

# 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**

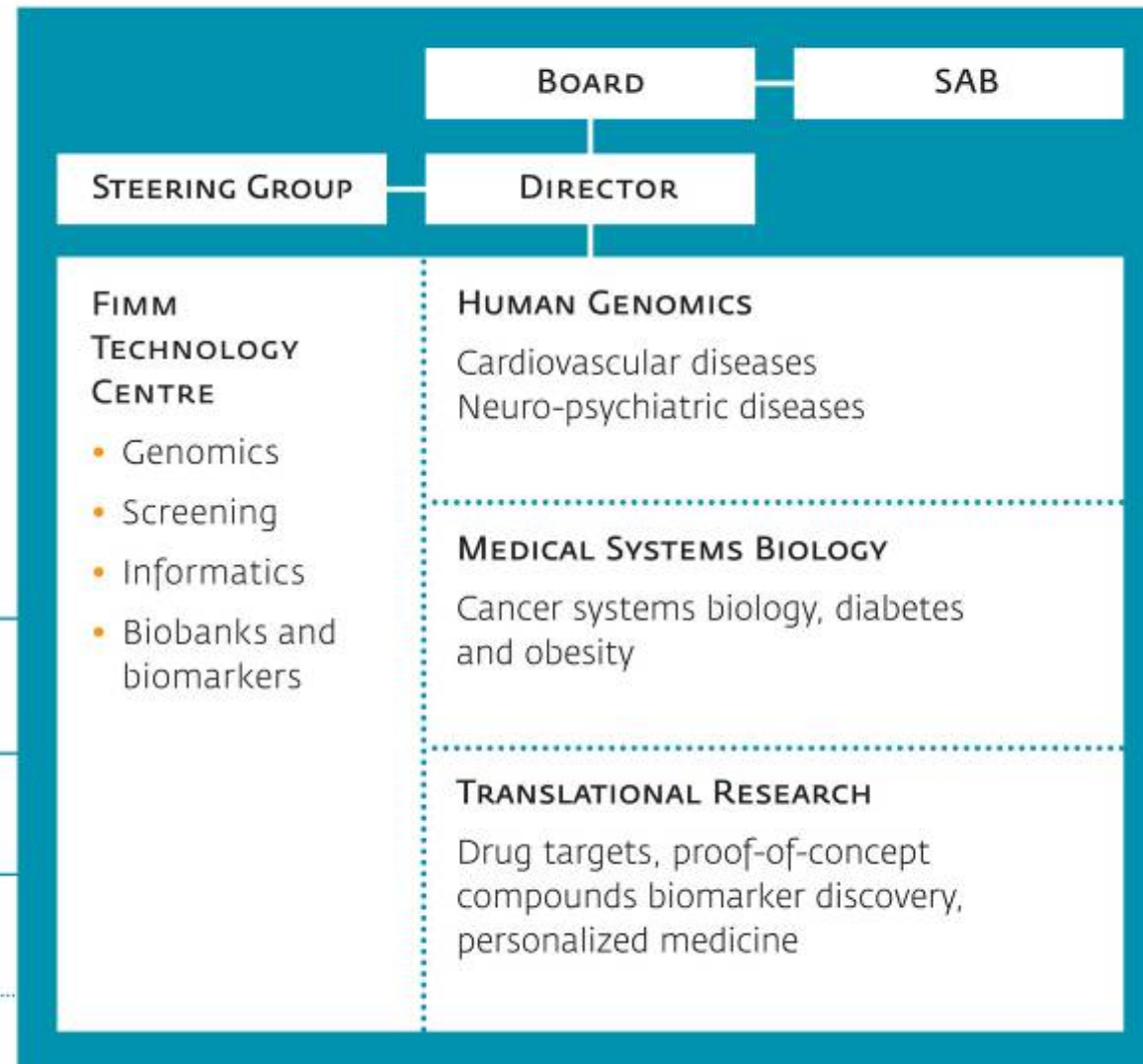
***Translational genomics:  
linking rare and common diseases***

CMSB, Leiden University  
Medical Center, NL



**Institute for Molecular Medicine Finland**  
Nordic EMBL Partnership for Molecular Medicine

*Building a bridge from discovery to medicine*



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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 father was a civil servant and subsequently a teacher. She had one brother who developed type 1 diabetes from which he died of complications. Leena recalled that this early intimate encounter with disease kindled her enthusiasm for medical research.

Leena graduated from high school top in her class, characterizing herself with typical self-deprecating humor as “a swot” (slang for an overzealous student), and proceeded to study medicine in Oulu. Following graduation, she obtained her first taste of research studying inherited collagen disorders, subject that was an abiding interest for many years. Between 1978 and 1980 she undertook the first of several interludes of work outside Finland, a period of postdoctoral study at Rutgers University, New Jersey where she attempted to characterize mutations in the inherited bone disease osteogenesis imperfecta through protein analysis. Transplanted from the relative quiet

of 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

isolated populations. Leena 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

### OBITUARY

## Leena Peltonen-Palotie (1952–2010)

A visionary in medical genetics.

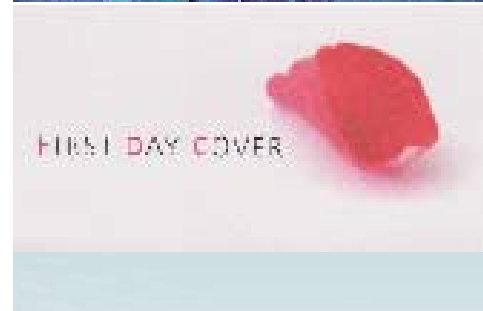
On 11 March, science lost an inspiring leader with the death of Leena Peltonen-Palotie, at the age of 57, following a two-year battle with bone cancer.

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



Academy of Finland (where she was 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



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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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