sup>, Sharon F Freedman<sup>4</sup>, Peng T Khaw<sup>5</sup>

<sup>1</sup>Pfizer Inc, La Jolla, CA, USA, <sup>2</sup>University of Utah, Salt Lake City, UT, USA, <sup>3</sup>Ophthalmology, University Hospital of Amiens, Amiens, France, <sup>4</sup>Duke University, Durham, NC, USA, <sup>5</sup>NIHR BRC Moorfields Eye Hosp & UCL Inst Ophthalmology, London, UK

*Presented by Tomoko Maeda-Chubachi*

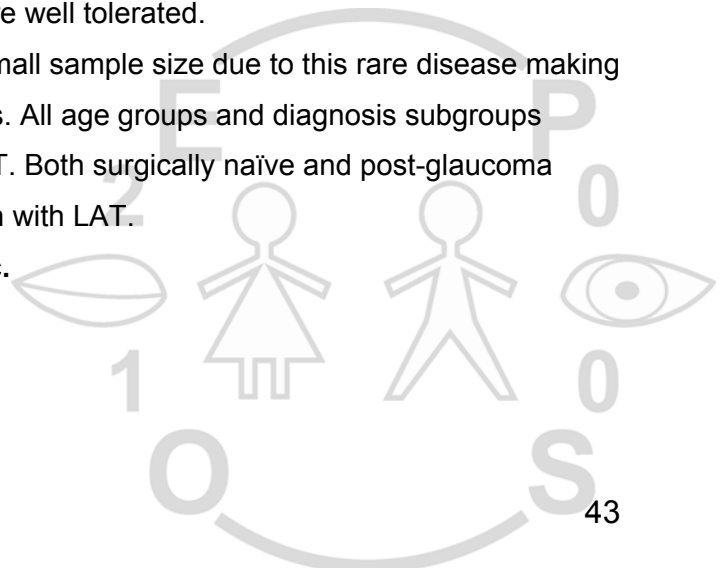
**Purpose:** To provide clinical guidance to ophthalmologists treating pediatric glaucoma from results of a clinical trial.

**Methods:** Subjects ≤18 years old with glaucoma in ≥1 eye were stratified by age (0 <-3; 3-<12; 12-18 yrs), diagnosis (primary congenital glaucoma [PCG] vs nonPCG), and IOP, and were randomised (1: 1) to LAT vehicle (8 AM) and LAT 0.005% (8 PM) or TIM 0.5% (or 0.25% for those <3 years old; 8 AM and 8 PM). At baseline, weeks 1, 4, and 12, IOP and ocular safety assessments were performed, and adverse events were recorded. Exploratory analyses were performed in age- and diagnosis-specific groups (ITT population).

**Results:** 137 subjects were treated (0-<3 yrs, n=34; 3-<12 yrs, n=55; 12-18 yrs, n=48). Mean IOP reductions (% reduction) at week 12 were LAT 7.0 mmHg (25%) vs TIM 6.0 mmHg (22%). Mean IOP reductions in PCG subgroup were LAT 6.0 (22%) vs TIM 5.1 mmHg (19%) and in non-PCG subgroup were LAT 7.9 (28%) vs TIM 6.7 mmHg (24%), respectively. Mean IOP reductions by age group were: LAT 5.8 (21%) vs TIM 2.6 mmHg (9%) for 0-<3 yrs; LAT 6.4 (23%) vs TIM 7.3 mmHg (26%) for 3-<12 yrs; LAT 8.5 (31%) vs TIM 6.8 mmHg (25%) for 12-18 yrs. For PCG subjects with a history of glaucoma surgery, mean IOP reductions were LAT 5.8 (22%) vs TIM 5.6 mmHg (22%) in those with prior glaucoma surgery (n=27), and LAT 6.1 (23%) vs TIM 4.6 mmHg (16%) in those without (n=35). For aphakic/pseudophakic subjects (n=17), mean IOP reductions were LAT 8.3 (27%) vs TIM 10.6 mmHg (31%); while for JOAG (n=40), mean IOP reductions were LAT 7.3 (28%) vs TIM 7.5 mmHg (28%). Both therapies were well tolerated.

**Conclusion:** The study is limited by its relatively small sample size due to this rare disease making it difficult to draw statistically significant conclusions. All age groups and diagnosis subgroups showed >20% IOP reduction when treated with LAT. Both surgically naïve and post-glaucoma surgical PCG subjects showed >20% IOP reduction with LAT.

**Conflict of Interest: Yes, Employed by Pfizer Inc.**



**T13: Preliminary Results of Superselective Ophthalmic Artery Chemotherapy (SOAC) in 14 Patients with Advanced Retinoblastoma**Francis L. Munier<sup>1</sup>, Maja Beck-Popovic<sup>2</sup>, Marie-Claire Gaillard<sup>1</sup>, Aubin Balmer<sup>1</sup>, Stefano Binaghi<sup>3</sup>

<sup>1</sup>Jules Gonin Eye Hospital, Lausanne, Switzerland, <sup>2</sup>Unit of Pediatric Hemato-Oncology, CHUV, Lausanne, Switzerland, <sup>3</sup>Radiology Department, Neuroradiology Unit, CHUV, Lausanne, Switzerland

*Presented by Francis L. Munier*

**Background:** Superselective ophthalmic artery chemotherapy (SOAC) has been proposed as an alternative to intravenous chemoreduction for advanced intraocular retinoblastoma. Results of a recent Phase I/II trial appear promising in terms of tumor control and eye conservation. However little attention was paid to ocular toxicity and visual prognosis. We report the preliminary results in our initial cohort of 14 patients.

**Patients and Method:** We retrospectively reviewed the charts of 14 consecutive retinoblastoma cases who received a total of 32 injections of Melphalan in the ophthalmic artery between November 2008 and July 2010. In these patients, SOAC was indicated only as an alternative to enucleation or external beam radiotherapy (EBR). Retcam and fluorescein angiography were performed at presentation and before each injection. Best corrected visual acuity was assessed at the latest visit following amblyopia therapy as necessary.

**Results:** Success rate of ophthalmic artery cannulation was 94.1% (32/34). The 2 failures to inject were due to transient internal carotid spasm and absence of ophthalmic artery originating from internal carotid artery respectively. There was no stroke or other systemic complications. Haematological toxicity did not exceed grade 1. No hair loss occurred. Enucleation and external beam radiotherapy could be avoided in all cases but one who, despite regression, required EBR for a persistent papillary tumor in his only eye. The mean follow-up of 7.5 months. Sectoral occlusive choroidal vasculopathy was observed in two eyes (14%), and retinal arteriolar emboli in one eye. Among the 10 eyes available for visual testing. At presentation the macula was affected in 4/10 eyes (vision range 0.012 to 0.2) and not affected in 6/10 eyes (vision range: 0.05-0.63).

**Discussion:** SOAC was effective and safe in all patients with no strokes or other systemic vascular complications, while systemic toxicity remained subclinical. Unlike intravenous chemoreduction, SOAC can be associated with sectoral chorioretinal atrophy secondary to subacute choroidal occlusive vasculopathy. Further studies are required to better appreciate the frequency and severity of SOAC-related vascular ocular complications and how they functionally correlate. In the mean time we strongly recommend that indications for SOAC should be performed in one eye only and restricted to advanced retinoblastoma, as first line or salvage alternative to enucleation or external beam radiotherapy.

**Conflict of Interest: None**



### T14: Orbital Tumors in Children

Anton Gerinec<sup>1</sup>, Kristína Husáková<sup>2</sup>

<sup>1</sup>Pediatric Ophthal.Dept., Children University Hospital Bratislava, <sup>2</sup>Pediatric Oncology Dept. Children University Hospital Bratislava

*Presented by Anton Gerinec*

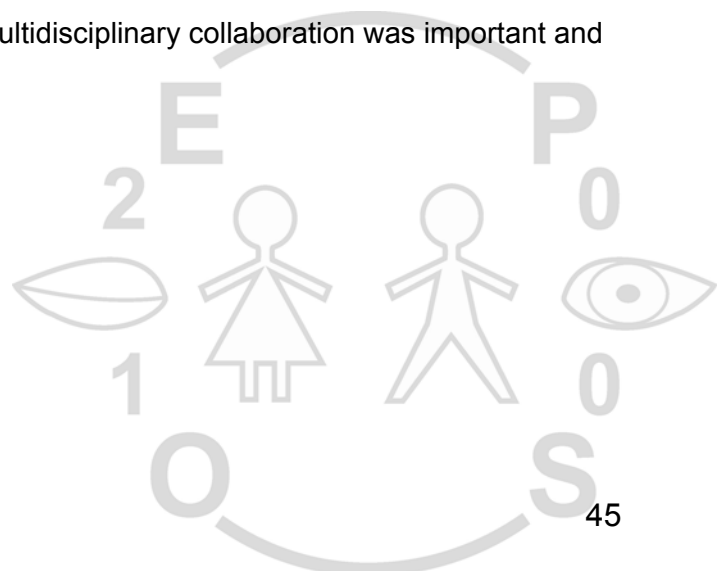
**Purpose:** To estimate the treatment efficacy in several main clinical forms of orbital tumors in children. The analysed tumors were capillary hemangiomas, optic nerve gliomas and rhabdomyosarcomas. Material and

**Methods:** The group of 63 patients treated on Pediatric Ophthalmology and Oncology Dept. in University Children Hospital in Bratislava from year 2000 has been analysed. In 25 capillary hemangiomas treated especially by steroids, surgery and alpha interferon was introduced in the year 2009 beta blocker propranolol. It was systemically administered in 12 small infants. Optic nerve glioma in NF-1 required multidisciplinary collaboration in 23 children. By its progression in to chiasma was performed in 10 older children radicaly optic nerve resection from chiasma. In 8 patients affected only by chiasmatic glioma was used chemotherapy and 5 small children with initial optic nerve glioma discovered by screening are followed without treatment. Orbital rhabdomyosarcoma was treated in 15 children by surgery, chemotherapy and radiotherapy.

**Results:** In hemangiomas were achieved very slowly size reduction by application of steroids in all tumors. Very rapid effect has been observed in infants by administering of propranolol. Resection of optic nerve from chiasma stopped progression of glioma and escaped visual function of the healthy eye. Chiasmatic gliomas by using chemotherapy are stable and initial optic nerve gliomas in small children during several years observation have negligible progress with good visual functions. All patients with rhabdomyosarcoma had embryonal type and during 10 years observation remain 87,3% survivors with preservation of the eyeball.

**Conclusion:** The good results in orbital tumors care in children were achieved by compliance of principle of the eye globe and motility preservation in orbital surgery and prophylaxy of amblyopia by rapid treatment of capillary hemangioma. The multidisciplinary collaboration was important and necessary.

**Conflict of Interest:** None





## T15: Screening for Retinopathy of Prematurity in Infants Born Before 27 Weeks of Gestation in Sweden

Dordi Austeng<sup>1</sup>, Ann Hellström<sup>2</sup>, Peter Jakobsson<sup>3</sup>, Kristina Tornqvist<sup>4</sup>, Agneta Wallin<sup>5</sup>, Gerd Holmström<sup>1</sup>

<sup>1</sup>Uppsala University, Sweden, <sup>2</sup>University of Gothenburg, Sweden, <sup>3</sup>Linköping University, Sweden, <sup>4</sup>Lund University, Sweden, <sup>5</sup>St. Erik's Eye Hospital, Sweden

*Presented by Dordi Austeng*

**Introduction:** The number of extremely preterm infants has increased the last decade giving us a larger population of infants at risk of developing severe retinopathy of prematurity (ROP). The purpose of this paper is discussing screening routines for ROP of infants born before 27 weeks of gestation.

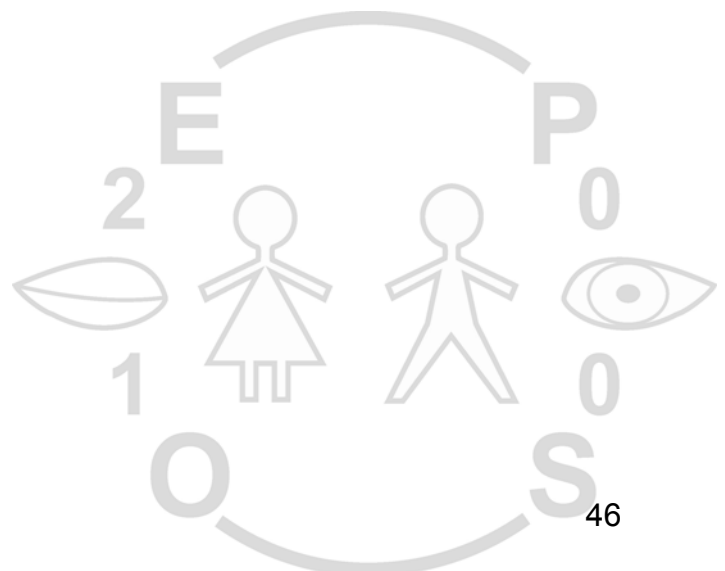
**Methods:** A national prospective study of neonatal morbidity in infants born before 27 weeks of gestation was performed in Sweden between 2004 and 2007. Screening for ROP was to start in the fifth postnatal week and to continue weekly until complete vascularization of the retina or until regression of ROP. The Early Treatment for ROP recommendations were followed.

**Results:** Onset of ROP 3 did not occur before postmenstrual age (PMA) 31 weeks and criteria for treatment were not reached before PMA 32 weeks. The age at onset of ROP and the risk of reaching treatment criteria were related to gestational age (GA) at birth. We also found that early onset of ROP was related to more severe ROP, even after adjustment for GA at birth.

**Conclusion:** The purpose of screening is timely detection of ROP 3. Based on our findings we propose modifications of guidelines for ROP screening, both regarding the start of screening and the screening intensity.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**



**T16: The Incidence of Visual Impairment Due to ROP and Their Concomitant Disabilities in the Netherlands. A Thirty Year Overview.**Nicoline Schalij-Delfos<sup>1</sup>, Arlette Van Sorge<sup>1</sup>, Jacqueline Termote<sup>2</sup><sup>1</sup>Dpt of Ophthalmology, LUMC, Leiden, the Netherlands, <sup>2</sup>Dpt of Neonatology, WKZ-UMCU, Utrecht, the Netherlands*Presented by Nicoline Schalij-Delfos*

**Introduction:** Retrospective study to determine the incidence of visual impairment (VI) caused by Retinopathy of Prematurity (ROP) and the incidence of associated disabilities in preterm neonates born between 2000 and 2009 in The Netherlands and compare data to three previous studies conducted from 1975 to 2000.

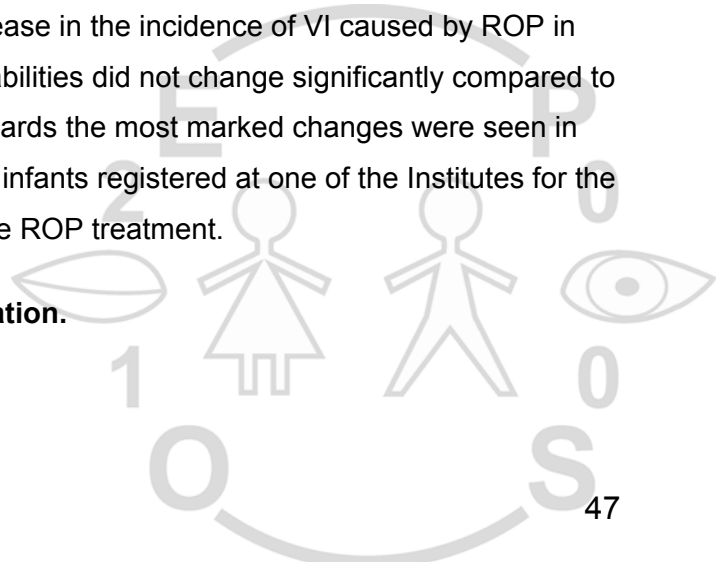
**Methods:** Neonatal, developmental and ophthalmological data of children born between January 1, 2000 and December 31, 2008 were retrieved from the Dutch institutes for the partially sighted and blind. To monitor trends data were compared to three previous Dutch studies conducted between 1975-1987, 1986-1994 and 1994-2000.

**Results:** Records of 43 infants born between 2000 and 2009 with VI due to ROP were found, 42 infants (97.7%) could be included in the study. In comparison with the previous surveys significantly less children were visually impaired due to ROP (1.84 per 100.000 live births per year in 2000-2009 vs 3.93 per 100.000 in 1994-2000  $p=0.000$ ). The incidence of infants that were completely blind declined from 27,5% in 1994-2000 to 7,1% in 2000-2009 ( $p<0,05$ ). Incidence of concomitant as well as multiple disabilities remained the same compared to the previous study. Comparing data of the four studies a gradual decrease of gestational age, birth weight and visual impairment over the years was found whereas an increase of duration of artificial ventilation and supplementary oxygen administration as well as bronchopulmonary dysplasia (BPD), developmental delay and behavioral abnormalities can be seen. The turning point being somewhere in the mid-eighties / nineties. The percentage of infants who received acute-phase ROP treatment increased from 56,9% in 1994-2000 to 66,7% in 2000-2009 (ns).

**Conclusion:** This study (2000-2009) shows a decrease in the incidence of VI caused by ROP in the Netherlands. The incidence of concomitant disabilities did not change significantly compared to the previous study. Comparing data from 1975 onwards the most marked changes were seen in the mid-eighties and the nineties. Still 33.3% of the infants registered at one of the Institutes for the partially sighted or blind did not receive acute-phase ROP treatment.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**



**T17: Preliminary Results of the NEDROP-Study: a National Inventory on Screening for Retinopathy of Prematurity.**

Arlette Van Sorge<sup>1</sup>, Jacqueline Termote<sup>2</sup>, Huib Simonsz<sup>3</sup>, Frank Kerkhoff<sup>4</sup>, Rene Van Rijn<sup>5</sup>,  
Nicoline Schalijs-Delfos<sup>1</sup>

<sup>1</sup>Leiden University Medical Centre, <sup>2</sup>Wilhelmina Children's Hospital / University Medical Centre Utrecht, <sup>3</sup>Children's Hospital Erasmus MC- Sophia, Rotterdam, <sup>4</sup>Maxima Medical Centre, Veldhoven, <sup>5</sup>VU University Medical Centre, Amsterdam

*Presented by Arlette van Sorge*

**Introduction:** Presentation of the preliminary year results of NEDROP-study, an inventory of the present situation of ROP screening in the Netherlands.

**Methods:** Pediatricians and neonatologists reported all infants that were born in 2009 and complied with the inclusion criteria of the old National Guideline for ROP-screening (GA < 32 wks and/or BW < 1500g). Ophthalmologists reported all children screened for ROP. A code was developed to enable anonymous data transmission and coupling to the National Perinatal Registry (PRN) to link neonatal risk factors for ROP with ophthalmological data, after completion of the inventory phase.

**Results:** Preliminary year results: a total of 2037 children were reported with a mean BW of 1324 gram and a mean GA of 30 weeks (wks). Data of 1790 infants were processed as 83 children (4.1%) were lost to follow up and 164 children died (9%). Of these infants 1655 were screened (92.4%). ROP was found in 317 children (19.2%): Stage 1 developed in 191 children (60.3%); mean BW 1066 g, mean GA 28 3/7 wks. Stage 2 was seen in 96 children (30.3%); mean BW 940 g, mean GA of 27 5/7 wks and 24 children (7.6%) with stage 3; mean BW of 948 g, mean GA of 27 wks. Two children (0.6%) developed stage 4 ROP; mean BW 1223 g, mean GA of 30 5/7 wks and 2 children developed a grade 5; mean BW 600 g, mean GA of 25 5/7 wks. Plus disease was found in 29 infants (9.2%). ROP was most seen between 26 and 29 weeks gestation (N=202) but there were also 4 children above 32 weeks of gestation (32-34 wks). 16 children were treated for ROP (5.1%).

**Conclusion:** Preliminary results of the NEDROP study: Only 4.1% of children were lost to follow up and 92.4% were screened for ROP. ROP developed in 19.2%. Severe ROP (stage ≥ 3) was found in 28 children (8.8%) and was most common between 26 and 29 weeks of gestation, but there was also 1 "big" child with stage 4. Sixteen infants (5.1%) were treated for ROP After closure of the inventory phase coupling with the PRN will give more insight in the perinatal risk factors for ROP.

**Conflict of Interest: None**



**T18: Five Year Visual Outcome of Children, Following Treatment with Diode Laser for Retinopathy of Prematurity Under Sub-Tenon's Local Anaesthesia.**

J Kate Barnes, Manoj V Parulekar, Chetan K Patel

Oxford Eye Hospital, John Radcliffe Hospital

*Presented by J Kate Barnes*

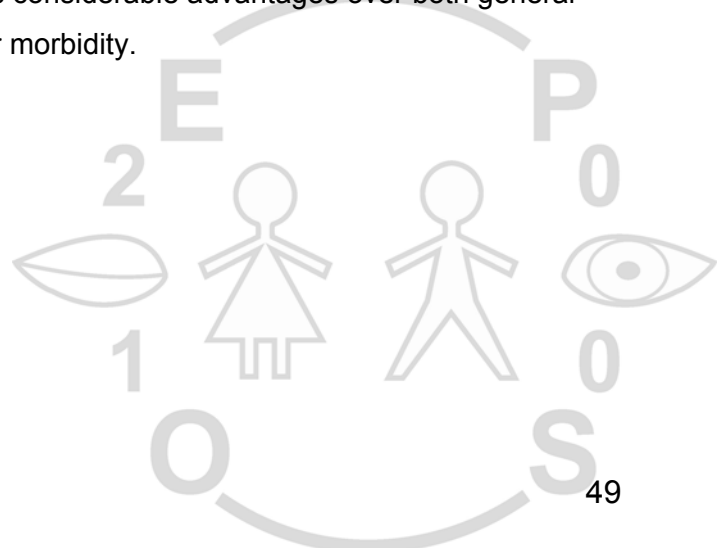
**Aim:** To audit visual outcome at 5 years of age following laser treatment for threshold retinopathy of prematurity (ROP) under sub-tenon's local anaesthetic.

**Methods:** A retrospective review of case notes was conducted, for 17 eyes of 9 neonates receiving diode laser peripheral retinal ablation for threshold ROP using oral sedation and sub-Tenon's anaesthesia over a period of 30 months. 5 years following treatment outcome measures recorded included logmar visual acuity, presence of myopia, astigmatism, strabismus, binocular function and poor structural outcome. Myopia was defined as spherical equivalent  $\geq 0.25$  diopters and high myopia  $\geq 5.00$  diopters. Astigmatism was defined as  $\geq 1.00$  diopter and high astigmatism  $\geq 2.00$  diopters. Educational and social competencies were also considered.

**Results:** Average logmar visual acuity was 0.359 (6/12 snellen equivalent), excluding one eye that had no light perception. Snellen equivalent was 6/12 or better in 70.6% of cases and better than 6/60 in 88.2% of cases. Myopia was seen in 5/17 (29.4%) of eyes of which 3/17 (17.6%) had high myopia. Astigmatism was seen in 5/17 (29.4%) of eyes of which 2/17 (11.8%) had high astigmatism. 4/9 (44.4%) of patients had a squint present at the 5-year follow-up. Steropsis was assessed using the Frisby test, with positive results in 5/9 (55.6%) of patients (results ranging from 85-300 seconds of arc). Structural abnormality was seen in 1 of the treated eyes. The affected eye developed an inoperable tractional retinal detachment with dragged macula, resulting in no light perception. Other ocular morbidity included 3 patients with nystagmus.

**Conclusion:** Sub-tenon's anaesthetic with oral sedation is a safe and effective technique for laser treatment of threshold ROP in neonates and does not compromise structural and visual outcomes, which are comparable to ETROP. We believe it has considerable advantages over both general and topical anaesthesia, with the potential for lower morbidity.

**Conflict of Interest: None**





**T19: Functional and Structural Ophthalmological Outcome in Cryo- or Laser Treated Premature Babies with Retinopathy of Prematurity (ROP) Between 1989 and 2008.**

Catherine Cassiman<sup>1</sup>, Peter Stalmans<sup>1</sup>, Joachim Van Calster<sup>1</sup>, Karel Allegaert<sup>2</sup>, Ingele Casteels<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, University Hospitals Leuven, <sup>2</sup>Department of Neonatology, University Hospitals Leuven

*Presented by Catherine Cassiman*

**Introduction:** To assess the evolution in postmenstrual age (PMA) at birth, birth weight, PMA at treatment, visual outcome, ophthalmoscopic appearance, refractive errors and strabismus in infants treated for ROP between 1989 and 2008 at the University Hospitals of Leuven, Belgium.

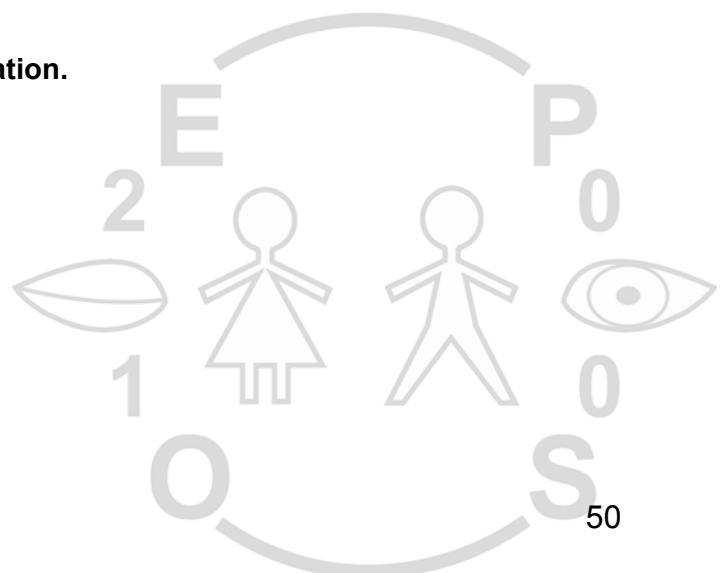
**Methods:** Retrospective analysis of the medical records of premature infants with ROP who were treated with cryotherapy or lasertherapy. The study population is divided into three groups. In group 1 (1989-1995) 26 cryo treated infants and in group 2 (1996-1999) 32 cryo treated infants are included. Group 1 was treated at the threshold stage whereas group 2 was treated at the prethreshold stage. Group 3 consists of 37 laser treated infants (2002-2008). Infants treated during the transition time from cryotherapy to lasertherapy (1999 - 2001) were excluded.

**Results:** Between the three groups differences in mean PMA at birth, mean birth weight and mean PMA at treatment were evaluated. Visual and refractive outcome, ophthalmoscopic appearance were compared. The presence of strabismus within the three groups, and involvement of the central nervous system were evaluated.

**Conclusion:** There is a tendency to better ophthalmological functional and structural outcome in infants treated with cryotherapy at the prethreshold stage (group 2) compared to those who were treated at the threshold stage (group 1). Ophthalmological functional and structural outcome is better in infants treated with lasertherapy (group 3) compared to those treated with cryotherapy (group 1 and 2). Subanalysis of the more recently laser treated group shows that poor visual outcome can mainly be attributed to central nervous system involvement. High myopia is mainly present in children who were diagnosed with ROP rush disease.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





## T20: Retinal Nerve Fibre Layer and Optic Nerve Head Parameters in Prematurely-Born Children.

Hanna Akerblom, Eva Larsson, Gerd Holmstrom

Dep. of Neuroscience, Uppsala University, Sweden

*Presented by Hanna Akerblom*

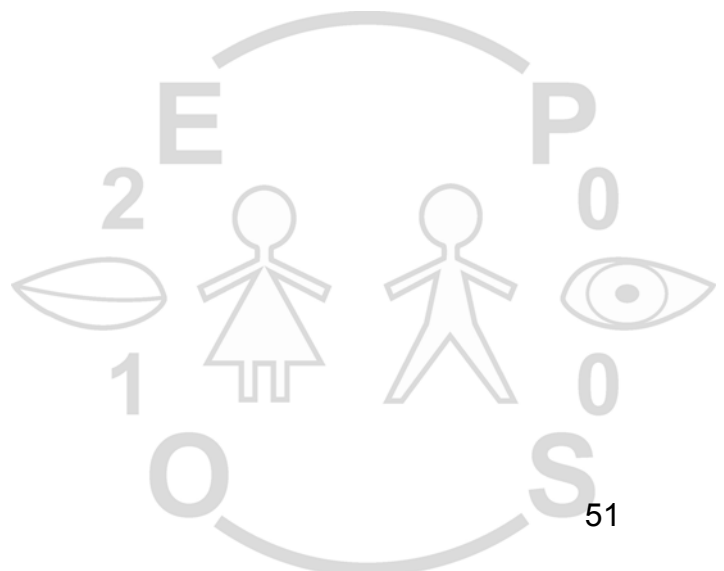
**Introduction:** To investigate the retinal nerve fibre layer (RNFL) and optic nerve head with optical coherent tomography (OCT) and Heidelberg retinal tomography (HRT) in prematurely-born children and children born at term.

**Methods:** Sixty-one children born before gestational age of 33 weeks together with a control group of fifty-five children born at term and with normal birthweights, were included in the study. Twenty-nine of the preterm children had ROP in the neonatal period and five were treated. RNFL was measured with Stratus OCT 3 and HRT III. The optic nerve head parameters were assessed with HRT and included; Disc Area (DA), Rim Area (RA), Cup Area (CA), Rim Volume (RV), Cup Volume (CV). The mean age at examination was 8.5 years in the preterm children and 10.1 years in the control group.

**Results:** The mean visual acuity was 1.0 in right (RE) and left eyes (LE) in the preterm group compared to 1.2 in the full-term group ( $p < 0,001$ ). In the preterm group the RNFL measured with OCT was significantly thinner in the nasal area, but not in the temporal, inferior or superior areas. The prematurely-born children had smaller DA but significant only in the right eye. The RA was significantly smaller in both eyes in the preterm group compared to the fullterm.

**Conclusion:** Prematurely-born children had lower visual acuity, but we could not prove that the reason for this was a reduced RNFL. However, in HRT, there was a trend for smaller DA and a significantly reduced RA. We have previously found thicker fovea in prematurely-born children. Thus the reason for reduced visual acuity may be due to both retinal and neuronal causes and remains to be elucidated.

**Conflict of Interest: None**



**T21: Which Psychophysical Colour Vision Test to Use for Screening in 3-9 Year Olds?**

Manca Tekavcic Pompe, Branka Stirn Kranjc

Univ. Medical Centre, Eye Hospital Ljubljana, Slovenia

*Presented by Manca Tekavcic Pompe*

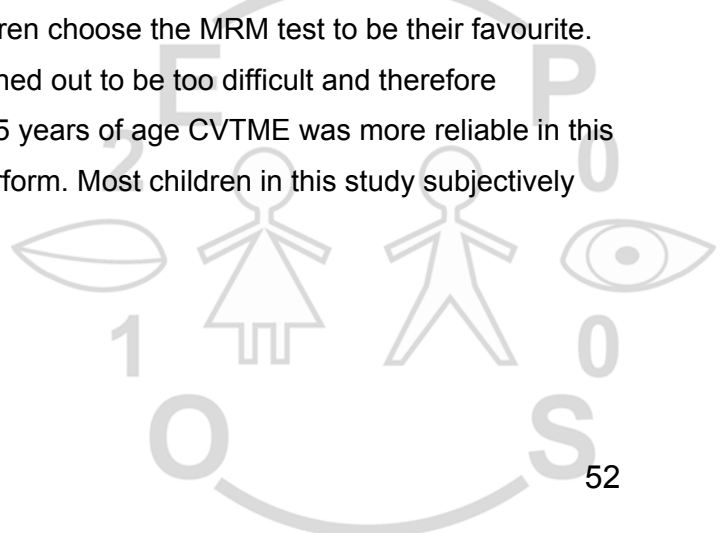
**Purpose:** To compare 4 psychophysical colours vision tests in children and to decide which test is most suitable for screening in pre-school and early school-age period.

**Methods:** The first test used was modified Ishihara plates (Handaya Co. Ltd., Tokyo). The second test »Colour vision testing made easy« (CVTME) (T.L. Waggoner, 1994) are also pseudoisochromatic plates with children friendly symbols. The third test used was »Neitz test of colour vision« (WPS, 2001) and the fourth test was »Mollon-Reffin Minimalist test« (MRM) (version 0.7, 1994). In the first three tests the child had to tell what is seen on the picture, whereas in the fourth test the child had to pick the coloured button among distracters. 37 children (19 girls and 18 boys) from 3-9 years of age with normal colour vision (at least one test should be performed without mistakes) were tested. The number of mistakes in each of the first 3 tests was registered, whereas in the MRM test the last button distinguished among distracters was registered. Every child was also asked which of the 4 tests he or she preferred.

**Results:** 16/37 children made one or more mistakes in the modified Ishihara test, among them all children under the age of 5 years. 9 made one mistake, 3 made two, 2 made three and 2 children made 5 mistakes. Altogether 31 plates were not correctly recognized, 21/31 plates were recognized as something else and 10 were not recognized at all. 8/37 children made one to three mistakes in CVTME test. 4 children made one mistake, 3 made two and 1 child made three mistakes. Altogether 13 plates were not correctly recognized. Since all of them are compositions of many motives, not all motives were recognized. All tested children made at least one mistake in the Neitz test. All gave wrong description for figure at position 6 of the test. Two 3-year old girls weren't able to perform the test. MRM test was performed in full by 14/37 children. Additional 8 children could not distinguish the faintest blue button among the distracters (which is still declared as normal colour vision by the authors). 28/37 children choose the MRM test to be their favourite.

**Conclusions:** Modified Ishihara colour test has turned out to be too difficult and therefore unreliable in youngest children. For children under 5 years of age CVTME was more reliable in this study. For all ages MRM test was the easiest to perform. Most children in this study subjectively preferred the MRM test.

**Conflict of Interest: None**



**T22: Evaluation of the Neonatal Macula Using Spectral Domain Optical Coherence Tomography (SD OCT)**

Ramiro S. Maldonado, Neeru Sarin, Rachelle O'Connell, David K. Wallace, Sharon F. Freedman, Cynthia A. Toth

Duke University Eye Center

*Presented by Ramiro S. Maldonado*

Introduction Optical coherence tomography (OCT) imaging of the retina plays an important role in the diagnosis of adult retinal diseases. With the exception of a few case reports, this technology has not been used for imaging the retina in the neonatal period. Limitations such as different optical properties of the neonatal eye and poor cooperation have prevented this technique from being used widely in infants. The purpose of this study was to evaluate the feasibility, safety and efficacy of an age-customized method of performing spectral domain OCT (SD OCT) for the evaluation of the macula in premature neonates. Methods Thirty-eight premature neonates, ages 31-42 weeks postmenstrual age (PMA) were enrolled in this IRB approved study. SDOCT was performed at the bedside using a portable system. SD OCT imaging parameters were customized for the infant age. . Feasibility was assessed by the time to complete each session and the number of scans needed to achieve macular imaging. Safety was assessed by documenting fluctuations in vital signs (oxygen saturation, heart rate and respiratory rate) and adverse events. Efficacy was determined by a) B-scan image quality adequate to identify retinal contour and differentiate retinal layers and b) ability to image the macula. Results A total of 305 individual imaging sessions were performed. It took an average of 11 minutes to image both eyes (with an average of 4 scans per eye), and the macula was imaged on average by the second scan. Vital signs did not change more than 20% from baseline values during 301 (98%) of the 305 imaging sessions, and no adverse events related to imaging were observed. The B-scan images met our quality requirements in 250 sessions (82%), and we were able to image the macula in 280 sessions (92%). SD OCT revealed sub-clinical pathology such as macular edema, pre-retinal tissue and sub-retinal fluid, not noted by indirect ophthalmoscopy. Conclusions SDOCT using an age-customized approach is a feasible, safe and efficient, imaging tool to evaluate the posterior pole of premature neonates. This imaging modality reveals sub-clinical pathology and might aid in understanding the pathogenesis of neonatal retinal diseases. 1. Maldonado et al. IOVS 2010, p2678

**Conflict of Interest: None, Dr. Cynthia Toth receives Research Financial support from Biogen. No financial interest.**

**T23: OCT and Clinical Features of Chorioretinal Colobomas in Children**

Jaume Català-Mora<sup>1</sup>, Annabella Zurita<sup>2</sup>, Jesús Díaz-Cascajosa<sup>1</sup>, Mariona Vidal-Santacana<sup>1</sup>, Rosa Navarro-Nomen<sup>3</sup>, Joan Prat-Bartomeu<sup>1</sup>

<sup>1</sup>Hospital Sant Joan de Déu-Barcelona. Spain., <sup>2</sup>Instituto Clínico La Florida-Caracas. Venezuela.,

<sup>3</sup>Hospital Universitari de Bellvitge. Barcelona. Spain

*Presented by Jaume Català-Mora*

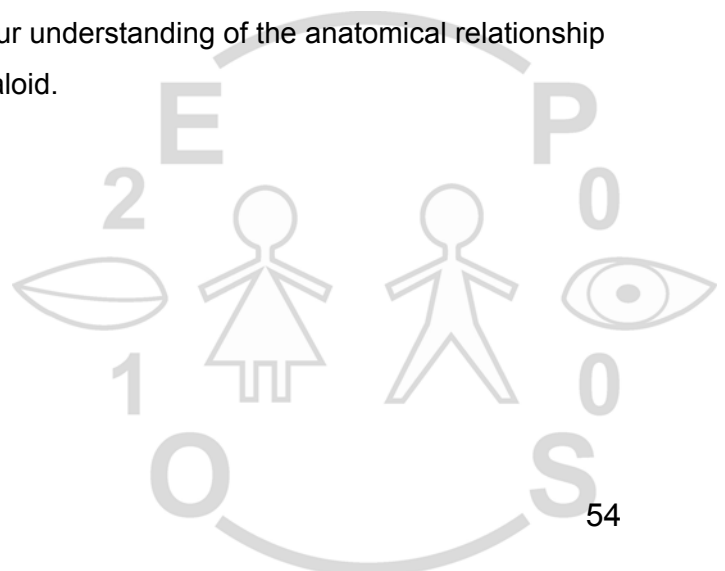
**Introduction:** Chorioretinal coloboma is a congenital defect caused by an incomplete closure of the embryonal fissure. The severity of the visual disability depends on the size and the involvement of optic disc and macula, and associated anomalies of the eye such as microphthalmos and nystagmus. Eyes with chorioretinal coloboma are at risk of detachment of the remaining retina.

**Methods:** We have reviewed charts from all patients with chorioretinal coloboma in our hospital between 2004 and 2009 collecting their best-corrected visual acuity, slit lamp examination, ophthalmoscopic examination, complications and any ophthalmological event. Then we selected patients suitable for Fourier Domain OCT imaging of the coloboma margin.

**Results:** 42 patients with coloboma, aged 1 mo-21 yo. 23 were boys and 17 were girls. 17 had unilateral coloboma (8 RE/9 LE) and 23 had bilateral coloboma. The iris was involved in 6 patients, papilla was affected in 10 cases and macula in 4 eyes. A combination of iris, papilla and/or macula was seen in the other 43 cases. We could find other ophthalmic malformations in 21 patients whereas systemic association was seen in 12 patients. 3 eyes developed retinal detachment that needed surgical repair while 2 other eyes developed retinal detachment resolved spontaneously. OCT was feasible in 15 patients. As described by Gopal et al. colobomas are covered or the intercalary membrane (ICM). Subclinical retinal detachments, retinoschisis and subretinal and sub-ICM spaces communication could be observed.

**Conclusions:** Chorioretinal colobomas are known as associated to other ophthalmic and systemic malformations. Retinal detachment is a relatively frequent complication that can spontaneously resolve in some cases although most of the patients require pars plana vitrectomy and silicone oil tamponade. OCT findings in these patients helps our understanding of the anatomical relationship between coloboma, normal retina and posterior hyaloid.

**Conflict of Interest: None**





**T24: Foveal Ultrastructure and Fundus Autofluorescence in Achromatopsia.**

Christoph Friedburg<sup>1</sup>, Julia Hoeges<sup>1</sup>, Susanne Kohl<sup>2</sup>, Birgit Lorenz<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Molecular Genetics Laboratory, Institute for Ophthalmic Research, Centre for Ophthalmology, University Clinics Tübingen, Germany

*Presented by Christoph Friedburg*

**Purpose:** To evaluate foveal morphology in achromatopsia.

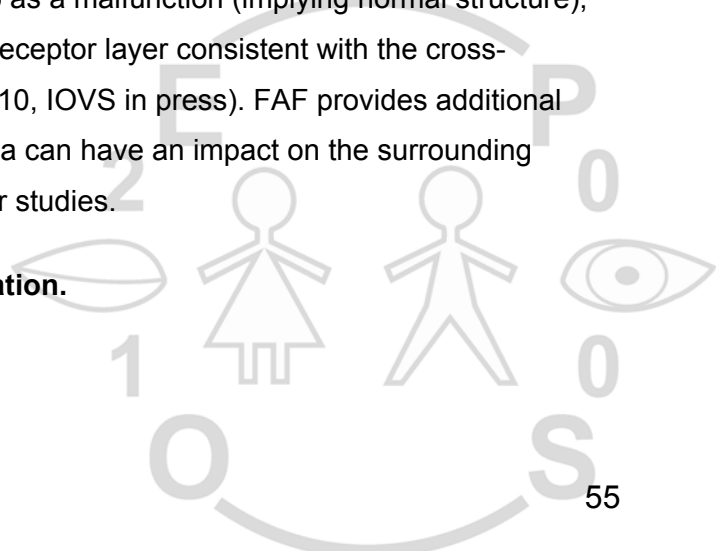
**Methods:** Fifteen patients, one aged 79, the remaining aged 6 to 34 (median 16 years) with a clinical diagnosis of achromatopsia were reassessed using optical coherence tomography (SD-OCT, volume scan if possible) and fundus autofluorescence (FAF; Spectralis, Heidelberg Engineering, Germany,). Ethic committee approval and written informed consent were obtained. Inclusion criteria were history of stable disease and plausible autosomal-recessive inheritance, congenital nystagmus, severely reduced visual acuity, no ophthalmoscopically visible signs of retinal degeneration, absent cone and normal rod electroretinogram. Four patients carried mutations in *CNGB3*, one in *GNAT2*, and one in *CNGA3*. In Patient #1, these 3 genes and blue cone monochromatism were excluded as causative. In 9 patients including the latter, mutations still have to be identified.

**Results:** OCT scanning was impeded by nystagmus. In some cooperative patients we used a contact ring similar to a gonioscopy lens, to reduce the amplitude of the nystagmus. Despite this, no OCT of the fovea could be obtained in 3 patients. Most patients demonstrated zones of reduced OCT-signal in the outer and inner segment photoreceptor layer. In some, this area appeared irregular. Surprisingly, two patients had dense reflexes in that layer, thickened in one of them (Patient #1). The FAF pattern of the macula appeared changed with a smaller than normal central zone of relatively lower signal. Some patients had irregularities or rarely a thin ring of clearly enhanced FAF. Three patients without a molecular diagnosis showed new features that casted doubts on the diagnosis of achromatopsia.

**Conclusion:** Although achromatopsia is referred to as a malfunction (implying normal structure), most patients show subtle defects within the photoreceptor layer consistent with the cross-sectional study of achromats by Thiadens et al. (2010, IOVS in press). FAF provides additional information. Whether structural changes of the fovea can have an impact on the surrounding retina, leading to changes in FAF will require further studies.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





## T25: Ocular Manifestations of Incontinentia Pigmenti

Pascal Dureau<sup>1</sup>, Florence Metge<sup>1</sup>, Christine Bodemer<sup>2</sup>, Georges Caputo<sup>1</sup>

<sup>1</sup>Fondation Rothschild, Paris, France, <sup>2</sup>Service de Dermatologie, Hôpital Necker, Paris, France

*Presented by Pascal Dureau*

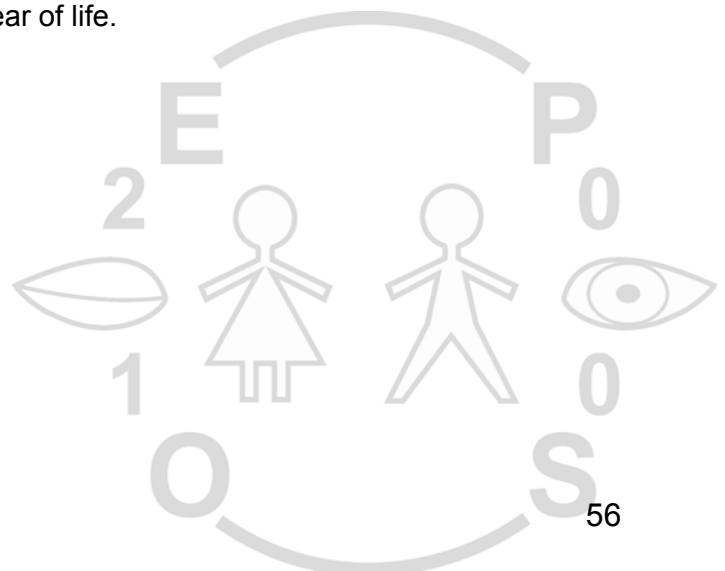
**Introduction:** Incontinentia Pigmenti (IP) is a neuro-oculo-cutaneous disease characterized by a congenital erythematous vesicular rash followed by hyperpigmentation and atrophy. Transmission is X-linked dominant. Neurologic complications result from vascular occlusions. The main ocular manifestation is retinal ischemia leading to neovascularization and detachment, resembling ROP. The aim of this study was to review ocular manifestations in a group of IP patients and discuss a follow-up program.

**Methods:** All IP patients referred for ocular screening or complications from 2003 to 2010 were retrospectively studied. The following points were noted: age at time of first examination, refraction, anterior and posterior segment status, neurological complications, surgical treatment, follow-up, final anatomical and functional outcome.

**Results:** A total of 13 patients were referred, 6 for screening and 7 for complications. Median age at the time of first examination was 2.5 months for screening and 4.2 months for complications. Mean refraction was slightly hyperopic. No patient referred for screening experienced complications during a median 8.7 months follow-up time. One adult patient had an optic neuropathy possibly related to IP. Six infants had tractional retinal detachment and surgery did not permit reattachment in any of these cases. Three of them had neurological complications.

**Conclusion:** The main ocular manifestation of IP is retinal detachment secondary to peripheral retinal ischemia and neovascularization. The anatomical and functional prognosis is poor despite early surgery. Anterior segment complications are often the consequence of posterior segment status. Neurological ischemic complications are often associated. For unknown reasons, these severe retinal complications are generally unilateral and occur in the first months of life. A careful screening program is strongly advised in the first year of life.

