European LGMDR9 Community Conference

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Patient registries: Supporting research & the patient community

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AskBio MAKING HISTORY

atamyo therapeutics

LGMD2iFund Limb Girdle Muscular Dystrophy 2I Research Fund

AFMTÉLÉTHON INNOVER POUR GUERIR

CURELGMD2i Limb Girdle Muscular Dystrophy

MUSCULAR DYSTROPHY UK OUR MUSCLES MATTER

The SPEAK Foundation

GLOBAL FKRP REGISTRY

The John Walton Muscular Dystrophy Research Centre

Newcastle University

The Newcastle upon Tyne Hospitals NHS Foundation Trust

What are registries?

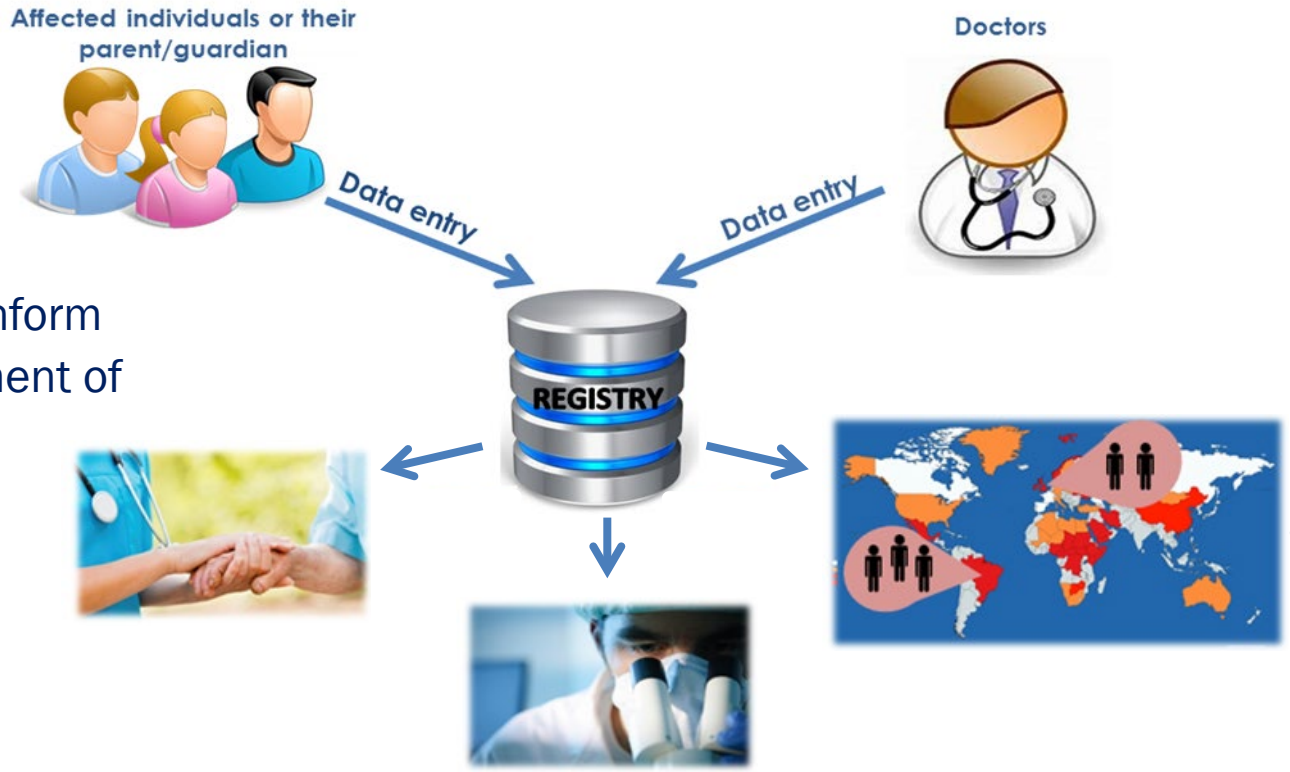
GLOBAL FKRP REGISTRY

- Research databases containing demographic, genetic and clinical information on individuals affected by a specific condition or gene mutation

- Can be regional, national or international depending on prevalence of specific condition
- Registration is frequently initiated by the patient. All registries must have patient consent to collect, store & share their data appropriately
- Registries can be either patient self-reported (usually via a secure website), clinically-reported (doctors provide information), or a combination of both

- Information collected should be reviewed & updated regularly

What are registries?



