

Understanding Fabry in Families Study – the availability of pedigree testing, genetic counselling and understanding of inheritance across Fabry International Network countries

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Introduction

Fabry disease is a multi-systemic, X-linked lysosomal disease arising from a deficiency of the enzyme alpha-galactosidase, that can affect multiple generations of the same family.

Pedigree analysis (the testing of relatives following a diagnosis) is a powerful tool to aid in the diagnosis, establish inheritance patterns, and ensure the early detection of potentially affected relatives.^{1,2}

Genetic counselling recommendations related to issues in Fabry include:³

- Explaining the natural history and inheritance pattern of the disease
- Genetic testing support (including pre-natal testing)
- Provision of resources about the disease

The *Understanding Fabry in Families Study* was conducted to identify gaps in the availability of pedigree testing and genetic counselling across Fabry International Network (FIN) member countries.

Aims

To gain insight into the accessibility of pedigree testing and genetic counselling, for individuals living with Fabry, around the world.

Methods

The online survey was open to any individual (≥ 18 years), or a parent/carer of a child, with a diagnosis of Fabry disease; consent was sought from all participants.

Individuals were recruited by individual country FIN patient organisations (n=52) via an email invitation with a link to the appropriate language(s) for their members.

Results

In total, **717** responses were received across **41 countries**, with the most responses coming from the USA (n=214), UK (n=83) and Australia (n=61), respectively.

Here we present data from the total study population and global regions

- Global regions were defined (Table 1) and regional sub-analyses were run for Europe

Table 1. Regions

Africa	Morocco, Tunisia, South Africa
Asia	Azerbaijan, India, Japan, Turkey, Hong Kong, Israel
Europe	Denmark, United Kingdom, Sweden, Finland, Republic of Ireland, Lithuania, Norway, Greece, Italy, Spain, Slovenia, Russia, Poland, Romania, Belgium, France, Germany, The Netherlands, Switzerland, Hungary
Europe (Eastern)*	Russia, Poland, Romania, Hungary
Europe (Western)*	Belgium, France, Germany, The Netherlands, Switzerland
Southern America	Argentina, Brazil, Chile, Costa Rica, French Guiana, Mexico, Venezuela, Peru
Northern America	United States, Canada
Oceania	Australia, New Zealand

Pedigree analysis

Pedigree analysis had been performed for 67% of respondent's families (Figure 1).

The highest rates of pedigree analysis were in Southern America (85%) and Asia (81%), conversely, the lowest rates were in Africa and Oceania (50% and 51%, respectively).

Similar rates were noted across Northern America (70%) and Europe (66%)

- Western European families were less likely to have undergone pedigree analysis than families in Eastern Europe (54% and 75%, respectively).

Genetic counselling

Approximately half (53%) of respondents had been offered genetic counselling (Figure 2).

