
BIOGRAPHICAL SKETCH

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NAME Leena Peltonen, M.D., Ph.D.	POSITION TITLE Head of Human Genetics, Wellcome Trust Sanger Institute, UK; Prof, Research Director, Institute for Molecular Medicine FIMM, Univ of Helsinki and National Inst for Health and Welfare, Finland; Visiting Prof., the Broad Inst of MIT and Harvard, USA		
eRA COMMONS USER NAME			
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
University of Oulu, Finland	M.D.	1976	Medicine
University of Oulu, Finland	Ph.D.	1978	Medical Biochem
University of Oulu, Finland	Docent	1982	Cell Biology
University of Helsinki, Finland	Docent	1991	Mol Genetics

A. Positions and Honors.

Positions and Employment

- 1978–1980 Postdoc Fellow, Dept. of Biochem (Chair Darwin J. Prockop), Rutgers Medical School, NJ, USA
1981–1984 Associate Professor, Department of Cell Biology, University of Oulu, Finland
1985–1987 Senior Scientist of The Academy of Finland, Recombinant DNA Laboratory, Univ of Helsinki
1987–1991 Head of the Laboratory, Laboratory of Molecular Genetics, Nat'l Public Health Institute, Finland
1991–1995 Professor of Molecular Biology, Director of the Molecular Biology Prgm, Nat'l Public Health Inst.
1995–1998 Professor of Medical Genetics, University of Helsinki and National Public Health Institute, Finland
July 1998–2002 Chairman and Professor of Department of Human Genetics, UCLA
2000 - Director of the Center of Excellence in Disease Genetics of the Academy of Finland
1999 - 2004 Gordon and Virginia MacDonald Distinguished Chair in Human Genetics, David Geffen School of Medicine at UCLA
July 2002- 2003 Professor, National Public Health Institute, Finland and Professor, University of Helsinki, Department of Medical Genetics
2003 -2007 Academy Professor, the Academy of Finland (comparable to Howard Hughes fellowship)
2004 - Co-ordinator, The Nordic Center of Excellence in Disease Genetics
2005 - Visiting Professor (with the right to hold grants) the Broad Institute, MIT, Harvard, Boston, USA
2007 - Professor, Research Director, Institute of Molecular Medicine Finland, (FIMM), University of Helsinki and National Institute for Health and Welfare (former: National Public Health Institute)
2007 - Head of Human Genetics, Wellcome Trust, Sanger Institute, UK

Other Experience and Professional Memberships

- 1990 – 1992 Chairman, Finnish Society of Medical Genetics
1991 European Molecular Biology Organization (EMBO), Member
1991 – 1999 Human Genome Organization, Member of the International Council, 2004 President-elect
1993 – National Academy of Sciences, Finland, Member
1998 – 2000 American Society of Human Genetics, Member of the Board of Directors
1999 – European Academy of Sciences, Member
2002 Societas Scienterum Fennica, Member
2004 – 2005 European Society of Human Genetics, President
1992 – 1996 Member, Steering Committee, Nordic HUGO
1995 – 1997 Chairman of the Medical Research Council of Finland
1996 – 1998 Chairman of the European Medical Research Council
1996 – 1998 Member, Public Affairs and Awareness Committee, EMBO
1996 – 1998 Member, Science and Technology Policy Council of Finland (headed by the Prime Minister)
1996 – Member, Scientific Advisory Board, Center for Molecular Medicine, Karolinska Institutet, Stockholm

- 1996-2002 Member, Scientific Advisory Board of the Genetics Program of the Universities of Leiden and Rotterdam
- 1998 – 2003 Member, International Bioethics Committee, UNESCO
- 1999 – 2005 Foundation for Strategic Research, Sweden
- 2001 – Member, Scientific Advisory Board of UK Biobanks
- 2002 – Member, Scientific Advisory Board of the Shanghai Genome Center
- 2002 – 2003 Member, Scientific Advisory Board, Genome Canada
- 2001 – 2003 Chair, Scientific Advisory Board Netherlands, Genome Initiative
- 2003 - Institutional Advisory Board of VIB, Ghent, Belgium
- 2005 – President, Council of HUGO
- 2005 - Member, Board of the European Research Council

