ENGAGE
European Network of Genomic and Genetic Epidemiology

Special Seminar

Tribute to Leena Peltonen-Palotie
Academician of Science

Biomedicum Lecture Hall 1
Monday, May 31 at 9-11 am

Olli Kallioniemi
Opening remarks
Institute for Molecular Medicine Finland FIMM, FI

Mark McCarthy
The elegance of brute force: large-scale genetic analyses and metabolic disease
OCDEM, Oxford University, UK

Anna-Elina Lehesjoki
Rare diseases - pioneers for common ones?
Folkhälsan Institute, University of Helsinki, FI

Antti Sajantila
From individuals to populations – now and then
Hjelt Institute, Department Forensic Medicine, University of Helsinki, FI

Gert-Jan van Ommen
Translational genomics: linking rare and common diseases
CMSB, Leiden University Medical Center, NL

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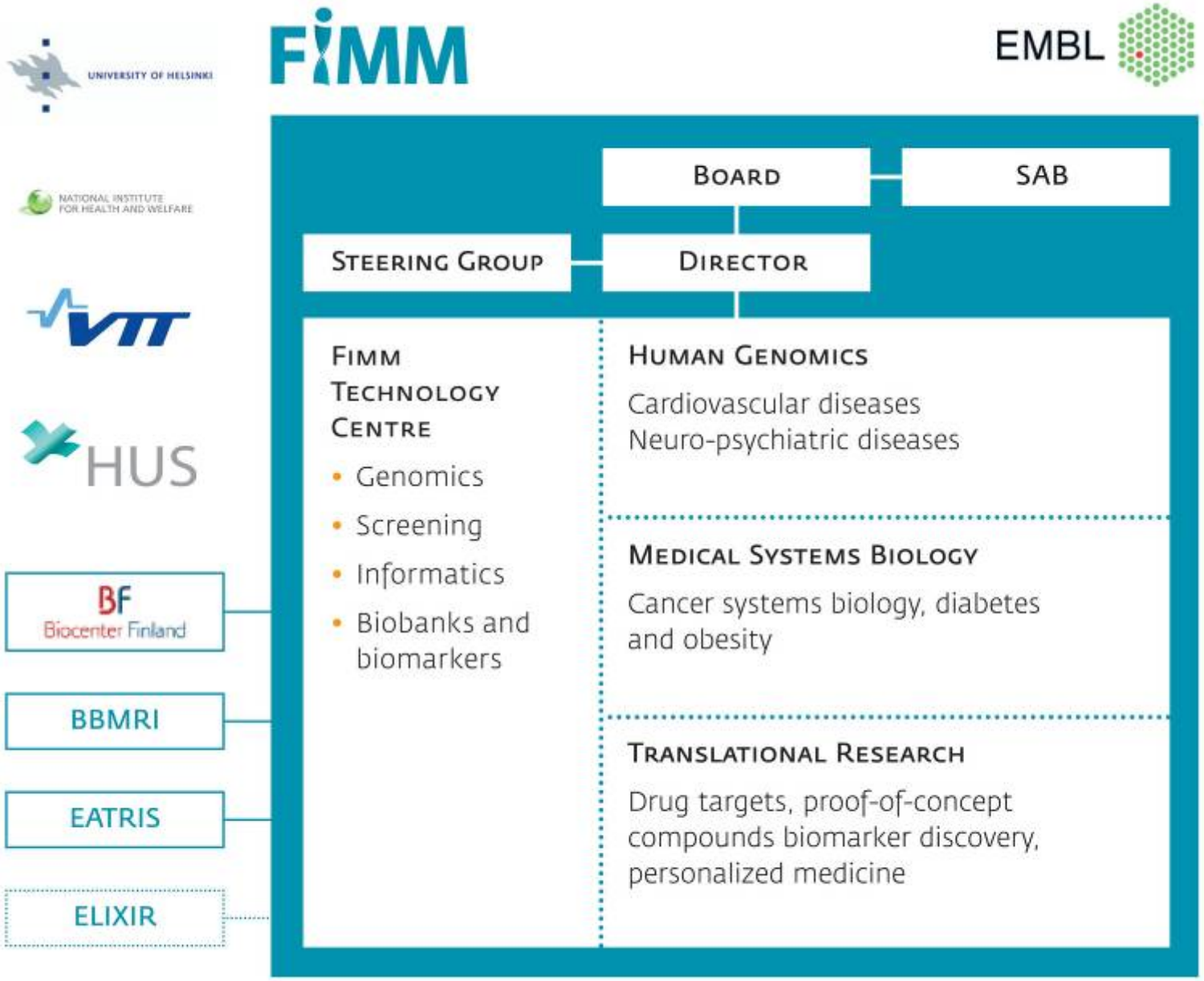
Institute for Molecular Medicine Finland
Nordic EMBL Partnership for Molecular Medicine
Building a bridge from discovery to medicine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Neuro-psychiatric diseases
MEDICAL SYSTEMS BIOLOGY
Cancer systems biology, diabetes and obesity
TRANSLATIONAL RESEARCH
Drug targets, proof-of-concept compounds biomarker discovery, personalized medicine

