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
- Lluis MONTOLIU (Madrid, Spain): The structure and function of the mouse Tyr locus – 20min
- Carolina MARÇON (São Paolo, 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

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

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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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