

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	Prof Saul Herranz Martin, Complutense
in FXN deficient neurons to generate a	University of Madrid (Spain)
drug screening test	
Molecular mechanisms of R-loop-mediated	Prof Natalia Gromak, University of Oxford
frataxin gene silencing	(UK)
Investigating the role of bioactive	Dr Sara Anjomani Virmouni, Brunel University
sphingolipids in Friedreich's ataxia (FRDA)	(UK)
Etravirine as a potential therapeutic for	Dr Alessandra Rufini, University of Rome Tor
Friedreich ataxia	Vergata (Italy)
Generating new Friedreich's Ataxia animal	Dr Filip Lim, Autonomous University of Madrid
models for validating HSV-1 FXN gene	(Spain)
therapy in Dorsal Root Ganglia	
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,	Dr Mark Baker, Prof Stuart Baker, & Prof
painless and simple screening test for the	Patrick Chinnery, Newcastle University,
onset of corticospinal tract disease/dorsal	Newcastle (UK)
root ganglionopathy in Friedreich's ataxia	
An investigation to determine the efficacy	Dr Mark Pook and Dr Michael Themis, Brunel
and safety of lentivirus mediated FXN gene	University, Uxbridge (UK)
delivery for the correction of Friedreich	
ataxia	

Ataxia UK, 12 Broadbent Close, London N6 5JW Office 020 7582 1444 Helpline 0800 995 6037 <u>office@ataxia.org.uk</u> Ataxia UK is a Charity registered in Scotland (SC040607) & England & Wales (1102391); & Limited Company (4974832)

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Professor Richard Festenstein, Department of
Medicine, Imperial College London (UK)
Professor Richard Festenstein, Department of
Medicine, Imperial College London (UK)
Dr Roberto Testi, Department of Experimental
Medicine, University of Rome Tor Vergata
(Italy)
Dr Michele Lufino and Dr Richard Wade-
Martins, Department of Physiology, Anatomy
and Genetics, University of Oxford (UK)
, , , ,
Dr Filip Lim, Autonomous University of Madrid
(Spain)
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)

recessive spastic ataxia of Charlevoix- Saguenay (ARSACS) and other ataxias using optical coherence tomography (OCT) Unravelling the pathophysiolgical mechanisms of ARCA 2 MOVE'n FUN: videogame-based coordinative training in children with degenerative ataxia Pharmacologic therapy for Machado- Joseph disease (SCA3): from a C elegans drug screen to a mouse model validation Pharmacologic therapy for Machado- Joseph disease (SCA3): from a C elegans drug screen to a mouse model validation Inherited Ataxias Neural regeneration in the cerebellum: Development of cell replacement strategies for the management of strategies for the reveal the disease mechanism underlying SCA19 Understanding the role of interruptions in polyQ diseases therapeutic approach for ataxias caused by a CAG repeat expansion Understanding the role of interruptions in polyQ diseases for the deleterious interaction between wildtype and episodic ataxia-2 mutant cava: 1 channels Improving hearing devices for Friedreich's valuate a new treatment option REFox proteins as critical determinants for Refor ataxia: exelutation of therapeutic coll coxicity in DRPLA and other Valuers (UK) Dresting the approach for Friedreich's valuate a new treatment option REFox proteins as critical determinants for Coell toxicity in DRPLA and other Cell toxicity		
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An investigation of the effects of Dynamic	Martin Watson, The University of East Anglia
Lycra Orthoses (DLOs) in the management	(UK)
of movement control problems caused by	
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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.