

## Main Sessions – Preliminary Program

Of note: Times indicated include Q&A.

### Tuesday January 21<sup>st</sup> Genetics

2pm-5pm (CET)\*

**Opening words:** Antoine GLIKSOHN – Global Albinism Alliance 5 min

**Session Chairs:** Benoît ARVEILER and David ADAMS

- **Defining the spectrum of pathogenic variants in oculocutaneous albinism 2 (OCA2)** 20 min  
Stacie LOFTUS (Bethesda, MD, USA)
- **OCA2 exon 10 splicing: of mice and men** 20 min  
Sophie JAVERZAT (Bordeaux, France)
- **Multiplex assays of variant effect and Re-classification of TYR variants in Chinese oculocutaneous albinism patients** 20 min  
Shushu LV (Beijing, China)
- **Update on the Albinism Variant Curation Expert Panel** 10 min  
David R ADAMS (Bethesda, MD, USA)

#### Break

- **Oculocutaneous albinism variants in 28 consanguineous families and functional classification of a pathogenic deep intron variant in TYR** 20 min  
Karen GRØNSKOV (Copenhagen, Denmark)
- **Insights into the genetic architecture of albinism** 20 min  
Panos SERGOUNIOTIS (Manchester, UK)
- **Digenic Inheritance of pigment genes** 20 min  
Yizhen WANG (Beijing, China)
- **TPCN2, a new albinism gene?** 10 min  
Cécile COURDIER (Bordeaux, France)
- **Understanding the genetic heterogeneity of albinism** 20 min  
Lluís MONTOLIU (Madrid, Spain)

# Wednesday January 22<sup>nd</sup>

## Genotype-Phenotype Correlations & Cell Biology

2pm-5pm (CET)\*

**Session Chairs:** Michael MARKS and Karen GRØNSKOV

- **New experimental approaches to better understand and diagnose oculocutaneous albinism** 15 min  
Vincent MICHAUD (Bordeaux, France)
- **Hermansky-Pudlak syndrome in Greece and Cyprus: The “Goldberg(s)” Rodopi and Troodos variations** 10 min  
Effrosyni MANALI (Athens, Greece)
- **Non-coding variants involved in the diagnosis of albinism** 20 min  
Modibo DIALLO (Bordeaux, France)
- **OCA7 or OA2?** 10 min  
Charlotte KRUIJT (Zeist, The Netherlands)
- **Control of skin pigmentation by TMEM138 and the primary cilium** 20 min  
Michael MARKS (Philadelphie, PA, USA)
- **Phenotype-genotype correlation in a cohort of Israeli patients with OCA1 albinism** 5 min  
Or SHMUELI (Jerusalem, Israel)

### Break

- **The Albinism International Databank Project** 10 min  
Donna APPELL and Karen BLY (USA)
- **Griscelli syndrome in vitro model provides insights into melanin transfer pathways** 20 min  
Duarte BARRAL (Lisbon, Portugal)
- **Photoprotection of keratinocytes: Cytoskeletal mechanics in pigment organelle positioning** 20 min  
Cédric DELEVOYE (Paris, France)
- **Characterization of novel pH-sensitive probes of the melanosome** 10 min  
Tyler WILLS (New York, NY, USA)
- **Impact of the R356Q mutation on stability and function of human recombinant intra-melanosomal domain of TYRP1** 5 min  
Waleed SABIR (Bethesda, MD, USA)
- **Human recombinant tyrosinase and OCA1-related variants reveal mutations' molecular destabilization mechanism** 5 min  
Sarah TOAY (Bethesda, MD, USA)

# Thursday January 23<sup>rd</sup>

## Visual function and pathophysiology, Rehabilitation & Therapies

2pm-5pm (CET)\*

**Session Chairs:** Brian BROOKS and Mervyn THOMAS

- **Quo Vadis? – Visual motion perception in albinism** 20 min  
Michael B. HOFFMANN (Magdeburg, Germany)
- **Selective visual function deficits and sparing in people with albinism** 10 min  
Nicole ROSS (Boston, MA, USA)
- **Altered retinal neurogenesis leads to decussation defects in the albino mouse:  
Role of the RPE?** 20 min  
Carol MASON (New York, NY, USA)
- **RPE and retinal communication in development** 15 min  
Paige HELMER (New York, NY, USA)
- **Evaluating the prevalence and associated factors of sleep problems in adults  
with albinism - First results of an international web-based study** 5 min  
Eila van STIPRIAAN (Amsterdam, The Netherlands)
- Break**
- **Unraveling the physiopathology of visual deficits in albinism and FHONDA  
syndrome in mouse and human cellular models** 20 min  
Alexandra REBSAM (Paris, France)
- **Soluble adenylyl cyclase inhibition as a potential therapeutic approach for  
Oculocutaneous Albinism Type 2** 20 min  
Jonathan ZIPPIN (New York, NY, USA)
- **Ampyrone (4-Aminoantipyrine) activates human tyrosinase and increases  
melanin production** 15 min  
Yuhong Anna WANG (Bethesda, MA, USA)
- **Gene therapy strategy for oculocutaneous albinism type 1A** 20 min  
Aman GEORGE (Bethesda, MA, USA)
- **Chromatic Aberration Reductive Lenses (CARL): preliminary results of the  
clinical trial involving 10 patients with albinism** 10 min  
Caterina TRAMONTANA (Pavia, Italy)

**Friday January 24<sup>th</sup>**  
**Albinism in Low and Middle-Resource Tropical Settings**  
**Skin Cancer & Health management**

**2pm-5pm (CET)\***

**Session Chairs:** To be confirmed

- **Outreach clinics for people living with albinism in Burundi** 15 min  
Doriane SABUSHIMIKE (Bujumbura, Burundi)
- **Validating a pilot study: The use of a sunscreen and educational program tailored for people with albinism (PWA) in low- and middle-resource countries** 15 min  
Shirley PARRAGA (Winston-Salem, NC, USA)
- **First clinic for albinism in the Americas** 5 min  
Bary BIGAY MERCEDES (Dominican Republic)
- **Demographic and clinical profile of people with oculocutaneous albinism seen in a tertiary hospital in Lagos, South-West Nigeria** 10 min  
Olufolakemi COLE-ADEIFE (Lagos, Nigeria)
- **Key parameter to develop outstanding sunscreen formulation for people with albinism in low-income settings** 10 min  
Stephanie ACKER (Grenach Wyhlen, Germany)
- **Skin cancer in persons with albinism: development of a research program at IARC** 10 min  
Valerie MCCORMACK (Lyon, France)

**Break**

- **Mapping persons with albinism in Togo in 2024** 5 min  
Malanna BIGMA (Lomé Togo)
- **Children living with albinism In Africa, A Kenyan perspective** 10 min  
Prabha CHOKSEY (Nairobi, Kenya)
- **Health-related quality of life and factors associated with the prevalence of depression among people with oculocutaneous albinism in Jinja, Uganda. A cross sectional study** 10 min  
Gaylord INENA WA INENA (Kisangani, DRC)
- **Freedom of religion and beliefs about people with albinism in Uganda: an action learning study for Mid-Western Uganda** 10 min  
Ninsiima PHIONAH (Kampala, Uganda)
- **Skin cancer detection, requirements and requests for a dermatologist consulting persons with albinism, a European point of view** 10 min  
Herbert KIRCHESCH (Cologne, Germany)
- **Bioptic driving: an overview, update, and comparison of driver training methods** 10 min  
Charles HUSS (Dunbar, WV, USA)

**Closing remarks:** Antoine GLIKSOHN – Global Albinism Alliance

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