**Conflict of Interest: None**



**T26: RD3 mutation in a consanguineous LCA family**

Markus Preising<sup>1</sup>, Nora Hausotter-Will<sup>1</sup>, Manuel Solbach<sup>1</sup>, Christoph Friedburg<sup>1</sup>, Franz Rüschemdorf<sup>2</sup>, Birgit Lorenz<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Max Delbrück Center for Molecular Medicine Berlin-Buch, Germany

*Presented by Markus Preising*

**Purpose:** To identify the underlying gene defect in a consanguineous Kurdish LCA family with several affecteds.

**Methods:** Linkage analysis was performed in a consanguineous 4 generation Kurdish family with 5 affected children consistent with autosomal recessive inheritance. The patients were clinically examined by BCVA, Goldman visual field, Ganzfeld ERG, VEP, and funduscopy. *GUCY2D*, *RPE65*, *LRAT*, *AIPL1*, *CRX*, *CRB1* and *CEP290* were submitted to mutation analysis by SSCP. Linkage analysis was performed by a SNP-microarray (Affymetrix Mapping 50k Xba 240 Array) assay. To limit the candidate region additional microsatellite markers were selected from the NCBI-dbSNP-Database according to the SNPs linked in the microarray assay. The microsatellite markers were amplified by standard PCR conditions. PCR products were analyzed on a QIAxcel capillary gel electrophoresis (Qiagen, Hilden). Alleles were determined from the electropherogram and haplotypes were built in Cyrillic 2.1. *RD3* was amplified by PCR in two amplicons and sequenced directly. 50 further patients are screened in an ongoing study in sets of increasing age at a visual acuity (VA) > 0.1 to describe further mutations in *RD3*.

**Results:** The patients presented with nystagmus, increased glare sensitivity, reduced BCVA since birth and progressive visual field constriction. VEP or ERG were below threshold in early childhood. Funduscopy at age 2 revealed constricted retinal vessels, pigment irregularities were seen in the periphery and the macula. Classical LCA genes were excluded. The SNP microarray assay was used to map the underlying gene defect to chromosome 1q31.3 – 1q32.3 covering known genes for retinal degenerations including *CRB1*, *USH2a*, and *RD3*. Linkage analysis reduced the candidate region to 3 Mb covering 138 genes including 10 genes involved in eye or brain function. *RD3* was chosen for candidate analysis since it had been reported once in a single Indian LCA family. Direct sequencing of *RD3* revealed a homozygous stop mutation (p.Y60X (c.180C>A)) in exon 2. Screening of the first sets of patients with VA > 0.1 before age 2 revealed no mutations.

**Discussion:** We identified the second family showing a mutation in *RD3*. It is the first nonsense mutation detected in *RD3*. Low detection rates in patients of VA > 0.1 before age 2 still making *RD3* a rare cause in LCA. Support: DFG Lo457/5, ReForM, Pro Retina Deutschland

**Conflict of Interest: None**



## T27: New Insight in Retinal Phenotype of Patient with *AiPL1* Mutations

Francesco Testa<sup>1</sup>, Settimio Rossi<sup>1</sup>, Valentina Di Iorio<sup>1</sup>, Sandro Banfi<sup>2</sup>, Alberto Auricchio<sup>2</sup>,  
Francesca Simonelli<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, Second University of Naples, Italy, <sup>2</sup>Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy

*Presented by Francesco Testa*

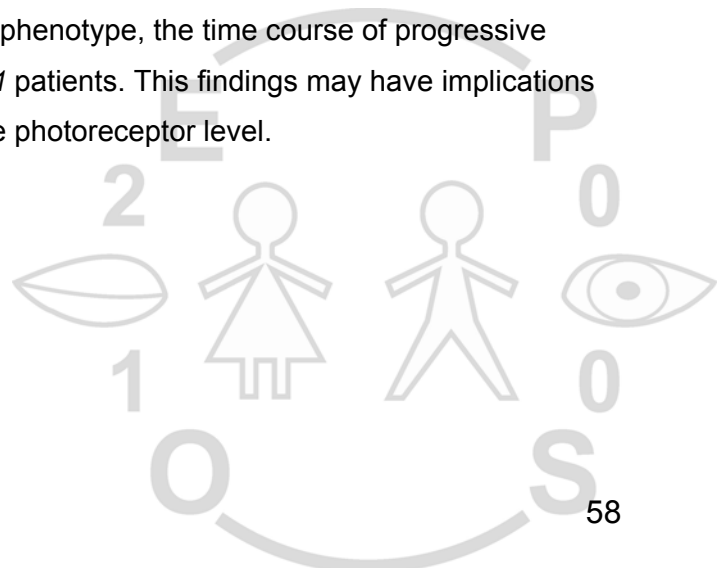
**Introduction:** To describe the retinal dystrophy phenotype associated with mutations in *AiPL1*, the gene encoding a aryl hydrocarbon receptor-interacting protein like-1 expressed in the photoreceptor cells and the pineal gland.

**Methods:** Ten patients with Leber congenital amaurosis (LCA) from 8 families with pathogenic *AiPL1* mutations on both alleles were studied. Retinal phenotypes were characterized by ophthalmic examination, including electroretinography, spectral domain optical coherence tomography (SD-OCT) and fundus autofluorescence.

**Results:** The molecular analysis revealed that 50% of *AiPL1* patients is homozygous for the most frequent p.Trp278X mutation, resulting in a null genotype, and 80 % had at least one allele with the p.Trp278X mutation. These individuals appeared to share a common clinical picture, independent of the type of mutation, characterized by poor, visual function in early life, due to both rod and cone degeneration. Keratoconus and cataracts were identified in 30% of patient. Marked pigmentary retinopathy, including bone spicules in the peripheral retina, was present in 80% of patients associated with maculopathy. Electroretinograms were extinguished in all patients. A unique view into the degree retinal degeneration was achieved by detection of fundus autofluorescence and partially retained retina lamellar structures with a preservation of the outer nuclear layer at OCT examination.

**Conclusion:** Ophthalmic findings in patients with LCA due to *AiPL1* mutations suggest that *AiPL1* loss-of-function results in a severe form of pigmentary retinopathy associated with maculopathy and in one third of cases keratoconus and cataract. From our autofluorescence and OCT data, it seems likely that, despite the severity of the retinal phenotype, the time course of progressive photoreceptor cell death may be slow in LCA *AiPL1* patients. This findings may have implications for future therapies designed to restore vision at the photoreceptor level.

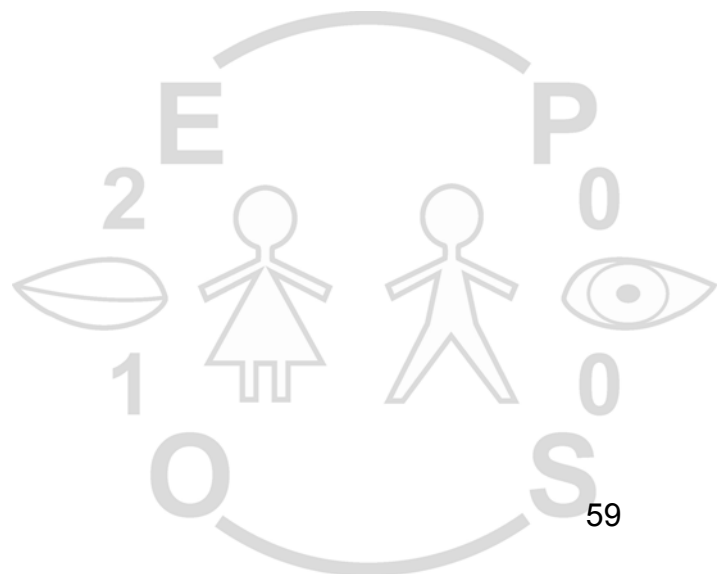
**Conflict of Interest: None**





# Poster Presentations

## Poster Presentations



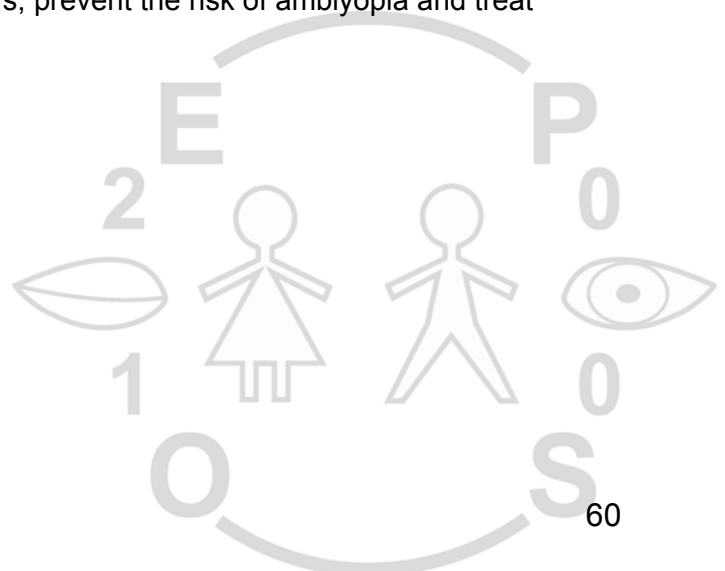
**P01: Preschool Visual Screening in the Municipality of Thessaloniki**Areti Vartholomeou<sup>1</sup>, T. Naoumidi<sup>2</sup>, M. Antoniadis<sup>2</sup>, A. Mandalos<sup>2</sup>, O. Leliopoulou<sup>2</sup>, N. Ziakas<sup>2</sup><sup>1</sup>Papageorgiou Hospital, Thessaloniki, Greece, <sup>2</sup>Ahepa Hospital, Thessaloniki, Greece*Presented by Nicolaos Ziakas*

**Introduction:** The Municipality of Thessaloniki organized a systematic routine health check-up in preschool children during which all children underwent physical, mental, and developmental checkups including dental, eye, and hearing examinations. The aim of this study is to assess the outcome and the importance of the preschool vision screening program.

**Methods:** A team of 7 ophthalmologists participated in a preschool visual screening program during the year 2009, in which 1025 children, aged up to 5 years old, from 19 day nursery schools of the Municipality of Thessaloniki, were included. The examination procedure consisted of 2 steps: 1. Visual acuity testing (only for  $\geq 2$  year-old children) using the Kays LogMar acuity test and 2. Orthoptic evaluation. Depending on the main outcome measures, those children that required further evaluation were referred to the Pediatric Ophthalmology Department of the Aristotle University of Thessaloniki in Ahepa Hospital, where a thorough ophthalmic examination was conducted to detect ocular anomalies, refractive errors, strabismus, and amblyopia.

**Results:** Of the total 1025 children that were included in the study, 779 (76%) were examined, 223 (22%) were absent at the time of visit and 23 (2%) did not cooperate. Distribution of ages is described as follows: 234 were aged 4-5 years old (30%), 336 were 3-4 years old (43%), 184 were 2-3 years old (24%) and 25 were  $< 2$  years old (3%). The 749 (97%) were normal, 11 (1%) already wore glasses and 19 (2%) required further evaluation. Of those referred: 12 children (63%) did not attend, 5 (26%) were diagnosed with astigmatism and 2 (11%) with mild hyperopia and esophoria. No cases of strabismus or amblyopia were detected.

**Conclusion:** The incidence of non diagnosed ophthalmic pathology in the preschool vision screening program of Thessaloniki, during the year 2009, was low. Nevertheless, the conduction of such programs is essential to detect refractive errors, prevent the risk of amblyopia and treat strabismus

**Conflict of Interest: None**



## P02: Place of Web-Based Visual Screening in Pediatric Eye Care

Patricia Domsa<sup>1</sup>, Zsuzsanna Alberti<sup>2</sup>, István Kiss<sup>4</sup>, Vilmos Kiss<sup>4</sup>, Samu Krisztián<sup>5</sup>, Katalin Sényi<sup>3</sup>

<sup>1</sup>Dept. Pediatric Ophthalmology HPCH Madarász Street Children's Hospital, <sup>2</sup>Faculty of Medicine, Semmelweis University, <sup>3</sup>Dept. of Ophthalmology, Semmelweis University, Budapest, <sup>4</sup>Tavkapcsolat Kft, <sup>5</sup>Dept. Mechatronics, Optics and Information Engineering, University of Technology and Economics, Budapest

*Presented by Patricia Domsa*

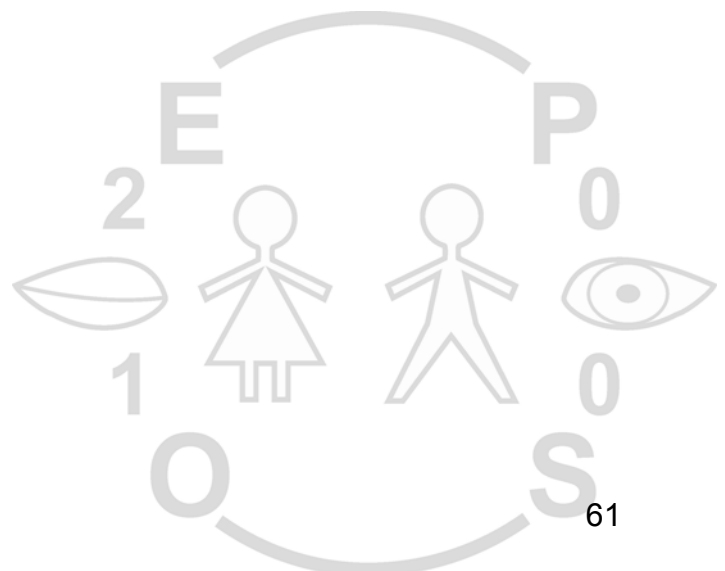
**Introduction:** Disparity of financial resources and lack of well-trained screening staff is an everyday challenge in pediatric eye care in Hungary. As web-based learning and self-testing is becoming more and more accepted, we decided to develop an easy to use web-based visual function screening test in Hungarian. Our goal was to develop a system for inexperienced users – for instance kindergarten teachers and health visitors -, and which is compatible with legacy hardware. The aim of this study was to evaluate the effectiveness of our new web-based test battery.

**Methods:** Forty-one children (age range 3-17 years) performed the computer based screening in our outpatient clinic, and were examined with widely used tests for comparison. Visual acuity was tested and compared using Snellen type symbol charts. Computer based color vision test was compared with the Ishihara Color Vision Test. Sensitivity, specificity, positive predictive value, negative predictive value and kappa test were evaluated.

**Results:** The sensitivity and the negative predictive value of all the visual acuity tests and the color vision test reached 100%, whereas the specificity and the positive predictive value of visual acuity tests were lower (Landolt: 66% and 60%; Valentine 62.5% and 70%; Valentine Multi: 71.4% and 75% respectively). The specificity of the web-based color vision test was 100%.

**Conclusion:** Our web-based tests are simple to use, offer high sensitivity and reasonable specificity in hands of non-trained personnel as well. Standard testing cannot be substituted by the method, but it helps to screen children at kindergarten, school or even at home, drawing attention to vision disorders.

**Conflict of Interest:** None



**P03: In Vitro Expression of the Anti-VEGF-F(Ab)-Fragment Ranibizumab**

Tobias Wimmer, Nina Wagner, Markus Preising, Birgit Lorenz, Knut Stieger

Department of Ophthalmology, Justus Liebig University, Giessen, Germany

*Presented by Tobias Wimmer*

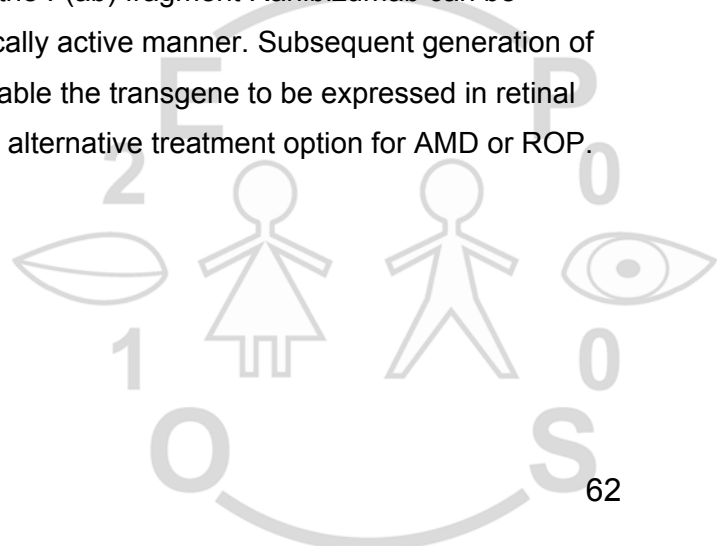
**Introduction:** Overexpression of VEGF (vascular endothelial growth factor) is the major growth factor involved in neovascular disorders such as AMD (age-related macula degeneration), but also the retinopathy of prematurity (ROP). In patients suffering from AMD, the treatment of choice comprises of repeated intravitreal injections with the VEGF-Inhibiting antibodies Bevacizumab and Ranibizumab. Recently, single injections of Bevacizumab in premature infants with advanced stages of ROP also showed significant success in limiting uncontrolled growth of blood vessels in the retina. The aim of this project is to clone and express the VEGF-inhibiting F(ab)-Fragment Ranibizumab (Lucentis®) in vitro, and the determination of its VEGF inhibiting activity in a proliferation-assay.

**Methods:** Ranibizumab is made of two antibody chains, the light and a part of the heavy chain of the VEGF-binding IgG Bevacizumab. Both chains were cloned into the expression-vector pIREShrGFP1a (Stratagene, Aglient Technologies Inc., Santa Clara CA). HEK293 (ATCC: CRL-1573) cell-cultures were transfected with this construct using Lipofectamin LTX (Invitrogen, Darmstadt). Expression of the chains was verified by western blot under reducing and non reducing conditions. The biological activity of the secreted protein was analyzed with a BrdU-Proliferation-Assay.

**Results:** We successfully cloned both genes into one expression-vector separated by an IRES (internal ribosomal entry site) to obtain the expression of this two genes simultaneously. The expression of both genes was shown by Western blot analysis under reducing and non reducing conditions. Both chains form a heterodimer, are secreted into the medium and showed significant biological activity in an in vitro assay.

**Conclusion:** The results of this study indicate that the F(ab) fragment Ranibizumab can be expressed from eukaryotic cells in vitro in a biologically active manner. Subsequent generation of viral vectors using this expression cassette may enable the transgene to be expressed in retinal cells in vivo after gene transfer into the retina as an alternative treatment option for AMD or ROP.

**Conflict of Interest: None**





**P04: Keratitis in Reiter's Syndrome in Childhood**

Nikolaos Kozeis<sup>1</sup>, Maria Trachana<sup>2</sup>, Eleni Pratsidou<sup>2</sup>, Nikolaos Ziakas<sup>3</sup>, Straton Tyradellis<sup>1</sup>

<sup>1</sup>Paediatric Eye department, Hippokration Hospital of Thessaloniki, Greece, <sup>2</sup>Paediatric Immunology and Rheumatology Referral Center, First Department of Pediatrics, Hippokration Hospital of Thessaloniki, Greece, <sup>3</sup>1st University Eye department of AUT, Thessaloniki, Greece

*Presented by Nikolaos Kozeis*

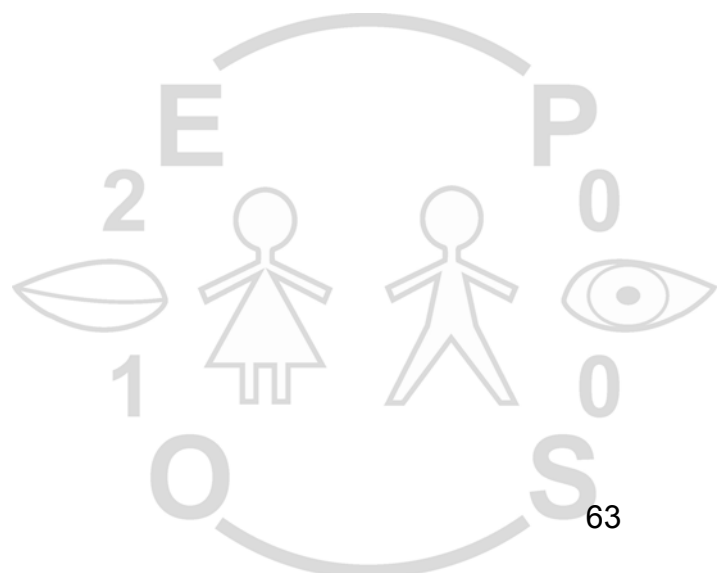
**Introduction:** To present a rare ocular manifestation of Reiter's syndrome in a child

**Methods:** A 10 year old girl who was admitted to our hospital with low grade fever, arthritis and aching left eye with blurred vision was diagnosed with Reiter's syndrome. At the time of admittance the ophthalmologic examination revealed keratitis, this mildly affected the vision

**Results:** Keratitis was resolved with treatment with topical steroids and antibiotic drops after one month, without scarring. Although 75% of the patients with Reiter's syndrome present ophthalmic manifestations, keratitis is a very rare finding in Reiter's syndrome and even rarer in children

**Conclusion:** It should be kept in mind that keratitis could be an ocular manifestation of Reiter's in young age

**Conflict of Interest: None**





**P05: Oculoglandular Syndrom Parinaud**

Aneta Skupin, Melanie Jaeger, Birgit Lorenz

Department of Ophthalmology, Justus Liebig University, Giessen, Germany

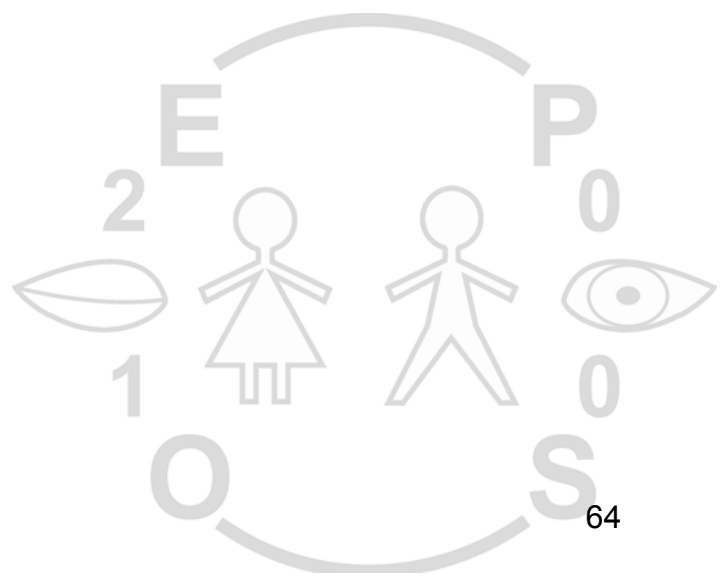
*Presented by Aneta Skupin*

**Background:** Parinaud Syndrome is a rare eye manifestation from a cat-scratch fever with conjunctivitis accompanied by nearby regional lymphadenopathy.

**Case report:** A 4-year-old boy with unilateral granulomatous conjunctivitis of the right eye (fig. 1), ipsilateral preauricular lymphadenopathy (fig. 2) and fever for 2 weeks was referred to our ophthalmology unit. Slit lamp examination showed conjunctival injection and chemosis (fig.3). There were no pathological findings in the fundus of both eyes (fig. 4). The blood tests showed an increase of IgG antibody levels to Bartonella henselae bacterium. There was a history of exposure to little cats. A local therapy was initiated with Ofloxacin, Erythromycin and Dexpanthenol. Systemically Rifampicin and Azithromycin were given thereafter the disease remained stable. After 2 weeks almost all symptoms had resolved (fig. 5).

**Conclusion:** Parinaud syndrome is the most common manifestation of atypical cat scratch disease. When the diagnosis is made early and treatment started immediately, the outcome of Parinaud syndrome can be very good.