Honors

- 1992 The Antoine Marfan Award (The Nat'l Marfan Foundation., USA)
- 1992 The Anders Jahre Prize for young scientist (Scandinavian Science Prize)
- 1992 The Poul Astrup Prize (Scandinavian Science Prize)
- 1994 Woman of the Year (Finland)
- 1996 Lennox K. Black Prize (Thomas Jefferson University, USA)
- 1997 Mauro Bachiroto Prize (European Society of Human Genetics)
- 1999-2002 Gordon and Virginia MacDonald Distinguished Chair in Human Genetics, UCLA
- 2000 Honorary Doctor of Medicine, Millennium promotion, University of Uppsala, Sweden
- 2001 Ribicoff Lecturer, Yale University
- 2002 Naomi M. Kanof Clinical Investigator Award lectureship, the Society for Investigative Dermatology
- 2002 Abbott Award for Distinguished Achievements in Molecular Diagnostics by International Federation of Clinical Chemistry and Laboratory Medicine
- 2004 The Margaret Pittman Lectureship Award, National Institute of Health, USA
- 2006 Prix van Gysel (European Research Prize)
- 2006 Erik K Fernstrom large Nordic Prize
- 2006 Foreign member of the Institute of Medicine of the US National Academy of Sciences
- 2008 Honorary Professor of the Faculty of Clinical Medicine, Univ of Cambridge, UK

B. Publications (in chronological order). (selected from 524 peer-reviewed publications)

- J. Vesa, E. Hellsten, L-A. Verkruyse, L.A. Camp, J. Rapola, P. Santavuori, S.L. Hofmann, L. Peltonen: Mutations in the palmitoyl protein thioesterase gene causing infantile neuronal ceroid lipofuscinosis. *Nature* 376:584-587, 1995.
- S. Kuokkanen, M. Sundvall, J.D. Terwilliger, P.J. Tienari, J. Wikström, R. Holmdahl, U. Pettersson, L. Peltonen: A putative vulnerability locus to multiple sclerosis maps to 5p14-p12 in a region syntenic to the murine locus Eae2. *Nature Genet* 13:477-480, 1996.
- J. Aaltonen, P..Björnses, J. Perheentupa, N. Horelli-Kuitunen, A. Palotie, Y.Su Lee, F. Francis, S. Henning, C. Thiel, H. Lehrach, M.-L. Yaspo, L. Peltonen: An autoimmune disease, APECED, caused by mutations in a novel gene featuring two PHD-type zinc-finger domains. *Nature Genetics*, 17:399-403, 1997.
- P.Pajukanta, M. Cargil, L. Viitanen, I. Nuotio, A. Kareinen, M. Perola, J. Terwilliger, E. Kempas, M. Daly, H. Lilja, J. Rioux, T. Brettin, J. Viikari, T. Ronnema, M. Laakos, E. Lander, L. Peltonen: Two loci on chromosomes 2 and X for premature coronary heart disease identified in early and late settlement populations in Finland. *Am J Hum Genet* 67:1481-1493, 2000.
- L Peltonen, A Palotie, K Lange: Use of Population Isolates for Mapping Complex Traits. *Nature Review* 1:182-191, 2000.
- N. Enattah, T. Sahi, E. Savilahti, J. D. Terwilliger, L. Peltonen, I. Järvelä: Identification of a DNA variant associated with adult type hypolactasia. *Nature Genet.* 30, 233-237, 2002.
- P. Pajukanta, H. Allayee, K. L. Krass, A. Kuraishy, A. Soro, H.E. Lilja, R. Mar, M-R. Taskinen, I. Nuotio, M. Laakso, J.I. Rotter, T.W.A de Bruin, R.M. Cantor, A.J. Lusis, L. Peltonen: Combined analysis of genome scans of Dutch and Finnish families reveals a susceptibility locus for high-density lipoprotein cholesterol on chromosome 16q. *Am. J. Hum. Genet.* 72 (4):903-917, 2003.