- Collect **real world data** to inform understanding & development of **standards of care**
- Provide **natural history** & **epidemiology** data
- Identify **participants** for clinical research & trials
- Inform **regulatory pathways** and monitor the **safety and efficacy** of new therapies
- Answer specific **research questions** from academics, healthcare professionals and industry
- **Link** the research community to the patients by offering a two-way flow of information

How does a registry work?

Patient (or parent/guardian) registers online

Patient completes online consent

Patient selects their doctor from Registry list of affiliated doctors

6 months/annually

Doctor completes questionnaires

6 months/annually

Patient completes questionnaires

How are registries used?

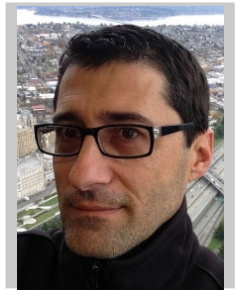
- In rare diseases, registries play a vital role in patient identification for research, study recruitment & subsequent therapy development

- **Who?** - Enquiries are received from academic researchers, industry, healthcare professionals & regulatory authorities

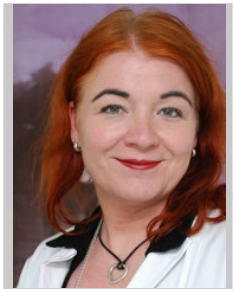
- **How?** - Submit an enquiry into the registry
  - Reviewed by registry Steering Committee
  - May be a cost associated with an enquiry

Steering committee






- **Dr Lindsay Alfano**, Nationwide Children's Hospital, USA
- **Kelly Brazzo**, CureLGMD2i Foundation, USA
- **Professor Nicholas Johnson**, Virginia Commonwealth University, USA
- **Dr Jean-Pierre Laurent**, LGMD2i Research Fund, USA
- **Dr Katherine Mathews**, University of Iowa, USA
- **Professor Volker Straub**, Newcastle University, UK
- **Dr Kristin Ørstavik**, Oslo University Hospital, Norway
- **Professor John Vissing**, University of Copenhagen, Denmark
- **Professor Maggie Walter**, Ludwig-Maximilians-University, Germany
- **Lacey Wood**, LGMDR9 patient, USA

The Global FKRP Registry

- Established 2011, coordinated at Newcastle University, UK
- International database of clinical and genetic data, with long-term follow-up
- Individuals with a confirmed diagnosis of a condition caused by changes in the **F**u**K**utin-**R**elated **P**rotein gene (FKRP)…

LGMDR9/2I	Limb Girdle Muscular Dystrophy, Type R9	*MDDGC5 (Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 5*
MDDGB5	Muscular Dystrophy-Dystroglycanopathy (Congenital with or without impaired intellectual development), Type B, 5	*MDC1C (Congenital muscular dystrophy-dystroglycanopathy)*
MDDGA5	Muscular Dystrophy-Dystroglycanopathy (Congenital with brain and eye abnormalities), Type A, 5	*FKRP-related Walker-Warburg Syndrome (WWS) and FKRP-related Muscle-Eye-Brain Disease (MEB)*

What is in the Global FKRP Registry?

GLOBAL FKRP REGISTRY

Patient-reported data

- Demographics
- Diagnosis
- Motor function
- Wheelchair use
- Myalgia
- Ventilation
- Family history
- Quality of Life

