5TH EDA | European Days of Albinism
Online conference • 4-7 Nov. 2020

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

- 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
- 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

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

2pm-5pm (CET)*

- Mathieu FIORE (Bordeaux): ***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 synthesis and maturation – Role of BLOCs*** – 30min

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- Michael MARKS (Philadelphia, USA): ***SLC45A2 and melanosome pH regulation: molecular basis of OCA4 and normal skin pigment variation*** – 30min
- Donna APPELL (New York, USA) – Founder and Executive Director of the HPS Network: ***Missions and activities of the HPS network*** – 20min

Friday November 6th – Ophthalmology

2pm-5pm (CET)*

- 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 (Sondrio, Italy): ***Anterior eye melanin deficiency and chromatic aberration in albino patients*** – 20min

Saturday November 7th – Cellular and animal models & Therapies

2pm-5pm (CET)*

- Vasiliki KALATZIS (Montpellier, France): ***Using iPSC-derived retinal organoids to model retinal dystrophies*** – 30min
- Muriel CARIO-ANDRÉ (Bordeaux, France): ***The potential of 3D reconstructed epidermis to model albinism*** – 30min
- Ana M. 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

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Flash presentations

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

Thursday November 5th, 2020

6.30pm-8:00pm (CET)*

- 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 (Paris, France): ***Phenotypic spectrum of Oculo-Cutaneous Albinism type 4 (OCA4)***.
- Kader BELAHDA (Paris, France). ***Ocular phenotype of oculocutaneous albinism type 7***.
- 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***.
- Tu ZHANHAN (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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