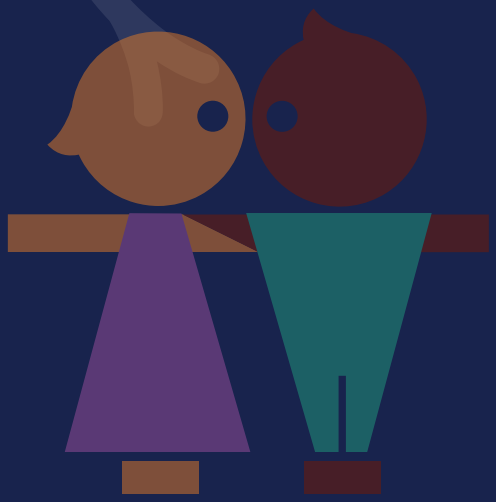
ABSTRACTBOOK



EPOS

9-11 October 2025 Leiden, the Netherlands

LUMC Boerhaave CME

Team- & Network in Paediatric Ophthalmology

Contents

Scientific programme EPOS 2025

Friday, 10 October 2025

08:00 – 08:30 **REGISTRATION**

08:30 – 08:45 **OPENING CEREMONY**
Prof. Henk-Jan Guchelaar, vice-chair, board of directors, LUMC
Dr. Darius Hildebrand, FRCS, FRCOphth, president EPOS
Prof. Nicoline Schalij-Delfos, organising committee

TEAM- AND NETWORK WITH THE GENETICIST (METABOLIC, GENETIC, SYNDROMES, RETINA)

Co-Chairs: C. Boon, A.Daruich

08:45 – 09:00 **KEYNOTE LECTURE**
Towards treatment of retinitis pigmentosa: developments and challenges
Camiel Boon, ophthalmologist, Leiden/Amsterdam, the Netherlands

09:00 – 09:15 **KEYNOTE LECTURE**
Clinical endpoints and patient-reported outcomes for IRD clinical trials
Joao Pedro Marques, ophthalmic geneticist and retina Consultant, Coimbra, Portugal

09:15 – 09:25 ***O-101 - Phenotypic Spectrum of Congenital Stationary Night Blindness: Associations with Foveal Hypoplasia***
Vanessa Rodwell, Leicester, United Kingdom

09:25 – 09:30 ***RF-102 - Bornholm Eye Disease: A Retinopathy in the Spectrum of Dysfunctional Opsin OPN1LW/OPN1MW Genes***
Dzenita Smailhodzic, Zeist, The Netherlands

09:30 – 09:35 ***RF-103 - The spectrum of retinal and skeletal phenotypes associated with variants in CFAP410***
Maria van Genderen, Zeist, The Netherlands

09:35 – 09:40 ***RF-104 - 12-month outcomes after voretigene neparvovec gene therapy in paediatric patients with RPE65-mediated inherited retinal dystrophy***
Alejandra Daruich, Paris, France

09:40 – 09:45 ***RF-105 - Beyond visual acuity: Evaluating functional vision in CRB1-Retinopathies using novel child-friendly tests***
Ana Catalina Rodriguez Martinez, London, United Kingdom

09:45 – 09:50 ***RF-106 - How Competent Are AI Models in Genetic Counseling for Inherited Retinal Disease in the Paediatric Patient? Evaluating Their Capacity to Inform Families About Gene Therapy Options***
Emine Tınkır Kayıtmazbatır, Konya, Turkey

09:50 – 10:10 **KEYNOTE LECTURE**
How effective use of a multidisciplinary team can achieve precise diagnosis in children with congenital cataract and anterior segment dysgenesis. A case-based presentation
Chris Lloyd, paediatric ophthalmologist & Sophie Marlowe, genetic counsellor, London, United Kingdom

10:10 – 10:15 ***RF-107 - The phenotypic and genotypic features of ADAMTSL4-related ocular disease***
Katie Williams, London, United Kingdom

10:15 – 10:25 ***O-108 - Genetic background of childhood glaucoma in the Amsterdam UMC***
Sarah Janssen, Amsterdam, The Netherlands

10:25 – 10:30 ***RF-109 - Ophthalmological manifestations in Aicardi-Goutières syndrome***
Priscille de Laage de Meux , Paris, France

10:30 – 11:15 **COFFEE BREAK, POSTER VIEWING & EXHIBITION**

TEAM- AND NETWORK FOR CHILDREN WITH CRANIOFACIAL OR ORBITAL DISEASES

Co-Chairs: S. van der Meeren, I. Casteels

11:15 – 11:35 **KEYNOTE LECTURE**
Joint ophthalmological care for children with congenital craniofacial anomalies
Irene Matthijsen, plastic surgeon & Sjoukje Loudon, paediatric ophthalmologist, Rotterdam, the Netherlands

11:35 – 11:45 ***O-110 - Optic neuropathy in patients with craniofacial fibrous dysplasia***
Stijn van der Meeren, Leiden, The Netherlands

11:45 – 11:50 ***RF-111 - Teamwork in the diagnosis and treatment of osteopetrosis***
Maria Szwajkowska, Olsztyn, Poland

11:50 – 12:10 **KEYNOTE LECTURE**
Joint ophthalmological care for children with orbital vascular malformations
Elyse Verboom, hemato-oncologist & Angela Arends-Tjiam, paediatric ophthalmologist, Rotterdam, the Netherlands

12:10 – 12:20 ***O-112 - Multidisciplinary Management of Orbital Langerhans Cell Histiocytosis: Insights from a Pediatric Case Series***
Ozlem Ural Fatihoglu, İzmir, Turkey

12:20 – 12:25 ***RF-113 - A periocular and intra-orbital congenital melanocytic nevus with a PRKCA-PLXNA1 fusion gene: a case report***
Ingele Casteels, Leuven, Belgium

12:25 – 12:30 ***RF-114 - Idiopathic Orbital Inflammatory Pseudotumor with Ocular Involvement: A Multidisciplinary Diagnostic Challenge***
Sofia, Lunardon, Milan, Italy

12:30 – 13:45 **LUNCH, POSTER VIEWING & EXHIBITION**

GOLDEN JUBILEE SESSION: LECTURES OF LIFETIME ACHIEVEMENT AWARD RECIPIENTS

Co-chairs: D. Hildebrand, N. Schalij-Delfos

13:45 – 14:00 ***Paediatric Ophthalmology in Europe: Past, present and future***
Darius Hildebrand, Cantonal Hospital St.Gallen, St.Gallen, Switzerland

14:00 – 14:30 ***Can we cure (childhood) glaucoma? A lifetime of lessons from patients and the laboratory***
Peng Khaw, Moorfields Eye Hospital London, London, United Kingdom

14:30 – 15:00 ***Do we really know what we are talking about? Naming does not always clarify***
Creig Hoyt, University of California San Francisco, San Francisco, United States

15:00 – 15:45 **COFFEE BREAK, MODERATED POSTER SESSION & EXHIBITION**

TEAM- AND NETWORK WITH NEUROLOGY (PAEDIATRIC BRAIN TUMORS)
Co-chairs: C. Cassiman, S. Loudon

15:45 – 16:05 **KEYNOTE LECTURE**
Optic pathway glioma in childhood: a national multidisciplinary approach for treatment and clinical research
Lisethe Meijer, paediatric neuro-oncologist, Utrecht, the Netherlands & Carlien Bennebroek, paediatric ophthalmologist, Amsterdam, the Netherlands On behalf of Shared care network of paediatric-oncology, the Netherlands.

16:05 – 16:10 ***RF-115 - Teamwork in diagnosing and monitoring neurofibromatosis type 1 - when does an ophthalmologist raise a red flag?***
Maria Szwajkowska, Olsztyn, Poland

16:10 – 16:15 ***RF-116 - Observation Series of 5 Cases of Intracerebral Lesions Causing Visual Impairment: Neuro-Ophthalmology and Minimal Invasive Neurosurgery (MIN)***
Susanna Maria Antal, Feldkirch, Austria

16:15 – 16:20 ***RF-117 - Suspected papilloedema referrals from the optometric community practice: diagnostic accuracy of OCT RNFL-thickness and B-scan ONSD measurements in a large paediatric cohort***
Anne Cees, Houtman, Glasgow, United Kingdom

16:20 – 16:30 ***O-118 - Ophthalmological findings in children with a primary brain tumor: the visual function up to two years after diagnosis***
Inge Corver, Utrecht, The Netherlands

16:30 – 16:40 ***O-119 - Retinoblastoma Look-Alikes: A Diagnostic Challenge in Pediatric Ocular Oncology***
Abdullah M. Khan, Riyadh, Saudi Arabia

TEAM- AND NETWORK FOR ALLERGIC AND INFLAMMATORY DISORDERS (ALLERGOLOGY, IMMUNOLOGY AND RHEUMATOLOGY)
Co-chairs E. Maka, A. Dahlmann-Noor

16:40 – 16:55 **KEYNOTE LECTURE**
Dual consultation in Ophthalmology and Allergy: field of expertise, practical tricks and tips
Jean-Luc Fauquert, ocular allergist and paediatrician, Clermont-Ferrand, France

16:55 – 17:05 ***O-120 - Management and outcomes of Paediatric Blepharokeratoconjunctivitis in a Tertiary care centre***
Anne-Marie Hinds, London, United Kingdom

17:05 – 17:15 ***O-121 - Retinal phenotypes in cerebral malaria may provide insight into underlying immunopathology***
Kyle Wilson, Liverpool, United Kingdom

17:15 – 17:25 **KEYNOTE LECTURE**
Spiroplasma ixodetis as the causative agent of unilateral uveitic congenital cataract
Manca Tekavcic Pompe, paediatric ophthalmologist, Ljubljana, Slovenia

17:25 – 17:30 ***RF-122 - Three Neonates With Congenital Cataract And Uveitis In 2 Years Caused By Spiroplasma Infection***
Ebba Ghyczy, Amsterdam, The Netherlands

17:30 – 17:35 ***RF-123 - A paediatric case of ocular toxocara and the value of metagenomics in undifferentiated uveitis***
Ameeta Kumar, London, United Kingdom

17:35 – 18:00 **CONCLUDING REMARKS / END OF FIRST DAY SCIENTIFIC PROGRAMME**

Saturday, 11 October 2025

08.00 - 08.30 **REGISTRATION**

TEAM- AND NETWORK WITH NEONATOLOGY & PANEL DISCUSSION 'THE BEST WAY TO TREAT AND FOLLOW UP ON ROP'
Co-Chairs: A. Mataftsi, A. Arends-Tjiam

08:15 – 08:30 **KEYNOTE LECTURE**
Applications of AI for ROP
Peter Campbell, ophthalmologist, retina surgeon, Portland, United States

08:30 – 08:35 ***RF-124 - Effect of image enhancement on agreement in the evaluation of plus disease features***
Pascal Dureau, Paris, France

08:35 – 08:40 ***RF-125 - Optimizing mydriasis for retinopathy of prematurity screening***
Aikaterini K. Seliniotaki Aristotle, Thessaloniki, Greece

08:40 – 08:45 ***RF-126 - Evaluating the Implementation and Impact of ROP Programs in Albania***
Eglantina Sinamati, Tirane, Albania

08:45 – 08:55 ***O-127 - The value of a national ROP register - experiences from the last two decades***
Gerd Holmström, Uppsala, Sweden

08:55 – 09:10 **KEYNOTE LECTURE**
Controversies and Concerns after ROP Treatment
Michael Blair, ophthalmologist, retina surgeon, Chicago Illinois, United States

09:10 – 09:20 ***O-128 - Anti-VEGF For Retinopathy of Prematurity in the Netherlands: National Collaboration and Shared Decision-Making***
L.A. Derks, Rotterdam, The Netherlands

09:20 – 09:25 ***RF-129 - The Red vs the Green Diode Laser for Treating ROP***
Mantsha Ahmed, Ivano-Frankivsk, Ukraine

09:25 – 09:40 **KEYNOTE LECTURE**
Treatment of stage 4 - 5 ROP
Şengül Özdek, ophthalmologist, Retina surgeon, Ankara, Turkey

09:40 – 09:45 ***RF-130 - A very unusual form of ROP***
Alicia Serra, Barcelona, Spain

09:45 – 09:50 ***RF-131 - Evaluation of timed dexamethasone eye drops to prevent proliferative retinopathy of prematurity: a study protocol for a randomized intervention, multi-centre, double-blinded trial (DROPROP)***
Pia Lundgren, Gothenburg, Sweden

09:50 – 09:55 ***RF-132 - The importance of teams in TEARDROPS 2: tear proteomics in infants at risk of Retinopathy of Prematurity (ROP) – a prospective observational study***
Megan Quinn, Glasgow, United Kingdom

09:55 – 10:20 ***Panel discussion: 'The best way to treat ROP'***
Michael Blair, ophthalmologist, retina surgeon, Chicago Illinois, United States
Peter Campbell, ophthalmologist, retina surgeon, Portland, United States
Stefan de Geus, opthhalmologist, retina surgeon, Nijmegen, the Netherlands
Asimina Mataftsi, paediatric ophthalmologist, Greece
Gerd Holmström, paediatric ophthalmologist Uppsala, Sweden
Şengül Özdek, ophthalmologist, retina surgeon, Ankara, Turkey

10:20 – 11:05 **COFFEE BREAK, MODERATED POSTER SESSION & EXHIBITION**

TEAM- AND NETWORK IN REHABILITATION FOR THE VISUALLY IMPAIRED

Co-Chairs: A. Van Sorge, M. van Genderen

11:05 – 11:25 **KEYNOTE LECTURE**
Strengthening bridges with ophthalmologic care by interprofessional counselling at the interface of health care and education in social pediatric centres in Germany
Verena Kerkmann, rehabilitation educationist, Bochum and Dortmund, Germany, & Thomas Becher, pediatric neurologist, Düsseldorf, Germany

11:25 – 11:45 **KEYNOTE LECTURE**
Differential Diagnostic considerations in Cerebral Visual Impairment - using new assessment tools.
Marinke Hokken, Neuropsychologist, specialized in Cerebral Visual Impairment, & Marlou Kooiker, researcher and pediatric (neuro)psychologist, Rotterdam, the Netherlands

11:45 – 11:55 ***O-133 - My CVI" psycho-education for children with cerebral visual impairment (CVI)***
Florine Pilon, Amersfoort, The Netherlands

11:55 – 12:10 **KEYNOTE LECTURE**
Why are they doing this to me? – the impact of visual impairment and medical procedures on the daily life of children and their families
Sandra van der Meijden, healthcare psychologist, specialized in rehabilitation, child & youth, Amsterdam, the Netherlands

12:10 – 12:20 ***O-134 - Prospective Open Label Randomized Controlled Clinical Study Evaluating the Efficacy of Perceptual Learning among Patients with Congenital Nystagmus***
Sigal Zmujack Yehiam, Beer Yaakov, Israel

12:20 – 12:30 ***O-135 - Evaluating Pediatric Eye-Related Quality of Life: Insights from the PedEyeQ Assessment at a Pediatric Ophthalmology and Strabismus Clinic***
Tal Koren, Haifa, Israel

12:30 – 13:00 **ASSEMBLY OF EPOS MEMBERS**

13:00 – 14:00 **LUNCH, POSTER VIEWING & EXHIBITION**

PAEDIATRIC OPHTHALMOLOGY NEVER WORKS EYE-LONE (TEAMWORK WITH OTHER SUBSPECIALISTS IN OPHTHALMOLOGY - SURGERY)

Co-Chairs: C. Gehrt-Kahlert, M. Robert

14:00 – 14:10 ***O-136 - Comparison of the efficacy of Defocus Incorporated Multiple Segments spectacle lenses and 0.01%atropine in slowing the progression of myopia in European children–a one year randomised clinical trial***
Emilia Wnękowicz-Augustyn, Zabrze, Poland

14:10 – 14:15 **RF-137 - Myopia-X-1: phase 2 randomised controlled trial of blue-light blind spot stimulation to reduce myopia progression in children: 6-month results**
Alicia Fothergill, London, United Kingdom

14:15– 14:20 **RF-138 - Perforating scleral vessels in pediatric myopia: prevalence, characteristics and association with disease progression**
Pascal Dureau, Paris, France

14:20 – 14:30 ***O-139 - Incidence of postoperative retinal detachment and bacterial endophthalmitis in the Swedish National Pediatric Cataract Register (PECARE) and associated risk factors***
Ulrika Kjellström, Lund , Sweden

14:30 – 14:35 ***RF-140 - Early Onset Cataracts Related to NHS gene***
Marta Morales, Barcelona, Spain

14:35 – 14:40 ***RF-141 - The use of the iris claw intra-ocular lens in the rare case of absence of capsular support in children; The Dutch network; historical overview, organization, teamwork, outcome and future perspectives.***
Marije Sminia, Haarlem, The Netherlands

14:40 – 14:50 ***O-142 - Refractive Error Profile in Infantile Nystagmus***
Yash Mehta, Leicester, United Kingdom

14:50 – 15:00 ***O-143 - Transition programmes in the UK and Europe - an international survey of practice***
Ameeta Kumar, London, United Kingdom

15:00 – 15:05 ***RF-144 - Ubi liquidus, ibi evacua: A novel approach in the treatment of recurring iris cysts***
Tom Collignon, Leuven, Belgium

15:05 – 15:10 ***RF-145 - Conjunctival Amelanotic Melanoma in a pediatric patient: A case report and management***
Veronica Zendejas-Sánchez, Leon, Mexico

15:10 – 15:45 **COFFEE BREAK, POSTER VIEWING & EXHIBITION**

TEAM- AND NETWORK FOR DIAGNOSTICS (FROM IMAGING TO DIAGNOSIS)