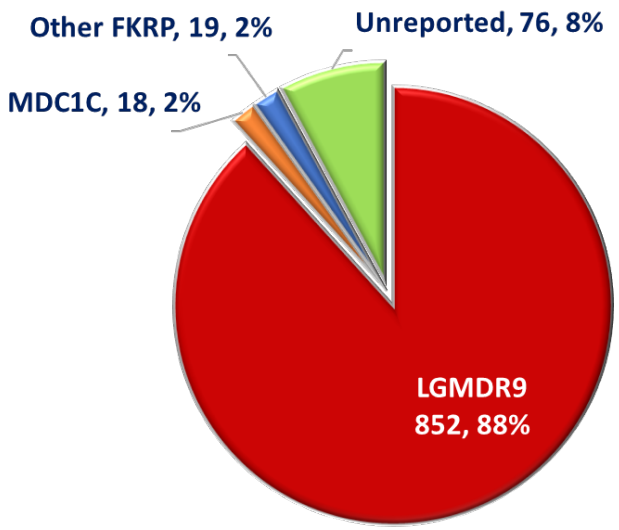
Doctor-reported data

- Disease onset
- Respiratory function
- Cardiac function
- MRI
- Cognitive function
- Contractures
- Other medical problems
- Function assessment scores
- Genetic mutation

Registration numbers

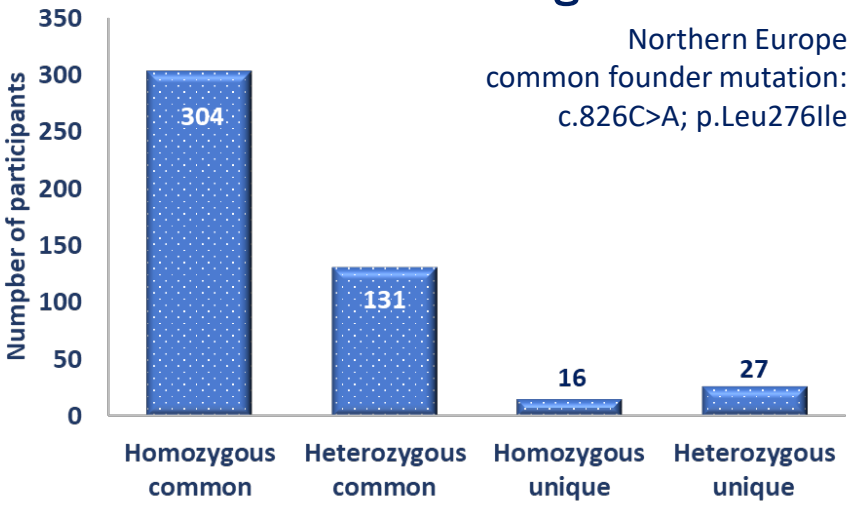
	Male	Female	Total
Patients	430	535	**965**
Genetically confirmed patients	217	261	**478**

