



Research funded by Ataxia UK that has ended in the last few years

Friedreich's ataxia Archive

Research Project	Principal Researchers
Finding a treatment for Friedreich's ataxia: identification and characterisation of frataxin up-regulating compounds from the Pfizer's small molecule library	Prof Paola Giunti, University College London and Prof Richard Wade-Martins, University of Oxford, UK
Effect of vitamin D in frataxin-deficient DRG neurons from rat and iPS cells obtained from FA patients	Dr Joaquim Ros, Dr Jordi Tamarit & Dr Marta Llovera, Universitat de Lleida, Spain
Exploring Ca²⁺ signalling in a Friedreich's ataxia mouse model	Professor Paola Giunti, Dr Sandip Patel & Mr Alexander Brown, University College London, UK
Beta-band EMG-EMG coherence: a novel, painless and simple screening test for the onset of corticospinal tract disease/dorsal root ganglionopathy in Friedreich's ataxia	Dr Mark Baker, Prof Stuart Baker, & Prof Patrick Chinnery, Newcastle University, Newcastle, UK
An investigation to determine the efficacy and safety of lentivirus mediated FXN gene delivery for the correction of Friedreich ataxia	Dr Mark Pook and Dr Michael Themis, Brunel University, Uxbridge, UK
Pharmacodynamic studies of a histone deacetylase inhibitor in Friedreich's ataxia	Professor Richard Festenstein, Department of Medicine, Imperial College London
Identifying additional sensitive targets in Friedreich ataxia	Professor Richard Festenstein, Department of Medicine, Imperial College London
Identification of the E3 ligase that ubiquitinates frataxin	Dr Roberto Testi, Department of Experimental Medicine, University of Rome, 'Tor Vergata,' Italy
Visual dissection of GAA-mediated mechanisms of FRDA repression and identification of novel candidate factors involved in frataxin function	Dr Michele Lufino and Dr Richard Wade-Martins, Department of Physiology, Anatomy and Genetics, University of Oxford.
Gene Therapy of Friedreich's Ataxia: Generation of Herpesvirus FXN Vectors for use in humans	Dr Filip Lim, Department of Molecular Biology Universidad Autónoma de Madrid (Spain)
Early and Longitudinal Assessment of Neurodegeneration in the Brain and Spinal Cord in Friedreich's Ataxia	Pierre-Gilles Henry, Assistant Professor, Dr Christophe Lenglet, Assistant Professor, Center for Magnetic Resonance Research, Department of Radiology University of Minnesota Medical School (USA)
Targeting metabolic remodelling as a therapeutic strategy for cardiomyopathy in Friedreich's ataxia	Dr Christopher Carroll, St George's University, London (UK)

Ataxia UK, 12 Broadbent Close, London N6 5JW

Office 020 7582 1444 Helpline 0800 995 6037 office@ataxia.org.uk www.ataxia.org.uk

Co-chairs: Richard Brown MBE and William Littleboy Chief Executive: Sue Millman

Patrons: Kim Wilde Dom Joly Paul Coia Prof Bob Williamson Jamie Raven James Moore

Ataxia UK is a Charity registered in Scotland (SC040607) & England & Wales (1102391); & Limited Company (4974832)

RNA-FISH evaluation of the FXN gene transcription: focus on cell-to-cell and cell cycle modulation, and the role in origin and evolution of the Friedreich's ataxia GAA-repeat mutation	Prof Antonella Russo, University of Padova (Italy)
Studying G-CSF as a potential treatment for Friedreich's Ataxia	Dr Alastair Wilkins, Dr Kevin Kemp, & Professor Neil Scolding, University of Bristol (Bristol, UK)
Effectiveness of LSVT in improving communication in people with Friedreich's Ataxia	Prof Anja Lowit, Strathclyde University (Glasgow, UK)
An investigation to determine the efficacy and safety of lentivirus mediated FXN gene delivery for the correction of Friedreich's Ataxia	Dr Michael Themis & Dr Mark Pook, Brunel University London (London, UK)

