

# EPOS 2002

## Figueira da Foz, Portugal

### Final Program

#### L/P/C Author

#### Abstract-Title

**Friday, 12:00 - 2:00pm Registration**

**Friday, 2:00pm Opening**

**Friday, 2:15 pm**

L	Michael Baraitser	<a href="#">The eye in the Dysmorphology Databases.</a>
L	Kerstin Strömmland	<a href="#">Using ocular dysmorphology and associated anomalies in timing birth defects in syndromes.</a>
SP	Eduardo Silva	<a href="#">Defining the minimal critical region for the Peters plus syndrome.</a>
C	Badia Fahad	<a href="#">Cranio-ectodermal dysplasia - Sensenbrenner syndrome: A case presentation.</a>
C	Christopher Williams	<a href="#">A New Case of ocular abnormalities and "Apple-Peel" intestinal atresia with multiple variable aneuploidy</a>
C	Shauna Quinn	<a href="#">Autosomal dominant brachydactyly, coloboma and anterior segment dysgenesis.</a>
C	Jin Hong	<a href="#">Bilateral Peters anomaly, failure to thrive and primary hypoadosteronism.</a>
C	Lilianne Duarte	<a href="#">Hypomelanosis of Ito. Case report.</a>
C	Bettina Wabbels	<a href="#">An unusual case of complex ocular and eyelid malformations.</a>
C	Pedro Faria	<a href="#">Bilateral congenital anophthalmia with cyst associated with severe neurodegeneration. A new syndrome or association?</a>

**Friday, 4:45pm**

L	Veronica van Heyningen	<a href="#">Transcription factors and eye development.</a>
L	Elfride De Baere	<a href="#">FOXL2 AND BPES: mutational hotspots, intrafamilial variability and revision of genotype-phenotype correlation.</a>
P	Hélène Dollfus	<a href="#">FOXL2 and TWIST mutations in families with developmental anomalies of the eyelids.</a>
P	Stella Hornby	<a href="#">Aetiology of ocular coloboma in South India.</a>

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| C | Peter Hodgkins          | <a href="#">Papillo-renal syndrome: A new PAX2 mutation.</a> |
| C | João Rodrigues          | <a href="#">Coloboma: clinical cases.</a>                    |
| P | Mary J. van Schooneveld | <a href="#">Morning glory syndrome.</a>                      |

**Saturday, 8:00am Registration**

**Saturday, 8:30am**

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|---|---------------------|--|
| L | Dagmar Wieczorek    | <a href="#">Call for patients - The Oculo-Auriculo-Vertebral Spectrum (OAVS) - a clinical and molecular project.</a>                                       |
| C | Ana Xavier          | <a href="#">A case of Moebius-Poland sequence: where did it start?</a>   |
| C | Teresa Torrent      | <a href="#">Moebius-like syndrome or another diagnostic challenge.</a>   |
| C | Michael Michaelides | <a href="#">A Novel case of Hypertelorism, Hypospadias, Strabismus and bilateral Congenital Lacrimal Fistulae</a>  |
| C | Tae Yoon La         | <a href="#">A Case of Patau Syndrome with Congenital Ocular Anomaly</a>  |
| C | Rosa Sust           | <a href="#">Wolf-Hirschhorn-Like Syndrome associated with a complex rearrangement of chromosome 4.</a>   |
| C | Ingele Casteels     | <a href="#">Familial inherited microtia, ocular colobomas and imperforation of the nasolacrimal ducts with abnormal chromosome condensation on 4p16.1.</a> |
| C | Ligia Cardoso       | <a href="#">Just another case of cerebral palsy or beyond?</a>   |

**Saturday, 10:30am**

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| P | Irene Maumanee        | Finding Genes in Leber Congenital Amaurosis  |
| P | Markus Preising       | <a href="#">Screening of NUB1 in Patients with Leber congenital amaurosis.</a>   |
| P | Astrid Plomp          | <a href="#">Does autosomal dominant pseudoxanthoma elasticum exist?</a>  |
| P | Marie-Claire Gaillard | <a href="#">Heterogeneity of retinal degeneration in cobalamin C disease methylmalonic aciduria with homocystinuria.</a> |
| P | Hansjörg Soeldner     | <a href="#">Retinal dystrophy in long chain 3-hydroxy-acyl-coA dehydrogenase deficiency</a>                              |
| C | Michel Michaelides    | <a href="#">Oligocone Trichromacy</a>  |
| C | Augusto Magalhães     | <a href="#">Ocular aspects of Aicardi's syndrome.</a>  |