Co-chairs: M. Tekavcic Pompe, M. Thomas

15:45 – 16:00 **KEYNOTE LECTURE**
Paediatric OCT: novel approaches to improve diagnosis and prognosis
Mervyn Thomas, ophthalmologist, Leicester, United Kingdom

16:00 – 16:15 **KEYNOTE LECTURE**
Teamwork makes the (AS-OCT uveitis detection) dream work: bringing data scientists and clinicians together
Ameenat Lola Solebo, paediatric ophthalmologist, London, United Kingdom

16:15 – 16:20 ***RF-146 - The importance of MRI in the diagnosis of various eye diseases***
Zeljko Maras, Podgorica, Montenegro

16:20 – 16:25 ***RF-147 - Evaluation of the Virtual Reality Eye Tracker for Neuro-Ophthalmic Assessment: Feasibility, Reproducibility, and Reliability***
Irem Karaer, Leicester, United Kingdom

16:25 - 16:30 ***RF-148 - Torpedo maculopathy (TM): multimodal analysis of a rare congenital maculopathy in a paediatric case series and review of the literature***
José Do Vale, Lausanne, Switzerland

16:30 – 16:35 ***RF-149 - Feasibility and Clinical Utility of Hand-held Optical Coherence Tomography in Children with Retinoblastoma***
Zhanhan, Tu, Leicester, United Kingdom

16:35 – 16:45 ***O-150 - Functional and Structural Outcomes in Paediatric Myelin Oligodendrocyte Glycoprotein Antibody-Associated Disease (MOGAD): A Prospective Study***
Flavia Gericke, Zurich, Switzerland

16:45 – 17:00 **AWARDS AND CLOSING CEREMONY**

ABSTRACTS

Team- and network with the geneticist (metabolic, genetics, syndrome, retina)

O-101

Phenotypic Spectrum of Congenital Stationary Night Blindness: Associations with Foveal Hypoplasia

Vanessa Rodwell[1], Rebecca McLean[1], Michael Hisaund[1], Jinu Han[2], Smirnov[3], Richard Hertle[4], James Self Self[5], Fred Chen[6], Gail Maconachie[1], Zhanhan Tu[1], Zeitz[1], Irene Gottlob[1], Mervyn Thomas[1]

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[2] Institute of Vision Research, Yonsei University College of Medicine, Seoul, South Korea
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[5] Ophthalmology, University of Southampton, Southampton, United Kingdom
[6] Ophthalmology, Lions Eye Institute, Nedlands, Australia

Introduction:
Congenital Stationary Night Blindness (CSNB) comprises non-progressive retinal disorders causing impaired night vision due to disrupted retinal signalling. While genetic causes are known, associated visual features remain undercharacterised. The role of myopia control in CSNB is unclear. This study examines associations between CSNB subtypes and foveal hypoplasia (FH), nystagmus, and myopia, and explores links between myopia severity, genotype, and age.

Methods:
We analyzed 199 patients with confirmed CSNB from five centres (n=89) and the literature (n=110). Diagnoses were based on ERG and/or genetic testing, classifying cases as incomplete CSNB (iCSNB; CACNA1F) or complete CSNB (cCSNB; NYX and others). Clinical features were recorded, and FH graded using standardised OCT criteria. Chi-squared tests and a generalised linear model (GLM) were used for analysis.

Results:
FH was present in 34.6% of cases, significantly more in iCSNB than cCSNB (χ^2=9.05, p=0.026), and always low-grade. Nystagmus occurred in 64% overall, with higher prevalence in the NYX subgroup (85%) than CACNA1F (65%). Visual acuity did not differ between groups. Myopia was significantly worse in cCSNB (median difference = –2.75 D, p=0.0062), with no significant age interaction in the GLM.

Conclusion:
This study links CSNB genotypes to distinct clinical features, improving understanding of phenotype–genotype correlations. Nystagmus and low-grade FH were more common in iCSNB, supporting genotype-based prognosis and management. Although myopia was more severe in cCSNB, refractive error appeared stable with age, suggesting a cautious approach to myopia control. Longitudinal studies are needed to clarify myopia progression in CSNB.

RF-102

Bornholm Eye Disease: A Retinopathy in the Spectrum of Dysfunctional Opsin OPN1LW/OPN1MW Genes

Dzenita Smailhodzic[1, 2], Gerard de Wit[1], Lonneke Haer-Wigman[3], Martha Tjon-Fo-Sang[4], L. Ingeborgh van den Born[5], Maria M. van Genderen[6, 7]

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[4] Pediatric ophthalmology, Rotterdam Eye Hospital, Rotterdam, Netherlands
[5] Medical Retina, Rotterdam Eye Hospital, Rotterdam, Netherlands
[6] Pediatric ophthalmology, Bartiméus Diagnostic Center for complex visuals disorders, Zeist, Netherlands
[7] Pediatric ophthalmology, Utrecht Medical Center, Utrecht, Netherlands

Introduction:
Bornholm Eye Disease (BED) is an underrecognized X-linked cone dysfunction caused by mutations in the *OPN1LW–OPN1MW* gene cluster. It typically presents in boys with reduced visual acuity, high myopia, and red-green color vision deficiency, but remains underdiagnosed.

Methods:
This multicenter observational cohort study included 39 genetically confirmed BED patients from 31 unrelated families, comprising 27 children (mean age 5.85 years) and 12 adults (mean age 38.6 years). All participants underwent standardized ophthalmic evaluations including best-corrected visual acuity (BCVA), cycloplegic refraction, HRR color vision testing, multimodal retinal imaging (OCT, fundus autofluorescence, color photography), and full-field ERG according to an extended ISCEV protocol. Visual development and myopia progression were analyzed using linear mixed-effects models.

Results:
At diagnosis, mean BCVA was 0.46 LogMAR in children and 0.42 LogMAR in adults. High myopia was common, with mean spherical equivalents of –5.9 D in children and –5.25 D in adults. A protan-type red-green color deficiency was found in 87.5% of patients. Photopic ERG responses were reduced and delayed, while scotopic responses remained relatively preserved. Visual acuity improved in early childhood but remained subnormal and stable into adulthood. Myopia progressed linearly at approximately –0.25 D/year between ages 3 and 15. The LVAVA haplotype was associated with milder disease, while LIAVA led to more severe impairment.

Conclusion:
BED is a distinct cone dysfunction syndrome with characteristic clinical and genetic features. Early diagnosis is essential for prognosis, visual rehabilitation, and genetic counseling. BED should be considered in boys with unexplained visual impairment, high myopia, and color vision defects.

RF-103

The spectrum of retinal and skeletal phenotypes associated with variants in CFAP410

Maria van Genderen[1, 2], Harriët van Diemen[1]

[1] Diagnostic Center for complex visual disorders, Bartiméus , Zeist, Netherlands
[2] Diagnostic Center for complex visual disorders, University Medical Center Utrecht, Utrecht, Netherlands

Introduction:
We describe the genotypic and phenotypic spectrum of *CFAP410*-associated disease, a relatively rare ciliopathy characterized by retinal degeneration and skeletal abnormalities.

Methods:
Medical record review of 7 patients with *CFAP410*-retinal dystrophy, and comparison to the findings of a recent study of 49 *CFAP410* patients in which we collaborated (PMID:40246852).

Results:
Onset of symptoms occurred early, between 4-12 years of age. Electrophysiologic testing showed that 2/7 patients had isolated cone dystrophy, 4/7 cone-rod dystrophy (CORD), and 1 retinitis pigmentosa (RP). Four patients from a genetic isolate carried the homozygous p.(Arg73Pro) variant; 3 had short stature but otherwise no skeletal abnormalities, 1 mild cognitive impairment. The remaining 3 patients had different homozygous variants and no skeletal abnormalities. The findings in our patients were comparable to those of the multicenter study, where the majority of patients had CORD, and only about one third had a RP phenotype. 11/46 patients had skeletal abnormalities which were mostly mild; other clinical symptoms, for instance renal abnormalities, were rare. The p.(Arg73Pro) variant accounted for at least one altered allele in 40.8% of patients.

Conclusion:
CFAP410-associated retinal degeneration is characterized by an early onset of symptoms and a CORD pattern in the majority of patients. The p.(Arg73Pro) variant is the most common and patients with this variant often have skeletal abnormalities, albeit mostly mild. When a diagnosis of *CFAP410*-associated retinal dystrophy is made, it is important to refer a child to a geneticist or pediatrician for a skeletal exam.

RF-104

12-month outcomes after voretigene neparvovec gene therapy in paediatric patients with RPE65-mediated inherited retinal dystrophy

Alejandra Daruich[1, 2], Maxence Rateaux[2], Batté[2], Nathalie De Vergnes[2], Sophie Valleix [3, 4], Matthieu Robert[1, 2], Dominique Bremond-Gignac[1, 2]

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[3] Genomic Medicine, Paris Cité University , Paris , France
[4] Genomic Medicine, Genomic Medicine Department of systemic and organ diseases, Cochin Hospital , Paris, France

Introduction:
Aims:To report main outcomes and complications following voretigene neparvovec (Luxturna) treatment in paediatric patients.

Methods:
Records of patients under the age of 17 treated by subretinal administration of voretigene neparvovec for confirmed biallelic *RPE65*-mediated inherited retinal dystrophy were retrospectively reviewed. Best-corrected visual acuity (BCVA) and data from spectral-domain optical coherence tomography, ultra-wide-field fundus imaging and Goldmann visual field (VF) were analysed at 12 months follow-up.

Results:
12 eyes of six patients (mean age: 7.8 years) were analysed. No intraoperative complications occurred. BCVA significantly improved at 12-month follow-up (mean LogMAR (logarithm of the minimal angle of resolution) BCVA: 1.0±0.8 at baseline vs 0.6±0.3 at 12 months, p=0.001). Mean central macular thickness and central outer nuclear layer thickness did not change at 12 months follow-up. VF V4e isopter did not show significant changes. Postoperatively complications included: elevated intraocular pressure in two eyes of the same patient, a parafoveal lamellar hole at 3 months post-treatment and atrophy on the injection site observed in all eyes except one, which significantly enlarged during 12 months (p=0.008).

Conclusion:
Most paediatric patients treated by voretigene neparvovec showed a significant increase in visual function at 12 months follow-up. None of the postoperative complications prevented gains in visual function.

RF-105

Beyond visual acuity: Evaluating functional vision in CRB1-Retinopathies using novel child-friendly tests

Ana Catalina Rodriguez Martinez[1, 2], Vijay Tailor-Hamblin[3], Robert Henderson[4], John Greenwood[1], Peter Jones[1], Mariya Moosajee[1]

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[2] Ophthalmology, Moorfields Eye Hospital, London, United Kingdom
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[4] Ophthalmology , Great Ormond Street Hospital, London, United Kingdom

Introduction:
Mutations in the *CRB1* gene cause a spectrum of inherited retinal dystrophies, including Leber congenital amaurosis/Early-onset severe retinal dystrophy (LCA/EOSRD), retinitis pigmentosa (RP), cone-rod dystrophy (CRD), and macular dystrophy (MD). As treatment strategies advance, establishing reliable functional vision metrics is crucial. This study evaluates two novel child-friendly, computer-based tests—PopCSF for contrast sensitivity function (CSF) and VacMan for visual acuity and visual crowding—against gold-standard clinical measures to better characterise visual deficits in *CRB1*-retinopathies.

Methods:
A cross-sectional study was conducted on 20 patients with molecularly confirmed *CRB1* disease-causing variants, compared to age-matched controls. Contrast sensitivity was measured using the gold standard Pelli-Robson chart and the novel PopCSF iPad-based test, while visual acuity and crowding were assessed using gold standard ETDRS and novel computerised VacMan test.

Results:
Visual acuity measured with ETDRS strongly correlated with the child friendly VacMan test in both uncrowded ($r = 0.868$, $p < 0.001$) and crowded thresholds ($r = 0.748$, $p < 0.001$). Additionally, visual crowding was significantly elevated in *CRB1* patients compared to controls, independent of phenotype or age of onset. *CRB1* patients exhibited significantly reduced contrast sensitivity compared to controls, with the LCA/EOSRD group showing the greatest impairment. Contrast sensitivity correlated moderately between Pelli-Robson and PopCSF ($r = 0.53$, $p = 0.020$).

Conclusion:
Novel child-friendly tests not only match gold-standard measures but also provide additional insights into functional vision in *CRB1*-retinopathies. This offers promising endpoints for monitoring disease progression and evaluating treatment outcomes in paediatric inherited retinal diseases.

RF-106

How Competent Are AI Models in Genetic Counseling for Inherited Retinal Disease in the Paediatric Patient? Evaluating Their Capacity to Inform Families About Gene Therapy Options

Emine Tınkır Kayıtmazbatır[1]

[1] Ophthalmology, Selcuk University, Faculty Of Medicine, Department Of Ophthalmology, Konya, Turkey

Introduction:
Inherited retinal diseases (IRDs) are complex conditions that often require specialized genetic counseling to help families understand the diagnosis and potential treatment options, including emerging gene therapies. This study aimed to evaluate the competence of large language models (LLMs) in simulating genetic counseling communication for pediatric IRD patients from a parental perspective.

Methods:
Seven family-centered questions were constructed based on common inquiries made by parents of children with IRDs. Responses were generated by three prominent LLMs—GPT-4, Gemini, and Claude. Three experienced pediatric ophthalmologists independently evaluated the responses using a 5-point Likert scale (1 = very poor to 5 = excellent) across four criteria: scientific accuracy, up-to-dateness, scope of content, and clarity.

Results:
Claude and Gemini outperformed GPT-4 in overall mean scores. Claude received the highest scores for scientific accuracy (4.28) and scope (4.33), while Gemini ranked highest for clarity (4.58). GPT-4's average scores were lowest in the "scope" domain (3.62), although its clarity remained acceptable (4.00). Reviewers noted that while all models provided mostly accurate content, Claude and Gemini produced more comprehensive and accessible responses, combining medical depth with lay-friendly language.

Conclusion:
This pilot evaluation suggests that large language models can meaningfully support family education in pediatric inherited retinal diseases. Claude and Gemini demonstrated superior performance in clarity and clinical relevance, indicating their potential utility in augmenting genetic counseling. However, professional oversight remains essential to ensure accuracy, individualization, and ethical integrity in real-world practice.

RF-107

The phenotypic and genotypic features of ADAMTSL4-related ocular disease

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Introduction:
Biallelic pathogenic *ADAMTSL4* variants are an important cause of isolated ectopia lentis with an increasing number of genetically confirmed cases internationally.

Methods:
We performed a retrospective, multi-center case series across six tertiary referral centers across Europe. We identified 42 individuals from 32 families in which genetic testing confirmed *ADAMTSL4*-related disease. Phenotypic features including age at diagnosis, lens abnormalities, refractive error, intraocular pressure and requirement for surgical management.

Results:
Identified participants had a young age of diagnosis (median 1.3 years) and a highly myopic refractive error (mean SE -10.27). A diagnosis of ectopia lentis et pupillae was made in a third of cases. Individuals with this feature were even younger at diagnosis (median 0.5 years), as too were individuals diagnosed with spherophakia (median 0.5 years) which we identified in 26% of our series. Subluxation tended to be in the inferior direction (~33%) but all directions of subluxation were observed. Zonules were noted to be missing or absent in the majority of cases. Lensectomy was performed in 81%. Overall, 16 different, potentially pathogenic variants in *ADAMTSL4* were identified. Apreviously reported 20-bp deletion (c.767_786del) was highly prevalent (23/32).

Conclusion:
ADAMTSL4-related disease tends to present at a younger age and to be associated with a higher myopic refractive error than other forms of ectopia lentis. Early genetic testing and identification of typical phenotypic features can aid early precise diagnosis, prevent unnecessary connective tissue disease workup, and guide targeted management of this early onset ocular disease.

O-108

Genetic background of childhood glaucoma in the Amsterdam UMC

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Introduction:
The treatment of childhood glaucoma is multidisciplinary. A genetic basis is often present, making collaboration with clinical geneticists essential. At Amsterdam UMC, genetic testing has been increasingly standardized over the past 15 years for primary congenital glaucoma (PCG), and selectively for secondary glaucoma. We will summarizes our findings and clinical implications.

Methods:
Retrospective database study

Results:
Over the past 15 years, we have treated 84 patients with PCG, 7 with juvenile glaucoma, and 95 with secondary glaucoma. Genetic testing was performed in 42 patients with PCG, often using a glaucoma gene panel. A pathogenic mutation was identified in 26 of these patients. In most cases, this was a variant in the *CYP1B1* gene, but mutations in other genes were also found (such as *LTBP1*, *TEK*, *PTBP1*, *PITX2*, and *SLC4A11*). Patients with a *CYP1B1* mutation often present with a more severe phenotype, requiring more surgeries and showing poorer visual development. Unexpected genetic findings, such as a mutation in the *SLC4A11* gene, helped us establish the correct diagnosis, while a *MYOC* mutation within a family helped guide management for a younger sibling. In secondary childhood glaucoma, genetic testing supports diagnosis and understanding of the underlying disorder. Examples include anterior segment dysgenesis, Axenfeld-Rieger syndrome, Stickler syndrome, aniridia, and megalocornea.

Conclusion:
Collaboration with clinical genetics in childhood glaucoma significantly enhances diagnostic accuracy, helps predict disease severity, informs treatment planning, and supports genetic counseling for families.

RF-109

Ophthalmological manifestations in Aicardi-Goutières syndrome

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Introduction:
Aicardi–Goutières syndrome (AGS) is a rare monogenic encephalopathy with early-onset microcephaly, leukoencephalopathy, widespread intracranial calcifications, acral chilblain-like lesions, and elevated cerebrospinal fluid interferon-α (IFN-α). Although ophthalmological involvement, including congenital glaucoma, has been reported, it remains poorly characterized. We present the ocular findings observed in AGS patients followed in our department.

