CURRICULUM VITAE:

17 April 2013

Rita Horvath
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Qualifications

2000	PhD	Hungarian Academy of Sciences
1992	Neurology	Postdoctoral Medical University Budapest, Hungary
1988	MD	Semmelweis Medical University, Budapest, Hungary

Appointments

2010 - Senior Lecturer and Honorary Consultant, Institute of Genetic Medcine, Newcastle University
2007 - 2010 Lecturer in Mitochondrial Research, Mitochondrial Research Group, Newcastle University
2006 - 2007 Mitochondrial diagnostic and neurogenetics, Medical Genetic Center, Munich, Germany
2004 - 2006 Diagnostic work, Mitochondrial Diagnostic Laboratory, Hospital Schwabing, Munich
2003 - 2ßß6 Consultant in Neurogenetics, Jahn Ferenc Del-pesti Hospital, Budapest, Hungary
1999 - 2004 Research in mitochondrial diseases, Hospital Schwabing, Munich, Germany
1992 - 1999 Neurologist, Jahn Ferenc Del-pesti Hospital, Budapest, Hungary
1988 - 1992 Fellow of the Hungarian Academy of Sciences, Department of Neurology, Jahn Ferenc Del-pesti Hospital Budapest, Hungary

Postgraduate Awards and Prizes

2006 Felix Jerusalem Prize for outstanding research in neuromuscular disease
1995 Soros Foundation international scholarship (Montreal Neurological Institute)

Active Grant Awards

2013 – 2018 **European Research Council – Starter Grant** 309548
Project: Reversibility and tissue specificity of mitochondrial translation defects in early childhood (€1.432.075)

2011 – 2014 **Medical Research Council – New Investigator Research Grant** G1000848,
Project: Molecular basis of the spontaneous recovery in infantile reversible cytochrome c oxidase (COX) deficiency myopathy (£397.000)

Previous Grant Awards

2009 – 2011 **Academy of Medical Sciences and Wellcome Trust`s Starter Grants for Clinical Lecturers Scheme**: BH090164, Project: Identification of new disease genes in combined respiratory chain deficiencies. (30.000 GBP)
2008 – 2009 **Newcastle upon Tyne Hospitals NHS Foundation Trust**: AB/SS/08051, Project: Investigation of mitochondrial translation and coenzyme Q10 biosynthesis in childhood-onset respiratory chain deficiencies. (37.598 GBP)
2006 – 2009 **Deutsche Forschungsgemeinschaft**: HO2505/2-1, Project: New insights into the pathomechanism of mitochondrial depletion syndromes. (200.000 Euro)

Selected publications (Total = 118)

Neeve VC, Samuels DC, Bindoff LA, van den Bosch B, Van Goethem G, Smeets H, Lombès A, Jardel C, Hirano M, Dimauro S, De Vries M, Smeitink J, Smits BW, de Coo IF, Saft C, Klopstock T, Keiling BC, Czermin B, Abicht A, Lochmüller H, Hudson G, Gorman GG, Turnbull DM, Taylor RW, Holinski-Feder E, Chinnery PF, **Horvath R**. What is influencing the phenotype of the common homozygous polymerase-γ mutation p.Ala467Thr? ***Brain*** 2012;135:3614-3626.

Payne BA, Wilson IJ, Hateley CA, **Horvath R**, Santibanez-Koref M, Samuels DC, Price DA, Chinnery PF. Mitochondrial aging is accelerated by anti-retroviral therapy through the clonal expansion of mtDNA mutations. ***Nat Genet*** 2011:43:806-810.

Zeman J, Udd B, Taylor RW, Turnbull D, Suomalainen A, Zeviani A, Chinnery P. Phenotypic spectrum associated with mutations of the mitochondrial polymerase γ gene. ***Brain*** 2006;129:1674-1684.

Kemp JP, Smith PM, Pyle A, Neeve VCM, Tuppen HA, Chinnery PF, Chrzanowska-Lightowlers ZMA, Lightowlers RN, Taylor RW and **Horvath R**. Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. ***Brain*** 2011;134:183-195.

Horvath R, Kemp JP, Tuppen HA, Smith PM, McFarland R, Chrzanowska-Lightowlers ZM, Lightowlers RN, Hirano M, Lochmüller H, Taylor RW, Chinnery PF, Tulinius M, Dimauro S. Molecular basis of infantile reversible cytochtome c oxidase deficiency myopathy. ***Brain*** 2009;132:3165 -74.

Weraarpachai W, Antonicka H, Sasarman F, Seeger J, Schrank B, Kolesar JE, Lochmüller H, Chevrette M, Kaufman BA, **Horvath R**, Shoubridge EA. Mutation in TACO1, encoding a translational activator of COX I, results in cytochrome c oxidase deficiency and late-onset Leigh syndrome. ***Nat Genet*** 2009;41:833-837.

Ghezzi D, Goffrini P, Uziel G, **Horvath R**, Klopstock T, Lochmüller H, D'Adamo P, Gasparini P, Strom TM, Prokisch H, Invernizzi F, Ferrero I, Zeviani M. SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. ***Nat Genet*** 2009;41:654-656.

Bulst S, Abicht A, Holinski-Feder E, Müller-Ziermann S, Koehler U, Thirion C, Walter MC, Stewart JD, Chinnery PF, Lochmüller H, **Horvath R**. In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. ***Hum Mol Genet*** 2009;18:1590-1599.

Gempel K, Topaloglu H, Talim B, Schneiderat P, Schoser BG, Hans VH, Palmafy B, Kale G, Tokatli A,Quinzii C,Hirano M, Naini A, DiMauro S, Prokisch H, Lochmüller H, **Horvath R.** The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. ***Brain*** 2007;130:2037-2044.

Horvath R, Hudson G, Ferrari G, Futterer N, Ahola S, Lamantea E, Prokisch H, Lochmüller H, McFarland R, Ramesh V, Klopstock T, Freisinger P, Musumeci O, Mayr H, Santer R, Tesarova M,