- A. Liston, D.H. Gray, S. Lesage, A.L. Fletcher, J. Wilson, K.E Webster, H.S. Scott, R.L. Boyd, L. Peltonen, C.C. Goodnow: Gene dosage limiting role of Aire in thymic expression, clonal deletion and organ-specific autoimmunity. *J Exp Med* 200 (8): 1015-1026, 2004
- P. Pajukanta, H.E. Lilja, J.S. Sinsheimer, R.M. Cantor, A.J. Lusis, M. Gentile, X.J. Duan, A. Sorola, J. Naukkarinen, J. Saarela, M. Laakso, C. Ehnholm, M-R.Taskinen, L. Peltonen: Familial combined hyperlipidemia is associated with upstream transcription factor 1 (USF1). *Nature Genetics*, published online 04-02-29, 36 (4): 371-376, 2004
- K. Komulainen, M. Alanne, K. Auro, R. Kilpikari, P. Pajukanta, J. Saarela, P. Ellonen, K. Salminen, S. Kulathinal, K. Kuulasmaa, K. Silander, V. Salomaa, M. Perola, L. Peltonen: Risk alleles of USF1 gene predict cardiovascular disease of women in two prospective studies. *PLoS Genet* 2(5); e69, 2006.
- S. Service, J. DeYoung, M. Karayiorgou, J. Louw Roos, H. Pretorius, G. Bedoya, J. Ospina, A. Ruiz-Linares, A. Macedo, J. Almeida Palha, P. Heutink, Y. Aulchenko, B. Oostra, C. van Duijn, M-R. Jarvelin, T. Varilo, L. Peddle, P. Rahman, G. Piras, M. Monne, S. Murray, L. Galver, L. Peltonen, C. Sabatti, A. Collins, N. Freimer: Magnitude and distribution of linkage disequilibrium in population isolates and implications for genome-wide association studies. *Nat Genet* Apr 2; 38(5); 556-560, 2006.
- J. Saarela, S. P. Kallio, D. Chen, A. Montpetit, A. Jokiahho, E. Choi, R. Asselta, d. Bronnikov, M. R. Lincoln, A. D. Sadovnick, P. J. Tienari, K. Koivisto, A. Palotie, G. C. Ebers, T. J. Hudson, L. Peltonen: PRKCA and Multiple Sclerosis: Association in two independent populations. *PLoS Genetics* 2:42-48, 2006.
- Y.S. Aulchenko, S. Ripatti, I. Lindqvist, D. Boomsma, I. Heid, C. Pattaro, A. Cecile, J.W. Janssens, J.F. Wilson, I. Rudan, Å. Johansson, T. Spector, N.G Martin, N.L Pedersen, F. Marroni, K. Ohm Kyvik, J. Kaprio, A. Hofman, P. Elliott, C. Gieger, A. Isaacs, C. Hayward, V. Vitart, I. Jonasson, E.J.G. Sijbrands, N. Freimer, T. Meitinger, A.G. Uitterlinden, J. Saharinen, M. Perola, J.C.M. Witteman, P.P. Pramstaller, M-R. Jarvelin, U. Gyllensten, H. Campbell, A. Wright, N. Hastie, B.A. Oostra, F. Kronenberg, C.M. van Duijn, L. Peltonen for the ENGAGE consortium: Genome-wide association study in 16 European population cohorts: Major loci determining lipid levels and coronary heart disease risk. *Nature Genetics* 41(1)47-55, 2008.
- S. Kathiresan, O. Melander, C. Guiducci, A. Surti, N.P. Burt, M.J. Rieder, G.M. Cooper, C. Roos, B.F. Voight, A.S. Havulinna, B. Wahlstrand, T. Hedner, D. Corella, E.S. Tai, J.M. Ordovas, G. Berglund, E. Vartiainen, P. Jousilahti, B. Hedblad, M-R. Taskinen, C. Newton-Cheh, V. Salomaa, L. Peltonen, L. Groop, D.M. Altshuler, M. Orho-Melander: Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans. *Nat Genet*, Feb:40(2):189-97 (Epub Jan13), corrigendum: Nov;40(11):1384, 2008.
- I. Prokopenko, C. Langenberg, J. Florez, R. Saxena, N. Soranzo, G. Thorleifsson, R. Loos, A. Manning, A. Jackson, Y. Aulchenko, S. Potter, M. Erdos, S. Sanna, J-J. Hottenga, E. Wheeler, M. Kaakinen, V. Lyssenko, W-M Chen, K. Ahmadi, J. Beckmann, R. Bergman, M. Bochud, L. Bonnycastle, T. Buchanan, A. Cao, L. Coin, A. Cervino, F. Collins, L. Crisponi, E. de Geus, A. Dehghan, P. Deloukas, P. Elliott, N. Freimer, V. Gateva, C. Herder, A. Hofman, T. Hughes, S. Hunt, T. Illig, M. Inouye, B. Isomaa, T. Johnson, A. Kong, Noha Iim, U. Lindblad, C.M. Lindgren, O. McCann, K. Mohlke, S. Naitza, M. Orru, A. Pouta, J. Randall, W. Rathmann, J. Saramies, P. Scheet, L. Scott, A. Scuteri, S. Sharp, E. Sijbrands, J. Smit, K. Song, V. Steinthorsdottir, H. Stringham, T. Tuomi, J. Tuomilehto, A. Uitterlinden, B. Voight, D. Waterworth, H-E. Wichmann, G. Willemsen, J. Witteman, X. Yuan, J. Zao, E. Zeggini, D. Schlessinger, M. Sandhu, D. Boomsma, M. Uda, T. Spector, B. Penninx, D. Altshuler, P. Vollenweider, M-R. Jarvelin, E. Lakatta, G. Waeber, C. Fox, L. Peltonen, L. Groop, V. Mooser, L. Cupples, U. Thorsteinsdottir, M. Boehnke, I. Barroso, C. van Duijn, J. Dupuis, R. Watanabe, K. Stefansson, M. McCarthy, N. Wareham, J. Meigs, G. Abecasis: Variants in the melatonin receptor 1B gene (MTNR1B) influence fasting glucose level. *Nature Genetics* 41(1):77-81, 2008
- S. Kathiresan, C.J. Willer, G. Peloso, S. Demissie, K. Musunuru, E. Schadt, L. Kaplan, D. Bennett, Y. Li, T. Tanaka, B.F. Voight, L.L. Bonnycastle, A. U. Jackson, G. Crawford, A. Surti, C. Guiducci, N. Burt, S. Parish, R. Clarke, D. Zelenika, K. A Kubalanza, M. A Morken, L. J. Scott, H. M. Stringham, P. Galan, A. J Swift, J. Kuusisto, R. N. Bergman, J. Sundvall, M. Laakso, L. Ferrucci, P. Scheet, S. Sanna, M. Uda, Q. Yang, K. Lunetta, J. Dupuis, P.I. deBakker, C. J. O'Donnell, J. C Chambers, J. S. Kooner, S. Hercberg, P. Meneton, E.G. Lakatta, A. Scuteri, D. Schlessinger, J. Tuomilehto, F. S. Collins, Leif Groop, D. Altshuler, R. Collins, G.M. Lathrop, O. Melander, V. Salomaa, L. Peltonen, M. Orho-Melander, J.M. Ordovas, M.Boehnke, G. R. Abecasis, K. L. Mohlke & L. A. Cupples: Common variants at 30 loci contribute to polygenic dyslipidemia. *Nature Genetics* 41(1):56-65, 2008.