Cerebellar ataxia Archive

Research Project	Principal Researchers
Detecting retinal changes in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) and other ataxias using optical coherence tomography (OCT)	Professor Paola Giunti, University College London, UK
Unravelling the pathophysiological mechanisms of ARCA 2	Dr Hélène Puccio, Institut de Genetique et de Biologie Moleculaire et Cellulaire (IGBMC), France
MOVE' n FUN: videogame-based coordinative training in children with degenerative ataxia	Dr Matthis Synofzik and Dr Winfried Ilg, University of Tübingen, Germany
Pharmacologic therapy for Machado-Joseph disease (SCA3): from a C elegans drug screen to a mouse model validation	Dr Patricia Maciel, Dr Anabela Silva Fernandes and Dr Andreia Teixeira-Castro, University of Minho, Portugal
Endogenous Cerebellar Stem Cells and their Potential Neuroregenerative Role in Inherited Ataxias	Professor Silvia Marino, Barts and the London School of Medicine and Dentistry
Neural regeneration in the cerebellum: Development of cell replacement strategies for the management of spinocerebellar ataxias	Dr Gian Giacomo Consalez, San Raffaele Scientific Institute, Milan, Italy; Professor Ferdinando Rossi, University of Turin, Italy Physiotherapy research
Kv4.3 mouse model to reveal the disease mechanism underlying SCA19	Dr Anna Duarri, Department of Genetics, University Medical of Groningen
Antisense oligonucleotide mediated skipping of the CAG containing exon as a therapeutic approach for ataxias caused by a CAG repeat expansion	Dr W.M.C van Roon-Mom, Leiden University Medical Center, the Netherlands

Understanding the role of interruptions in polyQ diseases	Paola Giunti, Annalisa Pastore, University College London and National Institute for Medical Research, London
Coenzyme Q10 a potential therapeutic target for ataxia: Evaluation of therapeutic strategies	Dr. Iain P. Hargreaves, Dr Simon R.J. Heales, Dr. Shamima Rahman, Department of Molecular Neurosciences, Institute of Neurology, University College of London
Development of an assay for inhibitors of the deleterious interaction between wildtype and episodic ataxia-2 mutant Cav2.1 channels	Professor Annette Dolphin, University College London.
Improving hearing devices for Friedreich's ataxia and spinocerebellar ataxia	Dr Gary Rance, Department of Audiology and Speech Pathology, University of Melbourne (Australia), and Dr Kai Uus, School of Psychological Science, University of Manchester (UK)
Investigating cerebellar glutathione in SCA14 by MR spectroscopy in order to evaluate a new treatment option	Dr Sarah Doss & Dr Jan Leo Rinnenthal, Department of Neurology, Charité University Medicine Berlin (Germany)
RBFox proteins as critical determinants for cell toxicity in DRPLA and other Spinocerebellar ataxias	Dr Manolis Fanto, Department of Basic and Clinical Neuroscience, King's College, London, UK
The role of glia in the pathogenesis of the ataxic syndrome Dentatorubropallidoluysian Atrophy (DRPLA)	Dr Manolis Fanto, Department of Basic and Clinical Neuroscience, King's College, London, UK
An investigation of the effects of Dynamic Lycra Orthoses (DLOs) in the management of movement control problems caused by cerebellar ataxia	Martin Watson, The University of East Anglia (UK)
Discrimination of wild type and mutant ATXN3 mRNA levels by qPCR: protocol design and implementation	Dr Mafalda Raposo, Dr Manuela Lima, University of the Azores (Portugal)
Effectiveness of LSVT in improving communication in people with Friedreich's ataxia	Prof Anja Lowit, Strathclyde University (Glasgow, UK)
Novel pharmacological approach to treat Episodic ataxia type 1	Dr Paola Imbrici, University of Bari (Italy)

*For more support or information please contact: Ataxia UK, 12 Broadbent Close, London, N6 5JW
Website: www.ataxia.org.uk.
Helpline: 0800 995 6037 Tel: +44 (0)20 7582 1444
Email: helpline@ataxia.org.uk.*