



Scientific Conference – Preliminary Program

Main Sessions

Of note: times indicated include 5 mins of Q&A.

Wednesday November 4th – Genetics & Dermatology

2pm-5pm (CET)*

Opening words: Benoît ARVEILER – Scientific Committee of Albinism Europe
Béatrice JOUANNE – President of Genespoir
Antoine GLIKSOHN – Genespoir International Affairs Officer

Session chair: Fanny MORICE-PICARD

- Maria VAN GENDEREN (Zeist, The Netherlands): *The phenotypic spectrum of the FHONDA syndrome: an autosomal recessive disorder caused by mutations in the SLC38A8 gene* – 20min
- Vincent MICHAUD (Bordeaux, France): *Demographic, phenotypic and genetic analyses of a cohort of 692 patients affected with albinism* – 15min
- Helen KUHT (Leicester, UK): *Phenotypical characteristics in albinism carriers* – 15min
- Line KESSEL (Copenhagen, Denmark): *Phenotypic characteristics of individuals with albinism caused by a pathogenic haplotype in TYR* – 15min
- Perrine PENNAMEN (Bordeaux, France): *Identification of two new albinism genes* – 20min
- Lluís MONTOLIU (Madrid, Spain): *The structure and function of the mouse Tyr locus* – 20min
- Carolina MARÇON (São Paulo, Brazil): *Albinism in Africa: My experience in an Angolan community* – 15min
- Christophe PRZYBYLSKI (Lavaur, France): *Fondation Pierre Fabre Initiatives for an access to health care for patients with albinism in sub-Saharan region (including a proposed photoprotective formula)* – 15min
- Yolanda GILABERTE (Zaragoza, Spain): *Evaluation of the acceptance and efficacy of a sun protection package bespoke for Persons with Oculocutaneous Albinism living in Malawi* – 15min

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Thursday November 5th – Hermansky-Pudlak Syndrome & cellular mechanisms of OCA

2pm-5pm (CET)*

Session chair: Benoît ARVEILER

- Mathieu FIORE (Bordeaux, France): *Platelet function testing in Hermansky-Pudlak syndrome* – 20min
- Bernadette GOCHUICO (Bethesda, USA): *Clinical Features of Lung Disease in Hermansky-Pudlak Syndrome* – 30min
- Daniel CUTLER (London, UK): *Organelle plasticity; how size control can modulate cargo function and exocytosis* – 30min
- Cédric DELEVOYE (Paris, France): *Melanosome biogenesis and maturation – Role of BLOCs* – 30min
- Michael MARKS (Philadelphia, USA): *SLC45A2 and melanosome pH regulation: molecular basis of OCA4 and normal skin pigment variation* – 30min
- Donna APPELL (Oyster Bay, USA) – Founder and Executive Director of the HPS Network: *The mission and priorities of the HPS network* – 20min

Friday November 6th – Ophthalmology

2pm-5pm (CET)*

Session chair: Irene GOTTLOB

- Alexandra REBSAM (Paris, France): *The retinal origin of visual defects in albinism* – 20min
- Erica WOERTZ (Milwaukee, USA): *Toward an objective grading scheme of foveal hypoplasia in human albinism* – 20min
- Michael HOFFMANN (Magdeburg, Germany): *Cortical visual field maps in albinism revisited – integrity, diversity, connectivity* – 20min
- Robert PUŻNIAK (Magdeburg, Germany): *Advanced imaging of the optic chiasm and its relevance for albinism diagnostics* – 20min
- Mervyn THOMAS (Leicester, UK): *Using artificial intelligence to distinguish between normal and abnormal retinal development* – 20min
- Abdullah AAMIR (Leicester, UK): *Phenotypic Characteristics as a Diagnostic Tool for Albinism: Sensitivity and Specificity Analysis* – 20min
- Caterina TRAMONTANA (Pavia, Italy): *Anterior eye melanin deficiency and chromatic aberration in albino patients* – 20min

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Saturday November 7th – Cellular and animal models & Therapies

2pm-5pm (CET)*

Session chair: Lluís MONTOLIU

- Vasiliki KALATZIS (Montpellier, France): *Using iPSC-derived retinal organoids to model retinal dystrophies* – 30min
- Muriel CARIO (Bordeaux, France): *The potential of 3D reconstructed epidermis to model albinism* – 30min
- Ana GUARDIA (Madrid, Spain): *New animal models of ocular albinism generated with CRISPR tools* – 15min
- Andrea MONTERO (Madrid, Spain): *Generating and phenotyping CRISPR genome-edited mice to assess the pathogenicity of TYR S192Y and R402Q human genetic variants* – 15min
- Richard HERTLE (Akron, USA): *Visual rehabilitation of patients with oculocutaneous albinism type I (OCA1): Results in 85 patients* – 20min
- Aida SANCHEZ-BRETAÑO (Southampton, UK): *Oral human-equivalent L-DOPA/carbidopa dosages rescue retinal morphology and visual function in a mouse model of human albinism* – 20min
- Smail HADJ-RABIA (Paris, France): *Therapeutic patient education in albinism: validation sessions of the program* – 20min
- Swati PARIDA (Leicester, UK): *Characteristics of abnormal head posture and outcomes of surgical management in patients with albinism and idiopathic infantile nystagmus: a comparative study* – 20min

Flash presentations

Format: 3 min per presentation followed by 2 min of Q&A

Thursday November 5th, 2020

6.30pm-8:00pm (CET)*

Session chair: Karen GRØNSKOV

- Özden HATIRNAZ NG (Istanbul, Turkey): *TYR gene variations in patients with Oculocutaneous Albinism from Turkey.*
- Raphaël LEJOYEUX (Paris, France): *Cases of stage 1 foveal hypoplasia in the parents of patients with albinism: description, analysis and beginning of an explanation.*
- Pauline BATAILLE (Paris, France): *Clinical variability and probable founder effect in oculocutaneous albinism type 7.*
- Ester MORENO-ARTERO (Paris, France): *Phenotypic spectrum of Oculo-Cutaneous Albinism type 4 (OCA4).*
- Kader BELAHDA (Paris, France). *Ocular phenotype of oculocutaneous albinism type 7.*

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- Sophie JAVERZAT (Bordeaux, France) **Functional exploration of new candidate genes for albinism: illustration with DCT/TYRP2.**
- Reinier BAKKER (Amsterdam, The Netherlands). **RNAseq analysis of in vitro retinal organoids shows early expression of albinism genes.**
- Virginie CHOTARD (Paris, France): **Developmental origin of visual deficits present in albinism.**
- Zhanhan TU (Leicester, UK): **What effect does nystagmus have on electro-retinographical (ERG) responses?**
- Joseph CARROLL (Milwaukee, USA): **Assessing Rod Photoreceptor Structure in Human Albinism**

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