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

May 15th, 2017, Skin and allergy hospital, Helsinki, Finland

10.00 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)

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

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

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

11.50 How should dermatologists and clinical geneticists work in unison? - Patient case example Palmoplantar keratoderma, Katriina 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, Katriina 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, Katriina Hannula-Jouppi, Annamari Ranki

16.30 Adjourn