

GUIDE TO UNDERSTANDING MUCOLIPIDOSIS IV



What is Mucopolipidosis IV?

Mucopolipidosis IV (ML IV) is a rare genetic disorder belonging to a group of disorders known as the lysosomal disorders. It is most commonly seen in Jews of Eastern European background. Other names for this condition include Ganglioside Sialidase Deficiency and Sialolipidosis.

Whilst there is no cure for individuals affected by ML IV this fact sheet explores the disease's presentation and clinical management. This fact sheet is produced by the Society for Mucopolysaccharide Diseases drawing on the experiences of parents and doctors and with reference to medical literature.

What causes ML IV?

ML IV is a rare inherited metabolic disorder believed to be caused by alterations in a membrane protein due to mutations in the mucolin-1 gene. This results in the blockage of endocytic transport which is the absorption of materials, for example, into the cell. This prevents the lysosomes forming correctly which results in a storage of complex lipids within the lysosome. This can cause iron deficiency anaemia and the neurodegenerative features seen in ML IV.

How is ML IV inherited?

ML IV is an autosomal recessive disease; both parents must carry the same defective gene and each pass this same defective gene to their child. Where both parents are carriers of the ML IV gene there is a 25% (1:4) chance of having an affected child with each pregnancy. There is a 50% (1:2) chance of a child receiving only one copy of the defective gene and therefore being a carrier.

A carrier will not be affected but can pass the defective gene to his/her offspring. The remaining 25% (1:4) will be neither affected nor a carrier. Using information from an affected individual's DNA, it may be possible to determine whether brothers and sisters are carriers of, or affected by, ML IV.

For further information on the inheritance pattern of MPS and Related Diseases contact the MPS Society for a specialist booklet on inheritance.

Genetic counselling

All parents of children with a lysosomal storage disease should consider asking for genetic counselling before having other children. The counsellor should be able to provide non-directive advice on the risk to close relatives, reproductive choices available and to suggest whether the wider family should be informed.

Is there screening available for ML IV?

The presence of an increase in the level of the hormone gastrin in the blood is an important and relatively easy way of screening for the disorder.

Presentation of ML IV

ML IV is characterised by the following signs and symptoms which commonly appear in the first year:

- Pronounced developmental delays in gross motor skills, such as sitting, standing and walking;
- Pronounced developmental delay in fine motor skills;
- Pronounced developmental delays in speech;
- Vision problems of corneal clouding, retinal degradation, sensitivity to light;
- Low muscle tone (hypotonia)

Physiotherapy and hydrotherapy

Physiotherapy and hydrotherapy can be useful to help individuals with ML IV because of the difficulties with movement and activity due to the joint pain and stiffness associated with the condition. At other times it is common sense for the individuals to be as active as possible to improve their general health and the physiotherapist may be able to suggest ways of achieving this. The best forms of physiotherapy are exercises that are introduced through play in the younger individuals. In adults it is important to remember that passive stretching may be painful and should only be used with caution.

Treatment of ML IV

At present, there is no cure for ML IV, only treatment for the symptoms as they arise. As we learn more about the disease it is hoped that treatment may become available in the future.

About the MPS Society

The Society for Mucopolysaccharide Diseases is a voluntary support group, founded in 1982, which represents from throughout the UK over 1200 children and adults suffering from MPS and Related Diseases, their families, carers and professionals. It is a registered charity entirely supported by voluntary donations and fundraising and is managed by the members themselves. For further information about the work of the Society and the service we provide please contact us.

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