screening (false-negatives) is an important issue.

The actual timing of a diagnosis is generally not seen to be as important as the manner in which the diagnosis is communicated to the family. Access to comprehensive counselling by appropriately qualified and trained professionals at the time of diagnosis is important, as is community and professional education. Parents requested that the counselling process be transparent and that the counsellor be self-aware and honest about the process of newborn screening.

Some participants considered that early diagnosis allows decisions to be made about early intervention and exploring various treatment options (e.g. diet, physiotherapy, bone marrow transplantation, enzyme replacement), as well as offering reproductive choice for future pregnancies and the relief of avoiding unnecessary and prolonged medical investigations. The majority of participants valued the "power" and choice that knowledge would give them.

The issue of parental bonding time with a newborn child is an important one. Some parents expressed gratitude that they had the opportunity of enjoying the time with their affected child before diagnosis, and also the opportunity of having more children before a diagnosis has been made. It was recognised that having a diagnosis in the absence of symptoms makes decisions about reproduction difficult.

Group 3 participants made the point of asking why newborn screening for these disorders should be introduced at a time when research and enzyme production timelines are so very long.

LDA thanks the MPS Society and its members for agreeing to participate in these sessions. Our particular thanks go to:

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Ms. Margaret Sahhar,
Dr. Ravi Savarirayn,
Professor David Sillence, and
Dr. Ed Wraith

Discussion groups will also be held with support groups for other lysosomal storage disorders over the coming months. It is intended that the issues identified in these sessions will form the basis of Focus Groups for more in-depth discussion of particular issues raised. Position Papers will be formulated from these discussions and presented for endorsement by the Human Genetics Society of Australasia (HGSA).

The information gathered through this process will complement the data collected from the Health Related Quality of Life research study that is also being conducted by LDA.

LYSOSOMAL DISEASES AUSTRALIA

HEALTH-RELATED QUALITY OF LIFE STUDY

Assessment of lysosomal storage disorders has traditionally relied on clinical examination by doctors and the results of laboratory tests. However, these approaches provide only limited information about the impact of the disorders on the broader day-to-day lives of affected individuals and their families, and little is known about their quality of life.

As a first step to address this issue, Lysosomal Diseases Australia is funding a new study on the quality of life of parents caring for children with lysosomal storage disorders. For statistical significance, the study will include parents from Australia and the United Kingdom and will be conducted by the Research and Evaluation Unit at the Women's and Children's Hospital in Adelaide.

Before undertaking the main study, it is first necessary to develop a reliable questionnaire, which can be used to obtain information about the impact of lysosomal storage disorders on parents. Appropriately qualified research staff from the Women's and Children's Hospital will be contacting parents of children with lysosomal storage disorders over the coming months to discuss the content of the questionnaire.

The parental study is intended to be the first in a series of studies that assess quality of life issues for people with lysosomal storage disorders and their families. The breadth of information that will be examined makes it necessary to focus on particular groups at different times. However, this should not restrict anyone from participating in this or a later study if they so wish. This focus is designed to ensure that we obtain this important information in the best manner possible, and to enable us to produce high quality research that will benefit both affected individuals and their families.

For more information about the study, please contact the study coordinator, Ms. Fiona Arney (Tel: (08) 8204 7790).

FABRY

Fabry disease is a lysosomal storage disorder that affects at least 1 in every 117,000 Australians. It is due to a deficiency of the lysosomal enzyme, alpha-galactosidase (α-galactosidase), and has been described in people from most parts of the world. Fabry disease is inherited in an X-linked recessive pattern, which means that females carry the gene and males are affected (see X-linked recessive inheritance below)

The disorder was first described independently in 1898 by Dr. Fabry from Germany and Dr. Anderson from England. Both were dermatologists. It was not until 1965 that the inheritance pattern was recognised by an American geneticist, Dr. Opitz. The deficient enzyme was first recognised by Dr Roscoe Brady (who pioneered enzyme replacement therapy in Gaucher disease, another lysosomal storage disorder) in 1967, and in 1970 it was specified as α-galactosidase by Kint and co-workers. The gene was discovered in 1986 by Bishop and co-workers.

The enzyme, α-galactosidase, is responsible for the breakdown of glycosphingolipids (complex compounds composed largely of sugars and fats). When the enzyme is deficient, there is an accumulation of these glycosphingolipids in blood vessels, body fluids and the cells of many tissues, particularly the nerves, kidney, heart and eyes.

During childhood and adolescence the main features in males are pain and paraesthesia (burning and tingling) of the hands and feet; hypohydrosis (reduced sweating); and the development of angiokeratoma which are the characteristic changes in the blood vessels of the skin and mucous membranes. Angiokeratoma appear as clusters of tiny dark red to black dots around the umbilicus (navel), buttocks, thighs, genitalia, and back (see accompanying photograph). They can also occur on the inside of the mouth. The pain is very severe and can either be constant or occur in episodes (Fabry crises) which can last a few days. Clouding of the front of the eye (cornea) and the lens develop at a young age. With advancing age the kidneys become affected and protein leaks out in the urine (proteinuria), the blood pressure increases