

1. JÄRVELÄ (née JOKIAHO), IRMA ELISABET

<https://tuhat.halvi.helsinki.fi/portal/fi/>

Gender: female

date of writing the CV: 20.10.2016

2. Date and place of birth, nationality, current residence

26.09.1953-052C Lapua, Finnish, Espoo

3. Education and degrees awarded

Title of Docent: Molecular Genetics, University of Helsinki	06.11.1996
PhD, University of Helsinki	16.04.1991
MD, University of Oulu	24.10.1979

4. Other training, qualification and skills

Special Competence in medical education (The Finnish Medical Association, FMA)	2013
Leadership Education Program for Health Care Helsinki School of Economics	2002-2003
Specialist degree in clinical genetics, University of Helsinki	1996
Specialist degree in general practice, University of Helsinki	1986

5. Linguistic skills

Finnish (mother tongue), English fluent, Swedish good, German good

6. Current position: Principal investigator, clinical teacher in medical genetics, University of Helsinki. Start and end date of the position 01.09.2015, no end date.

7. Previous work experience

Clinical teacher in medical genetics, University of Helsinki	01.09.2010-31.08.2015
Acting professor in medical genetics, University of Helsinki	01.03.2010-31.12.2011
Researcher on the grant from the Jusélius Foundation	01.08-2008-28.02.2010
University lecturer (50%), University of Helsinki	01.01.2008-30.06.2008
Researcher on personal grant from Paulo Foundation	01.01.2007-30.06.2007
Senior Scientist, the Academy of Finland	01.01.2006-31.12.2006
Clinical geneticist, Helsinki university hospital	14.08.2000-31.12.2005
United Laboratories Ltd, Cytogenetic laboratory (part-time, 50%)	01.01.1995-31.12.1995
House Officer, Helsinki University Hospital	
- Department of Obstetrics and Gynecology, Medical genetics	01.07.1996-31.12.1996
Group leader, The Institute of Health and Welfare	01.01.1995-12.08.2000
Visiting scientist, Department of Neurogenetics, Massachusetts General Hospital, Boston, MA	01.11.1994-31.12.1994
Postdoctoral fellow, University College London, London, UK	01.09.1992-31.10.1994
House Officer, Helsinki University Hospital	
-Department of Obstetrics and Gynecology, Medical genetics	01.02.1992-31.08.1992
PhD-student, National Public Health Institute	01.02.1988-31.03.1991
Medical geneticist, The Family Welfare Federation, Helsinki	01.01.1986-28.02.1988
General Practitioner in Helsinki City Health Centre	01.09.1982-31.12.1985
House Officer, Helsinki University Hospital	01.05.1981-31.08.1982
General Practitioner Vaasa and Kauhava Health Centre	01.11.1979-30.04.1981

8. Research funding, leadership and supervision

1992-1994	The Welcome Trust and Sigrid Jusélius Foundation	83 300 €	Post doctoral grant to UCL, London
1995	The Academy of Finland	37 000 €	homing, Genetic mapping of NCLs
2000-2007	The Sigrid Jusélius Foundation	259 000 €	Molecular and functional studies on human lactase deficiencies
2001-2002	The Päivikki and Sakari Sohlberg Foundation	166 000 €	Molecular genetics of autism spectrum disorders
2001-2009	Helsinki University Hospital Fund	504 100 €	Molecular genetics of autism spectrum disorders and lactase persistence
2003-2006	The Finnish Cultural Foundation	225 000 €	Molecular genetics of musical aptitude
2004-2007	Academy of Finland	150 000 €	Molecular genetics of autism spectrum disorders
2006	Senior Research Fellow, Academy of Finland	90 000 €	Human lactase deficiencies
2006-2008	European Union Specific Targeted project	278 000 €	Using European and International populations to Identify Autism Susceptibility Loci (chief: Anthony Bailey, Oxford)
2010	The Sigrid Jusélius Foundation	50 000 €	Molecular genetics of autism and mental retardation
2005-2014	The Finnish Medical Society	158 200 €	Molecular genetics of autism spectrum disorders, glaucoma, and intellectual disability
2011-2014	The Academy of Finland	1,3 milj €	Biological basis of musical aptitude
2011-2013	Biocentrum Helsinki	134 000 €	Biological basis of musical aptitude
2015	University of Helsinki	40 000 €	Gene regulation in music perception and performance

The major research funding is shown. Total sum 3,5 M €.

Experience as officially appointed supervisor to doctoral students, main supervisor

1. Mari Auranen MD, Molecular genetics of autism spectrum disorders; University of Helsinki, 2002
2. Nabil Enattah MD, Molecular genetics of lactase persistence; University of Helsinki, 2005
3. Tero Ylisaukko-oja PhD, In search for susceptibility genes for autism spectrum disorders; University of Helsinki, 2005
4. Heli Rasinpera PhD, Adult-type hypolactasia: phenotype-genotype correlation; University of Helsinki, 2006

5. Mikko Kuokkanen PhD, Molecular genetics of lactase deficiencies; University of Helsinki, 2006
6. Sanna Seitsonen MD, Molecular genetics of age-related macular degeneration; University of Helsinki, 2008
7. Susanna Lemmela PhD, Molecular genetics of glaucoma and exfoliation syndrome; University of Helsinki, 2009
8. Katri Kantojärvi PhD, Exploring genetic background of autism spectrum disorders; University of Helsinki, 2013
9. Liisa Ukkola-Vuoti PhD, Search for genetic variants underlying musical aptitude and related traits; University of Helsinki, 2013
10. Chakravarthi Kanduri PhD, Genomics approaches to study music perception and performance; University of Helsinki, 2015
11. Jaana Oikkonen PhD, Genetics and genomics of musical abilities, University of Helsinki, 2016

Ongoing supervision of doctoral students, main supervisor

1. Preethy Nair MSc, DVet, High throughput genomic approaches to decipher music perception and production, scheduled 2017
2. Anju Philips MSc (biotechnology), Identification of causes for intellectual disability (ID) using exome sequencing, scheduled 2017

Experience as officially appointed supervisor to MSc students

A total of 26 BSc/MSc theses supervised.