Methods:
This retrospective observational study included children with genetically confirmed AGS followed in the ophthalmology department of Necker-Enfants Malades Hospital between March 2015 and March 2025. Data collected included mutation type, presence of hypertonia, glaucoma, strabismus, age at onset of ocular symptoms, final visual acuity, and any surgical interventions.

Results:
Thirteen children (6 girls, 7 boys) with genetically confirmed AGS were included. Mutations involved the autosomal recessive genes RNASEH2B (31%), SAMHD1 (31%), TREX1 (15%), and RNU7-1 (8%), and the autosomal dominant gene IFIH1 (15%). Glaucoma was identified in 5 patients (38%), bilateral in 80% of cases, with a mean age of onset of 4.8 years (range: 2 months to 14 years). All underwent trabeculectomy; one underwent diode cyclophotocoagulation. Exotropia was observed in 54% of patients, and nystagmus in 6 patients, including one case of spasmus nutans-type nystagmus leading to AGS diagnosis. Seven children presented with significant optic atrophy, including two without glaucoma.

Conclusion:
Ophthalmological manifestations in AGS are diverse and frequent. Early ocular signs, such as nystagmus, may aid diagnosis and should prompt neuroimaging for timely diagnosis and management. Children with AGS should be systematically and regularly checked for glaucoma, the diagnosis being easily overlooked, due to associated and expected optic atrophy.

Team- and network for children with craniofacial or orbital diseases

O-110

Optic neuropathy in patients with craniofacial fibrous dysplasia

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Introduction:
Optic neuropathy in craniofacial fibrous dysplasia often occurs during childhood or adolescence and can be irreversible. Our objective was to characterize the patients with craniofacial fibrous dysplasia (CFD) and optic neuropathy.

Methods:
From a cohort of 439 patients with fibrous dysplasia/McCune-Albright syndrome (FD-MAS), of whom 203 had craniofacial FD, optic neuropathy was identified in 13 patients, 15 eyes (bilateral in two patients) and 18 occurrences (recurrence in 3 eyes). Clinical characteristics and OCT data were collected.

Results:
Median age of optic neuropathy occurence was 9 years. Optic neuropathy was caused by compression of a stenosed optic canal in eleven times, four times by an aneurysmal bone cyst (ABC), and two times the result of iatrogenic damage after surgery. Visual acuity and visual fields of patients with ABC recovered completely after surgery. In patients with a stenosed optic canal, visual function improved after surgery in four eyes, stabilized in two and deterioated in one eye.

Conclusion:
It is useful to regularly monitor CFD patients with optic canal involvement and to perform prompt intervention in case of sudden visual loss.

RF-111

Teamwork in the diagnosis and treatment of osteopetrosis

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Introduction:
Osteopetrosis is a rare disease charakterized by abnormal bone growth. As a result of osteoclast dysfunction, they become excessively dense, thickened and sclerotic. This causes symptoms in the long bones, skull and teeth, but also changes in the bone marrow and, as a result, blood disorders. The first symptoms of osteopetrosis may be ophthalmological disorders - nystagmus, strabismus, decreased visual acuity.

Methods:
The aim of this work is to present the symptoms of osteopetrosis and the possibilities of diagnostic and therapeutic procedures. We present the case of a 3-year-old boy whose first symptom of the disease was nystagmus, further symptoms included face and skull dysmorphia, strabismus, optic nerve atrophy and bone fractures.

Results:
Osteopetrosis is a rare disease caused by a mutation in the CLCN7 gene encoding a protein for a chloride channel. Disturbance of the function of this channel causes improper absorption of calcium and phosphorus.

It can be inherited in an autosomal dominant, recessive or X-linked. The prevalence, severity of symptoms and the age of their occurrence depend on the type of mutationIt manifests with skeletal abnormalities, bone fractures, cranial nerve dysfunction, osteomyelitis, anemia, and dental malformations.

Conclusion:
Teamwork with a radiologist, orthopedist and neurosurgeon allows to make a diagnosis and plan the appropriate treatment. Narrowing of the optic nerve canals or other cranial nerves requires rapid neurosurgical intervention. The literature reports that such treatment allows for preserving vision in children.

O-112

Multidisciplinary Management of Orbital Langerhans Cell Histiocytosis: Insights from a Pediatric Case Series

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Introduction:
Langerhans Cell Histiocytosis is a rare disorder characterized by clonal proliferation of Langerhans cells, with potential multi-system involvement. Ocular manifestations, although uncommon, can be the initial presentation. This study aimed to evaluate the clinical characteristics, diagnostic processes and outcomes of pediatric LCH patients with orbital involvement.

Methods:
We retrospectively reviewed the records of 10 pediatric patients diagnosed with LCH and orbital involvement between January 1999 and December 2022 at a tertiary referral center. Data collected included demographic features, ophthalmologic findings, systemic involvement, imaging studies, histopathological confirmation, treatment modalities, and clinical outcomes. Patient management was coordinated by a multidisciplinary team comprising pediatric oncologists and ophthalmologists.

Results:
The mean age at diagnosis was 34.9 ± 26.4 months (range: 7–75), with a male predominance of 70%. Left eye involvement was seen in 70% of patients, and right eye involvement in 30%. Most patients (70%) presented initially with ocular symptoms, predominantly proptosis and periorbital swelling. Osseous lesions were identified in all patients, whereas bone erosion was detected in 90% of cases, and 60% had multisystem involvement. Diabetes insipidus was observed as a complication in 20% of the patients. Systemic chemotherapy was the main treatment, with radiotherapy or surgery in selected cases. The mean follow-up was 84.1 ± 58.1 months (range: 9–190), with disease recurrence observed in 5 patients.

Conclusion:
Ocular manifestations in pediatric LCH can be a crucial early sign. Multidisciplinary collaboration significantly enhances diagnostic accuracy and therapeutic outcomes, underscoring its importance in managing such complex cases.

RF-113

A periocular and intra-orbital congenital melanocytic nevus with a PRKCA-PLXNA1 fusion gene: a case report

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Introduction:
This case highlights the identification of a novel PRKCA-PLXNA1 fusion with a potential pathogenic role in a congenital intra-orbital rapidly growing melanocytic nevus. It also reveals the potential use of Trametinib, a MEK inhibitor, in stabilizing rapidly growing congenital melanocytic nevi.

Methods:
We report a 10-day-old female baby who presented with a congenital melanocytic nevus (CMN) on the right cheek, lower eyelid with extension into the orbit.

MRI imaging of the orbit and brain and biopsies of the skin, conjunctiva and intra-orbital mass with orbital decompression at the same time, were performed with histopathological and genetic analysis.

A multidisciplinary approach by ophthalmologists, dermatologists, paediatric oncologists, geneticists, and pathologists was set-up for the care and follow-up of this complex case.

Results:
Immunohistology reports showed a congenital nevus with proliferative nodules.

Genetic analysis revealed a novel PRKCA-PLXNA1 fusion gene.

Following significant growth of the congenital melanocytic nevus, Trametinib - a MEK inhibitor - was administered daily.

Follow-up after eight months shows overall stabilization of the congenital melanocytic nevus.

Conclusion:
This case report highlights the potential pathogenic role of a novel PRKCA-PLXNA1 fusion protein in a congenital melanocytic nevus. The crucial role of genetic and immunohistological analysis to rule out malignancy in proliferative melanocytic lesions is emphasized.

In this single case the potential use of Trametinib in stabilizing a rapidly growing congenital melanocytic nevus is described.

RF-114

Idiopathic Orbital Inflammatory Pseudotumor with Ocular Involvement: A Multidisciplinary Diagnostic Challenge

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Introduction:
Idiopathic orbital inflammatory pseudotumor (IOIP) is a rare, non-infectious, non-neoplastic cause of orbital inflammation, especially in children. Its presentation may mimic cellulitis or malignancy, leading to diagnostic uncertainty.

Methods:
A 6-year-old girl presented with a 5-day history of progressive left eyelid oedema and mild periocular discomfort, without systemic symptoms. She was initially treated with oral antibiotics, but there was no improvement. A CT scan requested by her paediatrician suggested orbital involvement, and she was referred to our unit. Visual acuity in the left eye was 3/10 and not correctable; ocular motility and pupillary reflexes were preserved. OCT showed macular subretinal fluid and optic disc swelling. A multidisciplinary evaluation was undertaken (paediatrics, ophthalmology, ENT, maxillofacial surgery, radiology). Brain and orbital MRI under sedation revealed lacrimal gland enlargement and posterior-inferior thickening of the left globe with uveal-retinal involvement, mild periorbital tissue reaction, and oedema of the lateral rectus insertion. No signs of abscess or intracranial extension were found.

Results:
Extensive workup included infectious and immunologic screening and lumbar puncture to exclude neoplastic or systemic inflammatory disease; all tests were negative. Given the suggestive clinical-radiological picture and the absence of red flags, a biopsy was not performed. Intravenous steroids led to progressive improvement. Serial ophthalmologic exams and fluorescein angiography confirmed the resolution of retinal changes. Follow-up MRI showed significant radiological improvement.

Conclusion:
This case highlights the diagnostic complexity of IOIP in children and the importance of a multidisciplinary, imaging-guided approach. Avoiding invasive procedures is feasible when clinical, laboratory, and imaging findings are concordant and closely monitored.

Team- and network with neurology (paediatric brain tumors)

RF-115

Teamwork in diagnosing and monitoring neurofibromatosis type 1 - when does an ophthalmologist raise a red flag?

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Introduction:
NF1 is a disease that requires multi-specialist cooperation. Among the diagnostic criteria, four of them concern the organ of vision - orbital neurofibromas, Lisch nodules, optic pathway gliomas and choroidal abnormalities. An ophthalmologist is often the first doctor to suspect the disease and plays a significant role in monitoring its course. In our hospital in Olsztyn, we conduct tests and treatment until the age of 18 or continue treatment until the age of 25.

Methods:
We present two cases of patients of different ages (3 and 18 years old) diagnosed with NF1 with different manifestations, in whom only ophthalmological symptoms indicated the development of the disease and allowed for quick diagnosis and initiation of treatment.

Results:
A patient with orbital plexiform neurofibroma presented with headache, which he reported nonverbally. After examination under general anesthesia and imaging studies, we diagnosed secondary glaucoma and optic nerve atrophy. After meeting with a radiologist, oncologist and neurosurgeon, the patient was qualified for selumetinib treatment. The second patient with optic chiasm glioma and myopia reported decreased visual acuity. Ophthalmoscopic examination revealed discrete pallor of optic discs, which correlated with OCT. After visual field examination, we found progression of the lesion, which was confirmed by MRI. The patient was transferred to an adult center and qualified for chemotherapy.

Conclusion:
NF 1 is a complex disease requiring teamwork. Cooperation between centers is equally important. Development of an ophthalmological examination protocol and photographic documentation allows for an objective assessment of the ophthalmological condition during follow-up visits.

RF-116

Observation Series of 5 Cases of Intracerebral Lesions Causing Visual Impairment: Neuro-Ophthalmology and Minimal Invasive Neurosurgery (MIN)

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Introduction:
Preservation of visual functions can be optimized in selective cases only by close interdisciplinary cooperation of ophthalmology and minimal invasive neurosurgery (MIN). The analysis of the ophthalmological outcomes in this series to prove the effect of functional recovery by minimal invasive neurosurgical procedures, enclosed 5 cases: 2 supra-sellar cysts, 1 cystic tumor of the posterior fossa, and 2 intracerebral angioma-bleedings, in all cases causing disturbance of visual functions.

Methods:
This MIN-concept combined 5 neurosurgical MIN-key techniques to assist microneurosurgery: high-end neuro-sonography with small probes, mouth-tracking of the microscope, endoscopy and LASER. Sealing technique was always used. Neuro-ophthalmological examination techniques were perioperatively used to meticulously measure and document visual and cerebral functions. Visual acuity, visual field, RNFL, orthoptic status and fundus were examined as soon as the patients´ condition did allow so.

Results:
In all cases visual functions were improved or preserved. Endoscopy in 3 cases and micro-surgical techniques in 2 cases were applied. Ultrasound navigation in 2 cases was needed. The combination was decided individually for each case. MIN techniques and ophthalmological examinations differed in relation to the patients´ individual conditions.

Conclusion:
Cooperation of neurosurgery and ophthalmology can preserve visual functions - even in emergency cases. Ophthalmology plays in this context the rule of an emergency indicator. Individual combination of MIN-techniques in each case is a key-concept of MIN.

RF-117

Suspected papilloedema referrals from the optometric community practice: diagnostic accuracy of OCT RNFL-thickness and B-scan ONSD measurements in a large paediatric cohort

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Introduction:
Our dedicated 'blurry disc' clinic for community optometry referrals for suspected papilloedema combines clinical assessment and OCT and B-scan imaging, escalating to MRI and LP via our Clinical Decision Unit (CDU) in selected cases. We audited long-term outcomes to investigate the diagnostic accuracy of OCT retinal nerve fibre layer thickness (RNFL) and B-scan ultrasound optic nerve sheath diameter (ONSD) for raised intracranial pressure (rICP).

Methods:
Audit of 337 consecutive referrals. Long-term outcomes (6–42 months) were used to classify children as with or without rICP. Receiver operating curves investigated diagnostic accuracy of RNFL and ONSD cut-off values. Regression modelling investigated potential predictive variables.

Results:
218 children (65%) were discharged forthwith, and 92 (28%) following review. 27 children (8%) were referred to CDU: 18 (5%) had rICP. RNFL thickness cut-off >144 µm at 95% specificity had 83% sensitivity (95% CI 61–100%) and ONSD cut-off >4.99 mm at 90% specificity had 82% sensitivity (95% CI 50–100%) for rICP. Increased RNFL, increased ONSD and the presence of typical symptoms all contributed to the prediction of rICP; acuity, age, sex and other OCT parameters did not.

Conclusion:
95% (319/337) of children referred with suspected papilloedema did not have rICP. Almost three-quarters of referrals (253/337, 70%) would have been more appropriately managed in the community. OCT RNFL and ONSD cut-offs show promising diagnostic accuracy for rICP in this population and setting. Teamwork guided by clear objectives and good communication enabled to develop the model clinic that led to valuable research outcomes on blurry disc referrals.

O-118

Ophthalmological findings in children with a primary brain tumor: the visual function up to two years after diagnosis

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Introduction:
Visual impairment can be a progressive and irreversible adverse effect in children with a brain tumor. The prevalence, types, and course of ophthalmological findings in these children are unknown, and there are no standardized international guidelines for screening and follow-up. In addition, it is unclear how visual acuity and visual field evolve during and after treatment. This study aims to describe the relationship between visual functions, disease status, and different treatments in children with a primary brain tumor.

Methods:
In this prospective cohort study, children (0-18 years) diagnosed with a primary brain tumor (2019 to 2021) were enrolled in four hospitals in the Netherlands. A standardized ophthalmological examination was performed within 4 weeks from brain tumor diagnosis, and repeated at 6, 12, 18, and 24 months. The main endpoints were visual acuity and visual field. Descriptive statistics were used for data analysis.

Results:
Among the 170 included children (median age 8.3 years), 134 (78.8%) had abnormal ophthalmological findings at diagnosis, including orthoptic abnormalities, papilledema, anterior segment anomalies, and visual impairment. Visual impairment involved binocular visual acuity loss in 13 children and visual field defects in 32 children. At 6, 12, 18, and 24 months, binocular visual acuity loss was present in 9, 6, 4, and 5 of the evaluable patients. Visual field examination was abnormal in 32, 25, 29, and 32 patients, respectively.

Conclusion:
These results confirm a high prevalence of abnormal ophthalmological findings in children with a primary brain tumor and emphasize the need for regular ophthalmological follow-up in this population.

O-119

Retinoblastoma Look-Alikes: A Diagnostic Challenge in Pediatric Ocular Oncology

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Introduction:
Retinoblastoma is the most common primary intraocular malignancy in children. However, several benign and malignant conditions can mimic its clinical and imaging features, making accurate diagnosis challenging. Misdiagnosis may lead to unnecessary enucleation or delayed treatment.

Methods:
We conducted a retrospective review of pediatric patients referred to our tertiary ocular oncology center over a 40-year period with a preliminary diagnosis of retinoblastoma or other masquerading conditions, such as Coats disease, persistent fetal vasculature (PFV), and uveitis. Confirmed cases of retinoblastoma or its mimickers were analyzed for clinical presentation, imaging findings, final diagnosis, and management.

Results:
Among 1,137 patients with confirmed retinoblastoma, the most common mimickers included Coats disease, PFV, uveitis, endophthalmitis, preseptal and orbital cellulitis, medulloepithelioma, glaucoma, and retinopathy of prematurity (ROP). Lack of experience in managing retinoblastoma was a key factor contributing to diagnostic delays and mismanagement. Expertise in ocular oncology and appropriate use of diagnostic tools—including fine needle aspiration, multimodal imaging, and clinical correlation—were crucial in guiding accurate diagnosis.

Conclusion:
Retinoblastoma look-alikes pose a significant diagnostic challenge. A high index of suspicion, multidisciplinary evaluation, and awareness of mimicking conditions are essential to prevent misdiagnosis and optimize patient outcomes.

Team- and network for allergic and inflammatory disorders (allergology, immunology and rheumatology)

O-120

Management and outcomes of Paediatric Blepharokeratoconjunctivitis in a Tertiary care centre

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Introduction:
Paediatric Blepharokeratoconjunctivitis (pBKC) is a condition characterised by lid margin inflammation and secondary ocular surface disease. Various topical and systemic antibiotics, topical and systemic anti-inflammatory medications, lid cleaning techniques and dietary supplementation are used in its treatment.

Methods:
This is a retrospective review of patients with pBKC presenting to Moorfields Eye Hospital from 2008 to 2024. We collected data on patient demographics, clinical presentation and treatments used.

