

Results of MPS II and MPS III study: A Brief Synopsis

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Amanda conducted this study as part of her University of North Carolina at Greensboro Genetic Counseling Masters Program requirements. Barbara Wedehase served on her project committee.

In early 2007, families of children with MPS II and families of children with MPS III who are members of the National MPS Society participated in a study designed to provide preliminary information about how treatment decisions are made within this population. Some health care providers and families of patients consider hematopoietic stem cell transplantation (HSCT) with different forms of MPS as a possible treatment option. HSCT is a procedure in which stem cells in one person are replaced by fresh stem cells from another person. A stem cell is an unspecialized cell that is capable of developing into other types of cells, such as blood cells.

For patients with MPS II (Hunter syndrome) and MPS III (Sanfilippo syndrome) studies suggest the risks inherent in HSCT appear to be greater than possible benefits of the treatment. This qualitative study attempted to document just how frequently families are seeking HSCT as a treatment for MPS II and MPS III, and to identify what factors are most important to families in making this decision. Surveys sent to families coping with MPS II also included a section regarding enzyme replacement therapy (ERT). ERT involves replacing the deficient or missing enzyme in a person with a MPS disorder. The enzyme, made in a laboratory using advanced technology, is given by intravenous (IV) infusion on a regular basis. ERT is now an available treatment option for MPS II.

As a brief overview, the majority of respondents to the MPS II survey reported knowledge of HSCT as a treatment option for the condition (66.7%) as opposed to about half of respondents to the MPS III survey (53.8%). Of those who had heard of it, 33% of MPS II survey respondents and 46.4% of MPS III survey respondents considered HSCT as a treatment option for their children. And of those who considered the option, 50% of

MPS II survey respondents (11.1% of all respondents) and 23.1% of MPS III respondents (5.8% of all respondents) actually chose the option for their child.

Other questions asked in the survey included the following: how did you initially hear about HSCT (or ERT) and whom have you discussed this treatment option with, what were you told about HSCT (or ERT), how did the availability of other treatment options affect your decision regarding HSCT (or ERT), what were the most important factors in making these decisions, and what information do you have now that you would have wanted to know while making these decisions?

The following quotes demonstrate the extreme range of responses families have to the same information regarding HSCT as a treatment option for MPS II or MPS III.

Our decision was based on talking to other families and knowing the ultimate outcome of this horrible disease if we did nothing. We decided we would rather die than never have tried at all. We understood that this was an extreme and very risky option; we decided to have the courage to try and fight this horrid disease.

We made this decision a few years ago. I went back online when I received this survey and what did I find? A very sad journal that started out hopeful and ended up with death due to transplant, NOT due to Sanfilippo. Parents go through many phases of grief and I feel this procedure takes advantage of their desperation. I'm sure all involved mean well, but on a deeply spiritual level, I feel this treatment is not only ill-advised but immoral.

Families coping with MPS II and MPS III are faced with challenging and often limited options when making decisions about treatments for their children with MPS. It is clear that each family reaches their own decision about pursuit of particular treatment options with the quality of life and potential risks and benefits in mind.

It is important to remember that HSCT has not been proven to effectively treat either MPS II or MPS III. If you have any questions regarding HSCT, you should ask your doctor who may provide you with answers or refer you to a geneticist, genetic counselor or other specialist equipped to speak with authority on these issues. If you have any

additional questions regarding the study specifically, please contact Amanda Padro at amanda.padro@duke.edu with further inquiries.