2017

International Ataxia Research Conference

Pisa, Italy 27-30 September

gofar

ATAXIA
Ataxia UK

FARA Friedreich's Ataxia Research Alliance

Dear All,

I would like to give you a warm welcome to the Second International Ataxia Research Conference.

GoFAR is proud to host the Conference in the ancient city of Pisa. Although it is known worldwide for its Leaning tower, the city is home to a number of architectural treasures. It conserves the memories of its glorious past as a Maritime Republic.

Pisa is a seat of one of the oldest universities in the world and famous for being the birthplace of, among others, Galileo Galilei, the father of a modern scientific thought. We hope that the example of Galileo and the magical atmosphere that surrounds Pisa, can generate new vigor among the participants to adequately respond to the great need of patients with ataxia of an effective treatment.

Our aim is to have a stimulating conference where participants will bring new ideas and fruitful collaborations paving the road for a cure.

Patients ask for a cure!

We thank you in advance for contributing to the success of the Conference by your attendance and participation.

Filomena D'Agostino
President of GoFAR

Dear Participants,

Welcome to the Second International Ataxia Research Conference.
This conference is co-organized by GoFAR (Italy), Ataxia UK and FARA (USA).
We are delighted to welcome you to Pisa, in Italy for three and half days of discussion of the ataxias.

The meeting has been designed as a comprehensive scientific review of new research from disease definition to therapeutic treatments across different types of ataxia.
We have also included a few select talks from people outside of the ataxia research community, who will bring new ideas to our field. We have academic researchers, clinicians, industry drug developers, regulators, patient group representatives and people with ataxia attending this meeting, and hope to foster collaborations between attendees from different areas.

We hope the meeting will provide you with many new ideas and new collaborations that will help your research and drug development efforts move forward. Speakers have been asked to leave time at the end of every talk for discussion, the idea being that you will use that time to ask questions and to provide constructive suggestions.
We believe that increased discussion, cross discipline learnings and collaboration will help us to move closer to treatments and cures for ataxias.

Patients are at the heart of everything we do as patient organizations.
We have included a few people with ataxia and family members in the conference program as we know that greater understanding of the disease from the patient perspective will only improve our collective research efforts. We hope you will take the opportunity to spend time with these individuals and learn from them what is important to them about their diseases, and how they perceive and value the research that you do.
The work you do every day represents hope to the patient community – one step closer to a treatment or cure.

We hope you enjoy the conference.

