BIOGRAPHICAL SKETCH

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NAME: Rita Horvath

eRA COMMONS USER NAME (credential, e.g., agency login): N/A

POSITION TITLE: Director of Research

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	Start Date MM/YYY	Completion MM/YYYY	FIELD OF STUDY
Semmelweis University Budapest	MD	1982	1988	Medicine
Semmelweis University Budapest	Neurology Training	1988	1992	Neurology
Hungarian Academy of Sciences	PhD	1995	2000	Mitochondrial Medicine

A. Personal Statement

I am a clinician-scientist with a 25-year career of diagnosing and treating families with mitochondrial disease. My research interest in mitochondrial diseases began during my PhD at the Montreal Neurological Institute and my earliest achievement was introducing the first mitochondrial diagnostic lab for mtDNA mutations in Budapest, Hungary. As postdoctoral scientist I did laboratory research in Munich, Germany and established a mitochondrial diagnostic service in the Medical Genetic Center (MGZ), before accepting my first academic position in 2007 as Lecturer in Mitochondrial Research at Newcastle University. Since 2018 I am Director of Research at the Department of Clinical Neurosciences, University of Cambridge. My academic research focus has been the identification of novel genes and disease mechanisms in mitochondrial disease, with the aim of developing treatments. I am glad that more recently I can translate my preclinical research into clinical trials. Demonstrating my involvement in international activities, I led the Mitochondrial Diseases Group in the European Reference Network for Rare Neuromuscular Diseases (EURO-NMD) (2016-2021), and I am organizing committee member of the Wellcome Campus Mitochondrial Medicine Conference. I have been UK representative for the European Mitochondrial Society (E-mit) from the start and I would be very happy to continue working as a Board Member in E-mit.

B. Positions, Scientific Appointments and Honors

1988-1992	Hungarian Academy of Sciences Fellow, Dept. of Neurology, Jahn Ferenc Dél-pesti Hospital, Budapest, Hungary
1992-1999	Neurologist, Dept. of Neurology, Jahn Ferenc Del-pesti Hospital, Budapest, Hungary
1999-2004	Clinical Research Associate, Hospital Schwabing, Munich, Germany
2004-2006	Clinician Scientist, Mitochondrial Diagnostics, Hospital Schwabing, Munich, Germany
2006-2007	Clinician Scientist, Mitochondrial Diagnostics, Medical Genetic Centre Munich, Germany
2007-2010	Lecturer in Mitochondrial Research, Newcastle University, UK
2010-2018	Honorary Consultant, Newcastle NHS Trust, UK
2010-2013	Senior Lecturer, Institute of Genetic Medicine, Newcastle University, UK
2013-2018	Professor of Neurogenetics, Institute of Genetic Medicine, Newcastle University, UK
Sept 2018 -	Director of Research, Department of Clinical Neurosciences, University of Cambridge, Honorary Consultant Neurologist, Cambridge University Hospitals NHS Trust, UK
1990	Neuromuscular fellowship, University of Oxford (Prof. John Newsom-Davis)

National and international recognition

1995	Soros Foundation fellowship, Montreal Neurological Institute (Prof. Eric A. Shoubridge)
2006	Felix-Jerusalem Prize of the German Muscular Dystrophy Association (DGM)
2016-21	Chair of the Mitochondrial Disease Group, European Reference Network (EURO-NMD)
2021-	Associate Editor of Brain
2021-	Organising Committee Member of the Wellcome Mitochondrial Medicine Conference

C. Contributions to Science

I published >320 papers including several with first/corresponding/senior authorships in high-impact journals (e.g., JAMA, Lancet Neurol, Nat Genet, Nat Commun, Brain, Genet Med, h-index 72, i10-index 259, citations 18360, 10/05/2023 Google scholar). I wrote 22 invited commentaries including journals such as Nat Rev Genet, Cell Metab, 7 book chapters and edited a new Handbook of Clinical Neurology published in 2023. I have played a key role in the identification of ~20 novel disease genes and improved the diagnostic yield in mitochondrial diseases. By identifying the mechanism of reversible infantile mitochondrial myopathy, I demonstrated that digenic interaction of variants in mitochondrial tRNAs and nuclear genes underly tissue specific mitochondrial disease. I linked treatable neuromuscular transmission defects recently with mitochondrial disease, enabling precision therapies for these rare conditions, previously thought untreatable. By conducting natural history studies and identifying biomarkers I contributed to trial readiness in mitochondrial disease and I am very keen to perform clinical trials to translate my preclinical findings to treatments.

