Delphi Survey for Updated MPS II Clinical Guidelines

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OVERVIEW

- MPS II, or Hunter Syndrome, is a rare X-linked genetic disorder where the body lacks the enzyme (iduronate-2-sulfatase) to break down sugar molecules (GAGs), causing them to build up in cells, leading to progressive physical and mental development issues, affecting organs, joints, and the airway, primarily in boys. The clinical guidelines for Hunter syndrome (MPS II) were last updated in 2003.
- To update these guidelines and gather best practices from around the world, a study was initiated to collect input on consensus statements that inform treatment guidance for MPS II/Hunter syndrome. A Delphi Survey was developed with statements on overall care for MPS II patients and specialized care across neurology, cardiology, orthopedics, gastroenterology, pulmonology, ENT, and other relevant fields.

SCOPE & PURPOSE

- The purpose of this study is to collect input on consensus statements to inform the latest treatment guidance for MPS II/Hunter syndrome.
- Participants were asked to complete an online survey containing 77 questions, which took approximately 1 hour.
- The survey covers the following topics: frequency of assessments, exams, studies, and imaging; enzyme replacement therapy; hematopoietic stem cell transplant; ongoing therapies; anesthesia; airway issues; audiology testing; ear, nose, and throat issues; carpal tunnel surgery; cardiology issues; and central nervous system issues.

METHODS

- An overview of the literature was presented to a group of MPS experts, including the principal investigator and co-investigators. Based on the latest guidelines for treating MPS II patients and this comprehensive literature review, Delphi statements were developed and included in a Qualtrics survey.
- The online survey consists of 77 questions, and practitioners were asked to respond with a yes or no or indicate the frequency of care where indicated. The survey takes approximately one hour to complete.
- Each question was evaluated to determine the consensus outcome: Yes, No, or frequency. Statements with 75% or more answered in the affirmative are considered positive consensus statements. Those that fall below that threshold will not be regarded as positive consensus statements and will be considered for revision. The data collected through the survey will be aggregated and analyzed in a paper submitted to a medical journal.
- MPS II is a rare disease with fewer than 2,000 patients worldwide. As a result, there are very few practitioners who are familiar with the disease. The study team identified 76 practitioners who care for and treat MPS II patients through prior knowledge and recommendations from other MPS II practitioners, including MDs, PhDs, APPs, and those holding advanced degrees in relevant fields.
- Participants were asked to answer a few demographic questions to determine whether they qualified for the survey. The survey link was sent by Dr. Joseph Muenzer via email. The Delphi Survey was open from September 2025 to December 2025.

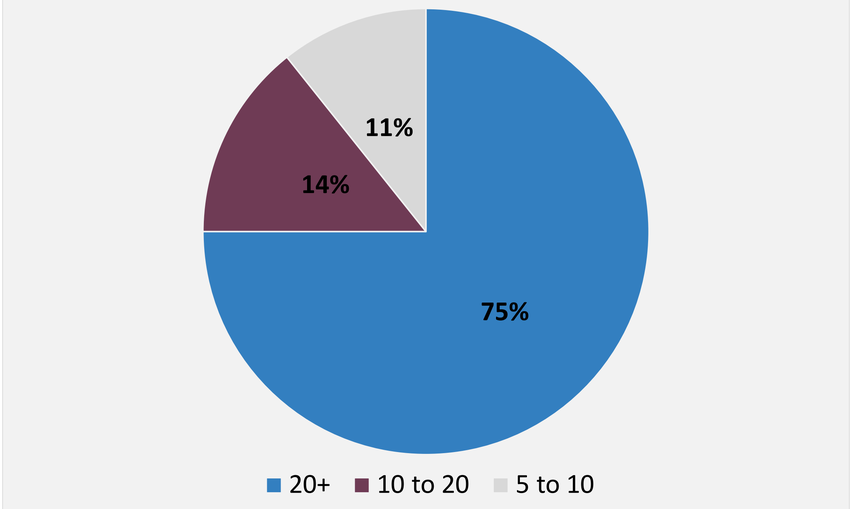
PARTICIPANTS

EDUCATION LEVEL	
MDs	20
MDs/PhDs	4
APP	4
PhD	1

SPECIALITY	
Geneticist	15
Pediatrician	7
Neurologist	4
Cardiologist	1
BMT Provider	1
Other	1

COUNTRY	
USA	15
Germany	5
UK	2
Mexico	2
Taiwan	1
Brazil	1
Argentina	1
France	1
Saudia Arabia	1

How many patients with MPS II have you seen in your practice?



