

European LGMDR9 Community Conference  
**SATURDAY 25<sup>TH</sup> MAY 2024**

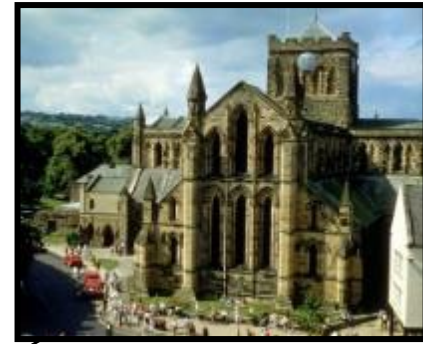
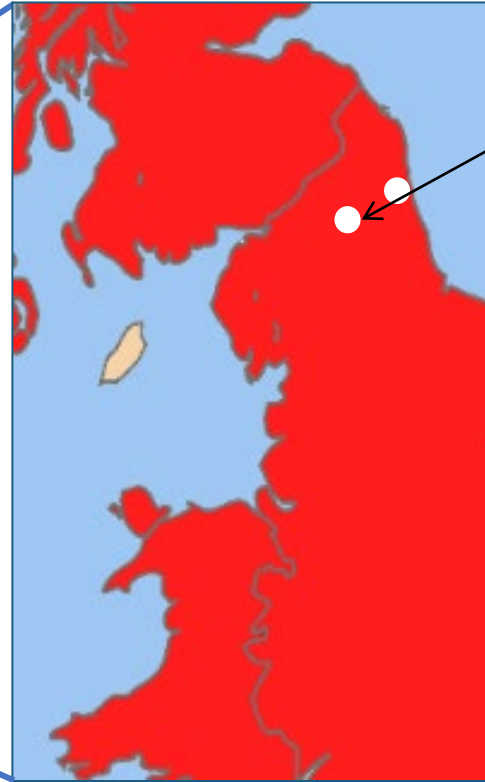
# Welcome!

Volker Straub

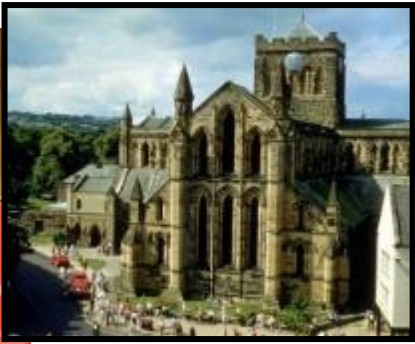




Newcastle



Hexham Cathedral



Hexham Cathedral

## The lost Cathedral

Around 674, work began on a stunning church and monastery at Hexham, inspired by the architecture that Wilfrid saw on his travels across Europe. He may even have brought continental masons to Hexham to rework stone taken from nearby Roman ruins.

Not since Roman times had there been such a building in this area: its grandeur bore witness to the majesty of God in the recently-converted kingdom of Northumbria. Within a few years, the church became a cathedral.

Yet just two hundred years after work first began, Hexham no longer had a bishop, and monastic life came to an end amid the chaos of Viking raids and settlement.





Territories and voyages of the Vikings

[https://commons.wikimedia.org/wiki/File:Territories\\_and\\_Voyages\\_of\\_the\\_Vikings\\_blank.png](https://commons.wikimedia.org/wiki/File:Territories_and_Voyages_of_the_Vikings_blank.png)

# Know your population

Viking founder mutations in Northumberland:

- LGMD2I/R9 FKRП-related
- LGMD2L/R12 anoctamin 5-related
- LGMD D4 calpain3-related



- |                            |        |
|----------------------------|--------|
| • LGMD R9 (FKRP)           | 21%    |
| • LGMD R12 (anoctamin 5)   | 15-20% |
| • LGMD R1 (calpain 3)      | 15%    |
| • LGMD R3-6 (sarcoglycans) | 13%    |
| • LGMD2B/R2 (dysferlin)    | 3%     |