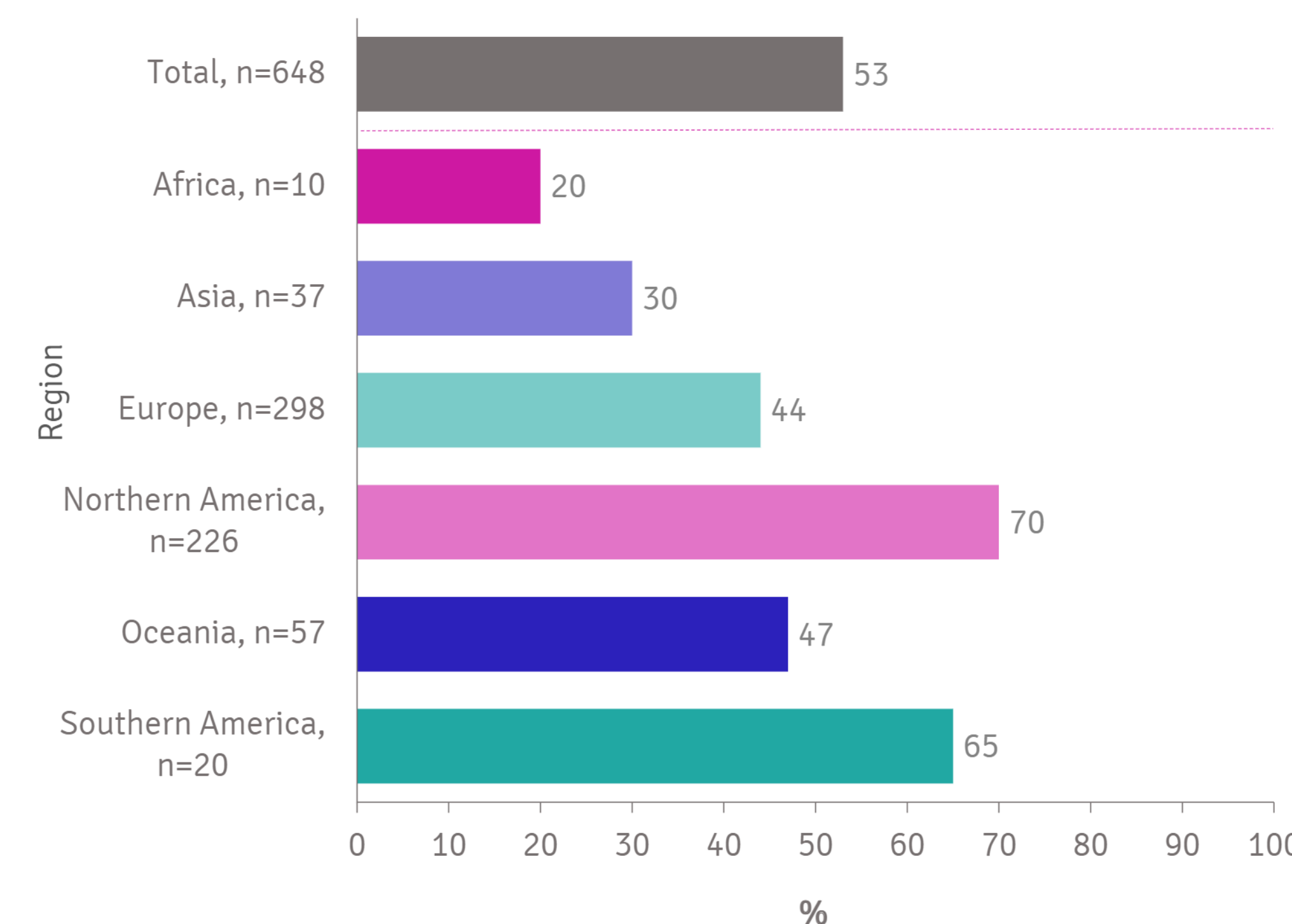


Figure 2. Genetic counselling offered

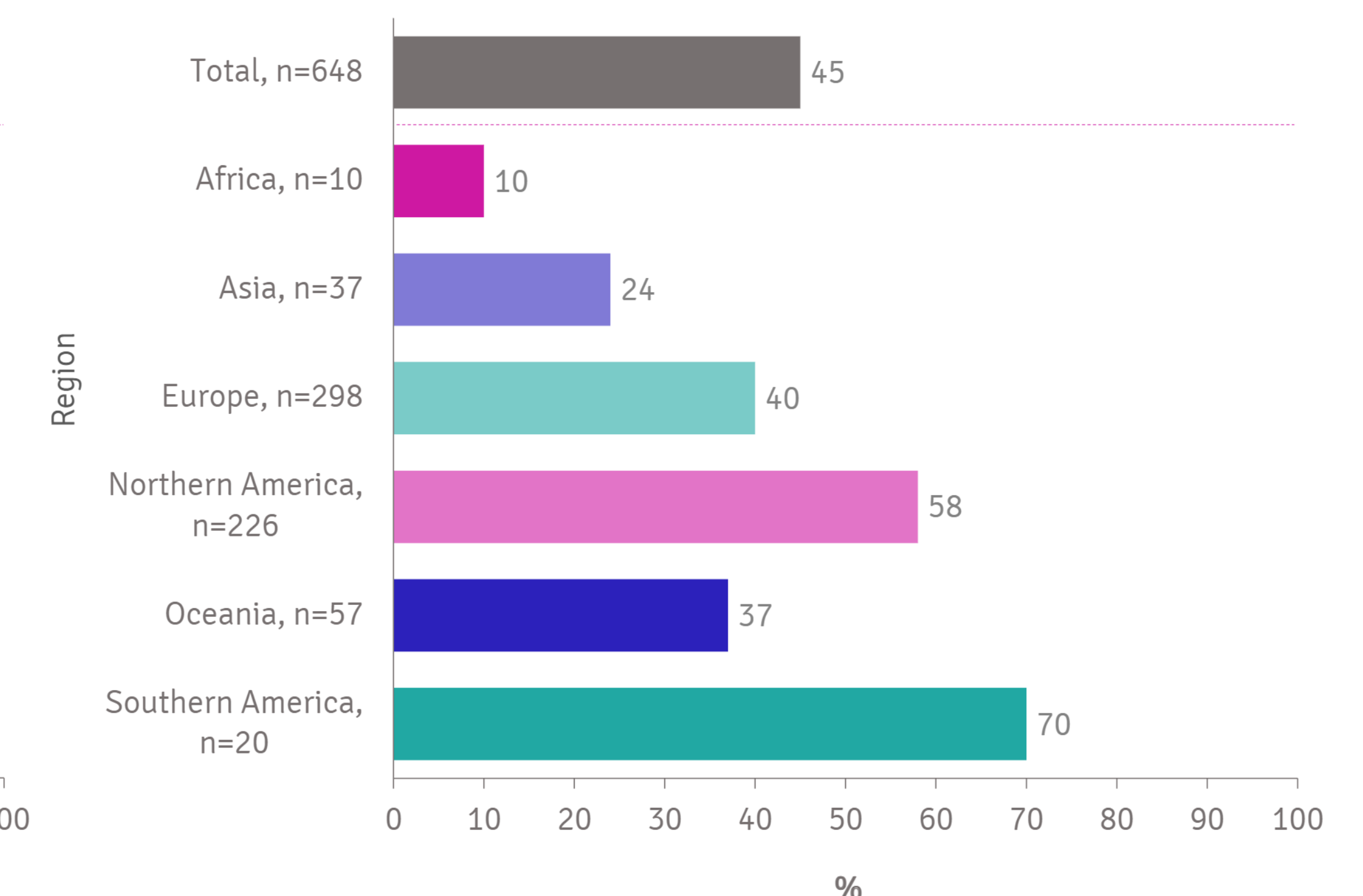


Figure 3. Genetic counselling accepted

Genetic counselling was more likely to be offered to individuals in Northern and Southern America (70% and 65%, respectively) than to those in Africa (20%) or Asia (30%).

Whilst genetic counselling had been offered to 44% of European respondents, rates were lower in Eastern Europe (30%) than Western Europe (51%).

Overall, of those who had been offered genetic counselling, 45% accepted the offer (Figure 3).

Acceptance of genetic counselling was highest in Southern America (70%) and Northern America (58%) and lowest in Africa (10%) and Asia (24%).

The majority (92%) had received an explanation on how Fabry disease is inherited

- This pattern was seen across: Europe (94%); Northern America (93%); Southern America (90%); Oceania (88%); and Asia (84%)
- Whilst still the majority, only 60% of respondents in Africa had been told about inheritance patterns

Take home information was available to 57% of the study population to consolidate what they had just been told about inheritance

- The availability of this information was highest in Southern America (72%) and lowest in Africa (0%)

Conclusions

This study identified regional gaps in the uptake of pedigree analysis and the provision/uptake of genetic counselling.

Further analysis of the data collected in this study may identify individual countries which would benefit from support to provide these services.

A further study to understand the reasons behind the low acceptance of pedigree testing and genetic counselling is warranted.

Acknowledgements

Christine Lavery, MPS Society, UK, the originator of this study.
Rebecca Brandon, Fabry Disease Advocacy Support Worker, MPS Society UK, for her input into the survey design.
Fabry International Network who distributed the survey to their members GfK Switzerland for the data analysis.
Tom Kenny, Rare Disease Research Partners, for his support in the development of this poster.

References

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2. Laney DA *et al.* 2008. *J Genet Couns*; 17:79.
3. Laney DA *et al.* 2013. *J Genet Couns*; 22:555.

Disclaimer

Shire awarded the MPS Society (UK) a grant to undertake this research; the MPS Society commissioned Rare Disease Research Partners to carry out this study. MPS Commercial (trading as Rare Disease Research Partners) is a wholly owned, not for profit subsidiary of the Society for Mucopolysaccharide Diseases (MPS Society UK) whose social objectives are to reinvest any profits to support the MPS Society mission of transforming the lives of patients through specialist knowledge, support, advocacy and research.