Results:
We reviewed the records of 493 patients. Of those meeting eligibility criteria, 50% were male. 65% declared their Ethnicity and 20% were Asian, 16% White, 6% Mixed, 1% Black and 21% Other ethnic group. Mean LogMAR vision at presentation was 0.2 (min -0.1, max 1.8) and at last follow-up was 0.1 (min -0.1, max 1.6). 70% had corneal involvement at presentation, improving to 55% at last follow-up. The most common treatment was topical steroid (92%). Topical antibiotics were used in 80% of cases. 73% of children had systemic antibiotics and 63% received lubricants. Cyclosporine was used in 50% of cases but Tacrolimus only rarely (3%). Lid hygiene was prescribed in 82%. Omega 3 was recommended in 15% of cases.

Conclusion:
Epithelial involvement was more responsive to treatment than corneal vascularization.

The incidence of cPBK presenting to our institution has increased over time, as it has in other parts of the world. There use of Ciclosporin has also increased with time and it is prescribed by general paediatric ophthalmologists as well as in specialist corneal clinics.

O-121

Retinal phenotypes in cerebral malaria may provide insight into underlying immunopathology

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Introduction:
Malarial retinopathy, a key finding present in two-thirds of paediatric cerebral malaria patients, has diagnostic and prognostic significance. However, the presence of the individuals signs of malarial retinopathy differ considerably among patients. Defining the different 'retinal phenotypes' is necessary to evaluate the immunopathogenetic processes which underpin these differing presentations.

Methods:
We retrospectively evaluated graded malarial retinopathy forms for 978 retinopathy-positive paediatric cerebral malaria patients admitted to the Queen Elizabeth Central Hospital in Blantyre, Malawi, between 1996 and 2024. Leiden clustering was applied to this health record data using the ehrapy package in Python to identify retinal phenotypes. To evaluate the underlying immune processes contributing to the different retinal phenotypes we used a high-plex plasma proteomics assay, Olink, to simultaneously evaluate 368 plasma proteins involved in inflammation.

Results:
Our data-driven analysis identified four novel retinal phenotypes in paediatric cerebral malaria, haemorrhagic, ischaemic, mild haemorrhagic and mild ischaemic, which can be broadly classified as two retinal phenotypes. Haemorrhagic cases had lower CCL17 and CCL22, implying reduced Th2 response and Treg recruitment in comparison to ischaemic cases. Haemorrhagic cases had higher FCRL6, suggesting CD8 T cell and NK cell responses are important to the development of the haemorrhagic phenotype.

Conclusion:
Differing retinal phenotypes may arise due to different underlying pathogenetic mechanisms in paediatric cerebral malaria.

RF-122

Three Neonates With Congenital Cataract And Uveitis In 2 Years Caused By Spiroplasma Infection

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Introduction:
The purpose of this abstract is to demonstrate the importance of considering Spiroplasma infection as a cause of congenital cataract and inflammation in neonates.

Methods:
Retrospective case series of neonates presenting within 2 years with cataract and signs of uveitis. Lens material was aspirated with vitrector and tested using the ISpro Molecular Culture method detecting a high load of bacterial DNA of the genera Spiroplasma confirmed by 16S metagenomic sequencing. Cases were evaluated by pediatrician for further organ involvement.

Results:
Three neonates presented with cataract and anterior segment inflammation in one or both eyes with posterior synechiae, cyclic membrane, uveitic nodules or keratic precipitates. Two patients had macular scarring in the otherwise unaffected eye. Four eyes underwent cataract operation, posterior capsulotomy and anterior vitrectomy and left aphakic. 16S RNA PCR on lens material aspirated with the vitrector identified Spiroplasma. All children were examined and followed by pediatrician and treated with systemic azithromycine. Visual axis opacification (VAO) and glaucoma required surgery in one eye of the first baby requiring surgery. One eye of the second baby had beginning VAO which resolved upon increasing topical steroids.

Conclusion:
This case-series represent the first identified Spiroplasma infections in the Netherlands causing cataract and uveitis. Two out of three patients had macular scarring which our center is the first to report. Little is known of the incidence, mechanism of infection, systemic and developmental effects on these children and is likely an underrecognized entity as diagnosing requires 16S RNA PCR on lens material.

RF-123

A paediatric case of ocular toxocara and the value of metagenomics in undifferentiated uveitis

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Introduction:
Ocular toxocariasis is a rare parasitic infection, which can manifest aschorioretinitis. This case report highlights the infrequentpresentation in a paediatric patientwho presented with panuveitis and chorioretinitissecondary to ocular toxocariasis.Metagenomics were performed fromvitreous fluid to identifyother contributing pathogens.

Methods:
A 14-year-oldfemale patientpresented with progressive visual deterioration in her right eye (uncorrected visual acuity 0.24). Clinical examinationidentified a panuveitis and chorioretinitis, demonstrated by anterior chamber inflammation, vitreous haze, tractional retinal folds, superonasalvessel dragging, and intraretinal fluid near the optic disc.

Results:
After a full uveiticwork up to exclude tuberculosis and toxoplasmosis, treatment included atwo-week course of oral albendazole, alongside tapering doses of systemic steroids. Due to deteriorating right visionto 0.80and retinal fibrosis, an urgent right vitrectomy, hyaloid and fibrotic membrane peel, dry vitreous biopsy and gas tamponade were performed. Intravitreal bevacizumabwas also administered. Metagenomic testing of vitreous fluid incidentally detected HHV6, which was deemed clinically insignificant.Vitreous fluid showed toxcaraon the Western blot. Positive ToxocaraIgG serology also supported ocular toxocariasis as the diagnosis.

Conclusion:
Surgical intervention stabilised retinal fibrosis, and improved visual acuity (postoperative right eye acuity 0.40 with pinhole improvement to 0.20). Incidental HHV6 finding is likely explained byits typically benign persistence after initial childhood infection.

This is a valuable rare case report ofpaediatric ocular toxocariasisand the benefit of early surgical intervention to reduce disease sequelae. Italsodemonstratesthe diagnostic aid of metagenomics in undifferentiated uveiticcases.

Team- and network with neonatology & panel discussion 'The best way to treat and follow up on ROP'

RF-124

Effect of image enhancement on agreement in the evaluation of plus disease features

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Introduction:
The presence of plus disease is a key factor in determining the need for treatment in retinopathy of prematurity (ROP). However, there is poor agreement in the assessment of plus disease, which remains a significant challenge in ROP management. This study aims to evaluate the impact of image quality improvement on the consistency of plus disease assessment in ROP images

Methods:
Five independent graders evaluated posterior pole images of preterm infants before and after the application of an image enhancement algorithm. For each retinal quadrant, arterial and venous dilation and tortuosity were independently assessed. Normal, preplus, plus disease and treated cases were included. Raw images (first session) and enhanced images (second session) were randomly graded one month apart to mitigate potential recall or learning bias.

Results:
A total of 45 cases were evaluated. Weighted Fleiss' kappa analysis demonstrated the greatest improvement in intergrader agreement for arterial tortuosity (raw: $\kappa = 0.662$; enhanced: $\kappa = 0.779$). The lowest agreement was observed for venous tortuosity (raw: $\kappa = 0.476$; enhanced: $\kappa = 0.540$). Overall, mean agreement improved from moderate to substantial following image enhancement. Across all features, the nasal quadrants (Q3 and Q4) showed the most notable gains. Pre-plus cases exhibited the highest increase in intergrader consistency after processing.

Conclusion:
Image quality enhancement can improve intergrader consistency in the assessment of vascular tortuosity and dilation, with the most pronounced effect observed for arterial tortuosity and in pre-plus cases. Nonetheless, residual variability in agreement highlights the inherent limitations of subjective evaluation of these vascular features

RF-125

Optimizing mydriasis for retinopathy of prematurity screening

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Introduction:
Mydriatic regimens used in retinopathy of prematurity (ROP) screening vary considerably and are usually unnecessarily overdosed, subjecting this fragile population to high risk of systemic complications.

Methods:
A cross-sectional online survey was conducted, using a self-report online questionnaire, to compile real-time data on the preferred mydriasis practice patterns for ROP screening across Europe. Additionally, a double-masked, non-inferiority, crossover, randomized controlled trial, the MyMiROPS Trial, examined whether microdrops (6.5μl) of combined phenylephrine 1.67% and tropicamide 0.33% in a mixture are non-inferior to standard drops (28-34μl), while minimizing systemic adverse events.

Results:
In 94.5%, the applied mydriatic regimen consists of phenylephrine with at least one muscarinic antagonist. About 54.5% of the reported mydriatic solutions are non-commercial, in-house preparations, i.e., either dilutions of commercial solutions or mixtures. The MiMyROPS trial found that microdrops are superior for induced pupil dilation at 45 minutes (Bonferroni-corrected 95%CI: 0.01, 0.23; p=0.008), and non-inferior at 90 and 120 minutes. Fewer cardiorespiratory adverse events were also observed after microdrop instillation during a 48-hour follow-up period. A one-compartment model with first-order absorption best described the pharmacokinetic data.

Conclusion:
There is considerable heterogeneity in the applied mydriatic regimens for ROP screening in Europe, reflecting the absence of universal guidelines. The wide use of in-house preparations underlines the gap in the pharmaceutical industry. The MiMyROPS trial is the first study establishing non-inferiority of microdrops compared with standard drops of a diluted mydriatic mixture, showing reduced systemic adverse events after microdrops, and determining the pharmacokinetic profile of phenylephrine eyedrops in preterm infants.

RF-126

Evaluating the Implementation and Impact of ROP Programs in Albania

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Introduction:
Retinopathy of prematurity (ROP) is a severe ophthalmological condition that can result in blindness in premature babies. In Albania, advancements in neonatal care and increased survival rates of preterm newborns have highlighted the critical need for early screening and timely intervention for ROP. This study examines the epidemiology, risk factors, and clinical outcomes associated with ROP within the Albanian context.

Methods:
A retrospective analysis was performed on preterm infants admitted to neonatal intensive care units (NICUs) across Albania over a two-year period. Clinical variables, including gestational age (GA), birth weight (BW), duration of oxygen therapy, and compliance with established screening protocols, were systematically collected. ROP screening was conducted in accordance with standardized guidelines, and disease severity was classified based on the International Classification of Retinopathy of Prematurity (ICROP). Statistical analyses were carried out to investigate correlations between identified risk factors and the development of ROP.

Results:
Key risk factors associated with the development of ROP included lower gestational age, reduced birth weight, and prolonged exposure to oxygen therapy. Furthermore, the study identified deficiencies in adherence to ROP screening protocols, which adversely affected early detection and timely management of the disease.

Conclusion:
ROP remains a significant public health concern in Albania, particularly among the most vulnerable preterm neonates. Despite improvements in neonatal care, this study underscores the urgent need for comprehensive screening programs and timely therapeutic interventions. Implementation of standardized protocols, nationwide professional training, and enhancements in healthcare infrastructure is needed to prevent avoidable blindness among Albanian preterm infants.

O-127

The value of a national ROP register - experiences from the last two decades

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Introduction:
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Methods:
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Results:
SWEDROP is a national quality register for ROP, initiated in 2006. The major aims are to collect national data on infants screened for ROP in Sweden, to provide information on various aspects on ROP, on the screening process, and on treatment, to be able to improve and optimize screening and reduce potential future visual problems for the prematurely born children.

The presentation will highlight various aspects, such as how the register was started, how patient data were collected and registered, what were the obstacles, how was research made possible and how did the results of research improve of the screening process.

Finally, some thoughts about the future will be presented.

Conclusion:
-

O-128

Anti-VEGF For Retinopathy of Prematurity in the Netherlands: National Collaboration and Shared Decision-Making

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Introduction:
Anti-VEGF treatment is an alternative to laser treatment for retinopathy of prematurity (ROP), although it has seen limited application in the Netherlands. We evaluate the use of anti-VEGF as primary treatment for ROP since the implementation of new guidelines and national collaboration.

Methods:
Eligible were infants treated with anti-VEGF for ROP in the Netherlands from October 2023 to October 2024. Ophthalmic data, treatment characteristics, timing of treatment and reactivation, and correspondence between ophthalmologists were collected after parental consent. This is a sub-analysis of a nationwide observational cohort study on ROP treatment.

Results:
All nine eligible infants were included. Treatment decision was reached through peer consultation in all cases. Reasons for anti-VEGF treatment included: very young post-menstrual age (PMA) at treatment indication (n=8), aggressive ROP (n=3), maximum vessel growth in zone I (n=3) or posterior zone II (n=5), the child being unfit for general anesthesia (n=2), and inadequate retinal visualization (n=2). Median PMA at primary treatment was 33 weeks and 4 days (IQR=10 days). All nine infants received ranibizumab 0.02ml (10mg/ml). N=1 reached full vascularization 97 days after primary treatment. Additional treatment was performed in N=8; median time to first additional treatment was 49 days (IQR=22 days). N=7 received laser as secondary treatment. N=1 was reinjected with anti-VEGF: 40 days post-reinjection, a vitrectomy (unilateral) and (endo)laser (bilateral) were performed.

Conclusion:
In the Netherlands, the incidence of indications for anti-VEGF as described in the new ROP-guidelines is low. Implemented national collaboration and shared decision-making increases exposure for ROP experts.

RF-129

The Red vs the Green Diode Laser for Treating ROP

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Introduction:
The purpose of this study is to compare the results of the treatment of threshold ROP with two different lasers; the red 810nm vs green 520nm diode lasers.

Methods:
The red laser (LightMed LIGHTLas 810nm laser, San Clemente, CA) was employed from January 2021 until March 8, 2023, at which point the green laser (Norlase LION 520nm laser, Copenhagen) was deployed.

Results:
The parameters assessed were number of laser spots, the total amount of energy applied and the total duration of the laser treatment. The total number of spots required to treat all eyes with the red laser were 14587 (mean 1216) and for the green laser 21122 (mean 1760). This amounts to 31% more spots, 184% less energy and a 400% decrease duration with the green when compared to the red laser

Conclusion:
Introduction of the green laser led to shorter treatment times, improved post-treatment eye conditions, clearer corneas, and better retina views.

Red laser: 1) consistent post-op corneal and retinal edema, 2) hot, sharp, rapidly occurring, deep retinal burns, 3) consistent post-op retinal hemorrhages, 4) slow retinal pigmentation, >1 month, 5) consistent loss of dilation, 6) rare post op cataract.

Green laser: 1) no post-op corneal and very little retinal edema, 2) soft, slow developing, more anterior (less deep and less white) retinal burns, 3) very rare post-op retinal hemorrhage, 4) rapid retinal pigmentation by 2 weeks, 5) no loss of dilation, 6) no post-op retinal edema, 7) no post-op cataracts.

RF-130

A very unusual form of ROP

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Introduction:
We present an unusual case of ROP associated with a delayed vasculogenesis

Methods:
Retinographies of all the fundus examinations will be presented

Results:
This is a premature boy 25+1 weeks of gestational age (wGA), 660 grams.

At first fundus examination (31+1 wGA) there were not vessels at the optic nerve head, nor at 33+3 wGA. Vessels appear at the examination of 35+1 wGA as short and narrow arriving at the midle of zone 1. Then the patient was transferred to another hospital, and was refered to us 5 weeks later (40+3 wGA) with ROP stage 3 + affecting 360º in zone 1.

Patient was treated with antiVEGF injection in both eyes, with regression of the lesions and slow progression of vasculogenesis, that was not completed at 54 w GA.

Conclusion:
The patient showed an important delay of the angiogenesis/vasculogenesis with an aggressive form of ROP that behaves as an AP-ROP but much more later than usual.

Possible association with other systemic findings will be discussed

RF-131

Evaluation of timed dexamethasone eye drops to prevent proliferative retinopathy of prematurity: a study protocol for a randomized intervention, multi-centre, double-blinded trial (DROPROP)

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Introduction:
As the survival rate of preterm infants increases, more are at risk for sight-threatening retinopathy of prematurity (ROP). Off-label use of dexamethasone eye drops may prevent moderate/severe ROP from progressing to the point of requiring treatment. This study protocol presents the first randomized controlled trial evaluating the efficacy and safety of dexamethasone eye drops in preterm infants with ROP.

Methods:
In a Swedish randomized, multicenter, double-blinded study, 100 infants born before 30 weeks' gestational age and diagnosed with moderate to severe ROP (i.e., Type 2 ROP, including stage 2 ROP in posterior zone II) will be included. Infants will receive either intervention with dexamethasone eye drops (1 mg/mL) (n = 50) or placebo, saline (n = 50). Depending on the severity of ROP, one eye drop per day or every other day will be administered for a maximum duration of 12 weeks. Treatment will continue until ROP is resolved or Type 1 ROP (ROP fulfilling treatment criteria) develops. The primary outcome is whether dexamethasone intervention reduces the proportion of infants developing Type 1 ROP compared to placebo. Adverse events and potential side effects will be recorded, like high intraocular pressure and growth restriction. Levels of cortisol in saliva and glucose in urine will be measured repeatedly. Secondary outcomes will be presented.

Results:
Not applicable.

Conclusion:
This protocol outlines the first randomized controlled trial to evaluate the efficacy and safety of dexamethasone eye drops to preterm infants in preventing the progression of moderate/severe ROP to requiring treatment, supporting its clinical use and national guidelines.

RF-132

The importance of teams in TEARDROPS 2: tear proteomics in infants at risk of Retinopathy of Prematurity (ROP) – a prospective observational study

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Introduction:
Retinopathy of Prematurity (ROP) is a sight-threatening condition of increasing prevalence worldwide. Diagnosis relies on repeated screening by ophthalmoscopy or fundus imaging. This is resource intensive and often distressing for babies. Investigations into alternative screening methods have largely focussed on retinal imaging and AI. The TEARDROPS feasibility study (2024), showed promising results in the application of mass spectrometry to analyse tears samples obtained by Schirmer strips in babies meeting UK ROP-screening criteria. The aim of this study is to investigate if there are biomarkers for treatment-requiring ROP and the optimal time frame for such testing can be identified.