Cell

Michael R. Stratton[1,*] and
Eric Lander[2]
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Leena Peltonen 1952–2010

Leena Peltonen, one of the leading lights of the human genetics research community, passed away on March 11, 2010 at her home in Finland. Leena was born in Oulu, Finland, a town of approximately 100,000 people about 700 km north of Helsinki, in 1952. Her fath... a civil servant and subsequently ... of the business school and her m... teacher. She had one brother who ... oped type 1 diabetes from which ... died of complications. Leena rec... that this early intimate encoun... with disease kindled her enthu... asm for medical research.

Leena graduated from hi... school top in her class, charact... izing herself with typical self-depr... cating humor as "a swot" (slang ... an overzealous student), and pr... ceeded to study medicine in Ou... Following graduation, she obtain... her first taste of research stud... ing inherited collagen disorders, ... subject that was an abiding intere... for many years. Between 1978 a... 1980 she undertook the first of se... eral interludes of work outside F... land, a period of postdoctoral stu... at Rutgers University, New Jers... where she attempted to charact... ize mutations in the inherited bo... disease osteogenesis imperfec... through protein analysis. Trar... planted from the relative quiet ... the South and the East) and who subsequently remained relatively isolated. As a consequence of this founder effect, certain inherited diseases that are rare in most outbred populations turn out by chance to be relatively common in ...

elaborated this strategy by implementing the approach known as homozygosity mapping, which depends on consanguinity of parents of individuals with recessive diseases, using it to identify the gene responsible for infantile onset spinocerebellar ataxia. In 1998, she moved to the University of California Los Angeles (UCLA) where she spent 4 years establishing a major genetic research center.

Following this successful decade of ...

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OBITUARY

Leena Peltonen-Palotie (1952–2010)

A visionary in medical genetics.

Gertjan van Ommen
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On 11 March, science lost an inspiring leader with the death of Leena Peltonen-Palotie, at the age of 57, following a two-year battle with bone cancer.

Known to those in the field as Leena Peltonen (she was married to her close collaborator Aarno Palotie), she was a key player in human molecular genetics. This was true both in the early days of identifying the genes and mechanisms underlying rare diseases, and more recently in trying to track the causes of common inherited disorders. Peltonen left an invaluable legacy to medical genetics by authoring more than 600 papers and mentoring some 70 PhD students. But perhaps her most enduring contribution was to show how understanding the causes of genetic diseases in isolated populations can offer clues for large-scale studies that probe the risk factors linked to common diseases such as diabetes, obesity and heart disease.

Peltonen completed her PhD at the University of Oulu in Finland in 1978. After postdoctoral work at Rutgers University in New Jersey, she returned to the National

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at the forefront of this fresh wave of research, identifying an osteoarthritis gene in 1989. But as the hunt for risk factors ramped up, geneticists became engaged in a fierce debate over whether isolated inbred populations held clues to the factors responsible for common diseases. Although she was convinced that they did, Peltonen hedged her bets and genetic studies in isolated populations proved prescient. The enrichment of a limited set of mutations yields a simpler picture than that obtained from the larger-scale hunts for the factors underlying complex diseases. Yet some of the components unearthed from the smaller studies, such as the disrupted gene underlying osteoarthritis, have been found to be involved in complex forms of the same disorders.

Peltonen's interest in complex genetics always went beyond merely mapping genes. In 2002, her team unravelled the mechanism of lactose intolerance. Then in 2004 and 2005, her group found the link between a variant of a gene called *USF1*, which causes a lipid disorder that clusters in families, and insulin resistance. She was particularly proud of these findings, as in both cases the variant genes altered regulatory functions. This suggested a fundamental mechanistic difference between the basic genetic defects typical of Mendelian disease and the more subtle regulatory alterations underlying complex disease.

It is up to us to make good on her legacy, by pursuing not only fame and fortune, but also real clinical utility.

Gertjan van Ommen

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