9. Merits in teaching and pedagogical competence

- six years responsible for teaching medical genetics at the Medical Faculty
- University Pedagogy courses I & II, 15 credits (405h) 2005-2008
- Problem based learning (PBL) basic course (1.5 credits, 40h) 2008
- special competence in medical education awarded by the Finnish Medical Association 2013
- a study about vertical integration of the medical studies: Immonen T, **Järvelä I**, Pöyhönen M, Peltonen LM, Stenberg T: Towards horizontal and vertical integration. Molecular biology & medical genetics and Gastroenterology & nutrition.
- a study about the needs for further education in medical genetics among Finnish physicians by Varilo T, Pyörälä E, Varilo S, Hietala M, **Järvelä I**. Suomen Lääkärilehti 68:1368-69, 2013 (In Finnish).

Production of learning materials

1. **Järvelä I**, Kolho K-L. Laktoosi-intoleranssi. Lääkäriin käsikirja 2014, 2016.
2. Course book in medical genetics, 2010 -2016.

10. Awards, prizes, honours

Award winning in International evaluation of scientific research and doctoral training at the University of Helsinki 2012.

Secretary at the Ministry of Health, Genetic screening in Finland 1997-1998

11. Other scientific or academic merits

Duties as a scientific expert: reviewer of the theses and associate professorships

1. MD Sirpa Ala-Mello "Nephronophthisis – medullary cystic disease in Finland. Genetic, epidemiological and clinical study" University of Helsinki 1998
2. MD Tuija Viitanen "Mapping of the lysinuric protein intolerance gene", University of Turku 1999
3. MD Susanna Ranta "Positional cloning of the progressive epilepsy with mental retardation (CLN8 gene)" University of Helsinki 1999

4. MD Pirjo Salomäki "Free sialic acid storage diseases; same gene –different mutations" University of Turku, 2001
5. MD Anu-Liisa Moisio "Predisposing and modifying genes in familial colorectal cancer" University of Helsinki 2001
6. MD Kati Asumalahti "Molecular genetics of psoriasis" University of Helsinki 2003
7. MD Miia Melkonieni "Genetic defects of collagen XI. The role of a minor cartilage collagen in chondrodysplasias, oral cleft defects and osteoarthritis" University of Oulu, 2004
8. PhD Maarit Lappalainen "Molecular genetic studies of candidate genes in inflammatory bowel disease", University of Helsinki 2008
9. MD Auli Siren, "Childhood absence epilepsy: an epidemiological, neuropsychological and molecular genetic study", University of Tampere 2011
10. PhD Terhi Peuralinna, "Genetics of neurodegeneration", University of Helsinki 2015

Evaluation of scientific competence

1. MD Mirja Somer, Docentship in Medical genetics, University of Helsinki 2000
2. PhD Vesa Juvonen, Docentship in Molecular genetics, University of Turku 2010
3. PhD Csilla Sipeky, Docentship in Pharmacogenetics, University of Turku 2014

Member of international per evaluation committees of funding applications

Austrian Science Fund 2008, Lithuanian State Science and Studies Foundation, Lithuania 2008
National Science Foundation, USA 2009, Wellcome Trust Senior Fellowship, UK 2009, Young investigator grant ERC 2015, Consolidator grant, ERC 2015

Referee for scientific journals: Am J Hum Genet, Hum Genet, World J Gastroenterology, PLoS ONE, Clin Chem Acta, Annals of Medicine, J Ped Gastroenterol & Nutrition, Clin Genet.

12. Scientific and societal impact of research

In total, 177 peer reviewed articles in scientific journals. HI 36.

Merits related to the application of the results: Identification of the genetic variant of adult-type hypolactasia, introduction of the genetic test of lactose intolerance. *Patents:*
<http://patents.justia.com/inventor/irma-jarvela>.

Meetings organized: The Annual meeting of the Finnish Society of Ophthalmology and the Finnish Society of Medical Genetics, 2008. "Musical aptitude, music listening and biology", seminar, Music House, Helsinki 2015

Recent expert assignments in the media:

<http://www.youtube.com/watch?v=Hh2PcUNiRMs>, <http://ohjelmaopas.yle.fi/1-2290181>,
<http://www.mylab.fi/fi/puheenvuoro/>, www.yle1.fi/news

Selected invited lectures:

Riken Institute, Tokyo 1992, University College London, London 1992, University of Leiden 2001, International Congress of Human Genetics, Vienna 2001, The American Society of Human Genetics, Baltimore 2002, The American Society of Human Genetics, Toronto 2004, Max Planck Institute, Berlin, 2005, The Proust Symposium, European Union, Tartu University, Estonia 2008, UEG Week Vienna 2014, European Human Behaviour and Evolution Conference, Helsinki 2015.