

# Patient organisations working in partnership to research the patient experience of rare diseases – the Sanfilippo (MPS III) survey

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## Background

- Patient organisations for rare diseases are frequently approached by companies to help describe disease natural history and the impact on patients and their families
- The UK Society for Mucopolysaccharide Diseases (MPS Society) has a long established history of conducting surveys to address these questions and also performing our own research
- When several companies are working on one disease many of their knowledge gaps will overlap
- Continued requests for families to provide the same information can be an unnecessary burden and affect both the quantity and quality of information that we are able to collect
- Given that a number of companies currently have MPS III research programmes we identified an opportunity for both patient organisations and pharmaceutical companies to work together on a study to provide further understanding of the disease

## Study aims

There are a number of key challenges facing future treatments of MPS III:

- Highly variable onset and severity makes predicting the clinical course of disease very difficult
- Steep cognitive decline in the second stage of disease means that early diagnosis is essential for optimum outcomes of new treatments<sup>1</sup>
- Delay in the diagnosis of MPS III is common, but there is little information available on the length of delay and barriers to diagnosis
- Early symptoms are often non-specific, such as recurrent ear, nose and throat infections and diarrhoea<sup>2</sup> and behavioural issues that may be mistaken for variants of normal childhood behaviour<sup>3</sup>

To address these challenges and support the development and reimbursement of future therapies, we designed a study with three broad aims (Figure 1).

## Collaboration

- MPS Commercial (the non-profit subsidiary of the MPS Society UK) approached companies with the proposed study
- We would develop a disease specific questionnaire and give sponsoring companies an opportunity to review and add any specific questions relevant to their research within the framework supplied
- A key feature of the study would be the involvement of the member patient organisations of the European and International MPS Networks who would work with us to identify patients and conduct the research in their own countries

## Methods

- Following review by the sponsoring companies and two medical experts, we finalised the content of our questionnaire
- Members of the MPS Network were invited to take part in the study
- The questionnaire and accompanying Patient Information and Consent forms were translated as appropriate
- Parents/carers of individuals with MPS III A, B, C or D were invited to take part via their local patient organisation
- Questionnaires were completed by face to face interview, telephone interview, by post or on-line
- Informed consent was sought from all participants
- All participating patient organisations determined whether ethics approval was required in their country before starting the study

## Conclusions

Collaborative working in this way benefits the whole MPS III community in a number of ways:

### Shared knowledge

Through collaborative working in this way we hope to develop greater insights into the diseases we study. Countries with very small populations of these rare diseases benefit from the insights gained from an international study.

### Support for smaller patient organisations

The patient organisations involved vary in size and composition, from those consisting of only a small number of volunteers to those employing full or part-time staff. Many of the smaller organisations would not have the resources to conduct a large survey.