Methods:
This multi-centre study includes four NHS health-boards: Greater Glasgow & Clyde, Lanarkshire, Manchester and Birmingham. In total, 58 extremely preterm infants will be recruited into a longitudinal phase, in which half the infants are expected to reach treatment criteria. Each infant will undergo tear sampling coincident with each ROP-screening as per UK guidelines. Samples will be analysed by mass spectrometry for significant proteome changes that correlate with ROP stages, at Glasgow Polyomics. A validation study will be carried out in a second cohort of 52 infants.

Results:
Analysis of longitudinal samples in phase 1 will determine a narrower sampling time frame for the phase 2 validation of possible biomarkers.

Conclusion:
Trial progression will rely on thorough communication within the multiple teams in respective NHS sites and Glasgow Polyomics. We will discuss the TEARDROPS protocol with a focus on teamwork which will be crucial for success in this prospective observational trial.

Team- and network in rehabilitation for the visually impaired

O-133

'"My CVI" psycho-education for children with cerebral visual impairment (CVI)

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Introduction:
Cerebral visual impairment (CVI) is a complex visual disorder to understand and explain to others. We developed the computerized psycho-educational programme 'MyCVI' to educate children and youngsters with CVI about their condition. 'My CVI contains interviews of children who have CVI themselves.

Aims of the study:to increase participants'knowledge of CVI, as well as their self-esteem, adaptive coping, and wellbeing.

Methods:
Thirty-nine children (aged 7-16 years; 51 % boys) with CVI participated together with their teachers in a pre-test, post-test and follow-up assessment consisting of questionaires about self-esteem (SDQ), adaptive coping (SCQ), and wellbeing (Cantril, PERIK, HRQoL). Qualitative interviews were conducted for a subjective evaluation of the programme. A mixed-methods analysis was used to evaluate the effect of the psycho-educational programme

Results:
Participation in the psycho-educational programme resulted in increased knowledge of CV, as well as a better teacher-reported social and academic self-concept and less self-reported feelings of social exclusion. Qualitative data revealed that children enjoyed participating in the programme; they learned that there are other persons with CVI, and gained more knowledge and practical tools on how to deal with their CVI.

Conclusion:
Participation in the psycho-educational programme 'My CVI' increases children's knowledge about their visual impairment an improves social outcomes and feelings of inclusion. We also noticed, as an unexpected benificial effect of this study, that orthoptists and ophthalmologist who watched the interviews of 'My CVI' got more insight in the problems of CVI children.

O-134

Prospective Open Label Randomized Controlled Clinical Study Evaluating the Efficacy of Perceptual Learning among Patients with Congenital Nystagmus

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Introduction:
Introduction: There are limited treatment options to improve vision in patients with congenital nystagmus. We examined the use of repeated visual stimulation with Gabor Patches to achieve that goal.

Methods:
Methods: In a prospective randomized (3:1) controlled open label study, patients aged 9-55 years with congenital nystagmus and best corrected visual acuity (BCVA) between 20/40–20/200 were included. The treatment group underwent visual stimulation for four months, controls were followed without treatment. BCVA, contrast sensitivity, and stereoacuity were assessed.

Results:
Results: 26 patients were treated and 10 served as control. Mean age was 22.47±12 years. 13/26 (50%) of treatment subjects achieved driving license BCVA (20/40) vs 1/10 (10%) of controls. Distance treated BCVA improved by 0.11±0.06 logMAR (one Snellen line) compared with 0.013±0.06 logMAR in controls (p=0.001). Near BCVA improved by 0.20±0.18 logMAR (2 Snellen lines) compared with 0.06±0.06 logMAR in controls (p=0.020). Mean stereoacuity and contrast sensitivity improved by 801±730’ and 292±391% in the study group as opposed to 246±376’ and 152±67% in controls respectively (p=0.031; p=0.157).

Conclusion:
Conclusions: Visual perceptual learning using Gabor Patches resulted in a significant improvement in near and distance visual acuity, which allowed 50% of patients to achieve a driving license BCVA. A corresponding improvement in stereopsis and contrast sensitivity was noted. Further studies will identify which patients will benefit most from this treatment.

O-135

Evaluating Pediatric Eye-Related Quality of Life: Insights from the PedEyeQ Assessment at a Pediatric Ophthalmology and Strabismus Clinic

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Introduction:
Eye-related quality of life (E-QL) is a fundamental component of pediatric ophthalmologic care. Visual impairments in children can affect developmental milestones, daily functioning, and social interactions. Although these conditions have a considerable impact, E-QL is not routinely assessed in clinical practice. Standardized tools like the Pediatric Eye Questionnaires (PedEyeQ) offer a reliable means to quantify the effects of pediatric eye conditions on quality of life.

Methods:
A prospective study is being conducted at the Pediatric Ophthalmology and Strabismus Clinic at Carmel Medical Center. Children aged 5–17 years with various eye disorders and their parents are invited to complete the PedEyeQ. The questionnaires evaluate multiple domains including Functional Vision, Bothered by Eyes/Vision, Social interactions, and Frustration/Worry for the children, and the Impact on Parent and Family for caregivers. Clinical data—such as diagnosis, treatment interventions, treatment duration, and demographic information—are collected for correlation analysis with E-QL outcomes. All data is collected by a secured web app for building and managing online survey and database called REDCap.

Results:
Preliminary results from over 100 participants indicate that a significant proportion of children report moderate E-QL, while parent assessments suggest higher levels of concern regarding their child's condition. Final data and a comprehensive literature review, will be presented at the conference.

Conclusion:
The study underscores the importance of incorporating E-QL assessments into routine pediatric ophthalmologic care. Recognizing the impact of eye conditions on children's daily lives is essential for guiding clinical decision-making and enhancing patient-centered care involving psychological help when needed.

Paediatric ophthalmology never works Eye-lone (teamwork with other subspecialists in ophthalmology - surgery)

O-136

Comparison of the efficacy of Defocus Incorporated Multiple Segments spectacle lenses and 0.01%atropine in slowing the progression of myopia in European children–a one year randomised clinical trial

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Introduction:
The rising global prevalence of myopia among children underscores the need for interventions aimed at slowing its progression. The aim of our study is to compare the efficacy of DIMS spectacle lenses with 0.01% atropine in slowing the progression on myopia in Polish children. The study continues to its second year.

Methods:
One hundred and ten Polish children aged between 6 and 16 years old with no significant ocular pathology were randomly assigned to either the group wearing DIMS spectacle lenses and receiving daily placego eye drops or to the group wearing conventional single vision spectacle lenses and using daily 0.01% atropine eye drops. The primiary variables measured at six-month intervals were the cycloplegic spherical equivalent of refraction [SER] and axial eye lenght [AXL].

Results:
The mean change of AXL in group A was 0.19(±0.03)mm, compared to 0.11(±0.02)mm in group B. When divided by age: for those aged 6-11 in gr.A:0.3(±0.05)mm, in gr.B:0.15(±0.02)mm. For 12-16 years old in gr.A:0.1(±0.02)mm, in gr.B:0.07(±0.02)mm. Mean change of SER was 0.37(±0.06)D in gr.A, compared to 0.23(±0.05)D in gr.B. With the division by age: for 6-11 years old in gr.A:0.6(±0.01)D, in gr.B:0.25(±0.05)D. For 12-16 years old in gr.A 0.19(±0.05)D, in gr.B:0.21(±0.08)D.

Conclusion:
DIMS spectacle lenses demonstrated higher efficacy than 0.01% atropine in controlling the progression of myopia in participants, particularly those aged 6-11 years. Our study contributes to the growing body of knowledge on myopia control within the European population.

RF-137

Myopia-X-1: phase 2 randomised controlled trial of blue-light blind spot stimulation to reduce myopia progression in children: 6-month results

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Introduction:
Blue-light stimulation of the optic nerve head triggers dopamine release, mimicking the effect of sunlight. Using a VR-headset, a new smartphone app, Myopia-X, selectively directs blue light at the optic nerve head, whilst engaging the child in a game to incentivise adherence. Here we explored safety, tolerability and effectiveness over 6 months.

Methods:
124 children age 6-12 years with myopia -0.75 to -5.00D took part in a phase-2 randomised controlled trial of Myopia-X 10 minutes twice daily vs active control (defocus-incorporated-multiple-segment-spectacles) at 11 sites across Europe. Primary outcomes: axial length (AL), cycloplegic spherical equivalent (SER).

Results:
Withdrawal rates were 28.8% vs 2.9% for Myopia-X/control. Mean AL change from baseline was 0.14 mm (95% CI, 0.12, 0.16) vs 0.08 mm (95% CI, 0.05, 0.10, p=0.004) for Myopia-X vs DIMS; mean change in SER was -0.19 D (95% CI, -0.26, -0.11) vs -0.16 D (95% CI, -0.26, -0.06, p>0.05). Median adherence was 57.3% (IQR 44.6 to 73.4). Adverse events included headache (5.9%), dizziness (2.5%), asthenopia, eye irritation/pain/pruritus, blurred vision, nausea, malaise (0.8% each).

Conclusion:
Initial safety data are as anticipated; similar adverse effects have been reported with other treatments delivered via VR-headsets. Low sample size means that statistical analysis of effectiveness is not possible; the observed effect size could be used to design further trials of Myopia-X. High attrition and low adherence indicate that more engaging/incentivising games are needed.

Myopia-X is a safe and novel treatment option to slow myopia progression; further development and evaluation are warranted.

RF-138

Perforating scleral vessels in pediatric myopia: prevalence, characteristics and association with disease progression

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Introduction:
Perforating scleral vessels (PSV) are more common among highly myopic patients suffering from neovascular complications and are often present beneath the site at which lacquer cracks form in pathologic myopia. It has been hypothesized that structural scleral instability at the entry area of PSV contributes to scleral expansion and myopia progression. The aim of our study is to assess PSV prevalence in a cohort of pediatric myopic patients and to establish their association with myopia progression.

Methods:
Myopic pediatric patients (age at baseline<16 years) were retrospectively recruited from the database of the Institut Français de Myopie. For all included patients B-scan optical coherence tomography(OCT) acquisitions, best corrected visual acuity (BCVA) assessment and axial length(AL) measurements were available at baseline and at 1 year follow up. Exclusion criteria were spherical equivalent(SE) >-1.0 D, syndromic myopia, congenital glaucoma and cataract, amblyopia and overlapping retinal diseases.

Results:
One hundred and thirty (130) eyes of 65 patients with a mean age at baseline of 8.3 ± 4.1 were included. Mean SE at baseline was -5.1 ± 3.7 D. PSV were detected on baseline acquisitions in 46/130 eyes (35.4%). Eyes showing PSV were characterized by a higher prevalence of staphyloma and a higher baseline AL. Moreover they showed a significantly higher increase in AL and decrease in SE during the follow up.

Conclusion:
PSV in pediatric myopia are associated to the presence of staphyloma and may predict a faster myopia progression

O-139

Incidence of postoperative retinal detachment and bacterial endophthalmitis in the Swedish National Pediatric Cataract Register (PECARE) and associated risk factors

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Introduction:
To investigate the incidence and risk factors of retinal detachment (RD) and bacterial endophthalmitis in a cohort of children who underwent cataract surgery before the age of eight.

Methods:
Data was retrieved from the Swedish national pediatric cataract register. All eyes with congenital or infantile cataract that underwent surgery between January 1, 2007, and December 31, 2023 with at least one follow-up were included. Cases associated with trauma, uveitis or RD at surgery were excluded. Parameters that could be important for complications were analyzed.

Results:
RD was found in seven of 1073 eyes reflecting an incidence of 0.65%. There were no statistically significant differences in age at surgery, presences of intellectual disability or general disease, cataract type, surgical technique, axial length, corneal diameter, previous glaucoma surgery or occurrence of persistent fetal vasculature (PFV), although the frequency of glaucoma surgery and PFV was higher in RD cases; 42.9% versus 13.2% and 57.1% versus 26.0%. Aphakia was significantly more common in RD patients; 71.4% versus 19.3% (p=0.042), as well secondary glaucoma; 57.1% versus 19.5% (p=0.032). No cases of endophthalmitis were observed.

Conclusion:
The incidence of RD was low compared to previous studies and no endophthalmitis was found. This might be a result of centralized pediatric cataract care with few but experienced surgeons. Aphakia and secondary glaucoma were associated with higher RD risk and those cases should be followed carefully. PFV and glaucoma surgery were found at a higher frequency in RD cases prompting comprehensive post operative care also for these children.

RF-140

Early Onset Cataracts Related to NHS gene

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Introduction:
To describe the natural history of pediatric patients with early onset cataracts related to NHS gene.

Methods:
We retrospectively reviewed the records of eight children (16 eyes) with early onset cataracts related to NHS gene. Records variables included patients and parent's clinical exome, age at cataract onset, age at cataract surgery and type of surgery, type of cataract, ocular biometry, visual behavior and visual acuity, and dental and neurological development.

Results:
Eight children, five females and three males were included. Four gene variants were classified as pathogenic, one probably pathogenic and three of them as VOUS. The age at cataract's diagnosis was less than 6 months in six patients, 4 years old, and unknown in an adopted child (he underwent surgery at 2 years old). The carrier was the mother in two males and one female, and the father in two female siblings. Three patient had a de novo mutation, and it was unknown in the adopted child. Six of them had micro cornea and five nystagmus. Two males and one female had intellectual disability and the two males had dental anomalies.

Conclusion:
This is the largest serie of patients with early onset cataracts related to NHS gene. Microcornea, nystagmus, bilateral congenital cataracts associated or not with neurological disorders and dental misdevelopment is highly suspicious. The pathology is in general more severe in males. Bilateral early onset cataracts should always undergo a whole exome sequencing.

RF-141

The use of the iris claw intra-ocular lens in the rare case of absence of capsular support in children; The Dutch network; historical overview, organization, teamwork, outcome and future perspectives.

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Introduction:
In the absence of capsular support, intraocular lens (IOL) implantation needs a personalized surgical approach and selection of the most suitable IOL for the individual patient. Worldwide different approaches are used. This paper reports the long term outcome of anterior chamber iris-claw lens implantation in the eyes of children in the Netherlands. An historical overview of the introduction and the use of the 'Artisan' IOL will be presented, with a special emphasis on (the organization of) care for children with Marfan syndrome in the Netherlands.

Methods:
Review of literature, overview of Marfan care in the Netherlands.

Results:
Main clinical outcome measures were endothelial cell count (ECC), visual acuity gain and complications after a mean follow up of more than 7 years. Historical and organisational overview on Marfan care in NL.

Conclusion:
Anterior chamber iris-claw IOL implantation results in good visual outcome and endothelial cell counts with an acceptable complication rate. Iris-claw implantation is a suitable and low complex option for surgical correction of aphakia in the absence of capsular support in children. Collaboration and specialised teamwork is needed to provide the best (eye) for children with Marfan syndrome.

O-142

Refractive Error Profile in Infantile Nystagmus

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Introduction:
Infantile nystagmus (IN) is characterised by involuntary, rhythmic eye movements typically presenting within the first six months of life. It may occur in association with albinism, idiopathic infantile nystagmus, photoreceptor dystrophies, optic nerve hypoplasia, and other conditions. Understanding the refractive error profile in IN is important for early correction, supporting optimal visual development. Previous studies, largely single-centre, have suggested a predominance of with-the-rule (WTR) astigmatism and hypermetropia. In this study, we aimed to characterise the refractive error profile in IN using a multicentre approach involving cohorts in Europe and East Asia, to assess generalisability of existing findings.

Methods:
Data were obtained from a multicentre study on foveal hypoplasia conducted by the Foveal Development Investigators Group (FDIG). Only eyes with complete refractive data were included. Refractive errors were classified as myopic, hypermetropic, or emmetropic based on spherical equivalent. Astigmatism was categorised as WTR, against-the-rule (ATR), or neither.

Results:
A total of 154 eyes were analysed. WTR astigmatism was observed in 84% (n=130), ATR in 12% (n=18), and 4% (n=6) showed neither pattern. Regarding spherical error, 66% of eyes were hypermetropic (n=102), 29% myopic (n=45), and 5% emmetropic eyes (n=7), indicating a significant hypermetropic bias.

Conclusion:
This study confirms that WTR astigmatism and hypermetropia are the predominant refractive features in IN across different populations. The findings, based on a multicentre dataset, reinforce current understanding and highlight the need for early refractive correction. Further research is warranted to explore genotype-specific refractive profiles.

O-143

Transition programmes in the UK and Europe - an international survey of practice

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Introduction:
Transition programmes for children and young people (CYP) with developmental and chronic conditions moving from paediatric to adult eyecare services improve clinical outcomes and readiness in adolescents, yet implementation varies widely. This study aimed to compare transition practices and barriers in the UK and Europe.

Methods:
Paediatric ophthalmologists in the UK and Europe completed an electronic survey. Responses were analysed to evaluate transition practices, factors influencing transition decisions and challenges faced.

Results:
We received 81 responses (UK: 45 Europe: 36), predominantly from hospital-based ophthalmologists.Structured transition programmes for typically developing CYP are in place in 19% of departments in Europe and 20% in the UK, and for those with additional needs in 33% vs 13% of services. The most common age at transition to adult services is 16 years in Europe and 18 in the UK for typically developing CYP, and 18 for those with complex needs in both Europe and UK.

Both regions prioritise developmental level (74%), disease stability (60%), and young person's readiness (59%) as critical factors influencing decisions about transition. The UK may have greater multidisciplinary team involvement and communication with the GP (50% vs XX). Common barriers to setting up transition programmes include ‘lack of resources/time/ funding’ (64%), ‘lack of standardised approach’ (63%) and ‘lack of organisation/ co-ordination’ (57%).

