



## 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 (Bordeaux, France) and David ADAMS (Bethesda, MD, USA)

- **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)

\*CET: Central European Time (UTC+1). Make the appropriate adjustment according to your time zone.

## Wednesday January 22<sup>nd</sup> Genotype-Phenotype Correlations & Cell Biology

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

**Session Chairs:** Michael MARKS (Philadelphia, PA, USA) and Karen GRØNSKOV (Copenhagen, Denmark)

- **New experimental approaches to better understand and diagnose oculocutaneous albinism**  
Vincent MICHAUD (Bordeaux, France)

15 min
- **Hermansky-Pudlak syndrome in Greece and Cyprus: The “Goldberg(s)” Rodopi and Troodos variations**  
Effrosyni MANALI (Athens, Greece)

10 min
- **Non-coding variants involved in the diagnosis of albinism**  
Modibo DIALLO (Bordeaux, France)

20 min
- **OCA7 or OA2?**  
Charlotte KRUIJT (Zeist, The Netherlands)

10 min
- **Control of skin pigmentation by TMEM138 and the primary cilium**  
Brice MAGNE (Philadelphia, PA, USA)

20 min
- **Phenotype-genotype correlation in a cohort of Israeli patients with OCA1 albinism**  
Or SHMUELI (Jerusalem, Israel)

5 min

### Break

- **The Albinism International Databank Project**  
Donna APPELL and Karen BLY (USA)

10 min
- **Griscelli syndrome in vitro model provides insights into melanin transfer pathways**  
Duarte BARRAL (Lisbon, Portugal)

20 min
- **Photoprotection of keratinocytes: Cytoskeletal mechanics in pigment organelle positioning**  
Cédric DELEVOYE (Paris, France)

20 min
- **Characterization of novel pH-sensitive probes of the melanosome**  
Tyler WILLS (New York, NY, USA)

10 min
- **Impact of the R356Q mutation on stability and function of human recombinant intra-melanosomal domain of TYRP1**  
Waleed SABIR (Bethesda, MD, USA)

5 min
- **Human recombinant tyrosinase and OCA1-related variants reveal mutations' molecular destabilization mechanism**  
Sarah TOAY (Bethesda, MD, USA)

5 min

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

### Visual function and pathophysiology, Rehabilitation & Therapies

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

**Session Chairs:** Brian BROOKS (Bethesda, MD, USA) and Mervyn THOMAS (Leicester, UK)

- **Quo Vadis? – Visual motion perception in albinism**  
Michael B. HOFFMANN (Magdeburg, Germany)

20 min
- **Selective visual function deficits and sparing in people with albinism**  
Nicole ROSS (Boston, MA, USA)

10 min
- **Altered retinal neurogenesis leads to decussation defects in the albino mouse:  
Role of the RPE?**  
Carol MASON (New York, NY, USA)

20 min
- **RPE and retinal communication in development**  
Paige HELMER (New York, NY, USA)

15 min

**Break**

- **Unraveling the physiopathology of visual deficits in albinism and FHONDA syndrome in mouse and human cellular models**  
Alexandra REBSAM (Paris, France)

20 min
- **Soluble adenylyl cyclase inhibition as a potential therapeutic approach for Oculocutaneous Albinism Type 2**  
Jonathan ZIPPIN (New York, NY, USA)

20 min
- **Ampyrone (4-Aminoantipyrine) activates human tyrosinase and increases melanin production**  
Yuhong Anna WANG (Bethesda, MA, USA)

15 min
- **Gene therapy strategy for oculocutaneous albinism type 1A**  
Aman GEORGE (Bethesda, MA, USA)

20 min
- **Chromatic Aberration Reductive Lenses (CARL): preliminary results of the clinical trial involving 10 patients with albinism**  
Caterina TRAMONTANA (Pavia, Italy)

10 min

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

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

**Session Chairs:** Elizabeth ROBERTS (Leicester, UK) and Fanny MORICE-PICARD (Bordeaux, France)

- **Outreach clinics for people living with albinism in Burundi**  
Doriane SABUSHIMIKE (Bujumbura, Burundi)

15 min
- **Validating a pilot study: The use of a sunscreen and educational program tailored for people with albinism (PWA) in low- and middle-resource countries**  
Shirley PARRAGA (Winston-Salem, NC, USA)

15 min
- **First clinic for albinism in the Americas**  
Bary BIGAY MERCEDES (Dominican Republic)

5 min
- **Demographic and clinical profile of people with oculocutaneous albinism seen in a tertiary hospital in Lagos, South-West Nigeria**  
Olufolakemi COLE-ADEIFE (Lagos, Nigeria)

10 min
- **Key parameter to develop outstanding sunscreen formulation for people with albinism in low-income settings**  
Stephanie ACKER (Grenach Wyhlen, Germany)

10 min
- **Skin cancer in persons with albinism: development of a research program at IARC**  
Valerie MCCORMACK (Lyon, France)

10 min

**Break**

- **Mapping persons with albinism in Togo in 2024**  
Malanna BIGMA (Lomé Togo)

5 min
- **Children living with albinism In Africa, A Kenyan perspective**  
Prabha CHOKSEY (Nairobi, Kenya)

10 min
- **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**  
Gaylord INENA WA INENA (Kisangani, DRC)

10 min
- **Freedom of religion and beliefs about people with albinism in Uganda: an action learning study for Mid-Western Uganda**  
Ninsiima PHIONAH (Kampala, Uganda)

10 min
- **Skin cancer detection, requirements and requests for a dermatologist consulting persons with albinism, a European point of view**  
Herbert KIRCHESCH (Cologne, Germany)

10 min
- **Bioptic driving: an overview, update, and comparison of driver training methods**  
Chuck HUSS (Dunbar, WV, USA)

10 min

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

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