Lea M. Urpa

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EDUCATION

PhD in Neurogenomics **Aug 2017 – Present**
Institute for Molecular Medicine Finland (FIMM), HiLIFE, University of Helsinki
FIMM-EMBL International PhD in Molecular Medicine.
Supervisors: Dr. Aarno Palotie and Dr. Mark Daly

FIMM-EMBL Rotation Program **Aug 2016 – Dec 2017**
Institute for Molecular Medicine Finland (FIMM), HiLIFE, University of Helsinki
Supervisors: Dr. Simon Anders, Dr. Matti Pirinen

International Masters in Translational Medicine (MSc) **Aug 2014 – July 2016**
University of Helsinki
Supervisors: Dr. Iiris Hovatta

Postbaccalaureate Research Education Program **July 2012 – July 2014**
University of Michigan
Supervisor: Dr. Gina Poe

BS (Hon), Biology **Sept 2005 – June 2011**
Early Entrance Program at California State University Los Angeles
Supervisor: Dr. Carlos Robles

PROFILE

I am a researcher with a keen interest in developing methods and projects to solve the diagnostic and treatment deficits for individuals with neuropsychiatric disorders. My research background is in neuroscience and genomics, most recently with a focus on the genomics of neurodevelopmental disorders and the integration of rare and common variation in a joint risk model for intellectual disabilites. I have experience in using cloud environments and tools such as Hail for analyzing large, heterogenous sequencing and SNP array datasets. I also have knowledge of RNAseq data analysis and of methods development for high-dimensional data. I am interested in learning to integrate large omics data and refined phenotypic data to better understand the etiology of neuropsychiatric diseases.

RESEARCH EXPERIENCE

Institute for Molecular Medicine Finland (FIMM), HiLIFE, University of Helsinki **Aug 2017 – Present**
Doctoral Researcher
Led the analysis of the genetic architecture of intellectual disability in the Northern Finland Intellectual Disability cohort.

- Focus on integration of rare and common variation to understand the differences in etiology between mild and more severe intellectual disability, and between individuals with and without psychiatric comorbidities.
- Rotation project in the lab of Dr. Simon Anders on methods development for visualization and exploratory analysis of high-dimensional data.
- Four-year funded position from the Doctoral Program in population health, a competitive salaried funding based on research plan evaluation.

RESEARCH EXPERIENCE

University of Helsinki **June 2015 – June 2016**
Master's thesis research
Master's thesis research focusing on differential expression of transposable genetic elements and anxiety-like behavior in mice.

- Transitioned from wet lab work to bioinformatics with a focus on bulk RNAseq data analysis
- Faculty of Medicine prize for the best Master's thesis published in 2016

University of Michigan, Department of Anesthesiology **July 2012 – July 2014**
Postbaccalaureate researcher
Postbaccalaureate scholar focused on the links between post-traumatic stress disorder and sleep disturbances in rodents.

- Performed animal behavioral experiments and analyzed sleep electrophysiological data

California State University, Los Angeles **June 2009 – June 2011**
Undergraduate researcher
Undergraduate thesis focused on the role of global warming-driven salinity changes on intertidal ecology. Performed field work and data analysis.

REFEREES

Professor Aarno Palotie (PhD advisor)
Research Director, Institute for Molecular Medicine Finland (FIMM), HiLIFE, University of Helsinki, Finland
Stanley Center for Psychiatric Research, The Broad Institute of MIT and Harvard, Cambridge, MA, USA
Psychiatric and Neurodevelopmental Genetics Unit, Massachusetts General Hospital, Boston, MA, USA
Analytical and Translational Genetics Unit, Massachusetts General Hospital, Boston, MA, USA
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Professor Mark Daly (PhD advisor)
Stanley Center for Psychiatric Research, The Broad Institute of MIT and Harvard, Cambridge, MA, USA
Analytical and Translational Genetics Unit, Massachusetts General Hospital, Boston, MA, USA
Psychiatric and Neurodevelopmental Genetics Unit, Massachusetts General Hospital, Boston, MA, USA
Institute for Molecular Medicine Finland (FIMM), HiLIFE, University of Helsinki, Finland
mjdaly@broadinstitute.org

Professor Iiris Hovatta (previous advisor)
Department of Psychology and Logopedics, University of Helsinki, Finland
iiris.hovatta@helsinki.fi

Professor Gina Poe (previous advisor)
UCLA Brain Research Institute, Los Angeles, CA, USA
ginapoe@ucla.edu

AWARDS/HONORS

Biomedicum Helsinki Young Investigator Grant
Grant for doctoral researchers at the University of Helsinki, 2022

Best Poster Award
Nordic Society of Human Genetics and Personalized Medicine 2021

'Human Heredity' Best Student Poster Award
European Mathematical Genetics Meeting 2017

SCIENTIFIC COMMUNITY

Founder, The Science Basement
Founding chair of a non-profit organization focused on providing opportunities for scicomm training in English in the greater Helsinki area. Coordinated and led the registration of the group as an official non-profit with the Finnish authorities.