**Conflict of Interest: None**





**P06: Bilateral Traumatic Optic Neuropathy in Child - Case Report**

Nikolaos Ziakas<sup>1</sup>, Areti Vartholomeou<sup>2</sup>, Nikolaos Kozeis<sup>3</sup>

<sup>1</sup>Ahepa Hospital, Thessaloniki, Greece, <sup>2</sup>Papageorgiou Hospital, Thessaloniki, Greece, <sup>3</sup>Hippokratio Hospital, Thessaloniki, Greece

*Presented by Nikolaos Ziakas*

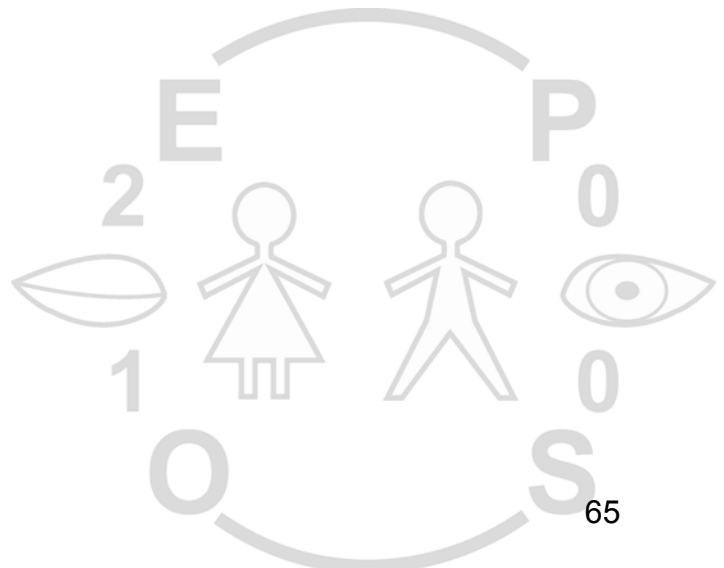
**Introduction:** Traumatic optic neuropathy usually occurs laterally in association with blunt skull trauma involving fractures of the skull and optic canal and signs of local or systemic contusion. Rarely it occurs after blunt ocular trauma. We report an atypical case involving bilateral traumatic optic neuropathy in a child after minor blunt ocular injury.

**Methods:** A seven year old boy presented to our department after having been injured by an elastic rope in both eyes. The patient underwent a thorough ophthalmological examination and therapy was set according to findings. Follow up examination data that were recorded after two days, 2 weeks, 4 weeks and 3 months later are being presented. Therapy was also adjusted accordingly and the patient is still being followed.

**Results:** Initial examination revealed bilateral corneal epithelial defect, elevated IOP and hyphema which obscured fundoscopy. Therapy was set with Timolol-Dorzolamide as well as Tobramycin-Dexamethasone eye drops. After two days bilateral iridoplegia was also observed but the ocular ultrasonography was normal. After two weeks, examination was normal except for a remaining hyphema in the left eye and bilateral iridoplegia. Four weeks later bilateral optic nerve atrophy was noted. Neuroimaging revealed optic sheath swelling due to cerebrospinal fluid concentration. Three months later the patient had visual acuity of 1/20 in the right and light perception in the left eye.

**Conclusion:** Direct or indirect trauma may damage the optic nerve and result in permanent visual loss. The diagnosis of traumatic optic neuropathy is not always straightforward and is complicated by unfavourable circumstances for the examination. Questions are still being raised about the efficiency of current therapy for improving visual outcomes. To our knowledge such case of bilateral traumatic optic neuropathy in a child following blunt ocular injury has not been reported before.

**Conflict of Interest: None**



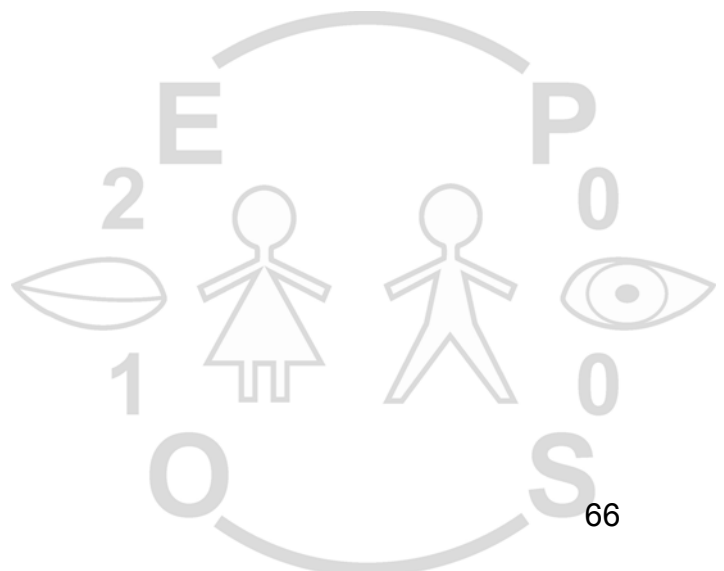
**P07: And where is Pathology at Oculomotor Pareses? New Approach to Old Challenge**

Igor Aznauryan, Victoria Balasanyan

Association of pediatric ophthalmology clinics "Yasniy Vzor"

*Presented by Victoria Balasanyan*

It is universally accepted that the oculomotor system paresis and palsy are due to the nucleus local pathology of either oculomotor, abducent or trochlear nerves, or their combination. However, the clinical presentation of these states often varies greatly within wide limits: the degree of paresis manifestation, the number of muscles involved into paresis, the degree of movement disorders in some muscles, occurrence of various combined nystagmoid movements, etc. From the aforesaid, it seems doubtful that the reason for the oculomotor involvement, oculomotor pareses, is the isolated nucleus lesion of the cerebral nerve innervating this or that muscle. We have studied from literature and in clinics the internuclear and supranuclear disorders, as well as the breaking of bonds in the vestibule-ocular system and those associated with the posterior longitudinal fascicle. Based on the results of our investigation we have concluded that in major cases the paralytic strabismus is not brought on by the direct injury of the respective cerebral nerve nucleus, but is initiated, first of all, by the breaking of the higher-order supranuclear and internuclear bonds, which determine control over the cerebral nerve final nuclei. From the findings of our investigation, we have developed the surgical procedure for treating the paralytic strabismus in children, which makes possible eye movement in all directions of the horizontal gaze.

**Conflict of Interest: None**

**P08: Visual Skills and Gross Motor Function in Spastic Diplegic Children**Nikolaos Kozeis<sup>1</sup>, Dimitrios Zafeiriou<sup>2</sup>, Nikolaos Ziakas<sup>3</sup>, Straton Tyradelis<sup>1</sup>

<sup>1</sup>Pediatric Eye department Hippokration Hospital of Thessaloniki, Greece, <sup>2</sup>1st University Paediatric department AUT, Thessaloniki, Greece, <sup>3</sup>1st University Eye department AUT, Thessaloniki, Greece

*Presented by Nikolaos Kozeis*

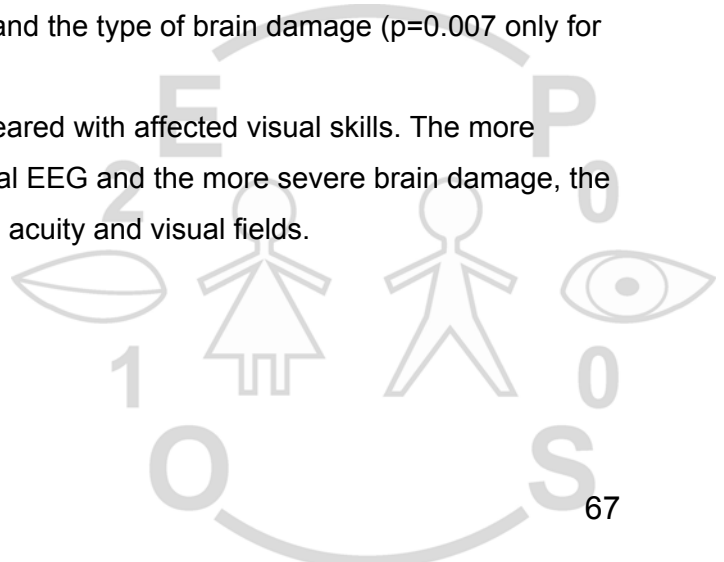
**Purpose:** In this study it was investigated the relationship between abnormal visual parameters and other parameters in children with spastic diplegia (SD) due to cerebral palsy (CP). Patients and

**Methods:** Twenty eight SD children (age range 24-54 months; mean age 38.86 months; std. 22.83) were enrolled in this study. Data, regarding pre-, peri- and postnatal events, history of epilepsy, were recorded retrospectively. Evaluation of the best corrected binocular visual acuity / resolution (BVA) (by using Lea's grating test), refraction (after cycloplegic refraction), functional visual fields, gross motor function (GMFCS), electroencephalogram (EEG) and brain MRI were performed in all patients.

**Results:** Seventeen children (60.70%) were classified as level GMFCS 1-2 (mild) and eleven (39.30%) as level GMFCS 3-5 (severe). One eye (3.60%) of them had BVA < 0.21 cpd (equivalent  $x < 6/120$ ), seven eyes (25.00%) had  $0.21 < BVA < 1.75$  cpd (equivalent  $6/120 < x < 6/18$ ) and twenty eyes (71.40%) had BVA > 1.75 cpd (equivalent  $x > 6/18$ , normal for the age group). Nine children (32.10%) appeared with hyperopia, three children (10.70%) with myopia and sixteen children (57.10%) with insignificant refractive error. For statistical analysis, only the results of the eye with better vision were considered. Three children (3.70%) manifested abnormal binocular functional visual fields. Three children (10.70%) were epileptic, while twenty- five (89.30%) demonstrated radiological evidence of periventricular white matter pathology and three (10.70%) demonstrated hypoxic-ischaemic lesions by brain MRI. Statistical analysis using the Kruskal-Wallis and Mann-Whitney U test (significant level 0.05), proved that abnormal visual acuity and functional visual fields were associated with: abnormal electroencephalogram ( $p=0.023$  &  $p=0.002$  respectively), the level of GMFCS ( $p=0,000$  &  $p=0.035$  respectively) and the type of brain damage ( $p=0.007$  only for the visual fields).

**Conclusion:** A significant incidence of CP SD appeared with affected visual skills. The more severely affected gross motor function, the abnormal EEG and the more severe brain damage, the more common to be associated with affected visual acuity and visual fields.

**Conflict of Interest: None**



**P09: Impaired Foveal and Peripheral Face Processing in Amblyopia**Judit Körtvélyes<sup>1</sup>, Éva Mária Bankó<sup>2</sup>, János Németh<sup>1</sup>, Zoltán Vidnyánszky<sup>2</sup><sup>1</sup>Dept. of Ophthalmology, Semmelweis Univ, Budapest, <sup>2</sup>Neuro and Infobionics Research Group, Hung. Acad. Sci. – Pázmány Péter Catholic Univ – Semmelweis Univ, Budapest*Presented by Judit Körtvélyes*

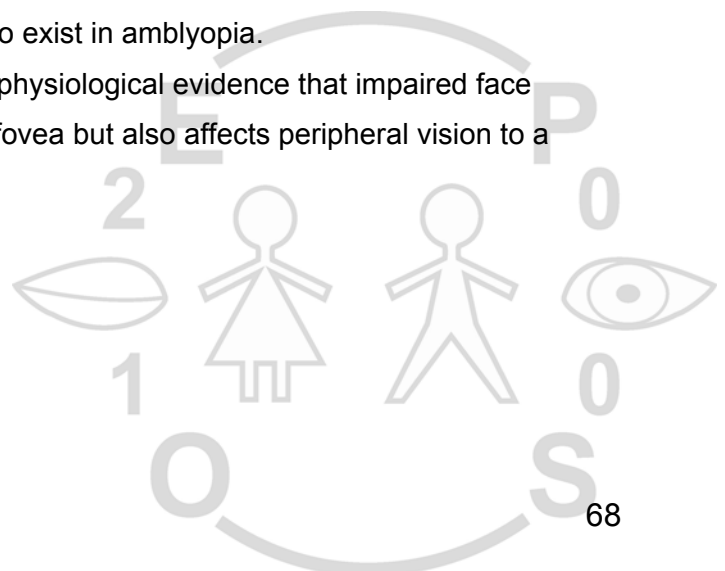
**Introduction:** Beyond developing new screening methods it is a major challenge to understand the neural substrate of this common disease. Based on the results of previous functional magnetic resonance imaging research it has been suggested that higher level object-specific visual processing – in particular processing of faces - is strongly impaired in the foveal, but not in the peripheral vision in amblyopia. However, direct comparison of foveal and peripheral object-related processing deficits in amblyopia requires that the cortical representation of the foveal and peripheral stimuli were matched according to the human visual cortical magnification factor, which was not the case in previous studies.

**Methods:** Fourteen amblyopic patients performed tilt discrimination in a two alternative forced choice task. Presentation sizes were foveal (2deg) and peripheral (4.7deg) with the cortical representation of the stimuli equated. We recorded neuronal activity using a high-density EEG electrode array.

**Results:** We found that the face-related N170 ERP component was significantly reduced and delayed in the amblyopic eye as compared to the fellow eye both in the case of foveal ( $p < 0,001$  and  $p < 0,0001$  for amplitude and latency, respectively) and peripheral ( $p < 0,01$ ;  $p < 0,001$ ) face stimuli, although the difference between the two eyes was more pronounced in foveal vision. Furthermore, we also showed that in peripheral vision the amblyopic effects found on the N170 component might primarily reflect the decreased power and inter-trial synchrony relative to prestimulus baseline in the alpha band, which suggest abnormal evoked alpha band oscillations in the case of the amblyopic eye. Additional analysis of the data as well as our control experiment showed that the amblyopic effects found on the N170 component cannot be explained based on the deficits of low-level feature processing, known to exist in amblyopia.

**Conclusion:** These findings provide the first neurophysiological evidence that impaired face processing in amblyopic vision is not limited to the fovea but also affects peripheral vision to a lesser extent.

**Conflict of Interest: None**





**P10: Visual Fields Defects by Sturge Weber Syndrome**

Giorgio Porro

Utrecht University Hospital

*Presented by Giorgio Porro*

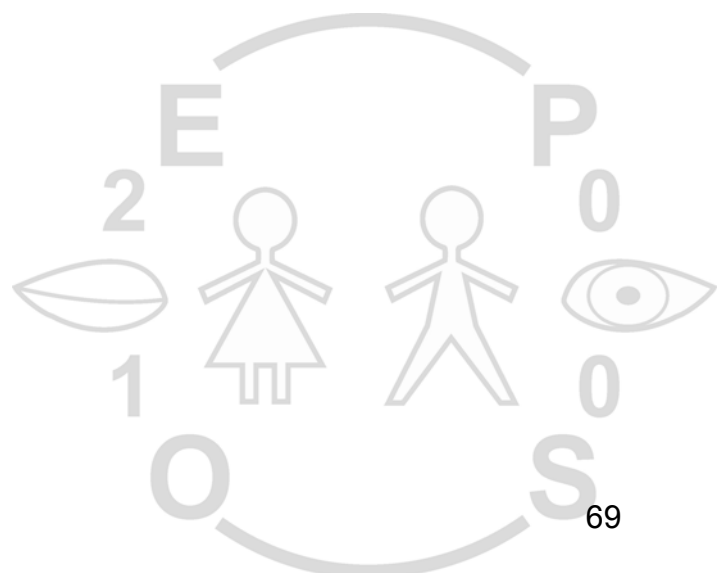
**Aim of the study:** Recording the visual field (VF) in children with Sturge Weber Syndrome (SWS) in relation to ophthalmological-, neurological- and neuro-imaging data. Material and

**Methods:** Above mentioned data, including behavioural VF were retrospectively analyzed.

**Results:** Seven children (4 males, 3 females) were examined. Mean age was 22 mo (range: from 7 mo to 5y 3 mo). Mean follow-up was 3 y 30 mo. Glaucoma was found in 2 children. Two children were treated with Vigabatrine and one underwent epilepsy surgery. Four children had a homonymous hemianopia contralateral to the leptomeningeal vascular malformation and one child had a concentric restriction probably caused by Vigabatrine therapy.

**Conclusion:** In SWS it is of paramount importance to monitor VF, since VF defects are not only caused by glaucoma in the ipsilateral eye or by side effects of Vigabatrine, but also by leptomeningeal vascular malformation, which affect the VF also in the contralateral “sound”eye

**Conflict of Interest: None**





**P11: Visual Function vs. Psychomotor Development of Premature Infants with No Severe Adverse Effects**

Nikolaos Kozeis<sup>1</sup>, Nikolaos Ziakas<sup>2</sup>, Straton Tyradelis<sup>1</sup>, Maria Mavromihali<sup>3</sup>, Vasiliki Soubasi<sup>3</sup>, Vasiliki Drossou<sup>3</sup>

<sup>1</sup>3<sup>rd</sup> University Eye department AUT, Thessaloniki, Greece, <sup>2</sup>1<sup>st</sup> University Eye department AUT, Thessaloniki, Greece, <sup>3</sup>1<sup>st</sup> University Neonatal department AUT, Thessaloniki, Greece

*Presented by Nikolaos Kozeis*

**Introduction:** To follow the visual development along with the psychomotor development of premature infants with no severe adverse effects

**Methods:** 50 premature infants (age range= 26-34 w; mean gestational age= 30.98 w; SD=1.56; range of birth weight= 860-2410 gr; mean birth weight= 1460 gr; SD=299.8) were enrolled in this prospective study. According to the protocol, at 10th and 18th adjusted months, the best corrected binocular visual acuity (BCVA) (by Lea's grating test), refractive status (cycloplegic refraction), contrast sensitivity (by Hiding Heidi test), strabismus (by Krimsky test), funduscopy, brain b-scan and DENVER test, were evaluated. Infants with IVH, PVL, ROP and eye dysplasias were excluded. A group of 50 full term controls was also included. Fisher exact test and Mann-Whitney for statistical analysis were used ( $p < 0.05$ )

**Results:** 46 infants (92%) and 47 infants (94%) appeared with normal BCVA at the 10th and 18th adjusted month of age respectively; 48 infants (96%) and 41 children (82%) appeared with insignificant refractive error at the 10th and 18th adjusted month respectively; 46 infants (92%) appeared with orthophoria at the 10th as well as the 18th adjusted month. All infants were able to recognize 25% contrast sensitivity Heidi faces at the 10th adjusted month, while 11 infants were able to recognize 10% contrast sensitivity Heidi faces at the 18th adjusted month. 39 infants (78%) appeared with normal DENVER score, while 11 infants (22%) appeared with abnormal DENVER score. 27 infants (54%) appeared with normal brain echo, while 23 infants (46%) appeared with enlarged ventricles. Significant statistically relation between prematurity and psychomotor development ( $p = 0.017$ ), but no between prematurity and visual function ( $p > 0.05$ ) was found; the visual acuity develops faster in full terms than in preterms during the first 10 months of life; much more pre terms appeared with significant refractive errors than the full terms during the second year of life; all preterms with pathological echo brain, at the age of 10 months, were myopic ( $p = 0.036$ )

**Conclusion:** Vision development is the same in pre- and full terms especially after the first year of life. Visual function affects but it is not significantly related to psychomotor development in preterms

**Conflict of Interest: None**



## P12: Overlapping of Alström and Bardet-Biedl Syndrome Early Phenotype Confirmed by Systematic High Throughput Ciliopathy Genes Sequencing

Konstantinos Aliferis<sup>1</sup>, Sophie Hellé<sup>2</sup>, Corinne Stoetzel<sup>2</sup>, Jean-Louis Mandel<sup>3</sup>, Hélène Dollfus<sup>1</sup>

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Presented by Konstantinos Aliferis

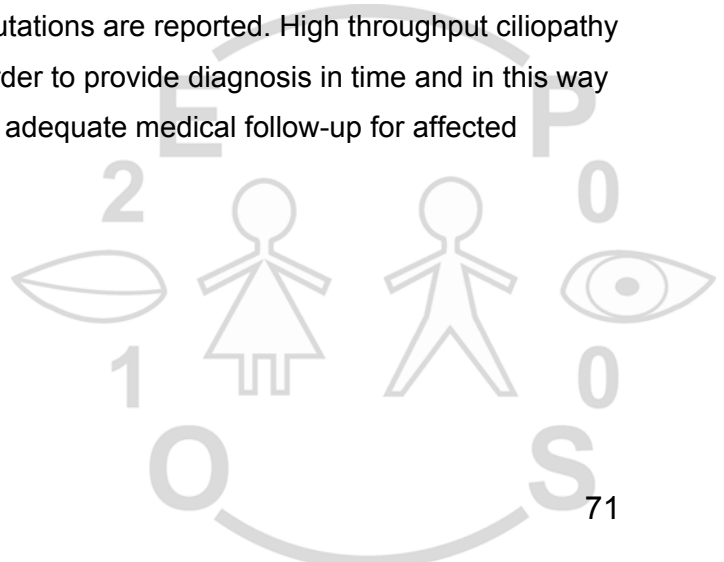
**Introduction:** Infantile retinal degeneration associated with obesity can present a diagnostic challenge in paediatric ophthalmology practice. Clinical overlap between Bardet-Biedl and Alström syndromes has already been pointed out, although they are genetically distinct syndromes. Up to date more than 14 genes are known to be associated with Bardet-Biedl syndrome (BBS1-14) and only one gene has been reported with Alström syndrome (ALMS1).

**Methods:** In collaboration with the French national center of sequencing (CNS, Evry), high throughput sequencing was conducted for 25 ciliopathy genes (*BBS1-12, MGC1203, TTC21b, AHI1, NPHP2-8 (NPHP6=BBS14), MKS1 (BBS13), MKS3, ALMS1*) in samples of 96 patients with no mutation for the 14 known BBS genes. *ALMS1* gene molecular analysis included all coding exons sequencing. *ALMS1* mutations were found in 4 cases.

**Discussion:** Among the four detected mutations, one (c.1131delAGAG) has already been reported whereas three were novel (c.6823insA, c.2293C>T, c.8410delA). All four mutations were predicted to cause the production of a truncated protein and were found in homozygous state in the patients, with positive family segregation. From a clinical point of view, all four patients presented severe retinal degenerative disease with early onset and congenital nystagmus associated with obesity.

**Conclusion:** Our study evaluates the rate of *ALMS1* mutations among suspected Bardet-Biedl patients with negative molecular test. The difficult early differential diagnosis between the two syndromes is pointed out and three new *ALMS1* mutations are reported. High throughput ciliopathy genes sequencing can be extremely important in order to provide diagnosis in time and in this way appropriate genetic counselling for the families and adequate medical follow-up for affected children.

**Conflict of Interest:** None



**P13: Development of a Humanized Mouse-Model for X-Linked Retinitis Pigmentosa Caused by a Point Mutation in the RPGR Gene**Jutta Hosch<sup>1</sup>, Stefan Günther<sup>2</sup>, Thomas Braun<sup>2</sup>, Markus Preising<sup>1</sup>, Birgit Lorenz<sup>1</sup>, Knut Stieger<sup>1</sup><sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Max Planck Institute for Heart and Lung Research, Bad Nauheim*Presented by Jutta Hosch*

**Introduction:** Mutations in the gene encoding the retinitis pigmentosa GTPase regulator (RPGR) are the most frequent causes for X-linked RP in humans, causing 70-80% of all XLRP cases and between 15 and 20% of all RP cases. Most of the responsible mutations can be found in a specific repetitive region of the ORF15, which is therefore called the “mutation hot spot” of RPGR. Point mutations in ORF15 cause a frame shift, leading to a modified C-terminal amino acid chain and thus causing a toxic gain of function of the mutated protein. The purpose of this study is to develop a mouse model that contains a 1 base pair deletion in the mutational “hot spot” region of ORF15 which provokes a change of the amino acids at the C-terminal end similar to the mutated human proteins.

**Methods:** To introduce the pathologic mutation into murine ES cells via homologous recombination, a targeting vector was used, containing mouse-DNA recombined from a BAC (bacterial artificial chromosome), the mutations as well as a Diphtheria-toxin-A cassette (DTA) as a negative selection side and a floxed Neomycin-cassette for positive selection. After the assembly of the final targeting vector, it was electroporated into the ES cells, where homologous recombination took place. PCR, restriction digests and sequencing were performed for analysis.

**Results:** The positive clones were transplanted into mouse blastocytes and then implanted into surrogate mother mice. The resulting chimeric mice as well as their offspring were screened. The chimeric animals were cross-breed with Cre-deleter mice to delete the Neomycin-cassette from the genome to prevent any negative effects. Resulting offspring is currently back-crossed into BL6 background.