Norwood et al., Brain, 2009  
Hicks, Sarkozy et al., Brain, 2010

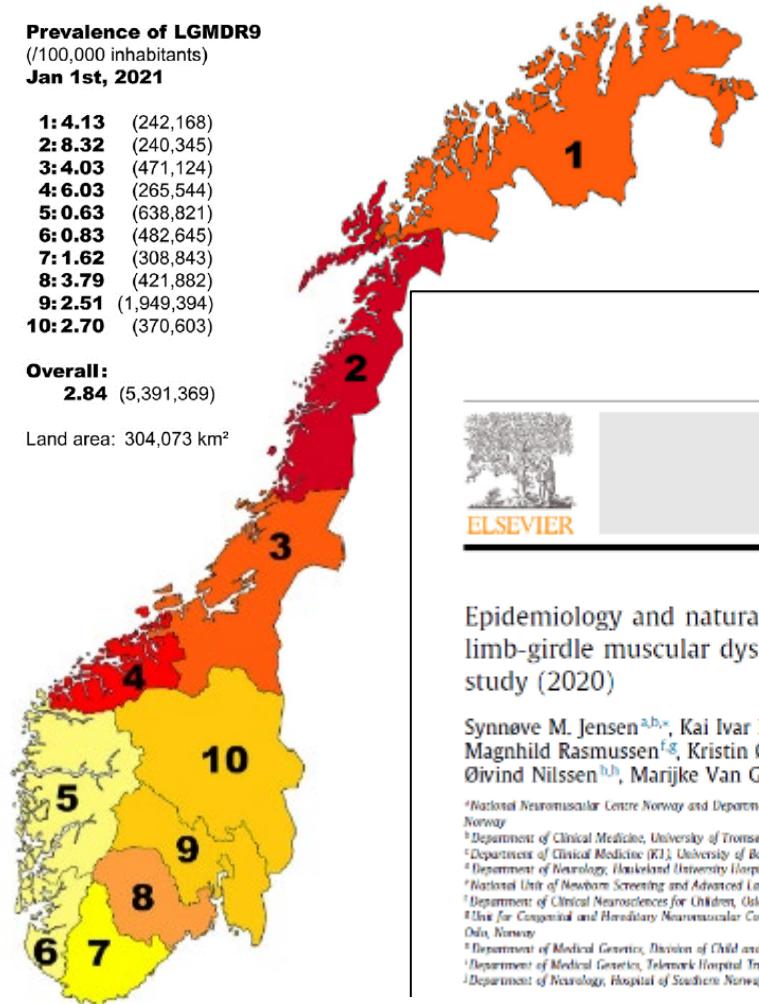
# LGMD R9 – natural history

## Prevalence of LGMDR9 (/100,000 inhabitants) Jan 1st, 2021

<b>1: 4.13</b>	(242,168)
<b>2: 8.32</b>	(240,345)
<b>3: 4.03</b>	(471,124)
<b>4: 6.03</b>	(265,544)
<b>5: 0.63</b>	(638,821)
<b>6: 0.83</b>	(482,645)
<b>7: 1.62</b>	(308,843)
<b>8: 3.79</b>	(421,882)
<b>9: 2.51</b>	(1,949,394)
<b>10: 2.70</b>	(370,603)

**Overall:**  
**2.84** (5,391,369)

Land area: 304,073 km<sup>2</sup>



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Contents lists available at ScienceDirect

**Neuromuscular Disorders**

journal homepage: [www.elsevier.com/locate/nmd](http://www.elsevier.com/locate/nmd)

**Epidemiology and natural history in 101 subjects with FKRP-related limb-girdle muscular dystrophy R9. The Norwegian LGMDR9 cohort study (2020)**

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## LGMD1

LGMD1A	5q31.2	<i>MYOT</i>
LGMD1B	1q22	<i>LMNA</i>
LGMD1C	3p25.3	<i>CAV3</i>
LGMD1D	7q36	<i>DNAJB6</i>
LGMD1E	2q35	<i>DES</i>
LGMD1F	7q32	<i>TNPO3</i>
LGMD1G	4q21	<i>HNRNPDL</i>
LGMD1H	3p23	?

