HELSINGIN YLIOPISTO
HELSINGFORS UNIVERSITET
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HUS

How to use new genetic methods in clinical diagnostics of rare skin diseases

May 14-15, 2017, Skin and allergy hospital, Helsinki, Finland

Sunday May 14th

17.00 **Welcome and introduction to the theme and ERN Skin network,** *Annamari Ranki, Professor and Chairperson, Department of Dermatology and allergology, University of Helsinki and Helsinki University Hospital (HUH)*

17.30 **Common and rare subtypes of Epidermylosis Bullosa: from clinic to molecular diagnosis,** Maya el-Hachem, professor Pediatric Dermatology Unit, Bambino Gesú Children's Hospital, IRCCS, Rome, Italy

18.15 **Discussion**

18.30 **Departure for Dinner on board and archipelago cruising**

Monday May 15th

09.00 **Rare Diseases – the need for registries, networking and collaboration,** *Dr. Mikko Seppänen, Director, HUH Rare Diseases Unit (HAKE)*

09.40 **How to identify disease causing mutations with novel genomics methods? - Clinical examples** *Janna Saarela, Research Director and Head of the FIMM Technology Center*

10.10 **Genodermatoses: not only genes,** *Smail Hadj-Rabia professor, Department of Dermatalogy, referral Center for Genodermatoses (MAGEC) Imagine Institute, Necker-Enfants maladies Hospital (AP-HP), Paris, France*

11.10 **Role of a clinical geneticist in genodermatoses,** *Sirpa Kivirikko, Ass.professor, Department of Clinical Genetics, University of Helsinki and HUH*

11.50 **How should dermatologists and clinical geneticists work in unison? - Patient case example Palmoplantar keratodermas,** *Katariina Hannula-Jouppi, Ass. professor, Section Chief, Department of Dermatology and allergology University of Helsinki and HUH and Sirpa Kivirikko*

12.30 **Lunch**

13.30 **Patient case examples: eczemas that turned out to be genodermatoses ,** *Katariina Hannula-Jouppi*

14.00 **Neurofibromatosis type 1 (NF1) gene: Beyond café au lait spots and dermal neurofibromas**, *Sirkku Peltonen, Ass.professor, Department of Dermatology, University of Turku and Turku University Hospital*

14.30 **Autoinflammatory syndromes with skin involvement**, Mikko Seppänen

15.00 **Short break**

15.15 **Skin transcriptomics as a new source of diagnostic markers in skin disorders,** *Juha Kere, Professor of Genetics & Molecular Medicine, King's College, London, and Professor of Molecular Genetics, Karolinska Institutet, Stockholm*

16.00 **Interactive discussion,** *Katariina Hannula-Jouppi, Annamari Ranki*

16.30 **Adjourn**