

GUIDE TO UNDERSTANDING

WINCHESTER SYNDROME

What is Winchester Syndrome?

Winchester Syndrome is an inherited bone disorder. Although it is not one of the mucopolysaccharidosis it has many features in common with MPS disorders.

Whilst there is no cure for individuals affected by Winchester Syndrome 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.

How common is Winchester Syndrome?

Between 1969-2001, only 12 cases of Winchester Syndrome were reported worldwide. It appears that Winchester is more common in females than males with a female to male ratio of 3:1 (nine females, three males).

What causes Winchester Syndrome?

Winchester Syndrome is caused by an alteration in a gene called MMP2. Two very similar conditions called Torg Syndrome and Nodulosis-Arthropathy-Osteolysis are caused by alterations in the same gene.

How is Winchester Syndrome inherited?

Winchester Syndrome 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 Winchester 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, Winchester.

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.

Can you test for Winchester Syndrome in pregnancy?

If you have a child with Winchester and the altered gene is known in your family, it may be possible to have tests during any subsequent pregnancy to find out whether the foetus is affected. It is important for the gene testing to be carried out before you become pregnant and you can then discuss these results with your local genetics team. If testing is possible in your family it is important to contact your doctor as soon as you suspect that you may be pregnant if you wish for tests to be arranged.

Clinical Presentation of Winchester Syndrome

Winchester Syndrome characteristically presents in the first year of life with limitations in movement of the large joints. There may also be symmetrical joint swelling.

Growth

Growth restriction and marked short stature is characteristic of individuals with Winchester. This is probably due to the degenerative changes taking place in the vertebrae of the backbone and the long bones of the limbs.

Bones and joints

The most characteristic feature of Winchester Syndrome is osteolysis, 'dissolving bones'. This is most noticeable in the small bones of the hands and feet and may lead to their total disappearance. There may also be significant bone deformities due to pathologic changes occurring in the joints as well as vertebral column. These changes can be found in the joints of the hands, feet, knees, shoulder, elbow and hip joints. They can cause pain especially during movement and limited mobility. Osteoporosis is also a significant feature.

Facial features

Individuals with Winchester Syndrome may have a protruding forehead, large nose, thickened lips and overgrowth of the gums. Facial features may become coarser as the disease progresses over time.

Dental hygiene

It is important that teeth are well cared for to avoid the need for extractions. If the water in your area has not been treated with fluoride, individuals with Winchester Syndrome should have fluoride tablets or drops daily. Cleaning around the mouth with a small sponge or a stick soaked in mouthwash will help keep the mouth fresh and avoid bad breath.

Regular checks at the dentist are important as tooth decay could be a source of pain. If your child is severely affected it may be safer for treatment to be carried out at a hospital. It is important that you inform the dentist if your child has a heart problem and you will probably be advised that s/he should be given antibiotics before and after any dental treatment. This is because certain bacteria in the mouth may get into the blood stream and cause an infection on the heart valves. If teeth need to be removed under anaesthetic, this should be carried out in a hospital under the care of an experienced anaesthetist and never in the dental surgery. It may be possible for the hospital to carry out other treatment or investigations under the same anaesthetic.

Skin

The skin may be thickened and brownish and may also have a leathery consistency. Particularly on the trunk, the skin may be thickened with pigmentation and excess body hair (hirsutism). The skin changes can be very variable.

Heart

Heart murmurs and ECG changes have been reported.

Enlargement of internal organs

No cases of enlarged liver (hepatosplenomegaly) have been reported.

Eyes

Clouding of the cornea is sometimes seen in individuals with Winchester and can lead to significant visual disability especially as the disease progresses. Severe corneal clouding may reduce sight, especially in dim light. Some individuals cannot tolerate bright lights as the clouding causes uneven refraction of light. A loss of night vision is common which may also be due to damage of the retina. Glaucoma, abnormally high pressure in the eye, may occur and cause damage to the retina and results in optic nerve atrophy. Cataracts are also common.

General management of Winchester Syndrome Anaesthetic

Giving an anaesthetic to an individual with Winchester requires skill and should always be undertaken by an experienced anaesthetist. Where a child is concerned this should be a paediatric anaesthetist. The airway can be very small and may require a very small endotracheal tube. Placing the tube may prove difficult and require the use of a flexible bronchoscope. In addition, the neck may be somewhat lax and repositioning the neck during anaesthesia or intubation could cause injury to the spinal cord. For some individuals, it is difficult to remove the breathing tube after surgery is completed. There is a more detailed explanation of this complex subject in the booklet on managing anaesthesia in MPS individuals available from the MPS Society.

Physiotherapy and hydrotherapy

Physiotherapy and hydrotherapy can be useful to help individuals with Winchester 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 Winchester Syndrome

At present, there is no cure for Winchester Syndrome, only treatment for the symptoms as they arise. As we learn more about the syndrome it is hoped that treatment may become available in the future. There have been trials to look at the use of drugs called bisphosphonates to see if they can help strengthen the bones. Results, so far, have been rather disappointing.

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.