**Discussion:** The newly generated mouse model will help to gain further insight into the pathological mechanisms involved in retinal degeneration, the expression pattern of mutated RPGR-ORF15 forms, the influence of point mutations in the ORF15 repetitive region on expression and splicing of the mRNA, and the biochemical reason for the toxicity of such proteins. In addition to the development of a classic viral mediated gene addition therapy, this humanized mouse model will potentially make a substantial contribution to the development of a new therapeutic strategy called targeted gene alteration, which is still in experimental stage but holds great promise to further retinal gene therapy trials.

**Conflict of Interest: None**



## **P14: Immunohistochemical Characterization of AAV Transduced Retinae Following Subretinal Injection in Rats**

Bert Constantin Giers<sup>1</sup>, Alexandra Mendes-Madeira<sup>2</sup>, Birgit Lorenz<sup>1</sup>, Fabienne Rolling<sup>2</sup>, Silke Haverkamp<sup>3</sup>, Knut Stieger<sup>1</sup>

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*Presented by Bert Constantin Giers*

**Introduction:** Ocular gene therapy studies have been developed for a variety of different diseases. In particular, gene therapy trials for RPE65 Mutations have recently started and more than a dozen patients have been enrolled so far. In contrast to murine and canine studies, where vision and objective electrophysiological measurements such as ERG improved dramatically, therapeutic benefit in human patients remained somewhat limited to improved light sensitivity and better ambulation of an obstacle parcours in single cases. Synaptic circuits in the outer (OPL) and inner plexiform layer (IPL) are dynamic and receptive to environmental alterations, as it has been demonstrated in nonhuman primates that changed from dichromatic to trichromatic vision following gene transfer of the missing chromophore. In this study, the synaptic architecture in the OPL is characterized in rat retinae following AAV mediated gene transfer.

**Methods:** Five rats were injected subretinally with AAV2/5.CMV.gfp in both eyes or one eye respectively (n= 7 transduced eyes). Two rats received a unilateral mock injection with fluoresceine (n = 2 eyes). Two rats that did not receive any injection as well as the remaining eye in animals that received only unilateral injection were used as control animals (n=9 eyes). Eyecups were fixed in 4% PFA and the neuroretina was prepared as flat mount to subsequently localize the GFP expressing area in transduced eyes. Trimmed blocs were cryoprotected and frozen for later use in immunohistochemistry. Different primary antibodies were used, including CtBP2, CaBP, DHP, PKCa and GFP. In addition, inflammatory and pro-apoptotic markers were tested and the Tunnel Assay for the detection of apoptotic nuclei was performed.

**Results:** The transduced area in the retina of rats can easily be detected and localized by anti-GFP immunolabeling. Pre- and postsynaptic structures are not altered and absolute numbers of synaptic ribbons do not differ significantly between injected and control retina.

**Conclusion:** The neuronal circuits in the OPL of healthy rat retinae undergoing AAV mediated gene transfer are not altered by the presence of viral particles or the expression of GFP as transgene. This observation likely requires further investigation in the dog model for RPE65 deficiency in order to determine the impact of RPE65 transgene expression on diseased retinae in animals and men.

**Conflict of Interest: None**

**P15: Liquid Implant in Congenital Glaucoma Surgical Treatment.**

Nadiya Bobrova

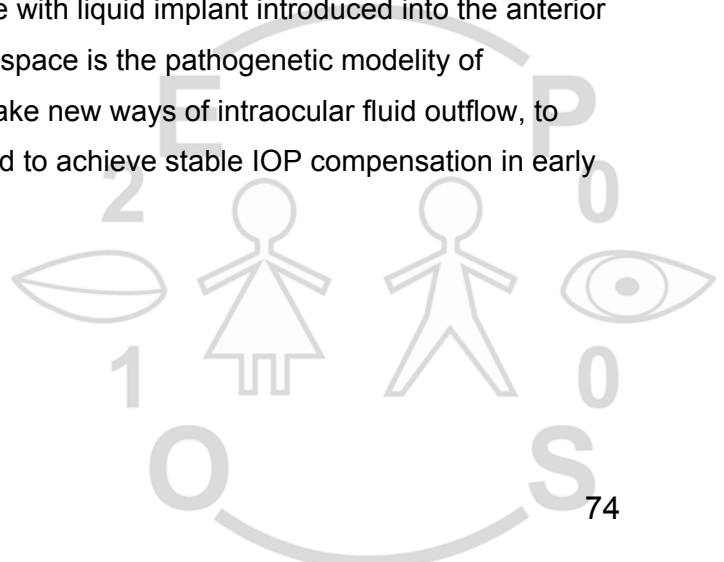
The V.P. Filatov Institute of Eye Diseases and Tissue Therapy

*Presented by Nadiya Bobrova***Introduction:** New congenital glaucoma surgical treatment development

**Methods:** The new technique of congenital glaucoma (CG) surgical treatment was elaborated, that differs from known before flap trabeculotomy by liquid implant introduction. Operation was performed at 29 children (47 eyes) with advanced congenital glaucoma at age 3 mo/o – 6,8 y/o (average 14,9 + 13,1 mo/o): bilateral – 18 patients, unilateral - 11 (3 from them with Sturge-Weber syndrome). Corneal edema and diffuse corneal opacities occurred on 39 eyes, corneal diameter exceeded age norm on all 47 eyes and varied from 12,5 to 16,5mm. IOP was elevated to 23,0 – 45,0 (mean 31,1 + 3,38) mm Hg. US-biometry showed deep anterior chamber 3,87 + 0,32 (range from 2,8 to 4,7) mm and enlarged axial length 24,5 + 1,84 (range 21,2 – 29,9) mm. Gonioscopy detected goniodysgenesis II-III stages with anterior iris attachment and goniosynechias in most cases.

**Results:** Distinctive features of new original technique are superficial and deep scleral flats formation; dispersive viscoelastic injection through lateral preliminary paracentesis into the anterior chamber, mainly in operative zone, which play role of liquid implant, its additional injection between superficial and deep scleral flats and between sclera and Tennon capsule. All operation proceeded without complication. In early postop anterior chamber shallowing was revealed in 6 cases and slight hyphema – in 4 eyes which resolved after medicamentous treatment with anterior chamber depth normalisation. In follow up 3 months IOP compensation observed in all cases (mean 20,4 + 2,49 mm Hg), the anterior chamber depth and eye axial length have decreased up to 3,6 (+ 0,31) mm and 24,1 (+ 1,9) mm accordingly, flat filtration pillow without cicatricial changes was formed in operative zone, a pupil was round without deformations and synechias.

**Conclusion:** Elaborated filtrative surgical technique with liquid implant introduced into the anterior chamber, between scleral flaps and in sub-Tennon space is the pathogenetic modelity of congenital glaucoma treatment which allowed to make new ways of intraocular fluid outflow, to perform operation without severe complications, and to achieve stable IOP compensation in early follow up.

**Conflict of Interest: None**



## P16: Infants Implantation Surgery – New Approaches

Nadiya Bobrova

The V.P. Filatov Institute of Eye Diseases and Tissue Therapy

*Presented by Nadiya Bobrova*

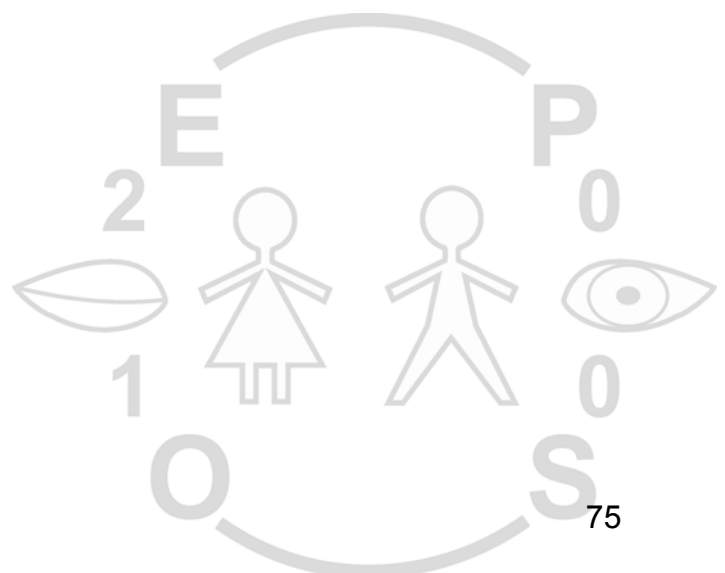
**Introduction:** To elaborate infants implantation surgery new technologies

**Methods:** 100 children (159 eyes) aged 1-24 months (ave  $9,9 \pm 5,3SD$ ) with congenital cataracts (atypical forms - 48,4%, total - 29,6%, zonular - 22,0%) were operated with primary IOL Acrysof in-bag implantation. Following new technologies were developed: Anterior capsule opening in atypical congenital cataract forms (knife, scissors and forceps) - 17 eyes (10,6%); Congenital cataract dry viscoaspiration in cases of posterior lenticonus - 11 eyes (6,9%); Posterior capsule opening and dry vitrectomy after IOL implantation in cases of posterior capsule congenital abnormality - 47 eyes (29,5%); Combination of primary IOL implantation and glasses in very little children with different refractive variation - 29 eyes (18,2%).

**Results:** Anterior capsule opening – helps to perform safety anterior capsule opening with necessary; Congenital cataract dry viscoaspiration helps to completely remove lens material from capsule bag at suspicion on primary posterior capsule disorders; Posterior capsule opening and dry vitrectomy after IOL implantation allow to perform safe posterior capsule opening at its central congenital pathology and prevent vitreous loss; Combination of primary IOL implantation and glasses allows to receive emmetropic refraction of pseudophakic infants eye after operation and in the remote terms of supervision.

**Conclusion:** The exploited methods allows to perform safe cataract phacoaspiration with primary in-the-bag IOL implantation in all cases, receive emmetropic refraction of pseudophakic infants eyes after operation and in the remote terms of the supervision that promote to improve visual function in infants.

**Conflict of Interest:** None





**P17: Secondary Infantile Cataract Associated with Presumed Intrauterine Infection**

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<sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Institute of Anatomy und Cellular Biology, Justus-Liebig-University, Giessen, Germany, <sup>3</sup>Institute of Medical Microbiology and Virology, Justus-Liebig-University, Giessen, Germany

*Presented by Birgit Lorenz*

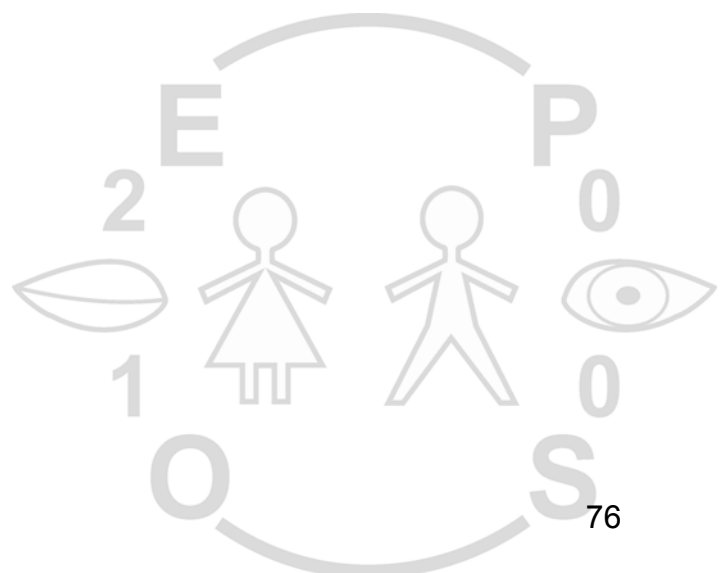
**Purpose:** To report on an under recognised aetiology of secondary cataract with uveitis in infants

**Methods:** Analysis of lens material and vitreous collected at the time of cataract surgery in infants with secondary cataract associated with uveitis with transmission electron microscopy TEM and PCR for DNA/RNA of prokaryotes, bacteria and viruses.

**Results:** Two infants, one premature, one born at term, presented with unilateral respectively bilateral cataracts associated with inflammatory signs at 4 respectively 6 months. Presurgery systemic erythromycine and corticosteroids together with local anti-inflammatory therapy and medical cycloplegia reduced the inflammatory signs within days. In both infants, TEM disclosed spiroplasma species confirmed by prokaryote PCR. Both mothers had experienced an upper respiratory tract infection during the third trimester, misdiagnosed as mycoplasma infection in one, and not further investigated in the second case.

**Conclusions:** Spiroplasma infection may be confounded with mycoplasma infection unless PCR is performed. Spiroplasma infection is a not yet recognised cause of secondary cataract in infancy. One of the two cases has been published previously (Lorenz et al. Graefes Archive Clin Exp Ophthalmol 2002, 240: 348-353).

**Conflict of Interest: None**





## **P18: Cataract Surgery and Postoperative Outcome in a Child with Hallermann-Streiff Syndrome with Dysproportional Microphthalmia**

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*Presented by Werner Schmidt*

**Background and Purpose:** Hallermann-Streiff syndrome (HSS) is characterized by malformation of the face, congenital cataracts, short stature, and dental anomalies. We report on a girl who first presented at age 3 months with typical symptoms of HSS. Because of dense bilateral cataract, lens extraction, discision of the posterior capsule, and anterior vitrectomy were performed O.U.. We report on extreme preoperative ocular biometric measurements and the postoperative outcome.

**Methods:** Biometry including 10 and 20 MHz sonography, ultrasonic biometry, videography of the retina, retinoscopy, orthoptics, optic rehabilitation with contact lenses, wide-angle digital imaging of the retina.

**Results:** Ocular findings at 3 months included O.U. microphthalmia, blue scleras, dense cataract, and pupillary membrane with circular posterior synechia, Due to steep corneal curvature automatic keratometry was not possible. Combining measurement of corneal diameter and high frequency sonography of the anterior segment, an idealized radius of corneal curvature of 5.3 mm was calculated. It corresponded well with the back curvature of the postoperatively fitted contact lenses (+49 dpt/+44 dpt). By means of 20 MHz sonography, the depth of the anterior chamber and the lens thickness were calculated to be 2.54 mm and 3.05 mm respectively (RE) with a central length of the vitreous of only 5.42 mm. The ultrasonic measurement of the axial length (AL) of 11 mm corresponded to the sum of the measurements of the individual compartments. The left eye showed similar results with even smaller dimensions. At 13 months, esotropia LE was associated with bilateral central retinal folds LE >RE likely associated with subretinal proliferation that remained unchanged during the follow-up of 2 years. Part time occlusion was started and well tolerated despite the pronounced central retinal changes. At 3-y, VA was 0.16 RE and 0.1 LE (LEA). Total refraction was 54 dpt RE and 53 dpt LE.

**Conclusions:** Microphthalmia is a common finding in HSS. Our girl had dysproportional microphthalmia with an AL of 11 mm and almost identical lengths of the anterior and posterior segments. Central retinal folds were associated with limited but still measurable visual acuity.

**Conflict of Interest: None**



### **P19: Uveal Effusion Syndrome in Hallermann-Streiff Syndrome**

Marta Morales, Alfonso Vasquez, Mariona Vidal, Alicia Serra

Hospital Sant Joan de Déu. Barcelona

*Presented by Marta Morales*

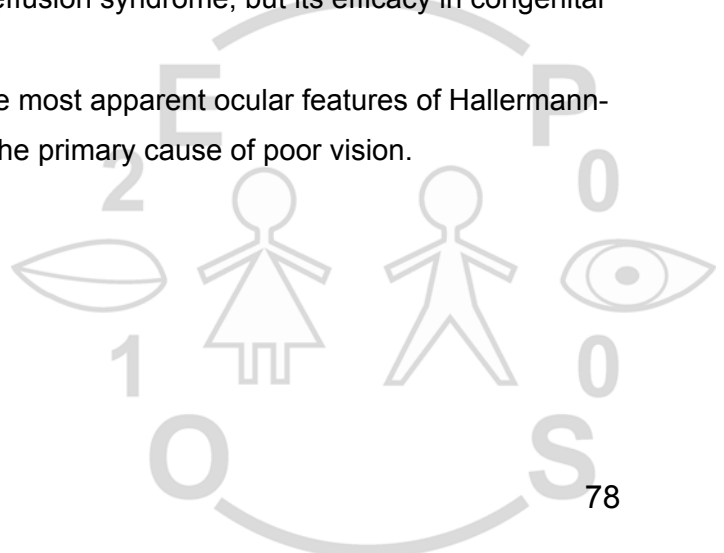
**Introduction:** To report the ophthalmological course of a children with a Hallerman Streiff syndrome.

**Methods:** On 25 august 2008, a 1- month -old girl, with a Hallerman-Streiff syndrome was referred for surgical treatment of congenital bilateral dense cataracts. Ophthalmologic examination revealed bilateral microphthalmia with horizontal corneal diameters of 7 mm and axial length of 15 mm and bilateral visually significant cataracts. Cataract extractions were performed at 8 and 9 weeks of age. Contact lenses of +29D were prescribed. She developed a central, stable and maintained vision. On September 2009 acute angle closed glaucoma was diagnosed in her right eye, and a limbal approach anterior vitrectomy with peripheral iridectomy was performed, which resolved the glaucoma. Intraocular pressure was normal in her left eye but the fundus exam revealed a serous retinal detachment. Oral prednisolone was prescribed. 3 weeks later, serous retinal detachment appears in her right eye. Both retinal detachments resolved 2 months later, except the subretinal fluid in the papillomacular bundle in the left eye, which still persists.

**Comment:** Hallermann-Streiff syndrome was first described in 1958, but uveal effusion syndrome in a nanophthalmic eye was not described until 1974 by Brockhurst. Pathophysiology is described. In these nanophthalmic eyes, the sclera is thicker. In one hand, this induces a congestion of the vortex veins and an extravascular leakage from choroidal vessels. In the other hand there is an impermeability of the transscleral outflow. Both induce fluid accumulation in the choroid and the subsequent dysfunction in the pump mechanism of retinal pigment epithelium which led to subretinal fluid accumulation. The high pressure in the right eye prevented it from the retinal detachment, but the subsequent resolution led to a serous retinal detachment in this eye. Sclerotomy has been advocated for primary uveal effusion syndrome, but its efficacy in congenital uveal effusion in microphthalmic eyes is unknown.

**Conclusion:** Cataracts and microphthalmos are the most apparent ocular features of Hallermann-Streiff syndrome, but retinal abnormalities may be the primary cause of poor vision.

**Conflict of Interest:** None





**P20: Treatment of Severe Vernal Keratoconjunctivitis with 1% Topical Cyclosporine in Childrens.**

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*Presented by Gianfranco Bellizzi*

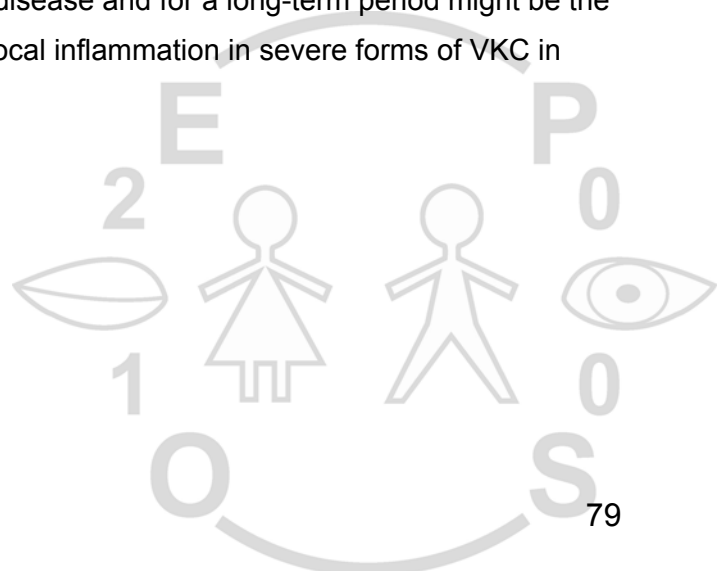
**Introduction:** Vernal keratoconjunctivitis is a seasonal ocular inflammatory disease, with greater prevalence in boys living in warm climate zones; although self-limited, VKC can induce severe corneal complications. VKC is characterized by the presence of cellular infiltrates mainly composed by eosinophils and T-lymphocytes. More recently, authors have proposed the direct activation of dendritic cells, bearing high-affinity receptors for IgE, as an alternative mechanism for initiating an allergic response in patients with or without evidence of specific IgE sensitization; moreover, the role of NK cells in the disease by other authors, based on the evidence of consistent NK infiltrates in the conjunctiva and low NK levels in blood of the patients.

**Methods:** An open trial involving 197 childrens (126M - 71F, age 5-14 y.o.) with severe VKC, who received topical cyclosporine 1% eyedrops for 4 months; ocular subjective symptoms and objective signs were scored in all children at enrolment, 2 weeks and 4 months. Skin prick tests and microscope endothelial cells evaluation were also performed; serum IgE and cyclosporine levels were assessed.

**Results:** The mean score values for severity of subjective symptoms and objective signs were significantly decreased after 2 weeks, and 4 months, compared with those at entry ( $p < 0.001$ ) in all children. Cyclosporine serum levels were not detectable at the end of therapy, no endothelial cells damages were evidenced. Patients who started the therapy at the beginning of the disease and/or received long-term regimen of treatment with cyclosporine had a faster improvement of ocular signs and symptoms, compared to all other patients.

**Conclusion:** Our findings suggest that 1% cyclosporine concentration, in artificial tears solution, employed topically at the beginning of the disease and for a long-term period might be the most effective treatment to control symptoms and local inflammation in severe forms of VKC in childhood.

**Conflict of Interest: None**





**P21: The Management of the Bilateral Palpebral Necrotizing Fasciitis in a Newborn with Agammaglobulinemia Bruton**

Stefanut Anne Claudia<sup>1</sup>, Vladutiu Cristina<sup>1</sup>, Chirica Ana-Maria<sup>1</sup>, Barsan Simona<sup>2</sup>, Georgescu Adrian<sup>2</sup>, Militaru Mihai<sup>3</sup>

<sup>1</sup>Cluj County Emergency Hospital, Ophthalmology Dept., <sup>2</sup>University of Medicine and Pharmacy Cluj-Napoca Plastic Surgery Dept., <sup>3</sup>University of Medicine and Pharmacy Cluj-Napoca Pediatric Dept.

*Presented by Anne-Claudia Stefanut*

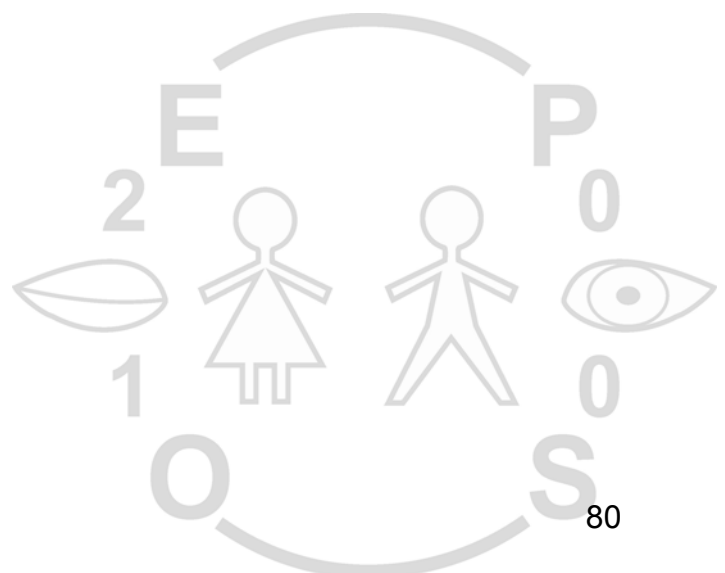
**Introduction:** Necrotizing fasciitis is a severe subcutaneous tissue infection with high mortality rate and sequelae. Agammaglobulinemia Bruton is a B-cell deficiency caused by tyrosin-kinase gene mutations. Both diseases are very rare in neonatal period.