- Sabatti, SK. Service, AL. Hartikainen, A. Pouta, S. Ripatti, J. Brodsky, CG. Jones, NA. Zaitlen, T. Varilo, M. Kaakinen, U. Sovio, A. Ruokonen, J. Laitinen, E. Jakkula, L. Coin, C. Hoggart, A. Collins, H. Turunen, S. Gabriel, P. Elliot, MI. McCarthy, MJ. Daly, MR. Järvelin, NB. Freimer, L. Peltonen: Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. *Nature Genetics* 41(1):35-45, 2008.

Overviews in articles in books and editorials:

- L. Peltonen, A. Palotie, K. Lange: Use of Population Isolates for Mapping Complex Traits. *Nature Reviews Genetics* 1:182-191, 2000.
- L. Peltonen, V.A. McKusick: Dissecting human diseases after the human genome project: *Science*, 291:1224-29, 2001.
- D. Boomsma, A. Busjahn, L. Peltonen: The classical twin studies and beyond. *Nature Reviews Genetics*. 3:872-883, 2002.
- L. Peltonen, J. Saarela, S. Kuokkanen: The genetic basis of common diseases: Multiple sclerosis (Eds R.A. King, J.I. Rotter, A. G. Motulsky). *Oxford monographs on medical genetics* no. 44, 2002
- L. Peltonen: GenomEUtwin: A Strategy to Identify Genetic Influences on Health and Disease. *Twin Research*, Vol. 6, No. 5, 354-360, 2003.
- T. Varilo, L. Peltonen: Isolates and their potential use in complex gene mapping efforts. *Current Opinion in Genetics & Development*, Vol.14, No. 3: 316-323, 2004.
- P. Peterson, L. Peltonen: Autoimmune polyendocrinopathy syndrome type 1 (APS1) and AIRE gene: New views on molecular basis of autoimmunity. *J Autoimmun* 25 Suppl: 49-55, 2005.
- I. Ulmanen, M. Halonen, T. Ilmarinen, L. Peltonen: Monogenic autoimmune diseases — lessons of self-tolerance. *Curr Opin Immunol*, 17(6):609-15, 2005.
- T. Varilo, L. Peltonen: Population selection in complex disease gene mapping. In: *Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics* (Ed. M. J. Dunn, L. B. Jorde, P. F. R. Little, S. Subramaniam), John Wiley & Sons, UK, 2005.

C. Research Support

Ongoing Research Support

Migraine: Assignment and Isolation of Predisposing Genes

2 RO1 NS037675-05 (Palotie, PI)	09/01/2004 – 08/30/09	
Co-Principal Investigator		5%
National Institute of Health/NINDS	\$250,000	

Center of Excellence in Complex Disease Genetics

The Academy of Finland	2006-2011	
	\$193,000/year	20 %

STAMPEED (Genetics of cardiovascular risk factors in large founder population birth cohorts)

1-R01-HL087679-01 (Peltonen)		
NIH/NHLBI	10/1/06-9/30/09	15 %
Principal Investigator	\$ 3 700 000	

ENGAGE – European Network for Genetic and Genomic Epidemiology

FP7-HEALTH-F4-2007-201413	01/01/08-12/31/12	
	\$258,000 /year	15 %

BBMRI – Biobanking and Biomolecular Resources Research Infrastructure

FP7-INFRASTRUCTURES-2007-212111	02/01/08-01-31/10	
	\$104,000 /year	15 %