GoFAR
Ataxia UK
FARA

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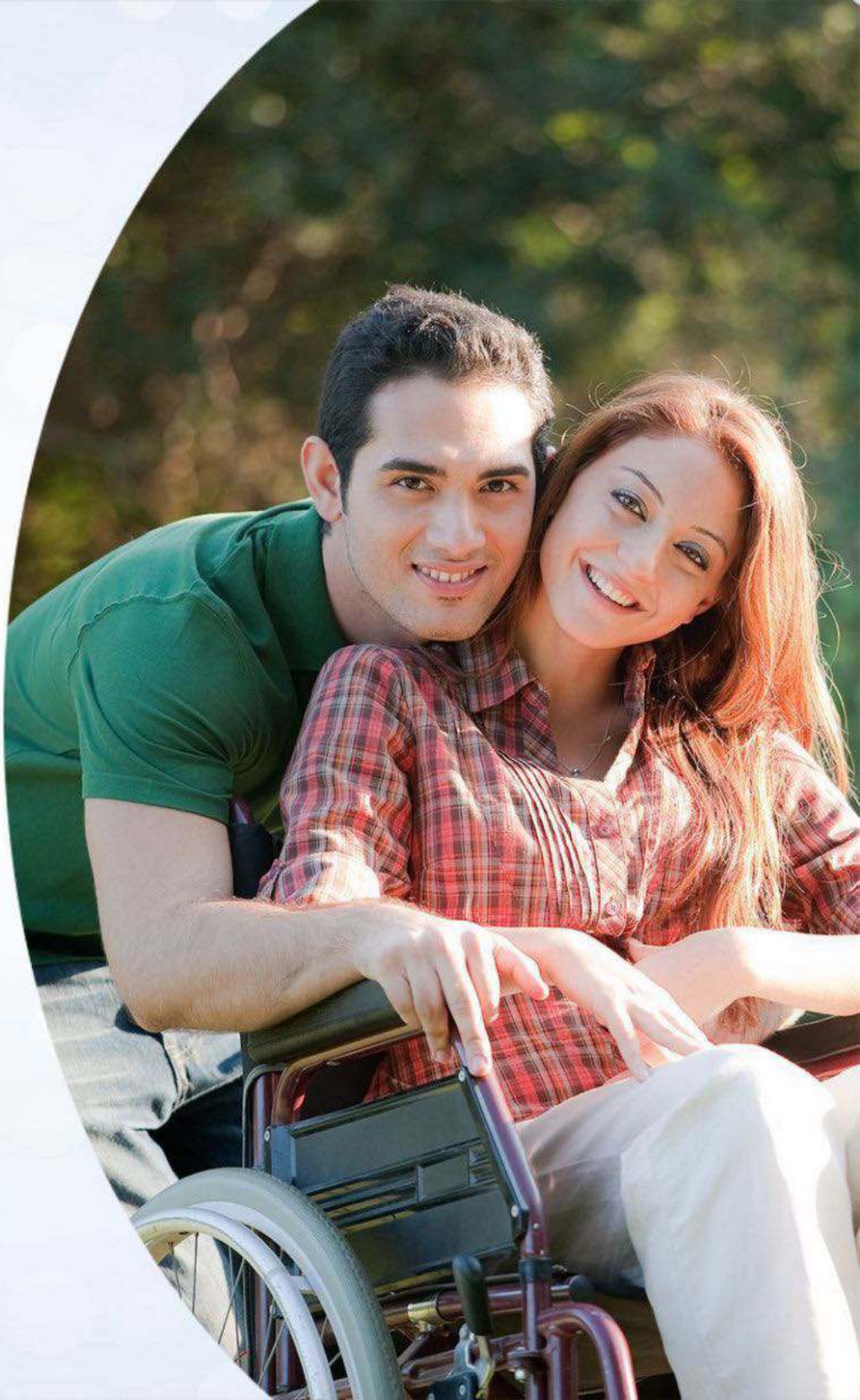
VISIT OUR TABLE IN THE EXHIBIT HALL FOR MORE INFORMATION ABOUT MOXIE, A STUDY IN FRIEDREICH'S ATAXIA

REATA PHARMACEUTICALS

MOXIe
a study in Friedreich's ataxia



Wednesday
27th September 2017

11.30 **Registration**

12.00-01.30 **Mentoring event for young investigators**

01.30-01.40 **Opening remarks - GoFAR**

01.40-06.35 **SESSION 1: MOLECULAR BASIS OF DISEASE**
Chairs: ***M. Napierala, M. Synofzik***

01.40-02.10 The rapid progress in next-generation genetics of ataxias: insights, challenges, and next steps - *M. Synofzik*

02.10-02.30 Elucidating the genetic background of childhood-onset ataxias - *E. Ignatius*

02.30-02.50 Genotype-phenotype correlation of mutant SLC25A46 disrupting mitochondrian fission in cerebellar degeneration - *J. Steffen*

02.50-03.10 Genes that affect synaptic excitability and trasmission identified by rare variant analyses in episodic ataxias - *V. Salpietro*

03.10-03.30 Novel SCA gene FAT Atypical Cadherin 2 is a regulator of autophagy *D. Verbeek*

03.30-03.50 Afg3l2 missense mutation p.Met665Arg impairs m-AAA protease function - new hints into a therapeutic strategy for SCA28 - *C. Mancini*

03.50-04.00 Discussion

04.00-04.15 **COFFEE BREAK**

04.15-04.45 The presence and relevance of autoantibodies to CNS proteins in patients with cerebellar ataxia - *A. Vincent*

04.45-05.05 Ataxin-2 regulated mitochondrial precursors to mantain nutrient balance and cellular energetics - *N. E. Sen*

05.05-05.25 Understanding the pathophysiological and molecular mechanisms underlying the recessive ataxia ARCA2 - *T. Jaeg-Ehret*

05.25-05.45 E3 ligase RNF126 directly ubiquitinates frataxin, promoting its degradation: identification of a potential therapeutic target for Friedreich ataxia *M. Benini*

05.45-06.05 Regulation of neuronal mRNA splicing by ATXN3 is disturbed in SCA3/MJD *A. Neves-Carvalho*