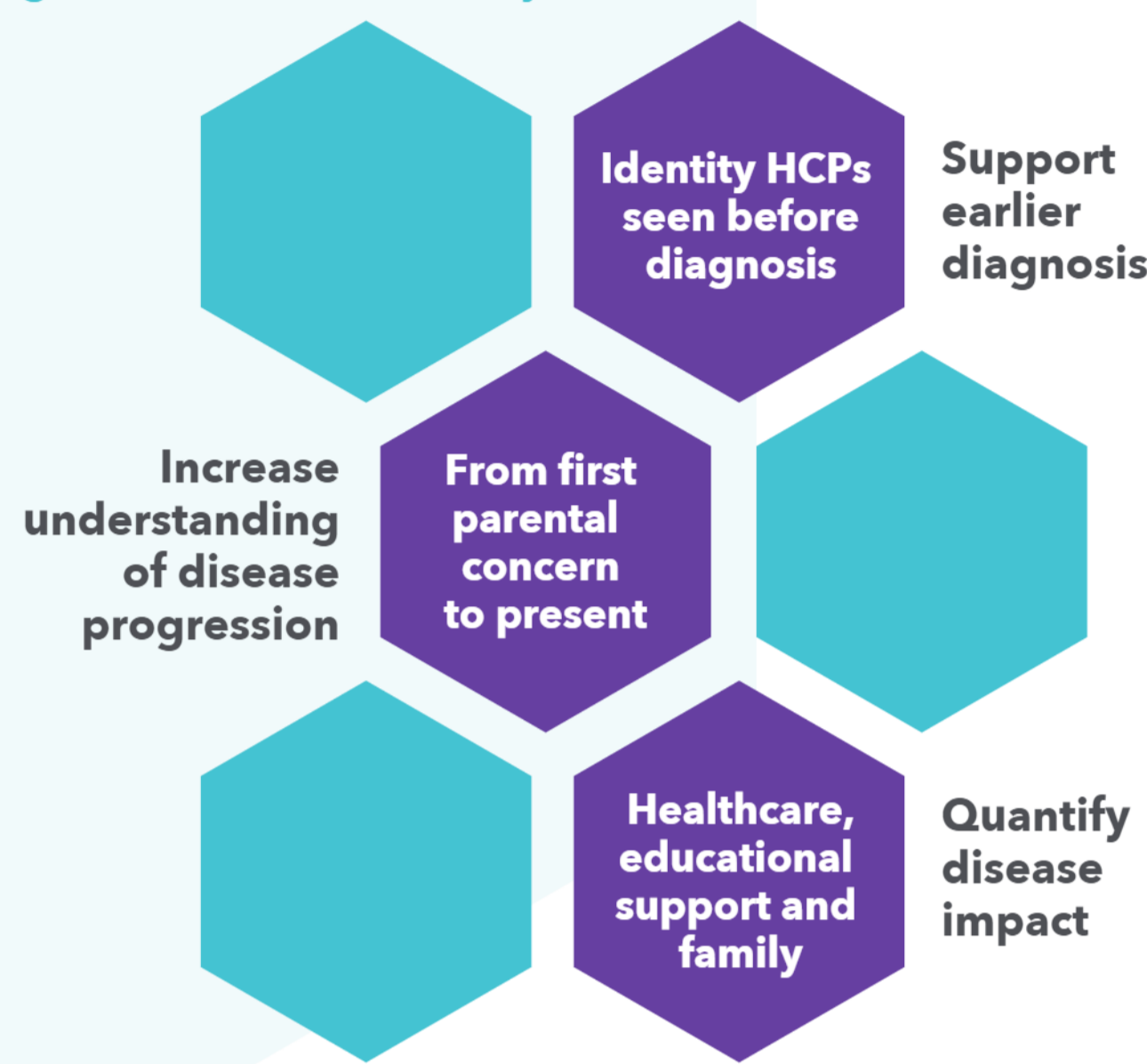
### Shared expertise

Some patient organisations are very experienced in conducting surveys, whereas for others, this was the first time they had undertaken a project of this nature.

### Fewer surveys for individuals and families to complete

Through collaboration we are able to share knowledge across countries, patient organisations and companies to reduce the overall burden of surveys on the individuals and families affected by MPS III.

Figure 1. Aims of the study



HCP: Health care provider

## Study progress

- The patient organisations from 13 countries have joined the project and collectively we have completed 133 questionnaires to date (Table 1)
- Data collection will be completed in the first quarter of 2018

Table 1. Responses to date

Country	Organisation	Number of responses to date
Austria	Gesellschaft für Mukopolysaccharidosen und ähnliche Erkrankungen	4
Australia	Mucopolysaccharide & Related Diseases Society Aust. Ltd	5
Canada	Canadian MPS Society	2
Germany	Gesellschaft für Mukopolysaccharidosen e.V	47
Greece	Greek Lysosomal Association	0
Indonesia	MPS and LSD Indonesia	1
Japan	The Japanese Society of the Patients and the Families with Mucopolysaccharidoses	0
New Zealand	Lysosomal Diseases New Zealand	0
Serbia	MPS Serbia	4
Spain	Asociación MPS España	11
Switzerland	MPS Schweiz	2
Turkey	MPS L H Derneği	16
UK	MPS Society UK	41

## References

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