



Main Sessions – Preliminary Program

Of note:

- Times indicated include 5 mins of Q&A.
- Chairs will be free to add a 15-minute break during their respective session when deemed necessary.

Wednesday November 16th – Genetics and Genotype-Phenotype Correlations

2pm-5pm (CET)*

Opening words: Antoine GLIKSOHN – Global Albinism Alliance
Muluka-Anne MITI DRUMMOND – UN Independent Expert on Albinism
Lluís MONTOLIU, Maria VAN GENDEREN – Scientific Advisory Committee

Session Chairs: Benoît ARVEILER, Karen GRØNSKOV

- **Stacie LOFTUS** (Bethesda, MD, USA): *Hiding in Plain Sight: A Rare Haplotype Comprised of Common Variants TYR p.Ser192Tyr and p.Arg402Gln is a Major Contributing Allele for OCA1B* – 15min
- **Panagiotis SERGOUNIOTIS** (Manchester, UK): *The Contribution of Common Regulatory and Protein-Coding TYR Variants to the Genetic Architecture of Albinism* – 20min
- **Vincent MICHAUD** (Bordeaux, France): *Unsuspected Consequences of Synonymous or Missense Variants in OCA2 can be Detected in Blood Samples* – 20min
- **Charlotte KRUIJT** (Leiden, the Netherlands): *The Remarkable Phenotypic Variability of OCA4* – 15min
- **Fanny MORICE-PICARD** (Bordeaux, France): *Oculo-Cutaneous Albinism Type 4 (OCA-4): Phenotype-Genotype Correlation* – 15min
- **Mervyn THOMAS** (Leicester, UK): *Genotypic and Phenotypic Spectrum of Foveal Hypoplasia: A Multicenter Study on behalf of the Foveal Development Investigators Group* – 20min
- **Anat BLUMENFELD** (Jerusalem, Israel): *Albinism in Israel: clinical and molecular evaluation* – 15min
- **Michelle ZEPPELIN** (Santiago, Chile): *Sociodemographic, Genotypic and Phenotypic Characterization of the Population with Albinism in Chile Through Self-Report* – 15min

Thursday November 17th – Cell Biology and Cellular & Animal Models

2pm-5pm (CET)*

Session Chairs: Lluís MONTOLIU and Michael MARKS

- Michael MARKS (Philadelphia, PA, USA): ***Melanosome Biogenesis and the Cell Biology of the Hermansky-Pudlak Syndromes*** – 20min
- Alistair HUME (Nottingham, UK): ***The Dynamics of Melanosomes in Melanocytes: Organelles Using Short Tracks for Long Trips?*** – 20min
- Jonathan ZIPPIN (New York City, NY, USA): ***cAMP Signaling and Melanosome pH in the Regulation of Pigmentation*** – 20min
- Elena OANCEA (Providence, RI, USA): ***The chloride/proton exchanger CLC7 functions in melanocytes as a negative regulator of human pigmentation*** – 20min
- Santiago DI PIETRO (Fort Collins, CO, USA): ***Role of LRMDA, the Product of the Gene Mutated in OCA7, in Pigmentation*** – 20min
- Cédric DELEVOYE (Paris, France): ***Cell Biology of Keratinocyte Pigmentation: From Pigment Positioning to DNA Photoprotection*** – 20min
- Lluís MONTOLIU (Madrid, Spain): ***Phenotyping New Mouse Gene-Edited CRISPR Lines of Albinism with Patient-Specific Mutations*** – 20min
- Brian BROOKS (Bethesda, MD, USA): ***Modeling of Human Oculocutaneous Albinism Using iPSC-Derived RPE*** – 20min

Friday November 18th – Visual Pathophysiology and Therapies

2pm-5pm (CET)*

Session Chairs: Maria VAN GENDEREN and Alexandra REBSAM

- Joseph CARROLL (Milwaukee, WI, USA): ***Advances in Treatment for Inherited Retinal Degeneration: Implications for Albinism?*** – 25min
- Carol MASON (New York City, NY, USA): ***CyclinD2-Mediated Regulation of Neurogenic Output from the Retinal Ciliary Margin is Perturbed in Albinism*** – 25min
- Alexandra REBSAM (Paris, France): ***Different Mouse Models to Unravel the Physiopathology of Albinism: What We Learn from 3D Retinal Projections*** – 20min
- Virginie CHOTARD (Paris, France): ***Functional Responses of the Retina and Structure of the Visual Projections in Albino Mice*** – 20min
- Helena LEE (Southampton, UK): ***Oral L-DOPA Rescues Retinal Morphology and Visual Function in a Murine Model of Human Albinism*** – 20min
- Sophie JAVERZAT (Bordeaux, France): ***Impaired Retinogenesis in Patients with Albinism: Does it all have to do with L-Dopa?*** – 25min
- Arthur BERGEN (Amsterdam, the Netherlands): ***The Retinal Pigmentation Pathway in Human Albinism*** – 25min

Saturday November 19th – Epidemiology, Ophthalmology, Dermatology and Healthcare Management

2pm-5pm (CET)*

Session Chairs: Fanny MORICE-PICARD and Irene GOTTLÖB

- Jennifer KROMBERG (Johannesburg, South Africa): ***Worldwide Prevalence of Oculocutaneous Albinism: A Systematic Review*** – 20min
- Maria VAN GENDEREN (Zeist, the Netherlands): ***The Influence of Different Types of Nystagmus in Patients with Albinism on the Pattern Reversal VEP*** – 20min
- Michael HOFFMANN (Magdeburg, Germany): ***Towards an OCT-Biomarker for Albinism Diagnostics*** – 20min
- Dayong BAI (Beijing, China): ***Evaluation of Visual Function in 84 Children with Albinism*** – 15min
- Dongsheng YANG (Jinan, China): ***Vision Rehabilitation for Patients with Albinism: Effect of Dark Exposure*** – 15min
- Panawé KASSANG (Lomé, Togo): ***Skin Cancers in Persons with Albinism: An Analysis of the Literature*** – 15min
- Herbert KIRCHESCH (Cologne, Germany): ***Skin Cancer in Patients with Albinism in a European Series*** – 15min
- Harry FREELAND (London, UK): ***A Model of Intervention to Strengthen Health and Wellbeing for People with Albinism in Africa*** – 25min

Closing remarks Antoine GLIKSOHN – Global Albinism Alliance
Lluís MONTOLIU, Maria VAN GENDEREN – Scientific Advisory Committee

With the financial support of:

