Research funded by Ataxia UK that has ended in the last few years (updated December 2024)

Friedreich's ataxia Archive

Research Project	Principal Researchers
Analysis of the mitochondrial dysfunction in FXN deficient neurons to generate a drug screening test	Prof Saul Herranz Martin, Complutense University of Madrid (Spain)
Molecular mechanisms of R-loop-mediated frataxin gene silencing	Prof Natalia Gromak, University of Oxford (UK)
Investigating the role of bioactive sphingolipids in Friedreich's ataxia (FRDA)	Dr Sara Anjomani Virmouni, Brunel University (UK)
Etravirine as a potential therapeutic for Friedreich ataxia	Dr Alessandra Rufini, University of Rome Tor Vergata (Italy)
Generating new Friedreich's Ataxia animal models for validating HSV-1 FXN gene therapy in Dorsal Root Ganglia	Dr Filip Lim, Autonomous University of Madrid (Spain)
European Friedreich's Ataxia Consortium for Translational Studies (EFACTS)	Massimo Pandolfo, Université Libre de Bruxelles (ULB), Brussels (Belgium), Jörg B. Schulz, University Hospital RWTH Aachen, (Germany), Paola Giunti University College London (London, UK), Alexandra Dürr, UPMC Université Paris (France), Mathieu Anheim, Hôpitaux niversitaires de Strasbourg (France), Sylvia Boesch, Medical University Innsbruck (Austria), Caterina Mariotti, Fondazione IRCCS Istituto Neurologico "C.Besta" (Italy), Enrico Bertini, Bambino Gesù Children's Research Hospital (Italy), Francesc Palau, Pediatric Institute for Rare Diseases – IPER (Spain)
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 (UK)
Identifying additional sensitive targets in Friedreich ataxia	Professor Richard Festenstein, Department of Medicine, Imperial College London (UK)
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 (UK)
Gene Therapy of Friedreich´s Ataxia: Generation of Herpesvirus FXN Vectors for use in humans	Dr Filip Lim, Autonomous University of 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)

Cerebellar ataxia Archive

Research Project	Principal Researchers
A joint model of online SLT intervention and peer support to enhance communication effectiveness and participation in people with progressive ataxia	Prof Anja Lowit, Strathclyde University (UK)
Preclinical development of an amelioration therapy for Dentatorubro-Pallidoluysian Atrophy	Dr Stevanin Giovanni, Institut du Cerveau et de la Moelle épinière (ICM), Paris (France)
Non-invasive transcranial cerebellar stimulation: double blind, randomised, sham-controlled study followed by an open label extension phase	Dr Barbara Borroni, University of Brescia (Italy)
DNA repair pathways underlie common genetic mechanisms that modulate onset in spinocerebellar ataxias and other inherited ataxias	Dr Conceicao Bettencourt, University College London (UK)
Modelling SCA11 in cultured cells using CRISPR/Cas9	Dr Mariana Graca, Insitute for Molecular and Cell Biology (Portugal)
Regulation of alternative splicing of voltage-gated Ca2+ channels by CRISPR/Cas9-mediated genome editing as potential genetic therapy for episodic ataxia type 2	Dr Lorenzo Cingolani, Center for Synaptic Neuroscience (NSYN), Fondazione Istituto Italiano di Tecnologia (IIT), Genoa (Italy)
Neuroprotective therapeutic approach for Spinocerebellar Ataxia type 2: pharmacological targeting of AMPK	Dr Clévio David Rodrigues Nóbrega, Universidade do Algarve (Portugal)
Modelling the molecular pathogenesis of ARSACS with patient cells: disrupted proteostasis in ARSACS neurons	Professor Paul Chapple, Queen Mary University of London & Professor Michael Cheetham, University College London (UK)

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 pathophysiolgical 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 (UK)
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 (Italy)
Kv4.3 mouse model to reveal the disease mechanism underlying SCA19	Dr Anna Duarri, Department of Genetics, University Medical of Groningen (the Netherlands)
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 (UK)
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 (UK)
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 (UK)
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)

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