



Main Sessions Program

Of note: Times indicated include Q&A.

Tuesday January 21st

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)

Wednesday January 22nd 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 23rd

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 24th
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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