**Saturday, 12:00am**

## **BOARD MEETING**

### ***Saturday, 2:30pm***

- P Caroline Gardiner [Cataract extraction +/- intraocular lens implantation in a series of infants and children with Down's syndrome.](#)
- P Joaquim N Murta [Traumatic cataract in children. Functional results.](#)
- P Isabel Prieto [Pediatric cataracts : our surgical experience.](#)
- SP Joaquim N Murta [Cataract surgery in children with uveitis.](#)
- P Filomena Ribeiro [Uveitis in children.](#)
- SP Branka Stirn Kranjc [Orbital myositis.](#)
- C Michael O'Keefe [Choroidal haemangioma associated with Sturge-Weber Syndrome treated with low dose ocular irradiation.](#)
- P M. Schittkowski [Treatment of congenital anophthalmos with self-inflating hydrogel expanders.](#)
- P Christoph Hintschich [Enucleation in childhood - a threat for bony orbital development in humans?](#)
- C Catherine G Brozou [Macrophthalmus associated with Neurofibromatosis type 1](#)

### ***Saturday, 5:00pm***

- P Nel Tijmes [A survey of the visual performance of mentally disabled people in institutions.](#)
- P Sajid Khan [Visual outcomes and amblyogenic risk factors in syndromic craniosynostosis - a review of 141 cases.](#)
- P Pierre Bitoun [Autism in congenitally blind children? Genetic or environmental.](#)
- P Marita Andersson Grönlund [Visual function and ocular morphology in healthy term children from 4 to 16 years.](#)
- C Eva Aring [Orthoptic findings In healthy term children from 4 To 16 years](#)
- P Susann Andersson [Visual acuity and visual perception in children with operated hydrocephalus.](#)
- P Elisabeth Moser [Cohen Syndrome - a possible differential diagnosis in retarded children with high myopia.](#)

### ***Sunday, 9:00am***

- P Susana Teixeira [Retinopathy of Prematurity: a retrospective case-control study in a neonatal intensive care unit.](#)
- P Natalya Fomina [The main risk factors for IV-V stages of Retinopathy of Prematurity.](#)

P	Gerd Holmström	<a href="#"><u>Incidence of ROP in two consecutive Swedish population-based studies.</u></a>
SP	Eva Larsson	<a href="#"><u>Peripheral and central visual fields in prematurely born children and in full-term controls</u></a>
C	Maria João Santos	<a href="#"><u>Coats' disease - a clinical case</u></a>
SP	Catherine Marsh	<a href="#"><u>Unilateral PHPV: A series of cases.</u></a>
C	Nicoline Schalijs-Delfos	<a href="#"><u>Uncommon presentation of retinoblastoma.</u></a>
<b>Sunday, 11:00am</b>		
C	Sharola Dharmaraj	<a href="#"><u>Ocular findings in proximal trisomy 1q mosaicism</u></a>
C	Susana Teixeira	<a href="#"><u>Facial and orbital haemangioma associated to an intracranial complex vascular malformation - Case Report</u></a>
C	Fernando Trancoso Vaz	<a href="#"><u>Unifocal Langerhans' Cell Histiocytosis</u></a>
C	Concepcion Ferrer Novella	<a href="#"><u>Hurler Syndrome: Early diagnosis starting from ophthalmological manifestations</u></a>
C	Sonia Campos	<a href="#"><u>Bilateral retinal dysplasia, developmental delay and autistic behaviour in two brothers. Another challenging case report.</u></a>
P	Ayd Shafiq	<a href="#"><u>Linkage analysis in a Liverpool family with dominantly inherited microcornea and cataract.</u></a>
C	Jorge Breda	<a href="#"><u>Ocular manifestations of congenital sleep disease.</u></a>
P	Susana Teixeira	<a href="#"><u>Paediatric AIDS</u></a>
C	Inmaculada González Viejo	<a href="#"><u>Ectropion uveae: Clinical manifestations</u></a>
<b>Sunday, 12:30pm Closing</b>		

*L = Lecture, P = Paper, C = Case report, SP = Short paper*