06.05-06.25 Epigenetic silencing in Friedreich ataxia is caused by hypermethylation of the FXN CpG island shore - *S. Bidichandani*

06.25-06.35 Discussion

06.45- 08.00 **WELCOME RECEPTION**

Thursday
28th September 2017

08.30-08.40 **Welcome - FARA**

08.40-10.40 **SESSION 1: MOLECULAR BASIS OF DISEASE**
Chairs: ***M. Napierala, M. Synofzik***

08.40-09.10 Spinocerebellar ataxia type 1 (SCA1): molecular basis of neuro-degeneration in the cerebellum (ataxia) and brainstem (lethality) - *H.Orr*

09.10-09.30 Transcriptional profiling of isogenic iPS-derived Friedreich's ataxia sensory neurons - *E. Soragni*

09.30-09.50 Early cerebellar mitochondrial biogenesis deficits and OXPHOS complex I and II deficiency in the KIKO mouse model of Friedreich ataxia - *H. Lin*

09.50-10.10 Addressing mitochondrial function in a mouse model of Friedreich's ataxia (FRDA) - *R. Abeti*

10.10-10.30 Mitofusin-dependent ER stress mediates degeneration in a Drosophila model of Friedreich's ataxia - *J. Navarro*

10.30-10.40 Discussion

10.40-10.55 **COFFEE BREAK**

10.55-03.00 **SESSION 2: TRANSLATIONAL MODELS OF DISEASE**
Chairs: ***P. Maciel, L. Petrucelli***

10.55-11.25 Targeting repeat expansion in cellular models of Friedreich's ataxia *M. Napierala*

11.25-11.45 Understanding Friedreich's ataxia neuropathology using a new conditional neuronal mouse model - *C. de Montigny*

11.45-12.05 A SCA7 mouse model showing multisystem phenotypes; new opportunities for pathomechanism studies and therapeutic development - *Y. Trottier*

12.05-12.35 Inducible and reversible phenotypes in a novel mouse model of Friedreich's ataxia - *V. Chandran*

12.35-12.45 Discussion

12.45-01.20 **LUNCH**

01.20-01.50 Repeat disorders: models, markers and more - *L. Petrucelli*

01.50-02.10 Voluntary running prevents onset of symptomatic Friedreich's ataxia in mice - *Z. Yan*

Engineering DNA Therapeutics for Rare Diseases

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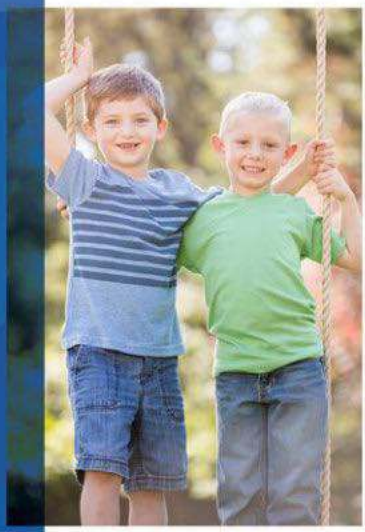
2017 International Ataxia Conference

