MUCOPOLYSACCHARIDOSES AND MUCOLIPIDOSES

Includes: MPS I (Hurler), MPS IS (Scheie), MPS IH/S (Hurler/Scheie), MPS II (Hunter), MPS III (Sanfilippo), MPS IV (Morquio), MPS VI (Maroteaux-Lamy), MPS VII (Sly), ML 1, ML 11 (I Cell Disease), ML III (Pseudo-Hurler polydystrophy)

FACTS

Mucopolysaccharidoses and mucolipidoses are lysosomal storage conditions. Within each cell in the body there are a number of different smaller units (organelles) which are involved in the function of the cells. A lysosome is one of these small organelles; they contain enzymes important in the working of the cells and therefore, the body. When there is a deficiency in one of the enzymes stored in the lysosomes, a lysosomal storage condition results.

In mucopolysaccharidosis and mucolipidosis, there is a deficiency or lack of enzymes which are important in the body’s metabolism, resulting in progressive physical and/or intellectual impairment. Several types exist with a range of severity. New treatments such as replacing the defective enzyme (enzyme replacement therapy or ERT) are being developed for several types of mucopolysaccharidoses.

Individuals with mucopolysaccharidosis conditions share many similar symptoms such as multiple organ involvement, distinctive ‘coarse’ facial features, and abnormalities of the skeleton, especially joint problems. In most cases, the mucopolysaccharidoses follow an autosomal recessive pattern of inheritance. Hunter syndrome, however, is inherited as an X-linked recessive condition. Genetic testing or enzyme testing is available for some forms of these conditions.

Support Groups can provide individuals and families with information about genetic conditions, community resources and an understanding and empathic ear.

If you are not able to contact the services listed below, please call the Association of Genetic Support of Australasia (AGSA) on (02) 9211 1462 or visit www.agsa-geneticsupport.org.au for support and assistance.

Further information and details of your local genetic counselling service can be found at www.genetics.edu.au, or please call The Centre for Genetics Education on (02) 9462 9599.