SELECTED PUBLICATIONS

EVIDENCE FOR THE ADDITIVITY OF RARE AND COMMON VARIANT BURDEN THROUGHOUT THE SPECTRUM OF INTELLECTUAL DISABILITY

Urpa L, Kurki M, Rahikkala E, ... Singh T, Kuismin O, Pietiläinen O, Palotie A, Daly M. *In submission.*

Created bioinformatic pipeline for quality control of heterogenous SNP array and WES data using Hail and Google Cloud platform. Co-designed and performed all analysis, helped coordinate sample collection and creation of phenotype database.

THE IMPACT OF RARE PROTEIN CODING GENETIC VARIATION ON ADULT COGNITIVE FUNCTION

Chen CY, Tian R, Ge T, Lam M, Sanchez-Andrade G, Singh T, **Urpa L** ... Daly M, Palotie A, Tsai E. Huang H, Hurles M, Gerety S, Lencz T, Runz H (2023). *Nature Genetics.* https://doi.org/10.1038/s41588-023-01398-8

Performed a replication analysis for the eight identified genes in the Northern Finnish Intellectual Disability cohort.

A NOVEL VARIANT IN *SMG9* CAUSES INTELLECTUAL DISABILITY, CONFIRMING A ROLE FOR NONSENSE-MEDIATED DECAY COMPONENTS IN NEUROCOGNITIVE DEVELOPMENT

Rahikkala E, **Urpa L** ... Daly M, Palotie A, Pietiläinen O, Moilanen J, Kuismin O (2022). *Eur J Hum Gen*. https://doi.org/10.1038/s41431-022-01046-5

Co-designed and led a secondary analysis of allele-specific expression in rare variant carriers.

FOCUSED MULTIDIMENSIONAL SCALING: INTERACTIVE VISUALIZATION FOR EXPLORATION OF HIGH-DIMENSIONAL DATA

Urpa L, Anders S (2019). *BMC Bioinformatics.* https://doi.org/10.1186/s12859-019-2780-y

Created an R package for novel visualization of high-dimensional data, with a focus on RNAseq data.

OTHER PUBLICATIONS

FUNCTIONAL CHARACTERIZATION OF SIX *SLCO1B1* (OATP1B1) VARIANTS OBSERVED IN FINNISH INDIVIDUALS WITH A PSYCHOTIC DISORDER

Hakkinen K, Kiander W, Kidron H, Lahteenvuo M, **Urpa L** ... Palotie A, Ahola-Olli A, Niemi M (2023). *Molecular Pharmaceutics.* https://doi.org/10.1021/acs.molpharmaceut.2c00715

POLYGENIC BURDEN HAS A BROADER IMPACT ON HEALTH, COGNITION, AND SOCIOECONOMIC OUTCOMES THAN MOST RARE AND HIGH-RISK COPY NUMBER VARIANTS

Saarentaus E, Havulinna A, Mars N, Ahola-Olli A, Kiiskinen T, Partanen J, Ruotsalainen S, Kurki M, **Urpa L** ... Hall I, Pietiläinen O, Kaprio J, Ripatti S, Daly M, Palotie A (2021). *Mol Psychiatry*. https://doi.org/10.1038/s41380-021-01026-z

THE BRADYKININ SYSTEM IN STRESS AND ANXIETY IN HUMANS AND MICE

Rouhiainen A, Kulesskaya N, Mennesson M, Misiewicz Z, Sipilä T, Sokolowska E, Trontti K, **Urpa L** ... Hyytiä P, Hovatta I (2019). *Scientific Reports*. https://doi.org/10.1038/s41598-019-55947-5

SLEEP ALTERATIONS FOLLOWING EXPOSURE TO STRESS PREDICT FEAR-ASSOCIATED MEMORY IMPAIRMENTS IN A RODENT MODEL OF PTSD

Vanderheyden W, George S, **Urpa L**, Kehoe M, Liberzon I, Poe G (2015). *Exp Brain Res.* https://doi.org/10.1007/s00221-015-4302-0

STRESS-FREE AUTOMATIC SLEEP DEPRIVATION USING AIR PUFFS

Gross BA, Vanderheyden W, **Urpa L** ... Poe G (2015). *J Neurosci Methods*. https://doi.org/10.1016/j.jneumeth.2015.05.010

SELECTED PRESENTATIONS

World Conference of Psychiatric Genetics 2022

Enrichment of rare, damaging variants in schizophrenia-related genes in intellectual disability patients with psychotic disorders (*Note: I was unfortunately unable to give this talk in person)*

Nordic Society of Human Genetics and Personalized Medicine Annual Meeting 2021

Multiple methodologies reveal novel picture of the genetic architecture of intellectual disability in Northern Finland. Best Poster Award.

The Stanley Center Symposium 2019: Severe Mental Illness- From Polygenicity to Biology

Genetic architecture of intellectual disability in Northern Finland.

The Biology of Genomes, Cold Spring Harbor Laboratory 2019

Rare and common variants in the genetic architecture of intellectual disability.

European Mathematical Genetics Meeting 2017

Focused multidimensional scaling: Interactive, intuitive visualization of high-dimensional data. Human Heredity Best Student Poster award.