## LGMD2

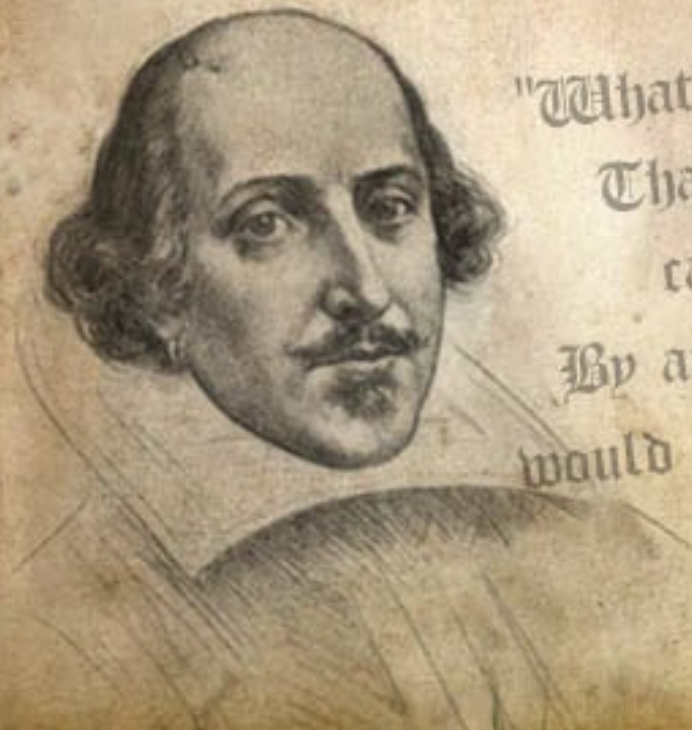
LGMD2A	15q15	<i>CAPN3</i>
LGMD2B	2p13	<i>DYSF</i>
LGMD2C	13q12	<i>SGCG</i>
LGMD2D	17q12	<i>SGCA</i>
LGMD2E	4q12	<i>SGCB</i>
LGMD2F	5q33	<i>SGCD</i>
LGMD2G	17q11	<i>TCAP</i>
LGMD2H	9q31	<i>TRIM32</i>
<b>LGMD2I</b>	<b>19q13</b>	<b><i>FKRP</i></b>
LGMD2J	2q	<i>TTN</i>
LGMD2K	9q34	<i>POMT1</i>
LGMD2L	11p13	<i>ANO5</i>
LGMD2M	9q3	<i>FKN</i>
LGMD2N	14q24	<i>POMT2</i>
LGMD2O	1p3	<i>POMGnT1</i>
LGMD2P	3p21	<i>DAG1</i>
LGMD2Q	8q24	<i>PLEC</i>
LGMD2R	2q35	<i>DES</i>
LGMD2S	4q35	<i>TRAPPC11</i>
LGMD2T	3p21	<i>GMPPB</i>
LGMD2U	7p21	<i>ISPD</i>
LGMD2V	17q25	<i>GAA</i>
LGMD2W	2q14	<i>PINCH2</i>
LGMD2X	6q21	<i>BVES</i>
LGMD2Y	1q25.2	<i>TOR1AIP1</i>
LGMD2Z	3q13	<i>POGLUT1</i>



1 = autosomal dominant  
2 = autosomal recessive

**90% LGMD2 (CK↑- ↑↑)**  
**10% LGMD1 (CK n - ↑)**

# What's in a name?



"What's in a name?  
That which we  
call a rose  
By any other name  
would smell as sweet."





**So, what's in a name?**  
**Everything!**  
**It's all we have!**

Stijn de Witt, web developer



you named me  
**WHAT!?**

## LGMD D

- LGMD D1 DNAJB6-related
- LGMD D2 TNPO3-related
- LGMD D3 HNRNPDL-related
- LGMD D4 calpain 3-related
- LGMD D5 collagen 6-related



**D = autosomal dominant**  
**R = autosomal recessive**

**90% LGMD R (CK↑- ↑ ↑)**  
**10% LGMD D (CK n - ↑)**

## LGMD R

- LGMD R1 calpain 3-related
- LGMD R2 dysferlin-related
- LGMD R3  $\alpha$ -sarcoglycan-related
- LGMD R4  $\beta$ -sarcoglycan-related
- LGMD R5  $\gamma$ -sarcoglycan-related
- LGMD R6  $\delta$ -sarcoglycan-related
- LGMD R7 telethonin-related
- LGMD R8 TRIM32-related
- LGMD R9 FKR-related**
- LGMD R10 titin-related
- LGMD R11 POMT1-related
- LGMD R12 anoctamin 5-related
- LGMD R13 fukutin-related
- LGMD R14 POMT2-related
- LGMD R15 POMGnT1 -related
- LGMD R16 dystroglycan-related
- LGMD R17 plectin-related
- LGMD R18 TRAPPC11-related
- LGMD R19 GMPPB-related
- LGMD R20 ISPD-related
- LGMD R21 POGLUT1-related
- LGMD R22 collagen 6-related
- LGMD R23 laminin  $\alpha$ 2-related
- LGMD R24 POMGnT2-related
- LGMD R25 BVES-related
- LGMD R26 POPDC3-related
- LGMD R27 JAG2-related
- LGMD R28 HMGCR-related