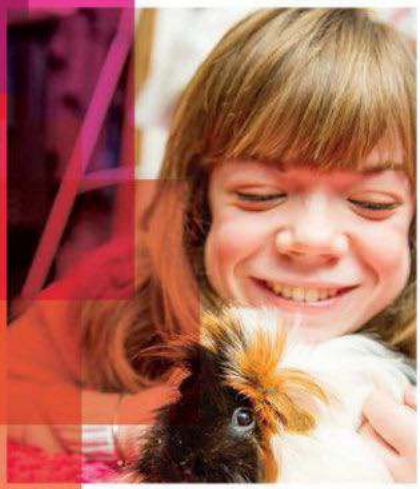
Thursday
28th September 2017

02.10-02.30	Using mouse models and BioID proteomic approach to understand ARSACS pathophysiology - *R. Lariviere*
02.30-02.50	Let-7 activates autophagy and alleviates motor and neuropathological deficits in pre and post-symptomatic Machado-Joseph disease mouse models-*S. Duarte*
02.50-03.00	Discussion
03.00-03.10	**COFFEE BREAK**
03.10-05.20	**SESSION 3: NATURAL HISTORY, BIOMARKERS AND ENDPOINTS** **Chairs: *P. Giunti, G. Manfredi***
03.10-03.40	Overview of natural history of Friedreich ataxia - *J. Schulz*
03.40-04.10	Natural history of the spinocerebellar ataxias (SCAs) - *T. Klockgether*
04.10-04.30	Autosomal recessive spastic ataxia of Charlevoix-Saguenay: a natural history study over a two year follow up - *C. Gagnon*
04.30-04.50	Detailing the natural history of Friedreich ataxia; loss of ambulation in the CCRN-FA study - *C. Rummey*
04.50-05.10	Long-term quality of life, depression and activities of daily living in the most common spinocerebellar ataxias (SCA1, SCA2, SCA3, SCA6) *H. Jacobi*
05.10-05.20	Discussion
05.30-07.30	**POSTER SESSION 1**

Friday
29th September 2017

08.30-08.40	**Welcome - Ataxia UK**
08.40-12.50	**SESSION 3: NATURAL HISTORY, BIOMARKERS AND ENDPOINTS** **Chairs: *P. Giunti, G. Manfredi***
08.40-09.10	Longitudinal MRS, MRI and DTI in the spinal cord in Friedreich's ataxia: 24-month follow-up - *P.G. Henry*
09.10-09.30	Basal ganglia and posterior fossa structural abnormalities in SCA3 stratified for disease stages - *J.L. Ribeiro de Paiva*
09.30-09.50	CCFS: a quantitative score of cerebellar dysfunction and evolution in Friedreich ataxia - *A. Durr*





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BioMarin is proud to support the 2017 International Ataxia Research Conference.

At BioMarin, we are inspired and driven by the patients who receive our therapies, and we will continue our efforts to help more patients living with rare conditions who have unmet medical needs.

We are dedicated to making a meaningful impact in the lives of patients affected by rare genetic disorders that are often underserved and ignored.

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Friday
29th September 2017

09.50-10.10	Corticokinematic coherence in patients with Friedreich ataxia correlates with GAA1 repeat expansion and SARA score - *G. Naejie*
10.10-10.20	Discussion
10.20-10.35	**COFFEE BREAK**
10.35-10.55	Exercise stress testing on adaptive equipment is feasible and reliable in Friedreich ataxia- *K. Lin*
10.55-11.15	Developing a clinically meaningful instrumented measure of upper limb function in Friedreich ataxia - *L.Corben*
11.15-11.35	Cardiac magnetic resonance T1 mapping as a window into the myocardium in Friedreich ataxia (FRDA) - *K. Lin*
11.35-11.55	Auditory dysfunction and its remediation in individuals with spinocerebellar ataxia - *K. Uus*
11.55-12.05	Discussion
12.05-12.50	Roundtable discussion on patient perspective on clinical trials and studies *M. Varbaro - F. Fortuna - C. Van Doorne - J. Dieusaert - A. Nadke*
12.50-01.30	**LUNCH**
01.30-05.35	**SESSION 4: THERAPEUTICS AND CLINICAL TRIALS** **Chairs: *N. Muzyczka, M. Pandolfo***
01.30-02.00	Summary and lessons learned from Friedreich's ataxia clinical trials - *F. Saccà*
02.00-02.30	Innovative trial designs for rare diseases, with focus on use of innovative endpoints and potential use of registry data - *K. Roes*
02.30-03.30	Roundtable discussion on clinical trial design for ataxias *P. Balabanov - K. Bryant - J. Cavagnaro - L. Benatti - P. Giunti - D. Jacoby D. Lynch - M. Pandolfo - S. Petraglia*
03.30-03.50	**COFFEE BREAK**
03.50-04.20	Activation of frataxin expression by duplex RNAs and antisense oligonucleotides - *D. Corey*
04.20-04.40	Gene-targeted synthetic molecules stimulate transcription through repressive GAA-repeats in patient-derived Friedreich's ataxia cells - *A. Ansari*
04.40-04.55	Class-I HDAC inhibitors with improved potency and drug-like properties for de-repressing frataxin production in Friedreich's ataxia - *S. Bhagwat*

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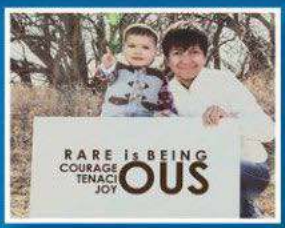

At Horizon Pharma, we understand that rare means many things to the millions of people affected by rare disease. One thing everybody shares is an urgency to accelerate the availability of new treatments. We salute the International Ataxia Research Conference and share their commitment to improving the lives of patients around the world.