Research funding

Principal Investigator	**start date**	**end date**	**amount**
Muscular Dystrophy UK, PhD fellowship	01/10/2023	30/09/2027	£119,871
Action for AT Research Grant (TREAT-AT)	01/01/2023	31/12/2025	£250,000
MRC Cellular and Molecular Biology MR/V009346/1	01/04/2021	31/03/2025	£855,816
Evelyn Trust Research Grant (19/14)	01/01/2020	31/12/2022	£184,378
Lily Foundation Research Grant	01/08/2019	31/07/2021	£62,432
Wellcome Investigator Award (109915/Z/15/Z)	01/04/2016	31/03/2022	£1,150,000
MRC Neurosciences Board Grant (MR/N025431/1)	01/10/2016	30/09/2020	£686,866
Newton Fund (UK/Turkey) MR/N027302/1	01/09/2016	30/09/2020	£253,515
Wellcome Trust Pathfinder Award 201064/Z/16/Z	01/11/2016	30/06/2018	£170.000
Starter Grant, European Research Council (ERC) (309548)	01/03/2013	28/02/2018	£1,139,280
MRC Confidence in Concept Fund	01/09/2014	28/02/2015	£19,457
New Investigator Research Grant, MRC UK (G1000848)	01/03/2011	28/02/2014	£397,000
AMS Starter Grant for Clinical Lecturers (BH090164)	01/10/2009	31/09/2011	£29,284
Newcastle NHS Foundation Trust	01/12/2008	01/12/2009	£37,598
German Research Council, DFG, HO2505/2-1	01/10/2006	30/09/2009	£200,000
Co-investigator	**start date**	**end date**	**total**
Wellcome Trust Discovery Award (1/3 PIs)	01/10/2023	30/09/2028	£4.3M
International MRC Centre for Neuromuscular Diseases	01/05/2019	30/04/2024	£3.6M
HORIZON 2020, Solve-RD (1/15 Centres) until 09/2018	01/01/2018	31/12/2022	£390.080
Wellcome Mitochondrial Research Centre (1/12 PIs)	01/05/2017	31/08/2018	£6,500.000
Wellcome Equipment Grant 208339/Z/17/Z (1/6 PIs)	01/10/2017	30/09/2020	£746,328
MRC Confidence in Concept Fund (1/2 PIs)	01/04/2016	01/11/2016	£37,852
FP7-PEOPLE-ITN (Marie-Curie Action 317433) (1/9 PIs)	01/01/2013	31/12/2016	£3,042,664
MRC Centre for Neuromuscular Disease (1/12 PIs)	01/01/2013	31/12/2017	£3,133,229
French Muscular Dystrophy Association (AFM) (1/8 PI)	01/01/2012	31/03/2013	£114,548

Teaching and researcher development

I have been the main supervisor of 19 PhD or MD PhD students (4 ongoing, 15 completed), 5 MSc, 6 MRes, 7 BSc students. I acted as examiner of 22 PhDs. I gave teaching lectures at national and international conferences and training courses (SSIEM Training Course, Recordati Course on mitochondrial disease).

D. Scholastic Performance

Invited speaker at >40 conferences and meetings in the last 5 years. Review Panel Member of the French Research Innovation Grants **(**Recherche Hospitalo-Universitaire en santé), the Neuroscience Centre, University of Helsinki and the European Research Chair Programme at the Dokluz Eylul University, Izmir.
Grant Reviewer for MRC UK, Welcome, ERC and many European Research Organisations and Charities.