



Guide to Understanding ML III

Mucopolipidosis III



Society for Mucopolysaccharide Diseases



Sarah (ML III)

Front cover photographs from top to bottom:
Sarah and Tessa (both ML III)

What is ML III?

Mucopolipidosis Type III (ML III) is one of the lysosomal storage disorders known collectively as mucopolipidoses. They are closely related to the mucopolysaccharidoses. ML III was sometimes referred to as Pseudo-Hurler Polydystrophy as it resembled a less severe form of Hurler Disease, MPS I. Polydystrophy means that many organs are abnormal. ML III was first described in 1966 by Dr Maroteaux and Dr Lamy from France.

Whilst there is no cure for individuals affected by ML III this booklet explores the disease's presentation and clinical management. The booklet is produced by the Society for Mucopolysaccharide Diseases (the MPS Society) drawing on the experiences of parents and doctors with reference to medical literature.

What causes ML III?

In the course of normal life there is a continuous recycling process of building new materials and breaking down old ones ready for disposal. This activity takes place in a special part of the body's cells called the lysosome and the process itself requires a series of biochemical tools called enzymes. Enzymes can only reach the lysosomes after a special signal has been attached to them.

In individuals with ML III the signal is not attached so the enzymes are unable to get to the right place and are therefore lost outside the cell. Babies may show little sign of the disease but symptoms start to appear as more and more cells become damaged by the accumulation of unwanted deposits.

Does ML III affect individuals differently?

Mucopolipidoses are a group of storage disorders displaying a spectrum of clinical symptoms. At the severe end these are labelled ML II. Less severely affected patients are considered to have ML III. Information on ML II is not covered in this booklet but a separate booklet may be obtained from the MPS Society.

How common is ML III?

The MPS Society which co-ordinates the Registry for Mucopolysaccharide and Related Diseases has shown that ML III is a rare condition. Between 1989 and 1999, 5 babies were born with ML III in the UK.

How is ML III inherited?

ML III is an autosomal recessive disease whereby 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 III 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.

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

Can you test for ML III in pregnancy?

For each pregnancy the chances of a baby inheriting ML III are totally independent of whether a previous child was affected by the disease. Prenatal tests can be arranged early on during a pregnancy for those families who already have had a child with ML III. Both amniocentesis and chorionic villus sampling (CVS) can be used to diagnose the disease in utero. Most laboratories do prenatal tests on cultured cells and it may take three to four weeks for a result.

Genetic counselling

All parents of children with a lysosomal storage disease should consider asking for genetic counselling before having other children. The counsellor will be able to advise on the risk to close relatives and to suggest whether the wider family should be informed.

Treatment of ML III

At present there is treatment for symptoms as they arise, but no cure for the underlying disease. Bone Marrow Transplant is not recommended for individuals with ML III. Research into bisphosphonate therapy is being carried out to investigate the effect of the drugs on skeletal problems and advice on this should be discussed with your, or your child's, consultant.

Life expectancy

Individuals with ML III live well into adult life. It is hard to be precise about life expectancy as the condition has been recognised for approximately only 30 years. There may, therefore, be even older adults who have never been given the diagnosis of ML III.