HORIZON PHARMA

Friday
29th September 2017

04.55-05.10 RNA/DNA hybrid interactome uncovers DHX9 as a novel regulator of pathological R-loops in Friedreich's ataxia - *N. Gromak*

05.10-05.25 Safety, efficacy and pharmacodynamics of omaveloxolone in Freidreich's ataxia patients (MOXIe Trial): Part 1 results - *D. Lynch*

05.25-05.35 Discussion

05.35-07.00 POSTER SESSION 2

08.30 GALA DINNER
Arsenali Repubblicani, Via Bonanno Pisano 2

Saturday
30th September 2017

08.30-03.10 SESSION 4: THERAPEUTICS AND CLINICAL TRIALS
Chairs: ***N. Muzyczka, M. Pandolfo***

08.30-09.00 Lessons learned from recent approvals of therapies for neuromuscular disorders *J. Larkindale*

09.00-09.30 Overview of viral gene therapy approaches for genetic diseases - *N. Muzyczka*

09.30-09.50 Role of microRNAs in Machado-Joseph disease: from pathogenesis to therapy *V. Carmona*

09.50-10.10 Docosahexaenoic acid (DHA) supplementation as a therapy for spinocerebellar ataxia 38 (SCA38) - *M. Manes*

10.10-10.30 Neurotrophic factor and cytokine mimetics as new potential therapeutic agents for Friedreich ataxia - *J. Diaz-Nido*

10.30-10.40 Discussion

10.40-11.00 COFFEE BREAK

Before it became a medicine,

It was 5,000 researched compounds.

87 different protein structures.

500,000 lab tests.

1,600 scientists.

80-hour workweeks.

14 years of breakthroughs and setbacks.

36 clinical trials.

8,500 patient volunteers.

And more problems to solve than we could count.

Before it became a medicine,

It was an idea in the mind of a Pfizer scientist.

Now it's a medicine

That saves lives every day.

Pfizer

Driven to discover the cure

Saturday
30th September 2017

11.00-11.30	Gene therapy for Friedreich's ataxia - *B. Byrne, M. Corti*
11.30-11.50	Targeting the intracellular localization of ataxin-3 as a novel treatment approach for spinocerebellar ataxia type 3 - *T. Schmidt*
11.50-12.10	Ataxin-3 exon skipping as a treatment strategy for spinocerebellar ataxia *L. Toonen*
12.10-12.30	Nicotinamide mononucleotide supplementation in a model of Friedreich ataxia cardiomyopathy improves cardiac function and bioenergetics in a SIRT3 dependent manner - *A. Martin*
12.30-12.40	Discussion and presentation of Best Poster Awards
12.40-01.20	**LUNCH**
01.20-01.50	Correction of sensory ataxia in a novel mouse model of Friedreich ataxia using gene therapy approach - *H. Puccio*
01.50-02.10	TALEN and CRISPR gene editing for treatment of Machado-Joseph disease *S. Lopes*
02.10-02.30	Phenotypic and functional characterization of sensory neurons derived from human pluripotent stem cells and examining their in vivo capability to integrate into adult dorsal root ganglia - *M. Dottori*
02.30-02.45	Intravenous delivery of AAV gene therapy to CNS and peripheral tissues critical for the treatment of Friedreich's ataxia - *H. Patzke*
02.45-03.00	Effects of acetyl-DL-leucine in cerebellar ataxias - *T. Bremova*
03.00-03.10	Discussion
03.10-03.20	**CLOSING REMARKS**

NOTES

ORGANIZING COMMITTEE

Filomena D'Agostino - *GoFAR*

Annaluisa Ponchia - *GoFAR*

Julie Greenfield - *Ataxia UK*

Barry Hunt - *Ataxia UK*

Jen Farmer - *FARA*

Jane Larkindale - *FARA*

SCIENTIFIC COMMITTEE

Vijay Chandran

Paola Giunti

Julie Greenfield

Barry Hunt

Jane Larkindale – *Co-Chair*

Patrícia Maciel

Giovanni Manfredi

Nick Muzyczka

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