Diagnosis



FKRP gene mutation

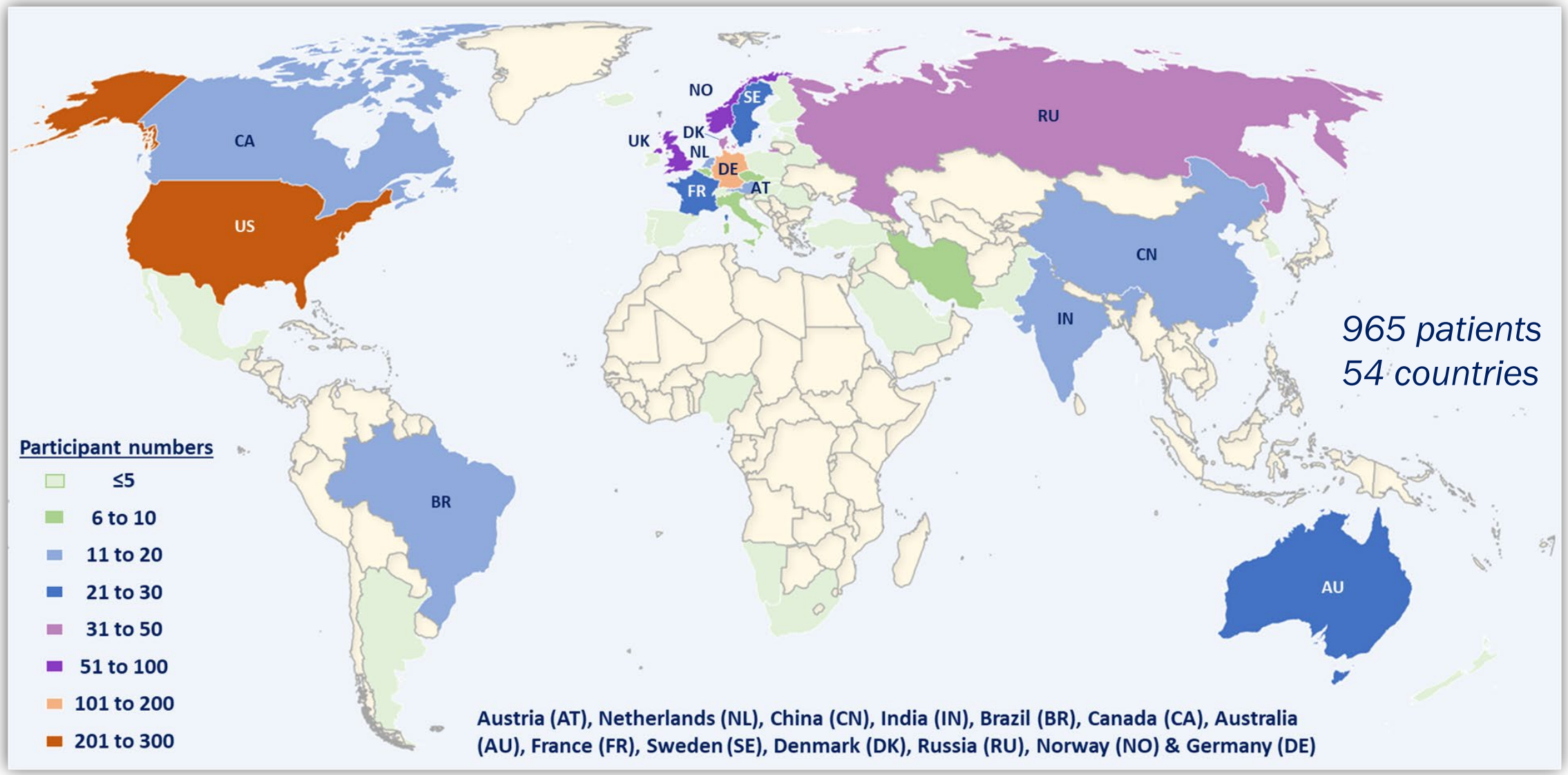
Northern Europe
common founder mutation:
c.826C>A; p.Leu276Ile



- 230 doctors from 35 countries
- New TREAT-NMD LGMD Core Dataset will be implemented this summer

Global distribution of global participants

GLOBAL FKRP REGISTRY



How is the Global FKRP Registry data used?

Recruitment of research participants

- Norwegian Natural History Study
- TREAT-NMD LGMD Standards of Care Survey
- Deflazacort Clinical Trial
- Experiences of Fertility and Pregnancy Survey
- Généthon Natural History Study
- ML Bio Solutions Natural History Study
- Wearable Devices and Outcome Measures Study
- GRASP Quality of Life Survey
- Gene Therapy Clinical Trial Atamyo Therapeutics

Data reports

- Longitudinal ventilation data, Généthon
- Longitudinal pain data, Newcastle University
- Longitudinal natural history data, ML Bio Solutions
- Current natural history data, Atamyo Therapeutics


ANNALS of Clinical and Translational Neurology
Open Access
RESEARCH ARTICLE
Global FKRP Registry: observations in more than 300 patients with Limb Girdle Muscular Dystrophy R9
Ana Topf[1], Pierre Laurent[8], venson[13],


Journal of Clinical Medicine
MDPI
Article
Prevalence of Pain within Limb Girdle Muscular Dystrophy R9 and Implications for Other Degenerative Diseases
Mark Richardson, Anna Mayhew, Robert Muni-Lofra, Lindsay B. Murphy and Volker Straub *

Benefits to patients and families

- **Newsletters**
- Find out about relevant **disease-specific news**
- Sense of **community**
- **Point of contact** with the curator - we will try to find someone to answer your questions if we can't!
- **Contribution** to research

WITH THANKS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AFMTÉLÉTHON
CURE THROUGH INNOVATION


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Limb Girdle Muscular Dystrophy


The
SPEAK
Foundation


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John
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MUSCULAR DYSTROPHY
RESEARCH CENTRE
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The Newcastle upon Tyne Hospitals
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