Conclusion:
Despite differences in transition timing, implementation and multidisciplinary involvement, tackling common challenges in resourcing, standardisation and coordination could facilitate the adoption of transition programmes to support CYP with taking responsibility for their healthcare.

RF-144

Ubi liquidus, ibi evacua: A novel approach in the treatment of recurring iris cysts

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Introduction:
Epithelial inclusion cysts of the iris are a rare complication following ocular surgery or trauma and typically exhibit a more aggressive course than primary iris cysts. Their management remains challenging due to their tendency for rapid growth, which can lead to obscuration of the visual axis, elevated intraocular pressure, corneal decompensation, and secondary glaucoma or uveitis. Particularly in pediatric and young adult patients, the high proliferative capacity of epithelial cells increases the risk of recurrence. Current treatment modalities, including Nd:YAG laser cyst disruption, sclerosing agent irrigation, and complete surgical excision, are associated with high recurrence rates.

Methods:
In this case series, we describe two patients with a posttraumatic recurrent iris cysts emerged in childhood, successfully managed with glaucoma drainage devices (Baerveldt tube and Paul tube) to provide continuous cyst fluid drainage.

Results:
In both cases, placement of the drainage device successfully controlled cystic growth and maintained the cyst outside of the visual axis. One patient experienced transient diplopia, which resolved spontaneously. The second case was complicated by scleral perforation at the surgical site following additional mechanical trauma but demonstrated a favorable outcome after surgical repair.

Conclusion:
These cases illustrate the potential role of glaucoma drainage devices as an alternative treatment strategy for recurrent iris cysts, particularly when conventional therapies have failed.

RF-145

Conjunctival Amelanotic Melanoma in a pediatric patient: A case report and management

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Introduction:
Conjunctival melanoma is a rare ocular neoplasm, representing 2–5% of all ocular tumors and 5–7% of ocular melanomas. It carries a five-years survival rate of approximately 83-84% and a recurrence rate of 39%. This malignancy typically arises in middle-aged adults and is exceptionally uncommon in the pediatric population, where available data remains scarce.

Methods:
A 13-year-old male with no relevant medical history presented in 2017 with a hyperemic lesion in the temporal region of the right eye. In 2022, due to progressive growth of the lesion, he went to an ophthalmologic consultation. A diagnostic and therapeutic excision was performed. Histopathological examination revealed positive immunohistochemical staining for HMB-45 and Melan-A, confirming the diagnosis of an ulcerated invasive melanoma with a depth of 1 mm and positive tumor margins.

Results:
Following the initial findings, a PET-CT scan and scintigraphy were performed, showing no evidence of tumor dissemination. A second surgical procedure was carried out, with conjunctival margin expansion of approximately 5 mm and adjunctive cryotherapy. Sentinel lymph node and parotid gland biopsies were also performed. A second histopathological analysis revealed no signs of malignancy. The patient has been followed up with OCT imaging, showing favorable progression with no evidence of recurrence to date.

Conclusion:
Due to the patient's socioeconomic limitations, there was a delay in receiving medical attention, and it was not possible to administer the recommended adjuvant pharmacological therapy. Therefore, management was limited to cryotherapy. Given the extreme rarity of this neoplasm in pediatric patients, further studies are needed to define optimal management strategies.

Team- and network for diagnostics (from imaging to diagnosis)

RF-146

The importance of MRI in the diagnosis of various eye diseases

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Introduction:
MRI is an important method for the diagnosis of a large number of diseases of the eye and orbits, including congenital anomalies, inflammatory conditions, tumors, brain diseases and demyelinating diseases. The use of MRI in children is of particular importance.

Methods:
The goal is to show various conditions and point out the importance of interdisciplinary cooperation between ophthalmologists and radiologists, especially in conditions where it is not possible to establish a diagnosis with certainty through clinical examination.

Results:
The paper presents the most interesting cases in the last 5 years in which MRI was the method of choice for diagnosis. MRI is the most important method for establishing the diagnosis of multiple sclerosis in a 15-year-old girl. This method is essential for diagnosis and monitoring the response to therapy in a five-year-old girl with orbital myositis. Congenital anomalies of the optic nerve, especially of the retroorbital part, as well as structural anomalies of the visual pathways can only be seen on MRI scans. This is the method of choice for dermoid cysts, especially for determining the exact localization, size and relationship with the surrounding structures.

Conclusion:
MRI is an important method for the diagnosis and monitoring of a large number of diseases of the eye and orbit.

RF-147

Evaluation of the Virtual Reality Eye Tracker for Neuro-Ophthalmic Assessment: Feasibility, Reproducibility, and Reliability

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Introduction:
Virtual Reality (VR) eye trackers offer portable, objective tools for neuro-ophthalmic testing. This study evaluated the feasibility, reproducibility and reliability of a VR eye tracker (BulbiCAM) compared towearable eye-tracking glasses (PupilLabs Neon glasses), highlighting its potential clinical utility and feasibility.

Methods:
A prospective study involving 39 healthy participants (mean age±SD: 30.0±9.5 years) assessed inter-visit reproducibility of BulbiCAM tests across two visits. Pupillary light reflex tests were conducted with both BulbiCAM (BulbiTech AS, Oslo, Norway) and PupilLabs Neon (Pupil Labs GmbH), enabling paired assessments. Reproducibility was analysed using intra-class correlation coefficients (ICC), reliability via Bland-Altman analysis, and participant experience through a survey evaluating test comfort and usability.

Results:
Participant feedback highlighted high acceptability for BulbiCAM: 89% found the test comfortable,92.6% approved of the duration, and 81.5% reported no eye strain or fatigue. Pursuit and pupil testsdemonstrated strong inter-visit reproducibility (ICC=0.81–0.88), whereas saccades showed variablereproducibility (best ICC was 0.62). Paired assessment between devices showed close agreement for keypupillometer metrics: baseline diameter (bias: -0.48mm ± 0.47), peak constriction diameter (bias: -0.56mm ±0.36), constriction velocity (bias: 0.22mm/s ± 0.58), and duration of constriction (bias: -0.052s ± 0.15).

Conclusion:
This study demonstrates BulbiCAM's clinical feasibility, with strong patient acceptability and reproducibility for pursuit and pupil tests. Paired assessments confirmed its accuracy in key pupillometric parameters, validating its reliability for clinical and research use. These findings are being extended to a paediatric cohort, where BulbiCAM shows promising potential as an all-in-one point-of-care tool in paediatric neuro-ophthalmology assessments, enabling efficient testing through pursuit, pupillometry, and additional integrated tests.

RF-148

Torpedo maculopathy (TM): multimodal analysis of a rare congenital maculopathy in a paediatric case series and review of the literature

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Introduction:
TM is a rare congenital macular lesion usually located temporal to the fovea.

Methods:
Data from children with TM were collected retrospectively since 2014. Demographics, VA, fundoscopy, fluorescein and ICG angiography, autofluorescence, OCT, OCT-A, VF, microperimetry and ERGs were analysed.

Results:
Seven children (5M:2F), i.e. 3 RE, 4 LE, were included. Median age at diagnosis was 5 years. VA was symmetrical with no significant amblyopia. Fundoscopy of most cases (5/7) revealed a hypopigmented torpedo-like lesion on the horizontal raphe pointing to the fovea. In one case, TM was located in the superotemporal arcade without ocular torsion. Disruption of the photoreceptors and RPE complex with thinning of the outer retina and hyperreflective choroid was seen on all OCTs. Four cases were classified as type 1 and three as type 2 according to Wong et al. classification. TM margins were hyperautofluorescent, FA and ICG angiography showed staining without leakage. OCT-A didn't show any variability of the multi-layered retinal vascular complexes. Visual field and microperimetry revealed a paracentral scotoma in one case. Multifocal ERG highlighted a wave amplitude depression next to the TM in two cases but no inter-ocular asymmetry on FF ERGs.Toxoplasma serology was negative in most patients.

Conclusion:
TM seems not to be always located exactly in the horizontal raphe. Whether individual factors or combined mechanisms at a precise time during foetal development of the RPE contributes toTM formation is still unknown. TM is usually non-progressive although it could be complicated by CNV. A regular follow-up is therefore recommended.

RF-149

Feasibility and Clinical Utility of Hand-held Optical Coherence Tomography in Children with Retinoblastoma

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Introduction:
Early tumour detection is of the highest importance for the preservation of vision and reducing the risk of metastasis in Retinoblastoma (Rb). We aim to investigate whether hand-held optical coherence tomography (HH-OCT) can improve diagnosis, treatment and outcomes in children with Rb compared to conventional investigations.

Methods:
In this prospective study, eligible participants with suspected Rb were consented and recruited. During examination under anaesthesia, HH-OCT was performed in addition to the standard clinical care and imaging (fundus photography and ultrasound). We recorded the: (1) success rates of imaging based on tumour location, (2) management plans blinded to the OCT scans and (3) change in management after analysis of OCT scans.

Results:
Ninety-six Rb eyes of 89 children (age range 2 weeks to 7.5 years old) were imaged in 136 OCT sessions. The scan acquisition success rate was 91% with the tumours located in Zone M (Macular, success rate 92%), Zone 1 (posterior pole, success rate 93%), Zone 2 (equatorial zone, success rate 56%) and Zone 3 (Anterior retina, success rate 7.4%). OCT enabled new tumour detection (92%), subclinical recurrence detection (84%), monitoring treatment (76%) and monitoring vitreous seeds (79%). HH-OCT altered management in 26% of all OCT sessions.

Conclusion:
HH-OCT provides high-resolution 3-dimensional images of the Rb, which can improve clinical judgement and monitor changes in the tumour, vitreous seeds and scar at microscopic resolution. We highlight the feasibility of this technology in Rb diagnostic and management workflows. However, further studies with larger numbers would be beneficial.

O-150

Functional and Structural Outcomes in Paediatric Myelin Oligodendrocyte Glycoprotein Antibody-Associated Disease (MOGAD): A Prospective Study

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Introduction:
Myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) is a rare autoimmune disorder, frequently manifesting with optic neuritis (ON) in children. While visual recovery is common, structural damage to the retinal nerve fibre layer (RNFL) may persist. This study prospectively assesses the diagnostic utility of functional and morphological measures in paediatric MOGAD.

Methods:
Twelve children with serologically confirmed MOGAD (10.9 ± 3.1 years; 13/23 ON+ eyes) and twelve age-matched controls (11.5 ± 2.8 years) were examined using optical coherence tomography (OCT), pattern-reversal visual evoked potentials (VEP; 14' and 62' check sizes), and visual functional testing. Outcome measures included peripapillary RNFL (pRNFL) thickness in global, temporal, nasal, and papillomacular bundle (PMB) sectors, and VEP P100 peak times. Diagnostic accuracy was assessed using receiver-operating characteristic (ROC) analysis.

Results:
ROC analysis demonstrated high discrimination for global (area under the curve (AUC) = 0.95), PMB (0.93), and temporal (0.92) pRNFL. ON+ eyes showed excellent separation (AUC = 1.00 global), while ON– eyes also showed strong separation in PMB (0.99) and temporal (0.96). P100 peak times showed moderate discrimination in ON+ eyes (AUC = 0.89 for 14′; 0.72 for 62′) and lower discrimination in ON– eyes (AUC = 0.73 for 14′; 0.60 for 62′). Visual acuity remained largely preserved.

Conclusion:
In our cohort, pRNFL thickness, particularly in the global, PMB, and temporal sectors, demonstrated strong discriminatory ability in paediatric MOGAD, including in eyes without previous ON.

Posters

Team- and network for allergic and inflammatory disorders (allergology, immunology and rheumatology)

P-151

Adalimumab in the treatment of pediatric noninfectious panuveitis

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Introduction:
Pediatric non-infectious panuveitis are rare sight-threatening conditions characterized by inflammation of the entire uveal tract. The pathophysiology is complex, and the etiology is multifactorial, with genetic, immune, and environmental factors.

Methods:
We will present patients with panuveitis treated with adalimumab.

Results:
The first patient is a 9-year-old boy who was hospitalized due to bilateral complicated panuveitis with visual acuity of 5/60 in the left eye, bacon-like precipitates on the cornea, dense opacities in the CV and exudative retinal detachment. After a complete diagnosis, he was treated with local and systemic therapy, corticosteroids and methotrexate. Due to the recurrence of the disease after 3 months, the patient was introduced to adalimumab therapy and a multi-year remission was achieved and the best corrected visual acuity in both eyes was 1.0 (Snellen). The second patient is a 13-year-old girl with bilateral panuveitis. After the applied KS therapy, methotrexate was included. After 6 months, stellate maculopathy develops and visual acuity drops, so adalimumab is introduced into the therapy. Four years later, the visual acuity in both eyes is 1.0 without correction with stable findings in the anterior and posterior segments.

Conclusion:
Adalimumab achieves cure of complications, remission and long-term stable visual acuity.

Team- and network for children with craniofacial or orbital diseases

P-152

Visual outcome in 5 years old children operated for non-syndromic craniosynostosis

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Introduction:
Craniosynostosis is a congenital condition characterised by the premature fusion of one or more cranial sutures, leading to cranial malformations. This study aimed to evaluate visual outcomes in children operated for non-syndromic craniosynostosis at pre-school age, in order to elucidate long-term ophthalmological effects and guide treatment and screening strategies.

Methods:
Eighty-nine children (63 boys) with various types of non-syndromic craniosynostosis, operated at Uppsala Craniofacial Centre between 2012 and 2019 and an age-matched control group of 33 healthy children underwent comprehensive ophthalmological evaluations at 5 years of age. Visual acuity (VA), crowding ratio, low-contrast visual acuity (LCVA), color vision and stereo acuity were examined using standardised protocols. Statistical comparisons across craniosynostosis subtypes and between the craniosynostosis group and control group were conducted.

Results:
Children who underwent operation for unilateral coronal craniosynostosis had lower distance VA and LCVA, mainly due to the higher prevalence of strabismus and refractive error on the contralateral side to the fused suture. Amblyopia and reduced stereoacuity were more frequent in this subtype. Crowding was common in craniosynostosis group, especially in children operated for sagittal craniosynostosis. Near VA was generally lower in all craniosynostosis subtypes, compared to controls.

Conclusion:
Long-term visual outcomes vary in children operated for non-syndromic craniosynostosis. Unicoronal craniosynostosis had the greater risk of visual impairment. Tailored screening along with optimised surgical strategies are needed in order to address the specific ophthalmological challenges associated with different craniosynostosis subtypes.

Team- and network with neurology (paediatric brain tumors)

P-153

A case of anorexic Idiopathic Intracranial Hypertension

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Introduction:
Idiopathic Intracranial Hypertension (IIH), is a rare disorder in the general population.There is a strong link between obesity and IIH. Rapid weight gain in patients with eating disorders and anorexia nervosa can increase the risk of developing idiopathic intracranial hypertension (IIH). It is a rare presentation in patients with low weight eating disorder.

Methods:
The community optometrist referred a 13-year old female patient to paediatric ophthalmology with blurry optic discs margins. She was diagnosed with eating disorder 11 months ago and referred to Child and Adolescent Mental Health Services (CAMHS) for counselling. Her initial weight was 30kgs (BMI 11.86) increased to 45.6Kg, (BMI 16.83) within one year as clinically improved. The patient was asymptomatic with normal visual acuities, colour vision and full visual fields. Ophthalmic examination was unremarkable apart from bilateral papilloedema. She had satisfactory blood tests including vitamin profile. Only her vitamin E lipid ratio was slightly raised 4.66 (2-4).She underwent emergency brain MRI with contrast, which was normal. She had a lumbar puncture with opening pressure of 36 mmH2O. Her CSF microscopy and PCR was unremarkable.

Results:
Treatment started with Acetazolamide orally 250 mg TDS with good response and without significant side effects. She remains under ophthalmology and paediatric review.

Conclusion:
Rapid weight restoration can trigger IIH and patients might not be symptomatic. Ophthalmic monitoring of these patients might be beneficial.

Team- and network with the geneticist (metabolic, genetic, syndromes, retina)

P-154

Coexistence of Legg-Calvé-Perthes and Stickler Syndrome in a Pediatric Case: A Rare Condition Presenting with Chronic Retinal Detachment

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Introduction:
In this study, we present the case of an 8-year-old male patient who was admitted to the Ophthalmology Clinic of Eskişehir Osmangazi University in March 2024 with complaints of decreased vision in the right eye and was diagnosed with chronic retinal detachment.

Methods:
The patient had a history of Legg-Calvé-Perthes (LCP) disease diagnosed by the orthopedics department. A family history revealed that both his father and uncle had experienced retinal detachment. On ophthalmologic examination, the visual acuity of the right eye was counting fingers at 2 meters, while the left eye had a visual acuity of 1.0 (Snellen). Fundus examination showed a macula-off chronic total retinal detachment in the right eye and peripheral lattice degeneration in the left eye. Pars plana vitrectomy was performed on the right eye, and 360-degree prophylactic laser photocoagulation was applied to the left eye.

Results:
In the postoperative period, proliferative vitreoretinopathy (PVR) developed in the right eye, requiring revision surgery. Genetic evaluation revealed a COL2A1 gene mutation, confirming the diagnosis of Stickler syndrome. Due to the delayed presentation, despite anatomical success, no significant visual improvement was achieved.

Conclusion:
This case highlights the importance of considering underlying Stickler syndrome in pediatric patients presenting with a diagnosis of LCP and emphasizes the critical role of early diagnosis in preventing avoidable blindness.

P-155

A Pediatric Case of Homocystinuria with Bilateral Ectopia Lentis and Anterior Staphyloma

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Introduction:
We present a 9-year-old girl who was admitted with severe photophobia and decreased vision and was later diagnosed with homocystinuria.