“LGMD, inheritance (R or D),  
order of discovery (number),  
affected protein”



Save  
the  
Dates

Iowa Wellstone  
Dystroglycanopathy  
Patient & Family  
Conference

July 12-13, 2024  
Ivins Agency COVID-19 Hall  
& Conference Center

CONNECTING  
FOR A CURE

September 28, 2024  
Multiple locations across the  
US & Canada

Stay tuned for more information at [cardgndz.com](http://cardgndz.com)



## LGMDR9 European Community Conference

Saturday 25<sup>th</sup> May 2024  
Doubletree by Hilton, Amsterdam Centraal Station

### Agenda

<i>Chair: Dr Kate Adcock, Muscular Dystrophy UK</i>		
09:00	Welcome and Keynote	Volker Straub (UK)
09:20	Patient advocacy	Mélanie Bordes (France), Kathryn Bryant-Knudson (USA), Kelly Brazzo (USA), Aleksandra Leijenhurst-Le Belle (The Netherlands)
09:40	Patient registries: Supporting research and the patient community	Lindsay Murphy (UK)
09:50	Physiotherapy session  Playing sports with a muscular dystrophy  The sailing experience!	Meredith James (UK)  Hedwige Van Steen (Belgium)  Laetitia Mercier & Ramsey Deforce (Belgium)
10:35 Coffee break (15 minutes)		
<i>Chair: Professor Volker Straub, Consultant Neurologist, Newcastle University, UK</i>		
10:50	Respiratory involvement and management	Robert Muni-Lofra (UK)
11:10	Cardio involvement and management	Karim Wahbi (France)
11:30	How do clinical trials work?	Gorka Fernández (France)
11:45	Clinical trial updates	John Vissing (Denmark)  Nick Johnson (USA)
12:15	Clinical trial readiness	Isabelle Richard (France)
12:30	Natural History studies	Andreas Rosenberger (Norway)
12:45 Lunch (1h 15 minutes)		

### Breakout sessions

	Room: Leeds 1 Parents with young children	Room: Birmingham 1 - Glasgow 2 Teenagers & young adults	Plenary Room and Leeds 2 Older adults	
<i>Patient leads:</i>	<i>Kelly Brazzo Julia Davage</i>	<i>Maud Cornelissen Cerys Davage Cadi Davage</i>	<i>Mélanie Bordes Kathryn Bryant-Knudson</i>	
<i>Clinicians:</i>	<i>Kathy Mathews Erik Niks Menno van der Holst</i>	<i>Lone Knudsen Sam Geuens Volker Straub Aleksandra Leijenhurst-Le Belle Lindsay Alfano Elise Dupitier (session 1)</i>	<i>Tracey Willis Nick Johnson Kristen Ørstavik Andreas Rosenberger Teresinha Evangelista Tanya Stojkovic Elise Dupitier (session 2)</i>	
14:00 Session 1	Strategies for school	Mental health & Strategies for school and college	Room: Leeds 2 Healthcare management (English)	Room: Plenary Healthcare management (French)
14:45 Session 2	Healthcare management	Free time & physiotherapy sessions if desired.	Room: Leeds 2 Managing pregnancy	Room: Plenary Managing work

15:30 Coffee & Connections (45 minutes)

### Plenary session

<i>Chair: Kathryn Bryant-Knudson, The Speak Foundation, USA</i>				
16:15	Expert panel:	Erik Niks Nick Johnson Andreas Rosenberger	Kathy Mathews Teresinha Evangelista Kristen Ørstavik	Volker Straub Tanya Stojkovic Elise Dupitier

16:55 Closing remarks and thanks (Volker Straub)

17:00 End

19:00-22:00 Conference Dinner



**You are the LGMD R9 experts!!!**

# European LGMDR9 Community Conference



# WITH THANKS



# Thank You!



Heather Hilsden



Lindsay Murphy



**Performing world class translational research to bring diagnosis, care and therapy to people with neuromuscular disease**