CONCLUSIONS

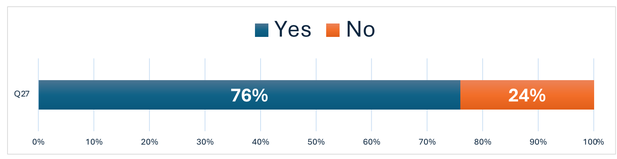
- The last MPS II treatment guidelines were published 23 years ago. Through research and practice, a tremendous amount of knowledge has emerged. With the rarity of the disease and the lack of specialists, new guidelines are needed to guide the treatment of those with MPS II. The insights from the Delphi Survey will guide practitioners with little to no MPS II experience and will provide updated guidelines to veteran MPS II practitioners. As a result, individuals with MPS II will receive the best possible care, informed by input from MPS II experts.

FUTURE DIRECTIONS

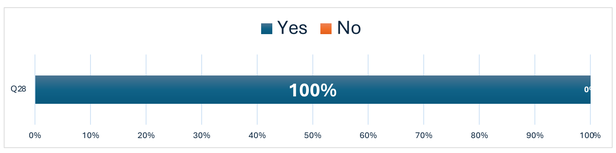
- The Delphi online survey will continue through March to allow for more participants. Once all the surveys have been completed, the study team will aggregate the data in Qualtrics. All results will be written up and submitted to an appropriate medical journal.

RESULTS

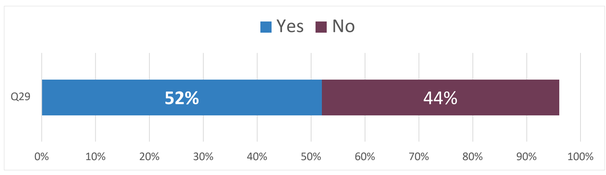
- Q27: Should ERT be started before the presentation of physical symptoms in an attenuated phenotype?



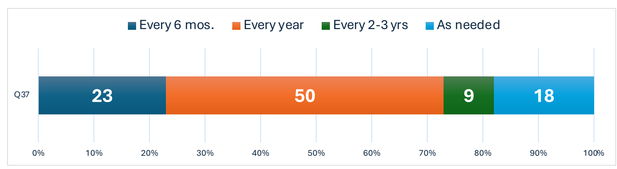
- Q28: Should ERT be started before the presentation of physical symptoms in a severe phenotype?



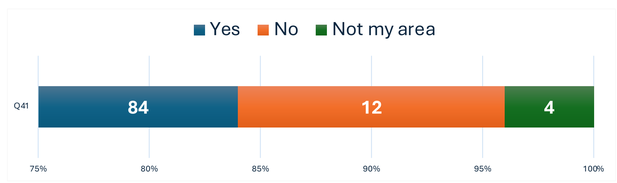
- Q29: In the absence of a definitive genotype, should IV ERT be started before the onset of physical symptoms?



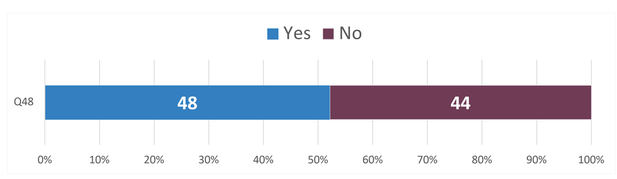
- Q37: How often should drug antibodies be tested in an MPS II patient receiving IV ERT?



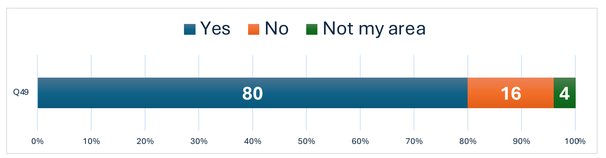
- Q41: Should HSCT be considered for patients with MPS II who are predicted to be severe?



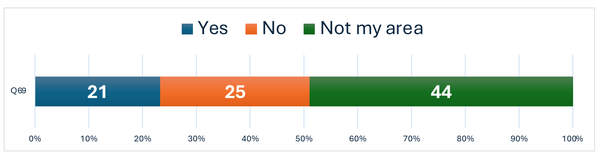
- Q48: Should neuromonitoring be considered for MPS II anesthesia cases that last less than 2 hours?



- Q49: Should neuromonitoring be used for all extended MPS II procedures/surgery under anesthesia over 2 hours in duration, regardless of involvement of the spine?



- Q69: Should tarsal tunnel release be recommended for patients who are toe walkers?



ACKNOWLEDGEMENTS

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REFERENCES

1. Neufeld E, Muenzer J. The mucopolysaccharidoses. In: C Scriver, A Beaudet, W Sly, eds. *The Metabolic and Molecular Bases of Inherited Disease*. McGraw-Hill; 2001: 3421-3452.
2. Muenzer J. Overview of the mucopolysaccharidoses. *Rheumatology (Oxford)*. 2011; 50 (Suppl 5): v4-v12.