**Methods:** This report describes a case of a 7 weeks-old boy, twin, after artificial insemination, presenting with bilateral fulminant palpebral necrosis, sepsis with *Pseudomonas aeruginosa* and pluriorganic failure.

**Results:** Accurate diagnosis and therapeutical management required interdisciplinary collaboration between ophthalmologist, paediatrician, genetician and plastic surgeon. Intensive medical treatment and gentle local debridement, partially restored the general status and preserve the eyeballs. Surgical palpebral reconstruction was delayed by the recurrent respiratory infections. The suspicion of the genetic immunodeficiency was confirmed later by a genetic analysis.

**Conclusion:** This case raised many issues due to the occurrence of this bilateral disease in full anatomical and functional development of the eyeball and of the adjacent structures in a newborn with severe immunodeficiency. It is important to have a high index of suspicion, since early recognition and multidisciplinary aggressive management offers the best chance for survival and favourable visual outcome.

**Conflict of Interest: None**





**P22: Infantil Palpebral Angioma Treated with Topical Betablockers**

Julia Escudero, Angel Vera, Francisco Jose Barrero

Hospital Regional Carlos Haya. Málaga, Spain.

*Presented by Julia Escudero*

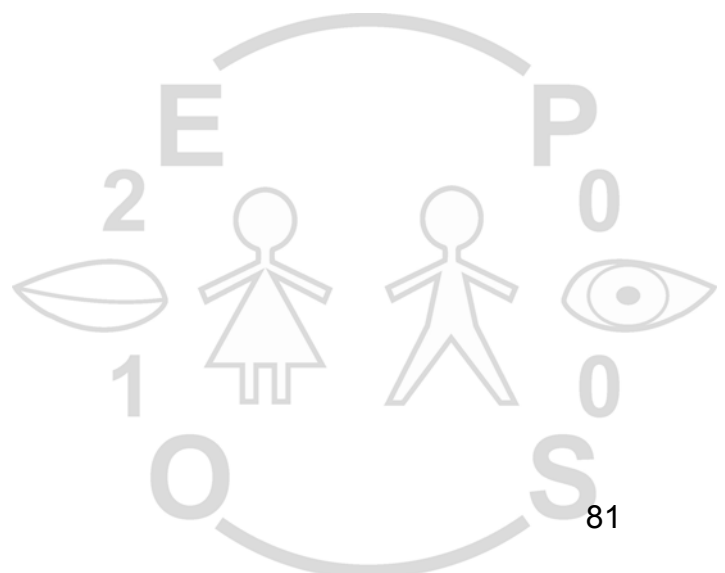
**Introduction:** There are several methods of treatment for infantil angioma. Treating with betablockers is replacing oral corticosteroids as first line of treatment and topical timolol has been recently used with good results.

**Case report:** We report two cases of infantil palpebral angioma treated with betablockers. One of the cases was treated with oral propranolol and the other with topical timolol.

**Results:** In both cases we observed a notable improvement, with no secondary effects.

**Conclusion:** It seems that treating infantil angioma with betablockers is replacing oral corticosteroids as first line of treatment, although there are no studies that clarify which betablocker must be used and the best way of application  
**KEY WORDS:** Infantil Angioma, propranolol, betablockers

**Conflict of Interest: None**



**P23: Unusual Unilateral Orbital Tumor in an Infant**Melanie Jaeger<sup>1</sup>, Ute Zake<sup>2</sup>, Gerhard Alzen<sup>3</sup>, Birgit Lorenz<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Department of Paediatric Hematology and Oncology, Justus-Liebig-University Giessen, Universitaetsklinikum Giessen and Marburg GmbH, Giessen Campus, Germany, <sup>3</sup>Department of Paediatric Radiology, Justus-Liebig-University Giessen, Universitaetsklinikum Giessen and Marburg GmbH, Giessen Campus, Germany

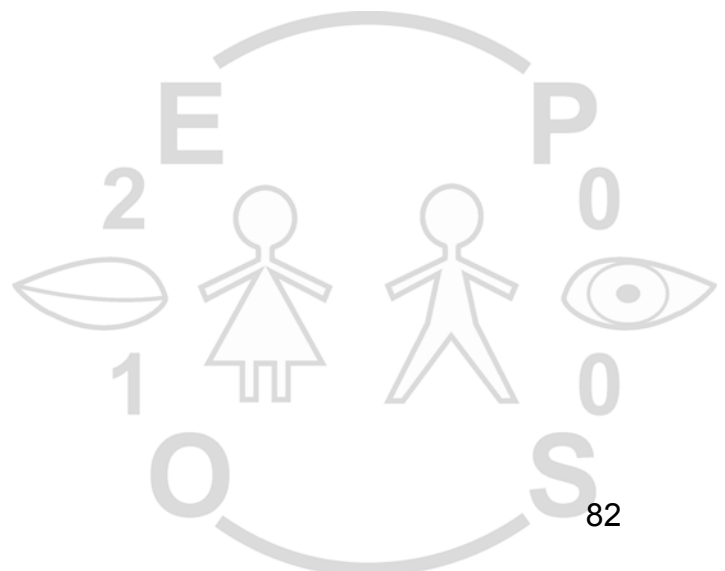
*Presented by Melanie Jäger*

**Introduction:** The differential diagnosis of unilateral tumors in infancy includes benign lesions such as hemangioma and lymphangioma, and malignant tumors such as rhabdomyosarcoma and neuroblastoma.

**Case report:** We describe the case of a 5-month-old girl who presented with a right progressive exophthalmos, ptosis and eyelid-swelling for less than one week. Motility was normal as well as the optic nerve. No other diseases were known. Ultrasonography disclosed a tumor with good perfusion and without involvement of muscles or bone. Cranial MRI showed an extensive tumor reaching into the apex of the orbit with enhancement on Gadolinium. On transconjunctival biopsy very brittle and highly vascularised tumor tissue was excised. The first histologic diagnosis was rhabdomyosarcoma, and one cycle of chemotherapy (I2VA) for rhabdomyosarcoma was started. As the final result of the histology was rhabdoid tumor, the treatment followed according to the European Rhabdoid Registry Study with one course Doxorubicin, two ICE courses followed by high dose chemotherapy (Carboplatin/Thiotepa) with autologous hematopoietic stemcell rescue. Under this therapy, the exophthalmos decreased and the tumor regressed on MRI. For local therapy proton radiation is planned.

**Conclusion:** Rhabdoid tumor of the orbit is a very rare and highly aggressive tumor that showed an incomplete response to chemotherapy and is a challenge for local therapy since the outcome is much dependent of the complete tumor eradication.

**Conflict of Interest: None**





**P24: Rhabdomyosarcoma Masquerade Syndrome: Pitfalls in Diagnosis**

Reshma Thampy<sup>1</sup>, Lucy Clarke<sup>1</sup>, L Irion<sup>2</sup>, Rajitha Ajit<sup>1</sup>, Sajjid Atallah<sup>1</sup>, Brian Leatherbarrow<sup>1</sup>

<sup>1</sup>Dept of Ophthalmology, Manchester Royal Eye Hospital, Manchester UK, <sup>2</sup>National Ocular Pathology Service, Department of Histopathology, Manchester Royal Infirmary, Manchester, UK

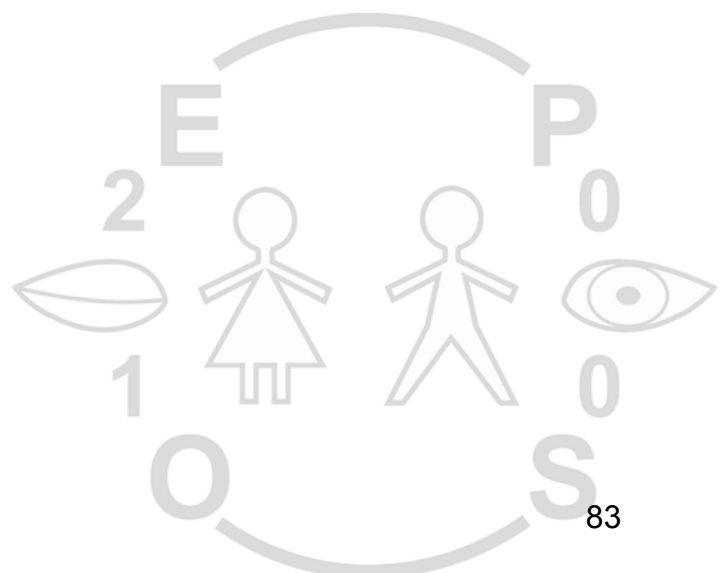
*Presented by Reshma Thampy*

**Introduction:** Rhabdomyosarcoma is the most common primary orbital malignant neoplasm of childhood. It may, however, be overlooked as a cause of orbital pathology. We present a series of five consecutive cases, each of which was initially misdiagnosed. By demonstrating the different clinical presentations and radiological images of these patients, we wish to highlight how rhabdomyosarcoma may mimic other orbital disorders, leading to potentially life-threatening delays in management.

**Methods:** Retrospective, comparative, consecutive, interventional case series with histopathological analyses of patients with orbital rhabdomyosarcoma from a single tertiary referral centre.

**Results:** Each case is presented with details of their clinical presentation including their initial diagnosis, subsequent histological subtype, and clinical progress. Case 1: Conjunctival papilloma Case 2: Eyelid papilloma Case 3: Langerhans cell histiocytosis Case 4: Orbital cellulitis Case 5: Chalazion Conclusion A high index of suspicion should be maintained when examining a paediatric patient with a history of a rapidly progressive orbital, eyelid or conjunctival lesion, with the diagnosis of rhabdomyosarcoma presumed until proven otherwise, in order to optimise the outcomes for this group of patients and to avoid unnecessary delays in diagnosis and appropriate management

**Conflict of Interest: None**





## P25: Screening of Retinopathy of Prematurity Using Sucrose As Analgesia

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*Presented by Nicolaos Ziakas*

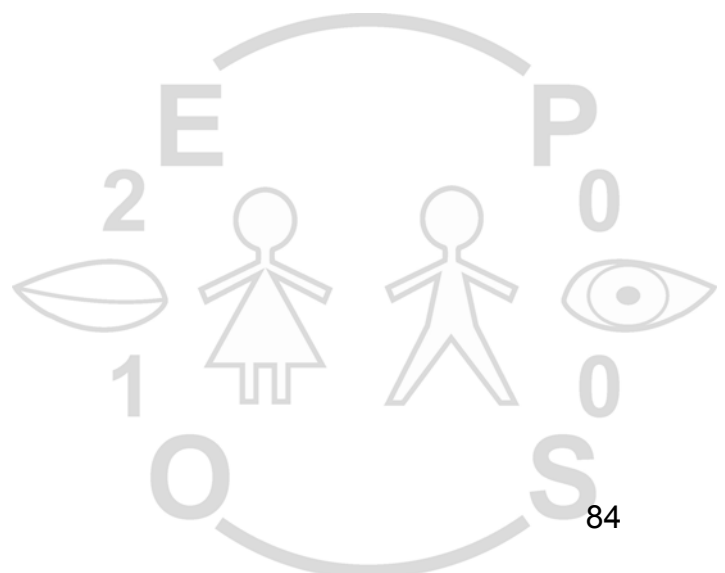
**Introduction:** The retinopathy of prematurity (ROP), is a serious condition that potentially induce blindness. The examination could be painful and some studies suggest the use of oral sucrose solution as analgesic.

**Methods:** Sucrose solution 24% was used according to the local and national protocols. We present the results of a prospective sequential study, that took place at Doncaster Royal Infirmary in two consecutive years. The first year babies were examined without sucrose. Pulse rate, respiratory rate, and oxygen saturation were monitored before, during and after the examination for ROP in both groups.

**Results:** Increased heart rate was more evident in neonates under the age of 36 weeks. There was less increase in respiratory rate in the sucrose group.

**Conclusion:** No significant differences in their responses were found in patients who had oral sucrose as analgesic.

**Conflict of Interest:** None





**P26: Early Neonatal Creatinaemia is an Indicator of the Subsequent Risk to Develop Threshold Retinopathy**

Isabel George<sup>1</sup>, Ingele Casteels<sup>2</sup>, Maissa Rayyan<sup>1</sup>, Djalila Mehkali<sup>1</sup>, Karel Allegaert<sup>1</sup>

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*Presented by Isabel George or Karel Allegaert*

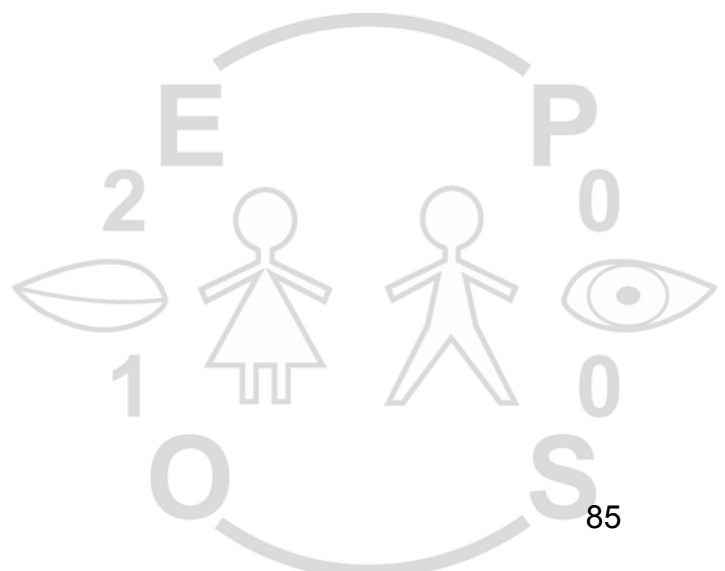
**Introduction:** Indicators available in early life might serve to predict the subsequent relative risk to develop threshold retinopathy (tROP) already associated with a given gestational age or weight. We therefore evaluated if creatinaemia - reflecting disturbed microperfusion – can be used as indicator for subsequent tROP in extreme low birth weight (ELBW, i.e. <1 000 g) neonates.

**Methods:** Retrospective analysis of clinical characteristics (gestational age, birth weight, maximal creatinaemia first week of life, duration of ventilation, supplemental oxygen or until full enteral feeding) of ELBW neonates in one unit between 2000 and 2005. Neonates who developed tROP (n=28) were compared with neonates who did not (n=119).

**Results:** tROP cases were younger (26 vs 27 wk), smaller (745 vs 820 g), remained longer on respiratory support (30 vs 7 days) and oxygen (66 vs 29 days). Duration until full enteral feeding (51 vs 32 days) was also longer. Maximal creatinaemia in the first week was higher in threshold ROP cases (1.3 vs 1.1 mg/dl) (all at least p<.001). In a logistic regression model, creatinaemia remained significant after introduction of birth weight or age.

**Conclusions:** In addition to immaturity (age, weight) peak creatinaemia seems to serve as an early available indicator for subsequent tROP. Disturbed microcirculation, both renal and retinal, might explain this association. This indicator, already available in early neonatal life might be of relevance for secondary prevention trials or selective screening programs.

**Conflict of Interest: None**





**P27: Oxidative Stress - a Biomarker in Retinopathy of Prematurity**

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*Presented by Anne-Claudia Stefanut*

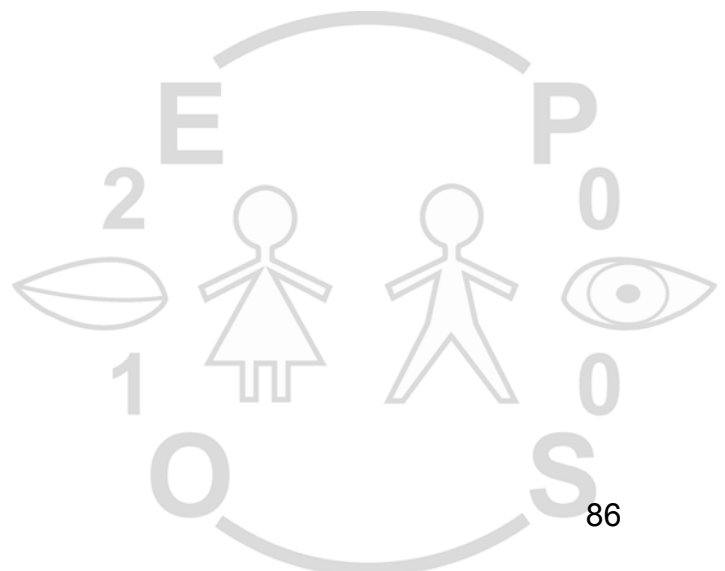
**Introduction:** Retinopathy of prematurity (ROP) is a multifactorial disease. Oxygen toxicity plays an important role in etiopathogeny of this disease. The oxidative compounds and the reduced ability to eliminate these compounds lead to oxidative stress. The purpose of this study was to evaluate the relationship between the oxidative stress parameters levels and the incidence ROP.

**Methods:** A prospective cohort study was designed. Twenty-six premature newborns of less than 32 wks GA and under 1500g BW were included. Reduced GSH and MDA serum levels were determined as a measure of oxidative stress. At 4 weeks of life samples were taken and ophthalmological examinations were performed. The results of samples were compared between infants who developed any degree of retinopathy of prematurity and those without it.

**Results:** The incidence of retinopathy of prematurity was 32,5 % (8/26). The mean values of GSH levels of the samples showed a significant difference ( $p=0,013$ ) between infants who developed retinopathy of prematurity ( $17,042\text{nmol} + 6,104$ ) and those who did not ( $26,616\text{nmol} + 9,536$ ). The mean values of MDA levels of the samples showed a significant difference ( $p=0,00007$ ) between infants who developed retinopathy of prematurity ( $1,294\text{nmol/ml} + 0,460$ ) and those who did not ( $0,561\text{nmol} + 0,181$ ).

**Conclusion:** There is a relationship between serum GSH and MDA levels, as a measure of oxidative stress, and the incidence of retinopathy of prematurity. These parameters could be biomarkers in screening and management of ROP.

**Conflict of Interest:** None





**P28: Oxidative Stress Parameters in a Model of Oxygen Induced Retinopathy**

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*Presented by Anne-Claudia Stefanut*

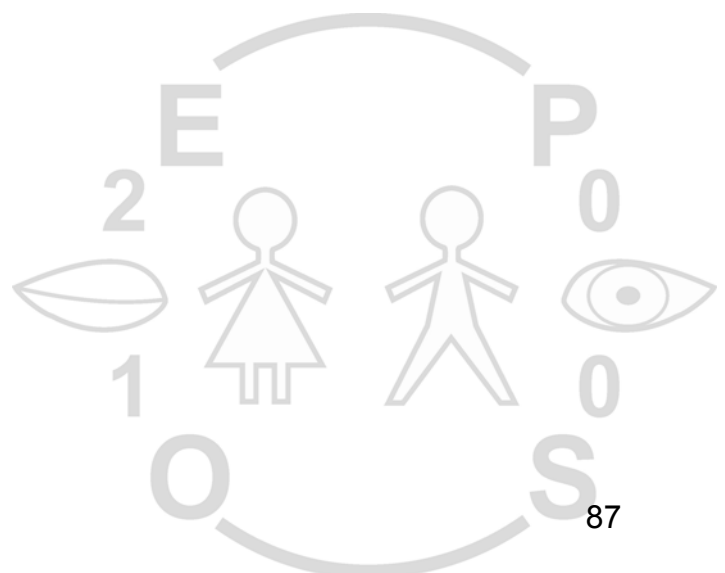
**Introduction:** The immature retina is highly susceptible to reactive oxygen species. The purpose of this study was to evaluate the retinal and serum levels of the oxidative stress parameters in a model of oxygen induced retinopathy.

**Methods:** The study has used two groups of newborn Wistar albino rats: control group(8 pups) was raised in room air (21% O<sub>2</sub>) and oxygen exposed group(7pups) was raised under varied levels of oxygen (80/10%) and then allowed to recover in room air. On postnatal day P21, malondialdehyde (MDA) and reduced glutathione (GSH) levels in serum and eye homogenates (without the lens) were determined. We have also recorded serial retinal histologic sections.

**Results:** All retinal histologic sections from oxygen exposed group 100% (8/8), revealed preretinal neovascularization and severe cytoarchitectural anomalies. The MDA levels was significantly increased in both the serum (1,950nmol/ml + 0,376; 1,502nmol/ml + 0,244; p= 0,014 )and the retina (0,121nmol/mg + 0,016; 0,037nmol/mg + 0,031; p=0,001)in oxygen exposed group and the GSH serum levels was significantly decreased in the serum(10,710nmol/ml + 0,956; 13,497nmol/ml + 1,766; p= 0,004) but not significantly in the retina(2,599nmol/mg + 0,748; 3,141nmol/mg + 1,567)

**Conclusion:** These results suggest the implication of lipid peroxidation processes and antioxidant capacity in the complex pathophysiology of oxygen induced retinopathy.

**Conflict of Interest: None**





## P29: Efficacy of Green Laser Photocoagulation for Aggressive Posterior Retinopathy of Prematurity

Nadiya Bobrova

The V.P. Filatov Institute of Eye Diseases and Tissue Therapy

*Presented by Nadiya Bobrova*

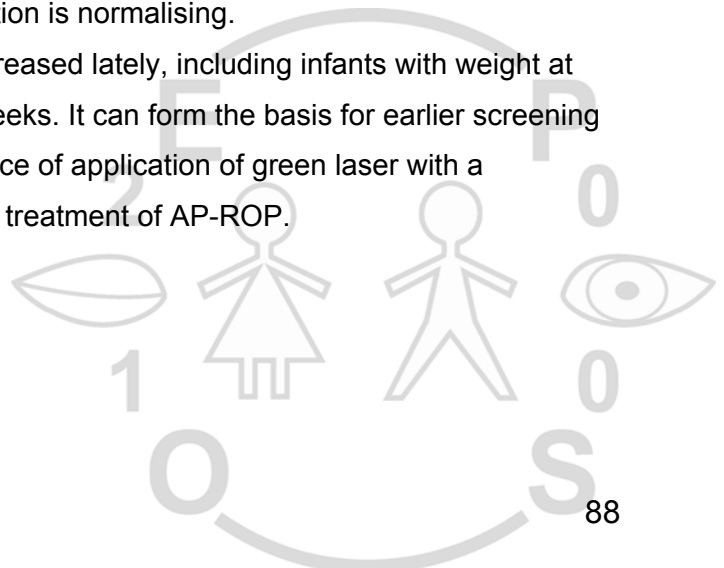
**Introduction:** To estimate the efficacy of laser coagulation for patients with aggressive posterior retinopathy of prematurity (AP-ROP).

**Methods:** During the period from May 2009 to June 2010, 15 premature infants (29 eyes) with AP-ROP were observed. Weight at birth was from 746 to 1680 g, the mean birth weight was 1133,267 + 282,16 g. Gestational age ranged from 24 to 31 weeks, mean gestational age was 26,8 + 2,5 weeks. In all cases the long oxygen therapy (more than 20 days) was carried out. In 4 cases (26.6 %) the newborns have been born after in vitro fertilization. To all infants laser coagulation of avascular retina with near-confluent laser burns has been executed. Average number of shots was 3100 per eye. The laser with wavelength of 532 nm was applied for the treatment. The mean interval between birth and carrying out of laser intervention was 7 + 2,94 (range 4-12) weeks. The mean follow-up time was 4,93 + 4,04 (range 1-12) months. Unfavorable structural result was determined in accordance with the criteria of The Early Treatment for Retinopathy of Prematurity study.

