Program

Saturday October 27th

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) Lluis 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 Taieb)
   (j) Alain Taieb, 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 Lluis Montoliu)

(q) Lluis 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

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