

# Program

## Saturday October 27<sup>th</sup>

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**9.30 -10.00** Opening session

### **Scientific conference**

**10.00 – 12.00 Genetics** (Chairman Benoît Arveiler)

- (a) Benoît Arveiler, Bordeaux: Oculocutaneous albinism and syndromic forms of albinism - general considerations
- (b) Fanny Morice-Picard, Bordeaux: Strategies for the identification of new genes
- (c) Thomas Rosenberg, Copenhagen: Identification of a new melanocyte differentiation gene underlying human autosomal recessive albinism
- (d) Lluís Montoliu, Madrid: An update on mouse models of albinism
- (e) Mónica Martínez-García, Madrid: Albinochip - a universal genetic diagnosis for all known mutations associated to albinism
- (f) Benoît Arveiler, Bordeaux: Next generation sequencing perspectives for the diagnosis of albinism
- (g) Lucia Mauri, Milan: Molecular Analysis of TYR, P, TYRP1, SLC45A2 and GPR143 Genes in the Italian Reference Centre for Patients with Oculocutaneous and Ocular Albinism

**12.00 – 12.15** coffee break

**12.15 – 13.15 Dermatology** (Chairman Alain Taïeb)

- (j) Alain Taïeb, Bordeaux: Oculocutaneous albinisms, the dermatologist's point of view
- (k) Herbert Kirchesch, Pulheim : Skin Tumours in Albinism, a challenge for scientific research: Presentation of recent findings

**13.15 – 14.30** lunch

**14.30 – 16.00 Ophthalmology** (Chairman Barbara Käsmann-Kellner)

- (l) Barbara Käsmann-Kellner, Homburg: The spectrum of oculocutaneous and ocular albinism
- (m) Alexandra Rebsam, Paris: Eye-specific projections of retinogeniculate axons are altered in albino mice
- (n) Maria van Genderen, Zeist: Recessively inherited misrouting and foveal hypoplasia not correlated with albinism
- (o) Alessandra Del Longo, Milan: Visual Function and Foveal Morphology in Albinism - Our experience

**16.00 – 16.30** coffee break

**16.30 – 18.00 Therapeutic aspects** (Chairman Lluís Montoliu)

(q) Lluís Montoliu, Madrid: L-DOPA and albinism

(r) Brian Brooks, Bethesda: Nitisinone, A Possible Treatment for Oculocutaneous Albinism, Type 1b

(s) Enrico Surace, Naples: Using AAV with TYR or GPR143 for transiently correcting retina of albino rodents

**18.00 – 19.00** Discussion on future collaborations – Round Table

***Meeting of representatives of patients' organizations***

**10.00 – 11.30** Presentations of attending patients' organizations

**11.30 – 11.45** Coffee break

**11.45 – 13.15** Roundtable discussion: Objectives pursued by the organizations, tools, successful experiences and challenges

**13.15 – 14.30** Lunch

**14.30 – 16.00** Networking between rare disease patients organizations in Europe (A. Helm)

**16.00 – 16.30** Coffee break

**16.30 – 18.00** Open discussion: What collaboration and networking between European organizations of people with albinism?

**Sunday October 28th**

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**9.15 – 9.20** Presentation of patients' organizations

Overview of existing organizations of people with albinism in Europe, F. Gliksohn

**9.20 – 10.50** Clinical diagnosis and lifelong medical care

- Milan Niguarda Hospital's Day Hospital, M-C Patrosso
- France's reference centers, A. Taïeb
- Workshop: Improving clinical diagnosis on newborn, infants and older patients; What explains late diagnosis? → towards an earlier and more accurate diagnosis of albinism
- Organizing medical care for people with albinism → towards a global approach

**10.50 – 11.20** coffee break

**11.20 – 12.50** Promoting scientific research on albinism in Europe

- Overview of research on albinism in Europe (inventory of research programs), B. Arveiler
- Genespoir's 15 years' experience, B. Jouanne
- Workshop:  
How can research on albinism be intensified?  
What role can patients' organization play?

**12.50 – 13.50** lunch

**13.50 – 14.30** Nitisinone, A Possible Treatment for Oculocutaneous Albinism, Type 1b, B. Brooks

**14.30 – 15.00** Closing session