Methods:
Ophthalmologic examination under general anesthesia included intraocular pressure (IOP) measurement and anterior and posterior segment evaluation. Bilateral central corneal opacities, segmental iris atrophy, microspherophakia, and lax eyelids were noted. The right lens was positioned normally but showed phacodonesis; the left lens was subluxated anteriorly with iridodonesis and bluish sclera in the upper quadrant. Axial length was 22 mm (OD) and 24.3 mm (OS). IOP measured 6 mmHg (OD) and 5 mmHg (OS). Fundoscopy revealed normal optic discs and retina.

Pars plana lensectomy, iridectomy, and anterior vitrectomy were performed in the left eye. Due to subsequent right lens subluxation, the same surgery was later performed on the right eye. Genetic analysis was initiated.

Results:
A homozygous CBS gene mutation confirmed homocystinuria with hyperhomocysteinemic thrombosis. The patient was referred to pediatric metabolism, hematology, and neurology. Despite a recommended methionine-restricted diet, adherence was poor, leading to inadequate metabolic control. Progressive anterior staphyloma and fluctuating IOP were noted during follow-up.

Conclusion:
Ocular involvement in homocystinuria may cause severe morbidity. Early diagnosis, timely surgical intervention, and effective systemic metabolic control are critical to prevent vision-threatening complications.

P-156

Morning Glory-Like Optic Disc Anomaly in a Neonate with PHACE Syndrome: A Case Report

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Introduction:
PHACE syndrome is a rare neurocutaneous disorder that may present with ocular anomalies. Previous reports have described infantile hemangiomas in association with optic disc anomalies. We present a case of a PHACE syndrome and a concurrent morning glory-like optic disc anomaly.

Methods:
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Results:
Case Presentation: A full-term female neonate presented at birth with segmental infantile hemangiomas on the right temporal region and right lower lip, corresponding to a segment measuring > 5 cm. Ophthalmologic examination revealed a morning glory-like optic disc anomaly with peripapillary staphyloma in the right eye; the left eye was unremarkable. Anterior segments and macular reflexes were normal. The infant showed reliable monocular fixation and tracking, full ocular motility and no relative afferent pupillary defect on both eyes. Cardiologic evaluation, including ECG and echocardiography, revealed no abnormalities.

A craniocervical MRI at six weeks of age revealed an excavation of the right optic disc, consistent with a morning glory-like optic disc anomaly. An additional thickening of the right optic nerve was detected, which has been described in combination with a morning glory-like optic disc anomaly. No additional intracranial or vascular abnormalities were identified. The diagnostic criteria for PHACE syndrome were met. Propranolol (Hemangiol®) was initiated at six weeks, resulting in regression of the hemangioma.

Conclusion:
Early ophthalmologic evaluation is essential in patients with hemangiomas and suspected PHACE syndrome, as ocular involvement may be present. In this case, the preserved macular reflex indicates a favorable visual prognosis.

P-157

Restoring PAX6 expression via CRISPR activation: a novel therapeutic strategy for Aniridia-Associated Keratopathy

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Introduction:
Aniridia is a rare pan-ocular genetic disease primarily characterised by iris and/or foveal absence or hypoplasia. Common complications include nystagmus, glaucoma, cataract, and aniridia-associated keratopathy (AAK). This autosomal dominant disorder is linked to heterozygous variants in the *PAX6* gene, ultimately causing haploinsufficiency. Currently, no universal vision-sparing treatment exists for aniridia, though several therapies have been proposed to counter its complications. AAK is a major therapeutic focus, as the progressive opacification of the cornea represents a rather painful condition that cannot be treated surgically.

Methods:
We exploited the clustered regularly interspaced short palindromic repeats (CRISPR)-mediated transcriptional activation (CRISPRa) system to target the *PAX6 P1* promoter region aiming to restore its expression and exploring this strategy as a potential AAK treatment. Three different single guide RNAs (sgRNAs) were designed for this purpose

Results:
The ability of the CRISPRa system to trigger *PAX6* expression was tested in primary cells derived from a patient harboring a balanced translocation involving the *PAX6* locus [t(11p13;22q11.23)]. PAX6 mRNA and protein levels were assessed 24 and 48 hours post-transfection, respectively. Chromatin immunoprecipitation (ChIP) confirmed PAX6-binding to its known targets. Among the sgRNAs tested, SG2 yielded the best results, doubling PAX6 expression while maintaining protein functionality.

Conclusion:
Although preliminary, our data highlight that the CRISPRa system is able to restore PAX6 expression and functionality *in vitro*. These results support further exploration of CRISPRa-based therapies for haploinsufficiency-related diseases.

P-158

Two different clinical presentations of X-linked retinoschisis

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Introduction:
X-linked retinoschisis (XLRS) is the most frequent inherited retinal disease presenting in young male patients and is caused by mutations in *RS1* gene, which encodes retinoschisin protein. Foveoschisis has been reported in 80% and foveal atrophy in 20% of eyes with XLRS. Here, we present two different clinical presentations of XLRS.

Methods:
We report the ophthalmologic evaluation of two different families followed by detailed phenotypic evaluation by clinical geneticist and genetic panel testing of X-linked retinoschisis-associated gene.

Results:
The first case concerns three brothers of a family with ten children (six boys, four girls). Their mother noticed her three sons have reduced vision before the age of 5. Two oldest brothers were diagnosed with myopia, however, their visual acuity did not improve with optical correction. A thorough ophthalmoscopic examination was performed. OCT imaging revealed macular retinoschisis in all patients. The oldest patient underwent ERG and genetic testing, both of which supported the diagnosis of XLRS. A hemizygous mutation in *RS1*, c.637C>T (p.Arg213Trp) was detected.

The second case describes a boy whose visual acuity at the age of four did not meet the normative age criteria. OCT imaging revealed foveal atrophy and VEP testing showed delayed visually evoked responses. The patient was referred to a clinical geneticist. A hemizygous mutation in *RS1* gene c.305G>A (p.Arg102Gln) was detected. The patient's mother also underwent genetic testing, which confirmed the diagnosis of XLRS.

Conclusion:
X-linked retinoschisis can be phenotypically highly variable and collaboration with a clinical geneticist is crucial for its early diagnosis.

P-159

Intraocular Pressure and Risk of Glucocorticoid-Induced Ocular Hypertension are Increased in Children with Congenital Adrenal Hyperplasia

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Introduction:
Children with classical congenital adrenal hyperplasia (CAH) require lifelong systemic glucocorticoid (GC) therapy. A significant but often underrecognized side effect is GC-induced intraocular pressure (IOP) elevation, which may lead to irreversible glaucomatous damage if undetected. This study aimed to assess IOP and the incidence of ocular hypertension in pediatric CAH patients under GC therapy.

Methods:
A cross-sectional analysis was performed on 26 CAH patients (aged 4–21 years) receiving hydrocortisone and 45 age- and sex-matched healthy controls. Ocular examinations, including IOP measurement via Icare tonometry, were conducted. Intraocular hypertension was defined as a peak IOP ≥21 mmHg. Data collected for patients included GC dosages, auxological parameters, and serum levels of androstenedione and 17-hydroxyprogesterone over the past year. IOP levels were compared between patients on high-dose (≥15 mg/m²/day) and maintenance-dose (<15 mg/m²/day) hydrocortisone.

Results:
IOP was significantly higher in CAH patients than in controls (20 ± 3 mmHg vs. 13.8 ± 3 mmHg, $p<0.001$). Intraocular hypertension was detected in 53% of CAH patients. IOP was unrelated to age, sex, or hormonal control but correlated positively with BMI SDS changes over the past year ($r=0.5$, $p=0.008$). No significant difference in IOP was found between high- and maintenance-dose groups ($p=0.5$).

Conclusion:
Elevated IOP is a significant complication in children with CAH receiving systemic GC therapy, even with physiological hydrocortisone doses adjusted to circadian rhythm, indicating an intrinsic ocular vulnerability. These findings emphasize the need for regular IOP monitoring, particularly in patients with BMI SDS increase, and interdisciplinary follow-up to prevent glaucomatous damage.

P-161

Occurrence of rhegmatogenous retinal detachment (RRD) in a cohort of genotyped stickler patients: Impact of age and sex, and implications for preventive treatment

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Introduction:
Stickler syndrome is a genetically inherited vitreoretinopathy that can lead to retinal detachment from early childhood. There is currently no consensus preventive treatment. We aimed at determining whether the sex influence the age of onset of retinal detachment (RD) in COL2A1 Stickler patients.

Methods:
Retrospective study including 46 Stickler patients with COL2A1 mutation from two tertiary centers who underwent an RRD. We analyzed the age differences of retinal detachment in males and females. A multivariate analysis was used to analyze risk factors for RRD including sex, presence of preventive treatment, congenital myopia or other ophthalmologic or extra-ophthalmologic conditions.

Results:
Among girls with the COL2A1 mutation, the average age of RRD onset was later (21.3 (+/-14.5) years; range: 5.4–66.0), and the age distribution was broader than in boys (10.07 (+/-5.7) years; range: 0.5–23.6; $p<0.0001$). Analyzing RRDs occurring before age 18, we identified two peaks of incidence in girls, around 9 years and 13 years. Only a preventive treatment (360° laser or rail band) was an independent protective factor for RRD (RR = -0.49 (+/-0.002); $p<0.001$).

Conclusion:
Males tend to experience retinal detachment at an earlier age whereas RRD occurrence in females is more spread out across their lifetime. Hormonal fluctuations throughout life could play a role in the development of retinal tears. Based on these findings, a considered preventive treatment should be performed before puberty and earlier in boys.

P-162

Choroidal neovascularization secondary to Best vitelliform macular dystrophy (BVMD): a pediatric case series followed by OCT-A and review of the literature

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Introduction:
BVMD is due to mutation in *BEST1* expressed in RPE. It occurs in childhood and progress through six stages. Rarely, BVMD is complicated by CNV.

Methods:
Case-series

Results:
We present two cases of children (F, M), median age 3,5 years evaluated for RE amblyopia (median VA 0,32). Fundoscopy revealed bilateral egg-yolk lesions and RE hemifoveolar hemorrhages. Lipofuscin deposits were hyperautofluorescent with a masking effect due to perifoveal bleeding. OCT highlighted subfoveal hyperreflective material, SRD and RPE changes. RE OCTs confirmed a bulging type 2 CNV with dye leakage on FA. OCT-A enabled clearer viewing of CNV. Treatment with aflibercept IVT was administered: the girl received 7 IVT every 1 to 6 months for 18 months with VA improvement to 0,8 RE. CNV remained inactive for the following two years. The boy received 3 aflibercept IVT at 2-monthly intervals, achieving a VA of 0,63 in RE. Unfortunately, he recently developed acute LE-type 2 CNV (VA 0,08) treated by aflibercept IVT.

Conclusion:
BVMD can present with active CNV in children, and these CNV usually develop more quickly and severely than in adults. OCT-A appears to be a more efficient and less invasive than FA visualizing CNV and monitoring treatment efficacy. Several CNV treatments are listed in the literature: PDT, anti-VEGF and triamcinolone IVTs. So far, no standard treatment has been validated for children.CNV occurs rarely but insidiously at any stage in children with BVMD. As CNV can lead to severe visual impairment, early diagnosis and treatment are crucial in order to preserve optimal visual function.

P-163

Uncovering Cryptic Anterior Segment Anomalies in FHONDA Using Ultra-High-Speed Swept-Source OCT

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Introduction:
Mutations in SLC38A8 cause foveal hypoplasia, optic nerve decussation defects, and anterior segment dysgenesis (FHONDA). Diagnosing anterior segment anomalies in FHONDA is challenging due to phenotypic variability and nystagmus-related limitations in clinical assessment, and can lead to misdiagnosis. We report a novel application of ultra-high-speed swept-source anterior segment OCT (SS-AS-OCT) to detect subtle phenotypic features not seen on routine slit-lamp or imaging.

Methods:
A patient with infantile nystagmus, grade 3 foveal hypoplasia, and SLC38A8 mutation (VA: 0.6 and 0.7 logMAR) underwent anterior segment imaging using a 400,000 A-scans/sec SS-AS-OCT. Post-processing included realignment algorithms to account for nystagmus and 3D reconstruction of anterior segment structures.

Results:
Standard slit-lamp examinations and prior anterior segment imaging failed to clearly identify anterior segment dysgenesis. SS-AS-OCT with three-dimension reconstruction revealed subtle iris processes: in the left eye, fine iris strands extended to and contacted the posterior corneal surface (iridocorneal strands); in the right eye, iris strands were visible in the angle. These findings were consistent with minor anterior segment dysgenesis, undetectable by conventional methods.

Conclusion:
Ultra-high-speed SS-AS-OCT with nystagmus-compensated post-processing enabled the detection of cryptic anterior segment anomalies in a patient with FHONDA, highlighting its utility in expanding the phenotypic spectrum and improving diagnostic accuracy in challenging cases. Utilising this technology in the assessment of patients with FHONDA may result in a stronger association between SLC38A8 mutations and anterior segment dysgenesis.

P-164

Clinical and electrophysiological phenotype of patients with oculo-cutaneous albinism type 8

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Introduction:
Albinism is a genetic disorder characterized by a defect in melanin biosynthesis. Affected patients' phenotypes are extremely heterogeneous, from severe forms with a diagnosis at birth based on dermatological signs, to many milder forms presenting as infantile nystagmus syndrome, from isolated to syndromic forms. Recently, variants in the *DCT* gene were showed to be responsible for a new type of oculocutaneous albinism (OCA) named OCA8. We report the ophthalmological, electrophysiological, and dermatological characteristics of three patients with genetically confirmed OCA8.

Methods:
This is a retrospective study of three patients with OCA8. Complete dermatological, ophthalmological, and orthoptic examinations were performed with clinical exploration and multimodal imaging. Visual evoked potentials (VEPs) were performed to characterize chiasmal decussation in two of the three patients.

Results:
The dermatological phenotype was mild, whereas all three patients exhibited infantile nystagmus syndrome with reduced visual acuity, foveal hypoplasia (grade 3), macular hypopigmentation (graded from 2 to 1), and iris transillumination (grade 3). Two patients could undergo a VEP examination; they both exhibited signs of strong chiasmal misrouting.

Conclusion:
Recently, pathogenic variants in the *DCT* gene were proven to cause OCA. Whereas patients with OCA8 exhibit a milder dermatological phenotype than others, their vision was initially described as impaired. The present report confirms previous findings and suggests that chiasmal misrouting is present in OCA8. This, together with recent findings in the murine model, supports the hypothesis that DCT regulates levels of L-Dopa and downstream signaling in the developing retina. These results convey critical future therapeutic implications.

P-165

Genome-Wide Insights into the Genes and Pathways Shaping Human Foveal Development

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Introduction:
Abnormal formation of the foveal pit is a hallmark of foveal hypoplasia (FH). FH is a key feature of several genetic eye conditions including albinism and *PAX6*-related aniridia. While rare high-penetrance variants in these conditions have been well described, the broader genetic architecture of foveal development, including the role of common and rare variants is poorly understood. To better understand the genetic determinants shaping human foveal development, we conducted the first genome-wide association study (GWAS) of foveal pit depth.

Methods:
We applied a deep-learning pipeline to quantify foveal pit depth from right-eye OCT scans of 61,269 European individuals in the UK Biobank. A GWAS was performed to identify independently associated genetic variants ($P < 5 \times 10^{-8}$), followed by comprehensive variant-to-gene mapping. Additionally, rare single-variant association testing and rare-variant gene-based analysis were conducted using whole-exome sequencing data from 59,313 individuals.

Results:
We identified 123 genetic signals, including 47 not previously associated with macular development. Variant-to-gene mapping prioritised 128 putative causal genes, revealing 64 novel associations with foveal development and two additional genes were implicated through rare-variant discovery analysis. The implicated genes include those involved in known foveal pathways such as retinal pigmentation and photoreceptor development, and also highlight roles for retinoic acid metabolism and cell fate determination in foveal morphogenesis.

Conclusion:
This study substantially advances our understanding of the genetic architecture underlying human foveal development. Our findings establish a foundation for future functional studies into the molecular mechanisms of FH and related visual disorders.

P-166

Whole genome sequencing for inherited retinal disorders, cataracts, and structural eye disorders: managing the unexpected through effective multi-disciplinary team working

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Introduction:
Since 2018, the UK NHS Genomic Medicine Service has enabled access to whole genome sequencing (WGS) for patients with retinal disorders, cataracts, and structural eye disorders. While analysis of large gene panels increases diagnostic yield, this comes with a concomitant increase in both incidental findings and the potential for genetic findings associated with syndromic eye disorders in those presenting with isolated eye findings. Coordination of care by the ophthalmic genetics multidisciplinary team (OG-MDT) is therefore critical in optimising patient pathways.

Methods:
Here we present three examples of families managed by the Great Ormond Street Hospital ophthalmology department. All underwent trio WGS through the North Thames Genomic Laboratory Hub and had input from the OG-MDT, comprising ophthalmologists, geneticists, genetic counsellors, clinical scientists and psychologists.

Results:
All probands had an unexpected diagnostic finding or an incidental finding identified through WGS. Following the OG-MDT, the genetic variants were interpreted and management plans were agreed. Results were then fed back with fully integrated genetic counselling and pre-coordinated management plans, ensuring that families were optimally supported in receiving a diagnosis.

Conclusion:
These cases highlight the essential role of the OG-MDT in managing complex cases involving unexpected findings. Coordination of clinical services via an OG-MDT permits efficient evaluation of complex variants, increases diagnostic yield, and ensures families are optimally supported through the testing pathway.