**Results:** The positive result of laser treatment has been reached in 80 % of cases - 23 eyes (at 12 patients). On 20 eyes (10 infants) signs of regression of disease were defined on the second week of observation. Repeat laser coagulation was required for 3 eyes (2 patients), after which regression was also observed. In 20 % of cases - 6 eyes (3 patients) the disease progressed to stage 5. During carrying out of laser intervention on 3 eyes (2 infants) small haemorrhages on border vascular and avascular retina were noticed, which in the course of observation have spontaneously resolved. Hyphema and anisocoria was observed in 4 eyes (2 cases) after laser intervention. In the course of observation the condition is normalising.

**Conclusion:** The development of AP-ROP has increased lately, including infants with weight at birth > 1500 g and gestational age more than 30 weeks. It can form the basis for earlier screening and treatment of this group of infants. Our experience of application of green laser with a wavelength of 532 nm has shown high efficiency of treatment of AP-ROP.

**Conflict of Interest: None**





**P30: Benefit of Paint Diode Laser Coagulation in Treatment of ROP.**

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Paediatric Ophthalmology Department Children University Hospital Bratislava

*Presented by Dana Tomcikova*

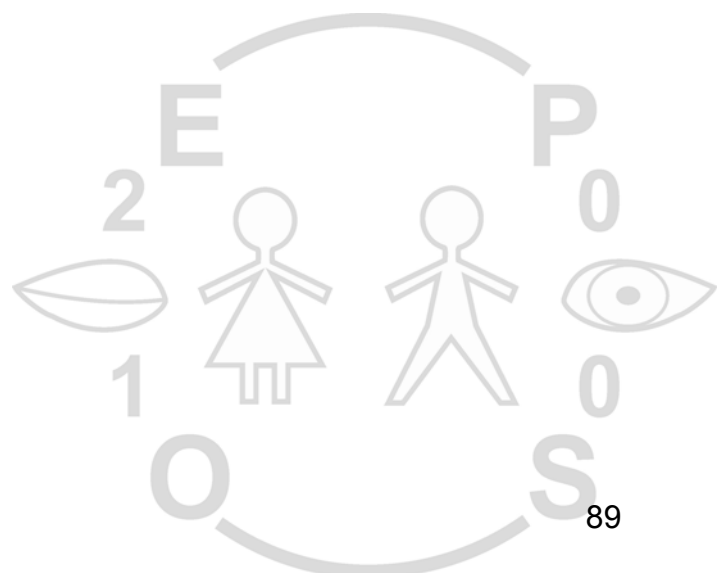
**Introduction:** Authors compare results of ROP treatment with a single spot diodlaserkoagulation (DLK) versus paint- DLK.

**Methods:** A retrospective chart review of patients with threshold retinopathy of prematurity treated between 2001-2010 was conducted. Single spot laser treatment, or paint laser treatment was applied anterior to the ridge extending to the ora serrata. Rate of progression, frequency of retreatment complications and structural outcomes werw evaluated.

**Results:** The single spot DLK was used from January 2001 to May 2008. In this group ( G I) there were 300 patients ( 500 eyes). In the second group ( G II) since June 2008 to June 2010 we have used the paint DLK in 159 patients ( 318 eyes ) . Authors compare need for reoperations to achieve regression of ROP in both groups. In G I was retreatment with DLK necessary in 83 eyes (13,85 %). Additional cryotherapy was used in 43 eyes (7,17 %). A scleral buckling was used in 84 eyes ( 14,02 %). In G II was DLK the retreatment used in 4 eyes ( 1,25 % ), the additional cryotherapy in 1 eye ( 0,3 % ) and the scleral bucling in 9 eyes ( 2,2 %)

**Conclusion:** According to our outcomes we have found out the paint DLK mode is more effective than the single spot DLK.

**Conflict of Interest: None**



**P31: The Challenge of the Relations Between the Parents of the Premature Babies and Paediatric Ophthalmologists.**

Natalya Fomina, Elvira Saidasheva

St.Petersburg's Postgraduate Medical Academy

*Presented by Natalya Fomina*

**Introduction:** The born of the premature baby is a heavy psychology stress of the family. All parents of these babies have the “syndrome of loss” because they waited healthy baby. This syndrome consists of 4 phases: the trouble; the negative; the anger; the adaptation. The ophthalmologist should know this syndrome and the aim of the normal relations between the parents of the premature babies and paediatric ophthalmologists is decreasing the manifestation of this syndrome.

**Purpose:** To improve the relations between the parents of the premature babies and paediatric ophthalmologists. The information about the possible Retinopathy of Prematurity (ROP) should be done at the birth of the premature babies or when baby was admitted to the NICU. If later – the parents can fill the trouble repeat. Usually severe ROP has been diagnosed at the age of 36 – 38 p.a. when some of baby’s systems work better and baby moves out of the reanimation department. The absence of the “RetCam” in the NICU forms the negative relations between the parents and the ophthalmologist. The parents can’t see the changes of the retina and they can’t believe. Usually they want to have the consultation of another ophthalmologist. Without registration changes of the retina by “RetCam” any results of the lasercoagulations will be “unfavorable”. If after surgery will be favorable outcome the parents can think that situation wasn’t so bad but laser coagulations presents. If after surgery will be unfavorable outcome – the reason of this may be mistake of the doctor – think parents. For improving the relations useful to show the photos of the healthy treated babies. These photos (with information of birth weight and gestational age) may be situated near the entrance of the reanimation department. For normal work all personals of NICU have to know the modern information about ROP but answer for the questions of the parents should the ophthalmologist.

**Conclusion:** Don’t wait good relations between the parents of the premature babies and ophthalmologists at once because there are 4 phases of the syndrome. For improving the relations the ophthalmologist have to be attentive and thoughtful in spite of the possible harm of the parents. “RetCam” can be expended the knowledge of the parents about ROP and helps to improve the relations.

**Conflict of Interest: None**



**P32: Retinal Nerve Fiber Layer Thickness in Children Measured by Spectral Domain OCT**

Christina Pieh-Beisse, Gesa Behnke, Daniel Boehringer, Wolf Lagrèze

University Eye Hospital, University of Freiburg, Germany

*Presented by Christina Pieh-Beisse*

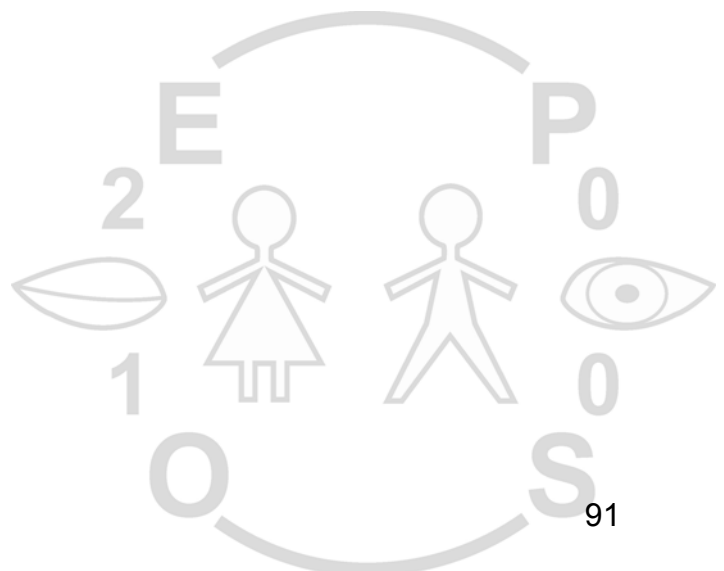
**Introduction:** Our aim was to determine normative values in children for peripapillary retinal nerve fiber layer (RNFL) thickness measured by spectral domain Optical Coherence Tomography (OCT). Further, we studied a possible correlation between the RNFL thickness and the examined age range and evaluated the correlation with clinical variables.

**Methods:** The peripapillary RNFL of 54 healthy, term-born children (23 males, 31 females) aged between four and nine years (mean 6.2yrs) was imaged with a high-resolution spectral domain OCT (Spectralis OCT, Heidelberg Engineering). RNFL thickness was measured around the optic nerve head using 16 automatically averaged, consecutive circular B-scans with 3.4mm diameter. The automatically segmented RNFL thickness was divided into 32 segments (11.25° each). One randomly selected eye per subject entered the study.

**Results:** Mean RNFL thickness in the study population was  $100.7 \pm 7.9 \mu\text{m}$ . Mean inferior RNFL thickness was  $130.1 \pm 34.6 \mu\text{m}$ , superior  $123.6 \pm 34.0 \mu\text{m}$ , nasal  $75.0 \pm 29.5 \mu\text{m}$ , and temporal  $73.8 \pm 21.3 \mu\text{m}$ . These measures are comparable to RNFL thickness reported in adults (Baleanu D, et al. J Glaucoma, online first 2009). There was no significant change of RNFL thickness over the examined age range ( $r = -0.143$ ,  $p = 0.3221$ ). Neither axial length ( $r = -0.348$ ;  $p = 0.1712$ ) nor refractive error ( $r = 0.265$ ;  $p = 0.1735$ ) showed a significant correlation with RNFL thickness.

**Conclusion:** RNFL thickness measured by spectral domain OCT in children is comparable to the results reported in adults. Measurements in children aged  $\geq 4$  do not require an age adjusted normative database but can be compared to normative adult values.

**Conflict of Interest: None**



**P33: Fluorescein Angiography-Guided Management of Retinopathy in Incontinentia****Pigmenti: A Case Series.**

Reshma Thampy, Sonia George, Jane Ashworth, Ian Chris Lloyd, Susmito Biswas

Department of Paediatric Ophthalmology, Manchester Royal Eye Hospital, Manchester, UK

*Presented by Reshma Thampy*

**Introduction:** Debilitating visual loss secondary to tractional retinal detachment is one of the most serious sequelae of Incontinentia Pigmenti (IP). Early signs of preceding ischaemic retinopathy can be clinically imperceptible. We describe our experience in three consecutive cases of IP where fundal fluorescein angiography elicited varying degrees of peripheral retinal ischaemia. This enabled individually tailored management with prompt treatment, where appropriate, to minimize sight threatening complications.

**Methods:** A retrospective, comparative, interventional case series of three paediatric patients with IP from a single quaternary referral centre. The clinical course and angiographic findings of each patient are described

**Results:** Case 1: This patient suffered rapidly progressive retinal ischaemia, which presented as peripheral fundal haemorrhages. Sudden subsequent retinal detachment in one eye, was surgically unsalvageable. In the fellow eye however, fluorescein angiography demonstrated extensive areas of abnormal vascular arborisation, neovascularisation and vessel leakage. Sectoral panretinal photocoagulation to these regions halted progression of tractional detachment, and preserved navigational vision. 2: Both retinæ appeared unremarkable until examination under anaesthesia with fluorescein angiography. This revealed peripheral fundal ischaemia demonstrated by a clearly identifiable non-perfused temporal zone with vascular pruning and peripheral leakage. Immediate sectoral panretinal photocoagulation was performed and to date the disease has remained quiescent. 3: This case also did not exhibit obvious ischaemia clinically. An examination under anaesthesia with fluorescein angiography revealed peripheral retinal ischaemia, however, there was no demonstrable vascular leakage. It was therefore concluded that interventional laser was not immediately warranted and that regular repeat examinations would suffice.

**Conclusion:** IP can result in devastating visual loss in early childhood. These cases illustrate how retinal disease and clinical course exhibit considerable variability and how fundus fluorescein angiography can detect occult disease and guide management. Digital photography and archiving systems are thus invaluable to compare serial examinations, highlight areas of progressive disease and facilitate prompt treatment. In IP, early diagnosis with regular fundoscopy enhanced by fluorescein angiography is imperative.

**Conflict of Interest: None**



### **P34: Macula and Nerve Fiber Layer Thickness in Amblyopia: an Optical Coherence Tomography Study**

Lígia Ribeiro, Eduardo Saraiva, Rosário Varandas, Marisa Oliveira

Centro Hospitalar Vila Nova de Gaia/Espinho EPE

*Presented by Lígia Ribeiro*

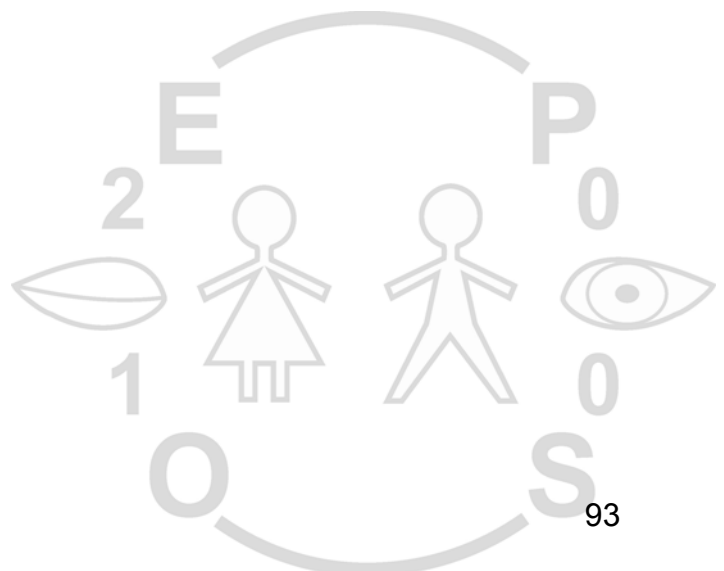
**Purpose:** 1. To determine whether retinal nerve fiber layer (RNFL), macular thickness (MT) and foveal volume (FV) in patients with unilateral amblyopia of different etiologies differ between the amblyopic and sound eye 2. To test the hypothesis that RNFL, MT or FV from eyes with established amblyopia differ from those with potentially reversible amblyopia 3. Review of the literature

**Methods:** Patients were prospectively recruited with the diagnosis of unilateral strabismic amblyopia, anisometric amblyopia, or both. They were divided in two groups: well-established amblyopia and under-treatment amblyopia. A detailed eye examination was conducted, including refractive error, determination of best-corrected visual acuity, ocular motility and alignment evaluation and optical coherence tomography (StratusOCT) through dilated pupils to obtain maps of macular and RNFL thicknesses of sound and amblyopic eyes.

**Results:** Twenty patients (40 eyes) were examined (10 male, 10 female; mean age  $13,45 \pm 12,6$  years; range 5-45). Mean foveal minimum thickness was 8% lower in the sound eyes than in the amblyopic eyes ( $169 \mu\text{m}$  vs  $156 \mu\text{m}$ ,  $p=0.006$ ). Amblyopic eyes also had slightly thicker central macula (1mm diameter region), although these differences were not statistically significant.

**Conclusion:** Histopathologic changes in the lateral geniculate nucleus and visual cortex have been reported but it remains unclear whether the retina is also affected in amblyopia. Studies that have observed the presence of retinal modifications in amblyopic eyes remain inconclusive and controversial. In our study foveal minimum thickness was significantly greater in the amblyopic than in the normal eyes.

**Conflict of Interest: None**





### **P35: Optical Coherence Tomography – Device Independent Automatic Segmentation of Intraretinal Layers**

Alexander Ehnes<sup>1,2</sup>, Erdmuthe Meyer zu Bexten<sup>2</sup>, Birgit Lorenz<sup>1</sup>

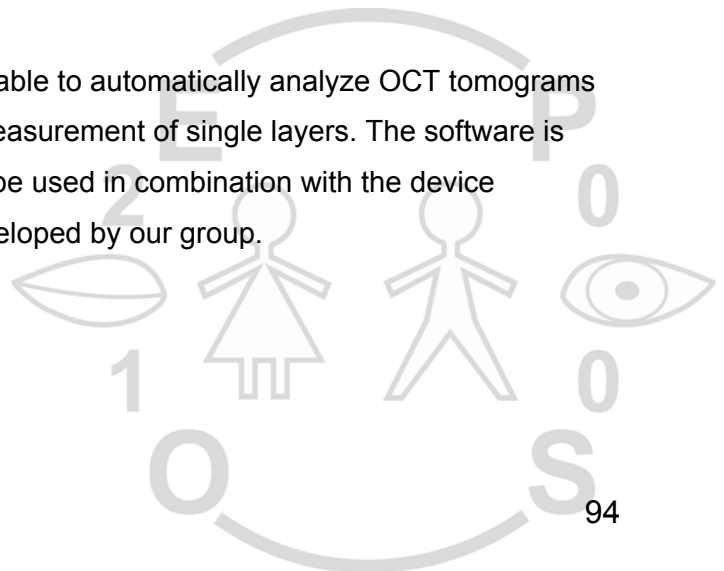
<sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Department of Informatics, University of Applied Sciences Giessen, Giessen, Germany

*Presented by Alexander Ehnes*

**Purpose:** Since the introduction of optical coherence tomography in 1996 it has become the most important non-invasive method for examining the retina. To date, mostly 3rd and 4th generation OCT devices from different manufacturers are in clinical use. However, the vast amount of data that is produced by the high definition live images requires the presence of an experienced examiner. The aim of this project is to develop an OCT-Device independent method for the automated segmentation of intraretinal layers. This application will support the examiner in routine clinical examinations and shorten decision making processes for the ophthalmologist. Method: An algorithm for automatic segmentation of retinal layers based on an active contour approach combined with iterative Region Growing was developed. In addition, preprocessing methods to reduce the noise, inhomogeneous contrast histories and artifacts such as vascular shadows were implemented. The software can use OCT data formats from the Stratus OCT (Zeiss Meditec) and OCT-Spectralis (Heidelberg Engineering). Raw data from healthy volunteers and from patients with early onset severe retinal dystrophy (EOSRD) were used for testing the software and generate basic values. Result: Currently, the analysis software can be used with data formats from time domain (Stratus III, Zeiss Meditec) and spectral domain (Spectralis, Heidelberg Engineering) OCT devices. Using Data from RtVue 100 (Optovue Corp) and Cirrus (Zeiss Meditec) is anticipated. The software is able to segment up to seven layers without user interaction (NFL, GCL, IPL, INL, OPL, ONL and RPE). Manual optimization of additional layer segmentation is possible. Thickness of the layers can be measured automatically. Raw OCT Data from healthy volunteers and from patients affected with EOSRD have been analyzed, and alterations between layer thickness and derivability of single layers can be observed.

**Conclusion:** The newly developed software is capable to automatically analyze OCT tomograms for intraretinal layer segmentation and thickness measurement of single layers. The software is independent of the source of the raw data and will be used in combination with the device independent OCT analysis (DIOCTA) software developed by our group.

**Conflict of Interest: None**





### **P36: Optical Coherence Tomography - Automatic Segmentation of Locally Limited Structures**

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<sup>1</sup>Department of Ophthalmology, Justus Liebig University, Giessen, Germany, <sup>2</sup>Department of Informatics, University of Applied Sciences Giessen

*Presented by Matthäus Pilch*

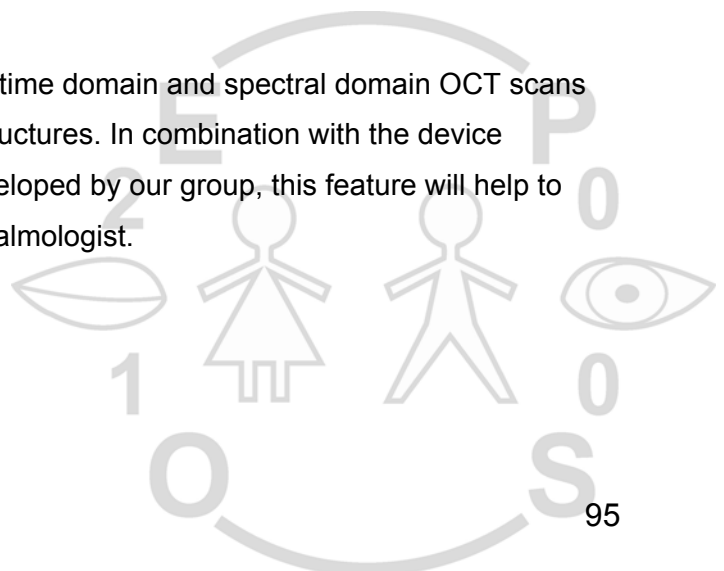
**Purpose:** With the development of spectral domain OCT technology, live images of the retina with high resolution have started to reveal pathological changes with high precision. However, manual detailed analysis of small and locally limited structures is time-consuming and requires a highly experienced examiner. This project aims at developing software solutions for the automated segmentation and subsequent identification of structures that occur in the normal retina (i.e. blood vessels) and during the process of various retinal disorders, such as hemorrhages, exudates, deposits, or other lipid inclusions.

**Methods:** Initially, two different methods for automatic segmentation of locally limited structures were combined: the statistical shape model segmentation, and the shape representation via point distribution model. In order to identify small irregularities within the located structure, a deformable contour model segmentation process has then been included. Volumetric analysis can be performed. Raw data performed with the Spectralis OCT (Heidelberg Engineering) from healthy humans and from subjects affected with early onset severe retinal dystrophies EOSRD due to mutations in different genes have been analyzed.

**Results:** The software can be used with OCT raw data from currently available OCT devices, e.g. the Spectralis OCT (Heidelberg Engineering), the Stratus III OCT (Zeiss Meditec), the RTVue100 (Optovue Inc.), and the Cirrus (Zeiss Meditec). The software readily identifies different locally limited structures, both, within normal and diseased retina. Blood vessels of different sizes were analyzed using raw data from OCT scans of normal subjects. In addition, pathological changes (exudates, drusen, other deposits) have been located and the size of the structures was calculated.

**Conclusion:** Using the newly developed software, time domain and spectral domain OCT scans can be automatically screened for locally limited structures. In combination with the device independent OCT analysis (DIOCTA) software developed by our group, this feature will help to facilitate the decision making process for the ophthalmologist.

**Conflict of Interest: None**



**P37: Introducing an Application for the Analysis, Segmentation and Interpretation of OCT Investigations Applicable for Multiple Devices**

Steffen Zahn<sup>1,2</sup>, Matthäus Pilch<sup>1</sup>, Alexander Ehnes<sup>1</sup>, Elisabeth Strohmayer<sup>1</sup>, Birgit Lorenz<sup>1</sup>, Erdmutha Meyer zu Bexten<sup>2</sup>

<sup>1</sup>Department of Ophthalmology, Justus-Liebig-University Giessen, Universitaetsklinikum Giessen and Marburg GmbH, Giessen Campus, Germany, <sup>2</sup>Department of Informatics, University of Applied Sciences Giessen, Giessen, Germany

*Presented by Steffen Zahn*

**Purpose:** The correct clinical diagnosis of retinal degenerative disorders normally requires the presence of experienced ophthalmogenetic specialists. The vast amount of data generated with current technologies requires the use of sophisticated applications for subsequent analysis. The aim of this project is to develop uniformly useable methods for the computer-aided analysis of OCT Data from patients with retinal dystrophies or cone dysfunctions, (i.e. early onset severe retinal dystrophy (EOSRD) or achromatopsia), which have been acquired with different OCT-Devices, in order to support and shorten the decision making process for the ophthalmologist.

**Methods:** Raw Data from currently available OCT devices can be imported into the new analysis software. The application provides methods for image enhancement, image manipulation and image analysis. Image quality can be enhanced by different B-Scan visualization modes and different techniques of noise reduction. In addition, automatic layer segmentation has been developed. Raw data from patients with EOSRD due to mutations in the RPE65 gene and with achromatopsia due to mutations in the CNGB3 gene have been analyzed with the software.

**Results:** Currently, raw data from the Spectralis OCT (Heidelberg Engineering) and the Stratus III OCT (Zeiss Meditec) are supported. Inclusion of RTVue100 (Optovue Inc.) and Cirrus (Zeiss Meditec) is anticipated. The fully-automated layer segmentation approach is able to segment up to 7 layers (NFL, GCL, IPL, INL, OPL, ONL and RPE) without the need of user interference.

Segmented layers and manually added shapes, such as area or distance measurements, can be manipulated in different ways. A uniform representation of the retina can be achieved by RPE alignment. When analyzing raw OCT data and additional clinical information from EOSRD and achromatopsia patients, typical pathological features can be visualized and highlighted.

