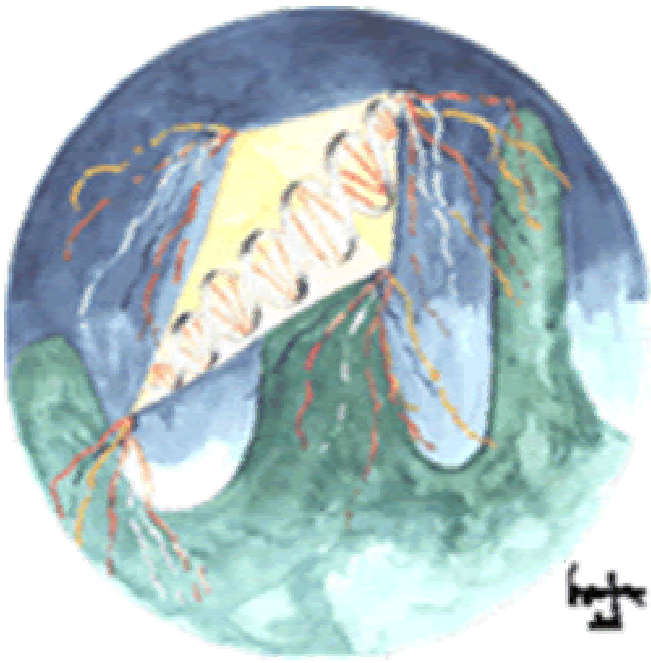
Co-ordination action - EU FP6 - LSHB-CT-2005-512117

geneSkin:

towards a better understanding and care of genetic skin diseases



Rome, 20 June 2008

Istituto Dermopatico dell'Immacolata

http://geneskin.idi.it/homepgs/news

8:30 – 9:00 Registration.
9:00 – 9:10 Giovanna Zambruno: *Welcome and introduction.*

Epithelial adhesion disorders
9:10 – 9:30 Leena Bruckner-Tuderman**:** *Epithelial adhesion disorders.*
9:30 – 10:00 Thomas Magin*: Modulating the chaperone machinery to treat EBS: a novel target for small molecule-based therapy*.

Connective tissue Disorders
10:00 – 10:20 Anne De Paepe: *Phenotypes and genotypes in cutis laxa and stiff skin syndromes.*
10:20 – 10:50 Uwe Kornak: *Disturbance of the secretory pathway can cause glycosylation abnormalities and cutis laxa.*

10:50 – 11:20 Coffee break

Keratinisation disorders
11:20 – 11:40 Alain Hovnanian: *Skin barrier defect and allergy by dysregulated protease activity: Netherton syndrome and mice models*.
11:40 – 12:00 Heiko Traupe: *Transglutaminase-1 and bathing suit ichthyosis: molecular analysis of gene/environment interactions*.

12:0 0 – 13:30 Lunch

DNA repair disorders
13:30 – 13:50 Alan Lehmann: *Heterogeneity and genotype-phenotype relationship in xeroderma pigmentosum.*
13:50 – 14:10 Miria Stefanini: *Trichothiodystrophy: recent advances and molecular insights.*
14:10 – 14:30 Koos Jaspers: *Nucleotide-excision repair deficiencies and ageing*.
14:30 – 14:50 Alain Sarasin: *Gene therapy in XPC cells by specific meganuclease homologous recombination.*

Ectodermal dysplasias
14:50 – 15:10 John A. McGrath: *p63: mutations and syndromes*.
15:10 – 15:30 Karl Heinz Grzeschik: *Deficiency of PORCN, involved in processing of WNT proteins, causes Goltz syndrome.*
15:30 – 16:00 Eli Sprecher: *Novel neurocutaneous ectodermal dysplasias: what's rare may be common*.

16:00 – 16:30 Coffee Break

Geneskin website
16:30 – 16:45 Giovanna Zambruno/Giandomenico Russo: *Geneskin website presentation.*

Standardization and validation of diagnostic tools
16:45 – 17:05 Judith Fischer: *ARCI mutation screening by Geneskin participants.*
17:05 – 17:20 Guerrino Meneguzzi: *Immunofluorescence analysis of villous trophoblasts: a new tool for prenatal diagnosis of inherited epidermolysis bullosa with pyloric atresia.*

Linkage to the community
17:20 – 17:40 Claire Robinson: *DebRA Europe: achievements and perspectives*.
17:40 – 18:00 Angelika Walser/Michael Willam: *Report from Geneskin ethical experts.*

18:00 Closing