Paediatric ophthalmology never works Eye-lone (teamwork with other subspecialists in ophthalmology - surgery), Glaucoma in corneal genetic dysgenesis

P-167

XEN® Gel Stent for Glaucoma in Peters Anomaly: A Surgical Alternative

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Introduction:
Peters anomaly is a rare congenital anterior segment dysgenesis, characterized by central corneal opacity, iridocorneal adhesions, and developmental abnormalities of the anterior chamber angle. Secondary glaucoma management is challenging in young patients as it requires multiple surgeries over their lifetime. Surgical strategies must consider the altered anatomy and ensure effective intraocular pressure (IOP) control.

Methods:
A 17-year-old patient with bilateral megalocornea, right eye central corneal opacity, and temporally stretched pupil was diagnosed with Peters anomaly type I at birth. Despite maximal medical therapy, IOP was 26 mmHg in the right eye (OD) and 18 mmHg in the left (OS). Gonioscopy showed an open angle superiorly (Shaffer grade 3). Due to thin sclera and anterior segment dysgenesis, a less invasive filtering procedure was chosen: XEN® Gel Stent implantation via an ab externo approach, combined with intraoperative subconjunctival MitomycinC 0.2 mg/ml.

Results:
At 10 days postoperatively, a functioning, slightly hyperemic bleb was observed with an IOP of 8 mmHg. Topical corticosteroid therapy (desamethasone 0.2% QID) was continued. At one month, corrected visual acuity improved to 5/10 (pinhole), and IOP remained stable at 8 mmHg. At three months, IOP control was maintained without additional surgical interventions.

Conclusion:
Due to anatomical alterations, managing secondary glaucoma in Peters anomaly is complex. In this case, a sufficiently open angle allowed for successful XEN® Gel Stent implantation, preserving the conjunctiva for future procedures. This is the first reported use of the device in Peters anomaly. Further studies are needed to confirm the safety and efficacy of this approach.

P-168

Unexpected Atalamia After Buckling Surgery in a Pediatric Eye with Marfan Syndrome and Scleral-Fixated IOLs

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Introduction:
To report the surgical management of a macula-off retinal detachment (RD) in a monocular pediatric patient with Marfan syndrome and scleral-fixated intraocular lenses (SF-IOLs).

Methods:
A 14-year-old male with Marfan syndrome presented with sudden, painless vision loss in his only functional eye. The left eye had no light perception due to longstanding RD. The patient had undergone bilateral lens extraction for lens subluxation and SF-IOL implantation at age 5. Fundus examination revealed in the right eye an inferior macula-off retinal detachment with pigmented clumps and suspected microholes, without visible breaks. A 360° scleral buckling surgery utilising the Drainage-Air-Cryotherapy-Explant (D-ACE) technique was performed.

Results:
Initial postoperative findings showed a flat retina with residual subretinal fluid. On day 3, the patient developed total atalamia without signs of wound leak or choroidal detachment. OCT imaging revealed diffuse posterior endothelial-iris contact with no visibility of the SF-IOL plate, suggesting air entrapment between the iris, IOL, and residual vitreous from a prior incomplete vitrectomy. Emergency reintervention included intracameral viscoelastic injection and 30-G needle aspiration of the trapped air. Postoperative hypotony and transient corneal oedema were resolved within days. Three weeks post-op, the subfoveal fluid was fully reabsorbed, the retina remained attached, and BCVA returned to baseline.

Conclusion:
Retinal detachment in paediatric Marfan patients with prior SF-IOLs presents unique anatomical and surgical challenges. Entrapped intraocular air between the iris and SF-IOL is an uncommon cause of anterior chamber flattening, and prompt surgical management is essential to preserve visual function.

P-169

Cost of myopia in UK and France

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Introduction:
Decisions about public funding for myopia control interventions in children and young people depend on long-term cost implications, f.ex. cost reductions for management of myopia-associated complications.

In order to support a health-economical argument, we modelled lifetime costs of myopia in UK and France for 5 scenarios: conventional management (Single-Vision-Lenses, SVL), low-concentration atropine, myopia-control spectacles (MCS) and contact lenses, and orthokeratology.

Methods:
Each modelled scenario began with an 8-year-old child with -0.75DS. We used natural progression data to determine the likelihood of possible refractive outcomes with risk of faster/slower progression. We collected care costs from published sources, key informants, and informal surveys. We estimated and compared lifetime cost under each scenario and calculated cost-ratios as myopia-control cost divided by conventional-care cost.

Results:
With SVL, estimated lifetime cost of myopia in the UK is US$48,170/US$29,664 with faster/slower progression (France: US$32,492/US$22,606). Cost ratios for myopia-control options in the UK range from 0.50-0.69/0.73-1.00 with faster/slower progression (France: 0.60-0.81/0.81-1.10). MCS provide the greatest cost savings. Girls/women incur higher lifetime costs due to higher contact lens wear rates, prevalence of vision impairment and longer life expectancy.

Conclusion:
Limitations: we excluded myopia-prevention measures, such as increasing time outdoors, which would reduce lifetime cost, and recent light-based interventions, due to lack of UK/European data.

Myopia control during childhood likely reduces total lifetime cost of myopia, probably by reducing progression, simpler corrective lenses and reduced risk of complications and vision loss. The economic advantage is greatest for those with fast progression.

P-170

CHAMP-UK: a phase 3 randomised controlled trial of 0.01% atropine for myopia in the UK

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Introduction:
Whilst optical myopia-control interventions see increasing uptake across Europe, low-concentration atropine (LCA) is less frequently used, in-part because of lack of data from European populations and lack of a commercial preparation. CHAMP-UK aimed to evaluate effectiveness, safety and tolerability of 0.01% atropine and explore its mechanism of action.

Methods:
289 children age 6-12 years with myopia of at least -0.50D in both eyes and best-corrected visual acuity of 0.2 logMAR or better took part in a randomised controlled trial with 2:1 allocation ratio to atropine/placebo at 5 centres across the UK. Adherence was measured using electronic MEMS-CAPS.

Results:
Mean age at randomisation was 9.3 years in both arms; most children were White (>70%). Mean change in cycloplegic spherical equivalent from baseline to 24 months was -0.39D ± 0.63 with atropine and -0.70D ± 0.67 with placebo ($p<0.001$), adjusted mean difference 0.33D (95% CI 0.17, 0.49), intention-to-treat-analysis. Adjusted mean difference in axial length was -0.14mm (95% CI (-0.21, -0.07). Progression of less than -0.25D over 2 years was observed in 40% of children on 0.01% atropine vs 19% on placebo. Mean adherence was 80% in both groups, with good tolerability.

Conclusion:
In the CHAMP-UK trial, Atropine 0.01% had greater efficacy than in the MOSAIC trial (Ireland) and WA-ATOM (Australia), and was comparable to that reported in the 2025 Cochrane meta-analysis. It was safe and well-tolerated. Future research should explore whether higher concentrations and combination with optical myopia-control interventions may increase efficacy.

Team- and network for diagnostics (from imaging to diagnosis)

P-171

The Role of Photobiomodulation Therapy (PBMT) in Paediatric Ophthalmology: A Scoping Review

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Introduction:
Photobiomodulation therapy (PBMT) is a non-invasive intervention that uses specific wavelengths of light to stimulate cellular chromophores, activating biological processes with potential therapeutic applications. PBMT has been used for various childhood eye/vision conditions. This scoping review maps the current evidence on applications, efficacy, and safety.

Methods:
We carried out a scoping review following PRISMA guidelines: systematic search of Ovid MEDLINE, Embase and Cochrane Libraries, staged review within Covidence software, with two independent reviewers screening titles/abstracts, then full texts for inclusion based on predefined criteria. We included primary research studies in English language with children under 18 years with eye, vision, lid, or orbital conditions treated with PBMT, low light therapy, or intense pulsed light therapy (400–1100 nm spectral range). Case reports, meta-analyses, systematic reviews, and studies using photodynamic therapy, cross-linking, or optogenetics were excluded.

Results:
Of 5,081 identified studies, 47 were included. The majority focused on myopia control, reporting reductions in axial elongation, some noted rebound effects upon cessation. Amblyopia, retinopathy of prematurity, meibomian gland dysfunction, chalazion, concussion, and visual fatigue were also investigated. Most studies were randomized controlled trials, with China as the predominant investigating region. PBMT demonstrated potential benefits across various conditions with a favourable safety profile.

Conclusion:
PBMT may have a role in the management of childhood eye conditions, particularly myopia and blepharitis. It appears generally safe, with transient adverse effects such as mild photophobia and dry eye. Further research is needed to optimize protocols and assess sustained efficacy and safety for widespread clinical adoption.

P-172

Hyperautofluorescent parafoveal ring in children with rod-cone dystrophy and literature review

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Introduction:
Autofluorescence is an increasingly useful tool in the paediatric ophthalmologist's diagnostic toolkit. A blue light is used to excite natural fluorophores in the retina which then further emit light. Patterns of autofluorescence have the ability to aide diagnosis and determine prognosis, as has been reported in retinitis pigmentosa. We report on a series of three children with hyperautofluorescent parafoveal rings with three different genetic variants for retinal dystrophy.

Methods:
This is a retrospective review of 3 children from a paediatric ophthalmology clinic in the UK. Clinical data and results of retinal imaging including autofluorescence and OCT macula are reported. Review of the literature using Medline database was completed. Genetic variants are tabulated.

Results:
All 3 children with exhibited a similar pattern and location of parafoveal hyperautofluorescent rings, specifically around the central macula but non-foveal involving. Vision ranged between -0.14 to 0.42 LogMAR in the better seeing eye of each patient. Fundoscopy in two patients demonstrated peripheral hyperpigmentation in keeping with bone spicule retinopathy found in retinitis pigmentosa. In our third patient, there were no peripheral retinal changes found and the macula appeared healthy on slitlamp examination. This patient had a diagnosis of Roifman syndrome associated with cone-rod dystrophy.

Recent reports in the literature have highlighted the hyperautofluorescent ring as a prognostic marker for slower progression in the context of retinitis pigmentosa.

Conclusion:
Our report demonstrates the utility of autofluorescence in children. Particularly the cohort who report unexplained reduced vision in the dark or light conditions with minimal retinal findings.

P-173

Battered child syndrome in the practice of an ophthalmologist - a case report.

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Introduction:

Battered child syndrome refers to the clinical and psychological condition observed in children who have experienced repeated and severe physical abuse, often from a caregiver. We would like to present the case of a 4-month-old child who experienced sudden cardiac arrest and, due to the cooperation and examination of an ophthalmologist and other specialists, was diagnosed with injuries resulting from domestic violence.

Methods:
Case description

A 4-month-old child was brought to emergency department after sudden cardiac arrest at home. It turned out that the patient had left the pediatric neurology department 10 days earlier due to intracerebral hematomas and minor preretinal hemorrhages of both eyes. There was no suspicion of battered child syndrome. Current examination revealed bruising of the right upper limb, bleeding in the projection of previous intracerebral hematomas and a new hematoma in the occipital lobe. Ophthalmological evaluation showed bleeding into the vitreous body and multiple pre- and intraretinal hemorrhages in both eyes.

Results:
After receiving appropriate treatment, including anticoagulants, corticosteroids and anti-inflammatory drugs, the child regained full function and normal status of the eyes. The parents were deprived custody of this child and his siblings.

Conclusion:
It is necessary to consider how to improve the effectiveness of early recognition of the battered child syndrome, so as to help our young patients at an early stage. All systems and organs can be affected, therefore, cooperation between doctors of different specialties is required.

P-174

Comparative Analysis of Autorefractometry Measurements Before and After Cycloplegia Across Age Groups

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Introduction:
Accurate assessment of refractive errors is fundamental to the diagnosis and management of visual disturbances, especially in pediatric populations where accommodation can significantly influence refraction measurements.This study aimed to evaluate the changes in autorefractometry measurements before and after cycloplegia across different age groups and refractive error types, and to determine the clinical importance of cycloplegia in routine ophthalmic practice.

Methods:
This study included 80 participants aged 5 to 40 years, stratified into three age groups: children (5–12 years), adolescents (13–18 years), and adults (19–40 years). Each subject underwent autorefractometry using a NIDEK ARK-1 device, first without cycloplegia and then 30 minutes after instillation of 1% cyclopentolate hydrochloride.Data were analyzed to assess mean differences in SE before and after cycloplegia and were compared across age groups and refractive categories.

Results:
Results showed that cycloplegia significantly impacted refractive measurements, particularly in hyperopic eyes and pediatric patients. Mean SE increased by +1.25 D in hyperopic, +0.25 D in emmetropic, and +0.50 D in myopic eyes. Hyperopic children demonstrated the greatest changes, up to +1.75 D, while adults showed minimal variation. The findings emphasize the diagnostic value of cycloplegic refraction in detecting latent hyperopia and accommodative spasm in younger individuals.

Conclusion:
Cycloplegia is essential in pediatric ophthalmology for accurate refractive assessment and should be routinely used in this group. In adults, cycloplegia may be applied selectively based on clinical need. Age-specific protocols can enhance diagnostic accuracy and optimize vision correction strategies.

Keywords: Cycloplegia, autorefractometry, refractive error, hyperopia, myopia, pediatric ophthalmology, spherical equivalent.

P-175

Retinal Imaging Study of Children (RISC): Findings from the first 334 children

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Introduction:
This study aims to explore the occurrence and patterns of retinal haemorrhages (RHs) in children with various clinical conditions that are suggested as differential diagnoses to abusive head trauma.

Methods:
A Swedish prospective multicenter study involving Stockholm, Gothenburg and Lund. Children are consecutively included for fundoscopy and fundus photography, using a handheld fundus camera. The study will continue until 1000 children have been examined, and include children from the following three groups:

All infants ≤ 18 months of age who undergo an elective brain ultrasound examination on the indication increased head circumference.

All critically ill children < 15 years of age, with neurological symptoms, that require neuroradiological examination of the brain.

All children < 2 years of age, who seek care at the emergency department and undergo emergency neuroradiological examinations of the head.

Results:
Of the 334 children included so far, 27 (8%) had RHs. All RHs were few in numbers except in one child with Alagille syndrome. Seven (26%) of the children with RHs had an elevated intracranial pressure due to various conditions, and the RHs occurred with concomitant papilledema. Cerebral haemorrhage due to ruptured arteriovenous malformations, aneurysms and pesudoaneurysms was the most common associated condition (33%). Notably, no children with high-energy trauma, such as those involved in traffic accidents, exhibited RHs

Conclusion:
Retinal haemorrhages were rare, occurring in 8% of the children and, when present, were few in number. Clinical conditions associated with RHs were identified in all cases, except in two.

Team- and network in rehabilitation for the visually impaired

P-176

Visual Function in Paediatric Rehabilitation – A Retrospective Analysis

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Introduction:
One of the key factors for enhancing outcomes in neurorehabilitation of patients with congenital or acquired brain injury is optimal sensory function and identification of possible visual deficits. Awareness of cerebral visual impairment (CVI) is therefore of utmost importance in the rehabilitation process.

Purpose: To investigate the incidence and nature of vision disorders in children admitted to a rehabilitation centre over a five-year period.

Methods:
Retrospective analysis of inpatients at the Swiss Children's Rehab who received an ophthalmological consultation between January 2019 and December 2024. Electronic chart review was performed in patients with consent for the study. Analysed data: reasons for admission to the rehab centre, corresponding ophthalmologic findings, demographics. The study was approved by the local Ethics committee.

Results:
A total of 229 consultations from 163 patients were reviewed. The two most common reasons for admission were inflammatory (36/163) and traumatic (41/163) conditions. A congenital disease was present in 30 patients. Most of the children had an acquired disorder (133/163). Pathological ophthalmologic findings were present in 99/163 children examined. The most common findings were reduced vision (26/163) followed by CVI (22/163) and refractive errors (20/263). Traumatic causes were most often associated with oculomotor disorders (10/41) and CVI (6/41). Follow-up examination showed a change of the ophthalmologic findings in 13/66 patients.

Conclusion:
Pathological ophthalmological examination was present in 61% of the children examined at Swiss Children's Rehab. CVI was prevalent in 22% of this cohort. Ophthalmologic evaluation and low vision assessment is recommended as a routine part of the comprehensive rehabilitation process.

Team- and network with neonatology & panel discussion 'The best way to treat and follow up on ROP'

P-177

Follow-up Study on Ridge-Adjacent Laser Therapy with Imaging and Dexamethasone for Retinopathy of Prematurity: Outcomes at 2.5 Years of Age

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Introduction:
Retinopathy of prematurity (ROP) is a leading cause of childhood blindness. Retinal laser therapy, which involves applying a large number of laser spots to the entire avascular retina, is a well-established treatment. Since 2016, paediatric ophthalmologists at Skåne University Hospital in Lund have modified this treatment by restricting the laser spots to a limited avascular area of the retina near the ridge, sparing the periphery. This approach is known as ridge-adjacent laser therapy (RALT). RALT is used in conjunction with intraoperative wide-field imaging and postoperative dexamethasone eye drops. This follow-up study, conducted at 2.5 years of age, presents the structural and functional outcomes of eyes treated with RALT.

Methods:
A retrospective review of medical records was conducted for all infants treated with RALT for ROP between June 2016 and June 2021 in Lund. Data from the 2.5-year follow-up were extracted from the Swedish ROP quality register.

Results:
Sixty-one eyes from 31 infants were treated with RALT. However, data from three children were missing at the 2.5-year follow-up. The visual function was perceived as normal in 20 children (71%). An unfavourable structural outcome was observed in two eyes (4%). The mean spherical equivalent was -1.1 dioptres (SD 2.7D). Nine children (32%) developed strabismus, and 11 children (39%) required glasses.

Conclusion:
The majority of children treated for ROP with RALT had developed a favourable visual function at 2.5 years of age.


Team- & Network in Paediatric Ophthalmology