**Discussion:** The evaluation process of OCT investigations in patients with EOSRD and achromatopsia benefits already from the provided methods. Further improvements to the software, such as automatic analysis and interpretation of disease specific properties, automated segmentation and analysis of locally limited structures, and the creation of a knowledge base that covers information about disease specific properties, structures and phenotype genotype correlation will greatly improve quantification of retinal diseases and treatment effects in the near future.

**Conflict of Interest: None**



**P38: Best Disease - A Case Report**

Elzbieta Markowska, Jadwiga Bakunowska, Danuta Sielicka, Monika Ozieblo-Kupczyk, Alina Bakunowicz-Lazarczyk

Department of Pediatric Ophthalmology

*Presented by Elzbieta Markowska*

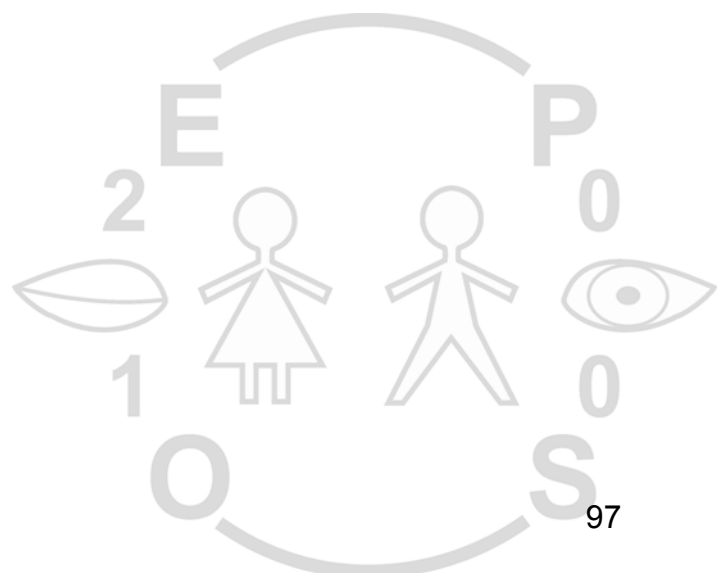
**Introduction:** Best disease (BS, Best vitelliform macular dystrophy) (OMIM#153700) belongs to a rare, slowly progressive, genetically determined disorders. It is transmitted in an autosomal dominant manner with variable expressivity and incomplete penetrance. *BEST1* gene, responsible for BS appearance, was localized on the long arm of chromosome 11q13. This gene codes the bestrophin-1 protein, which is found in retinal pigment epithelium (RPE). Mutation in *BEST1* gene leads towards lipofuscin accumulation in RPE, between RPE and Bruch's membrane, in external parts of photoreceptors and Muller's cells.

**Purpose:** The main aim of our work was to present a 9-years-old girl with ocular signs of Best syndrome

**Methods:** 9-years-old patient was admitted to our Ophthalmology Clinic on March 2009. First visual problems with unclear vision appeared at 2 years. The Best disease diagnoses was set up due to characteristic clinical findings in electroretinography (ERG), optical coherence tomography (OCT) and electrooculogram (EOG).

**Results:** Typical for BS signs on the eye fundus confirmed by EOG, OCT and ERG allowed us to set up a diagnosis of Best syndrome. The early diagnosis in our patient gives her an opportunity for further constant visits in Ophthalmology Clinic. This allows us to monitor changes in the eye fundus and then apply treatment then complications such as choroidal neovascularization will be visible. We postulated for the whole family ophthalmologic examination, as BS is usually of familial occurrence and may represent variable expressivity and incomplete penetrance.

**Conflict of Interest: None**



**P39: Severe Optic Neuropathy Mimicking Leber Hereditary Optic Atrophy in a Friedreich's Ataxia Patient.**

Yaumara Perdomo- Trujillo<sup>1</sup>, Fouzia Studer-Rezaiguia<sup>1</sup>, Valérie Pelletier<sup>1</sup>, Jonathan Letsch<sup>1</sup>, Jérôme De Seze<sup>2</sup>, Hélène Dollfus<sup>1</sup>

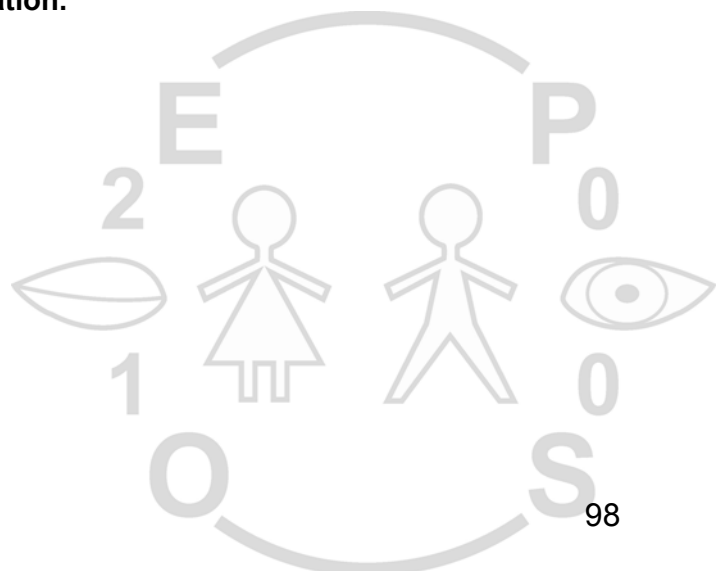
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*Presented by Yaumara Perdomo-Trujillo*

Friedreich's ataxia (FRDA) is an autosomal recessive neurodegenerative disease and the most frequent hereditary ataxia. The gene encodes a small mitochondrial inner membrane protein called frataxin. The most common mutation is a GAA triplet-repeat expansion within the first intron of the frataxin gene (*FXN*). Most patients carry two expanded GAA alleles. A small but significant number of FRDA patients are compound heterozygous with (GAA) n mutations on one allele and a micromutation on the other. A slowly progressive degenerative process involving both the optic nerve and the optic radiations has been reported but exceptionally cause severe visual loss. Optic neuropathy appears to occur late in the course of FRDA and occurs more frequently in patients with larger GAA repeats and also more frequently in compound heterozygotes than homozygotes. Severe optic neuropathy is common in mitochondrial disorders but however has very rarely been reported in FRDA. We describe here a 38-year-old man with sudden and markedly diminished visual acuity (light perception) associated with temporal optic disks pallor, bilaterally. He was diagnosed as having Friedreich's ataxia at 12 years old. Molecular genetics analysis showed two expanded GAA *FXN* alleles (2.4 Kb and 4.1 Kb respectively). The mitochondrial point mutations for Leber hereditary optic neuropathy were not identified. Electrophysiology test revealed normal retinal function. A subacute/ acute visual failure mimicking Leber hereditary optic neuropathy can be observed in FRDA in contrast to the classical very slow progression of FRDA optic neuropathy.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





**P40: Retinal Neovascularisation, Blepharokeratoconjunctivitis and Progressive Entropion in a Young Girl with Autosomal Dominant Severe Dyskeratosis Congenita (DC) Due to *TINF2* Gen**

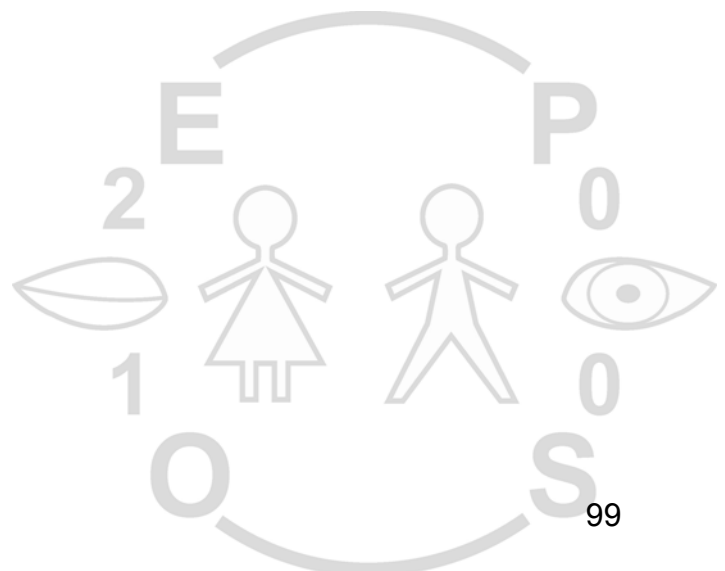
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*Presented by Francoise Roulez*

Dyskeratosis ongenita (DC) represents a genetically heterogeneous disease caused by defects in telomere biology. X-linked recessive forms are due to mutation in Dyskerin (*DKC1*) gene; autosomal recessive forms are due to mutations in *NOP10* or *NHP2* genes and autosomal dominant forms to mutations in *TERC*, *TERT* or *TINF2* genes. Affected patients developed leukoplakia, nail dystrophy, abnormal skin pigmentation together with bone marrow failure syndrome. We report the natural history of a 14 year-old girl, second child from healthy parents. She developed aplastic anemia at the age of 3 that required bone marrow transplantation. Her intelligence and development were normal. She developed few premature grey hair with small area of alopecia, oral leukoplakia, ungueal dystrophy and very large dyskeratosis on the perineal region . Ophthalmological features were right peripheral retinal ischemia and bilateral blepharitis with slight punctate keratitis and progressive entropion. Graft versus host disease or DC were suspected diagnosis. Combined DHPLC and direct DNA sequencing of *TINF2* gene identified a c.847C>G (p.Pro283Ala) mutation. This mutation occurred de novo since both parents were not detected as carrier. *TINF2* gene (TERC-1-interacting nuclear factor 2; 6 exons on 14q11.2) is a member of genes encoding for the shelterin complex proteins. It was recently identified responsible for severe DC and account for up to 11 % in patients with DC. We review the ophthalmological features of this multisystemic disorder.

**Conflict of Interest: None**





**P41: Nanophthalmos: A Family Case Report**

Eduardo Marinho Saraiva, Lígia Ribeiro, Rosário Varandas, Luís Agrelos

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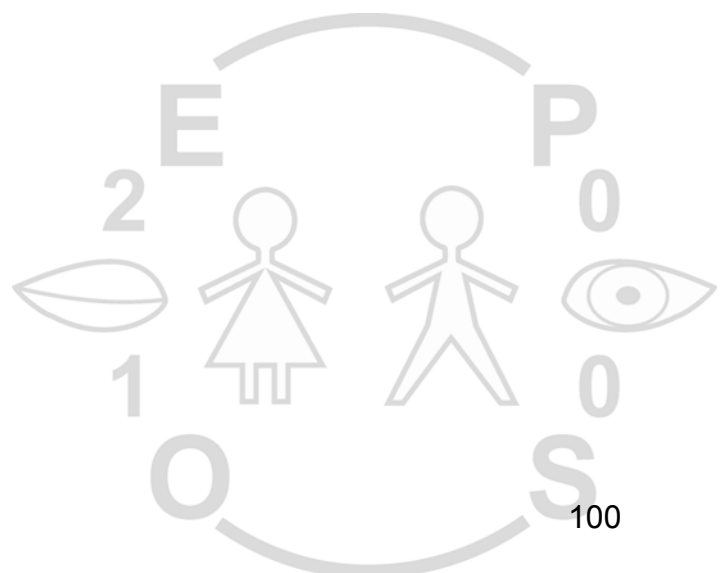
*Presented by Eduardo Marinho Saraiva*

**Introduction:** Nanophthalmos is a rare condition characterized by eyes with short axial lengths and high hyperopia, but without gross structural defects. Clinical Case: The authors present three cases of Nanophthalmos in a Portuguese family, consisting of a mother and two daughters (5 and 2 years old). All three have ocular axial lengths shorter than 16 mm, and hyperopia ranging from 9 to 16 diopters. The anterior segment was analysed with Orbscan and Optical Coherence Tomography (OCT): the mean keratometric value was 51.05 diopters, the mean pachimetry value was 564 µm and the anterior chamber depth and volume were normal. There was an increase in the corneal resistance measured by the Ocular Response Analyzer. Two of the family members had chorioretinal folds and a yellow macular pigmentation, evaluated by fundoscopy, retinography and OCT. The authors are currently waiting for the results of the genetic analysis. The older patient wanted refractive surgery, so the authors evaluated the different options available.

**Conclusion:** Nanophthalmos can result in significant visual impairment. If the diagnosis is late, it can lead to amblyopia. Nanophthalmic patients have a high risk of developing angle-closure glaucoma in the third to sixth decades of life. Refractive surgery in nanophthalmos remains challenging, because of the increased surgical risk and because of the lens power needed to correct the high hyperopia.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





**P42: Increased C/D Ratio in Noonan Syndrome**

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*Presented by Nikolaos Kozeis*

**Introduction:** A 14-year old child with characteristic findings of Noonan syndrome including short stature, mental handicap, and Kryptorchismus is presented.

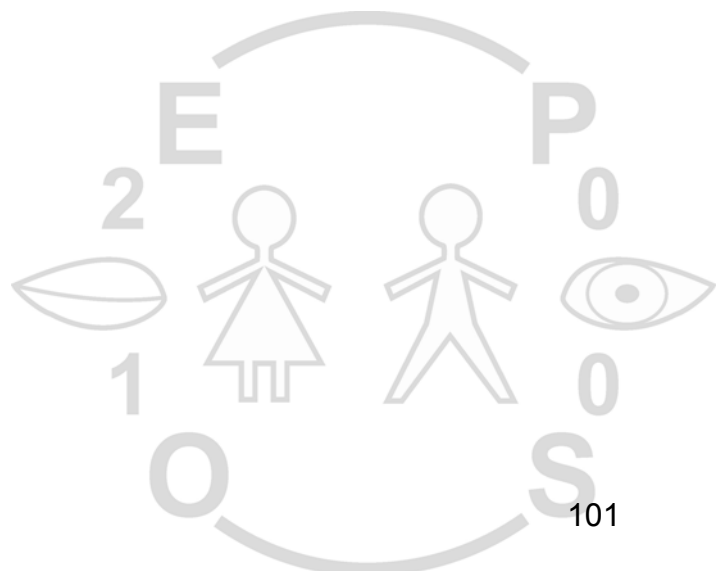
**Methods:** We did fundus photo and HRT.

**Results:** The ophthalmologic investigation resulted a increased C/D ratio with normal intraocular pressure (IOP).

**Conclusion:** To our knowledge this is the second published case of Noonan syndrome with increased C/D ratio and normal IOP.

**Conflict of Interest: None**

**Author does not allow recording of the presentation.**





**P43: Non-syndromic bilateral and unilateral optic nerve aplasia associated with microdeletion of 10q23.33q23.33: first familial case and potential role of *CYP26A1* and *CYP26C1* genes**

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*Presented by Françoise Meire*

Optic nerve aplasia (ONA, MIM #165550) is a very rare unilateral or bilateral condition that includes absence of the optic nerve, of the retinal ganglion cells and the retinal vessels with blindness of the affected eye. Bilateral cases often occur in association with brain malformations. Nevertheless unilateral and bilateral ONA has been reported in otherwise healthy patients. Both pathogenesis and genetic basis of ONA remain until now unknown. So far, there is no documented familial ONA. We report an autosomal dominant ONA with three-year-old dizygotic healthy blind twins (from non consanguineous parents) with normal appearing eyes and bilateral ONA and their father with left microphthalmos. The diagnostic value of neuro-imaging in uncovering ONA in microphthalmic patients is demonstrated. Ultrasonography with Doppler and MRI in the twins showed almost normal structure of both eyes but absence of optic nerves, central retinal vessels, chiasm and tracts. Ultrasonography in the father showed a normal right eye but left heterogeneous microphthalmos that prevented approach of the optic nerve. MRI demonstrated left ONA with chiasm and optic tracts asymmetry (L>R)). Anatomy of the brain and of the pituitary gland was normal in the 3 patients. Optic radiations on DTI (deterministic tractography) were present but decreased in size symmetrically in both children and asymmetrically (L>R) in the father. Mutation screening in three developmental genes *PAX6*, *OTX2* and *SOX2* revealed no pathogenic mutations. Genomewide copy number screening revealed a 249-363 kb microdeletion of chromosome 10q23.33q23.33 in all affected individuals and in unaffected grandmother, containing three genes *EXOC6*, *CYP26A1* and *CYP26C1*. The latter two encode retinoic acid degrading enzymes. Our findings implicate the *CYP26A1* and *CYP26C1* genes as potential novel candidate genes for non-syndromic ONA. Moreover, it is attractive to postulate that dysregulated RA metabolism caused by *CYP26A1* and *CYP26C1* haplo-insufficiency influenced by as yet uncovered modifiers and environmental factors might underlie the ONA phenotype presented here.



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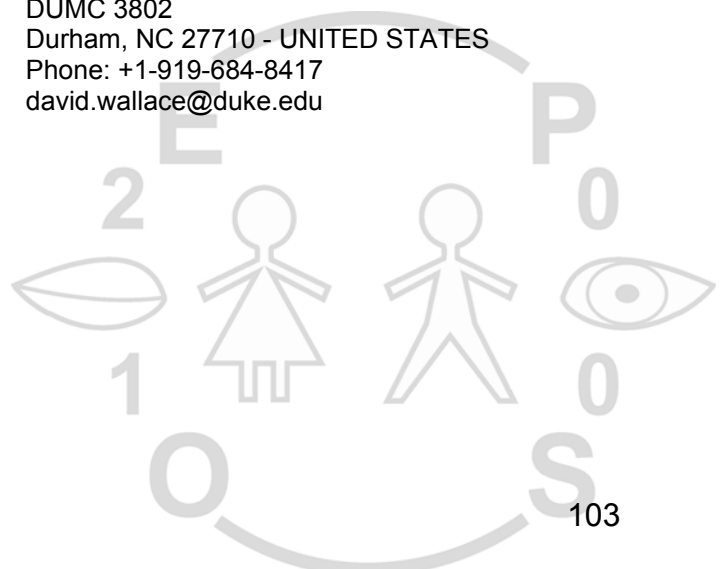
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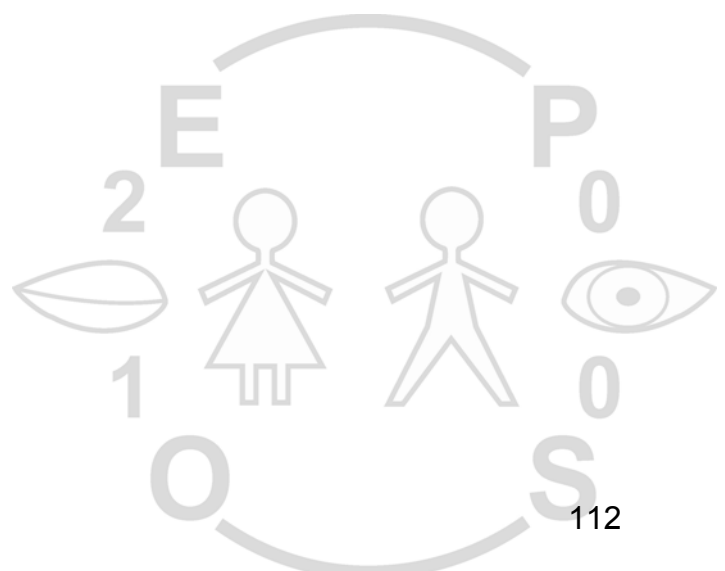
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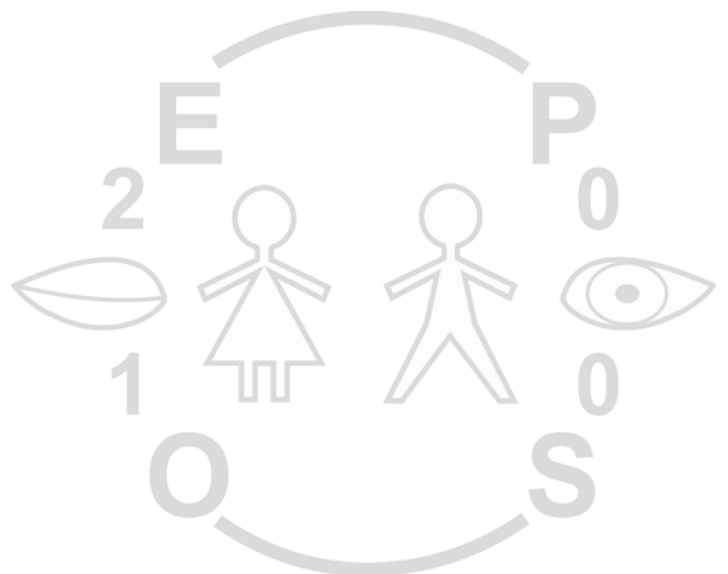
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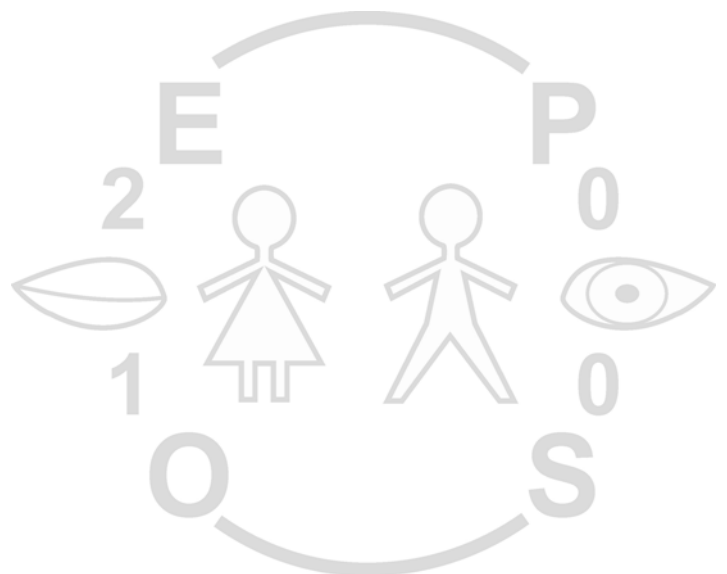
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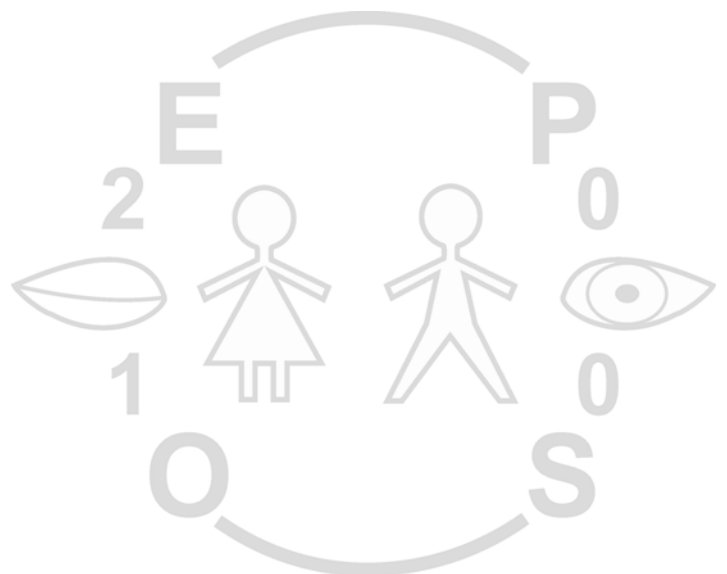
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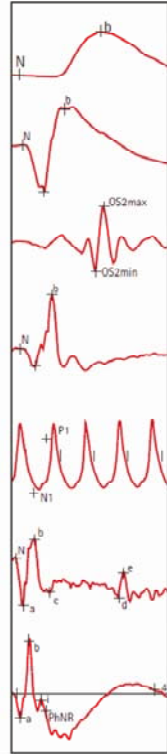
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*Functional Benefits of Sensory and Motor Evaluation Before Strabismus Surgery*, Katherine J. Fray, C.O.

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The John Pratt-Johnson Annual Lecture

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