HOW IS GENETIC TESTING DONE?

Genetic testing involves taking a sample of DNA from blood, saliva or tissue and analysing the genes that are most commonly known to cause MS.

When testing a family, we begin by testing a person known to have MS. If a mutation is identified in that individual, then testing can be performed on other family members to look for that specific mutation or mutations.

Genetic testing is complex; it takes place with genetic counselling and the process of informed consent.

If your family history suggests MS, please talk to your doctor. A referral to Curie Genetics can help you find out more about the condition and genetic testing.

GENETIC COUNSELLING

The diagnosis of MS has implications for other family members. A trained genetic counsellor will review your family history and discuss implications of the diagnosis with you and your family.

In addition, the genetic counsellor can serve as a resource to other family members who may have questions about the diagnosis. It is important to us that you understand all aspects of your condition and we are always available to answer questions.

REFERRAL INFORMATION

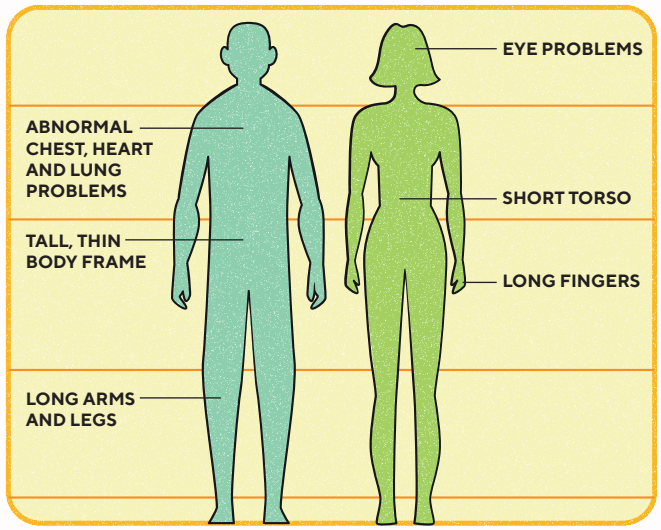
Address 📍: CURIE Genetics
(Mount Elizabeth Novena Hospital #10-37)
Singapore 329563

Telephone 📞: 6592 8615

Whatsapp : 8683 3767

Website : www.curiegenetics.sg

Adapted from NCBI (Genereview)



MARFAN SYNDROME

CURIE GENETICS

BY CURIE ONCOLOGY

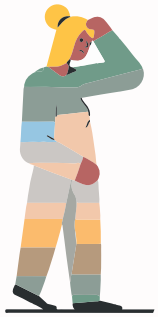
WHAT IS MARFAN SYNDROME?

Marfan syndrome (MS) is a disorder of connective tissue. Connective tissue holds all parts of the body together and helps control how the body grows. As connective tissue is found throughout the body, Marfan syndrome features can occur in many different parts of the body.

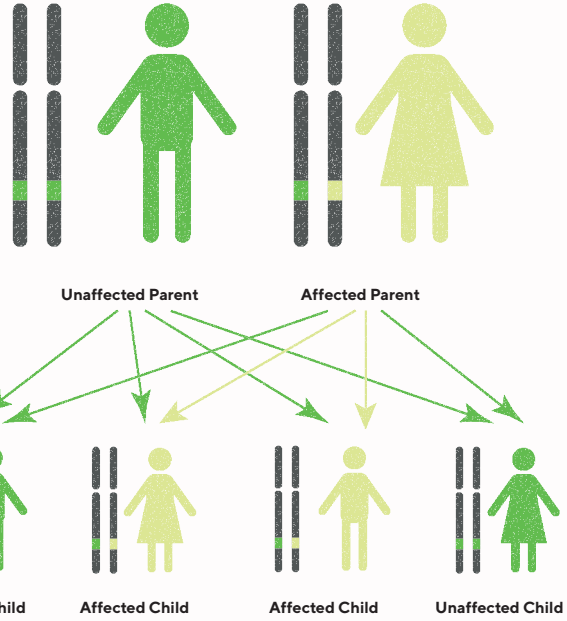
Most often the condition affects the heart, blood vessels, bones, joints, and eyes. Sometimes, the lungs and skin are also affected. Marfan syndrome does not affect intelligence.

MS is caused by changes (mutations) in the *FBN1* gene, located on the chromosome 15.

The protein made by the *FBN1* gene functions as a major building block of connective tissues. Absence or deficiency of the protein results in weakening of the structures of various organs.



MS follows an autosomal dominant pattern of inheritance. This means only one copy of the changed gene is needed to result in the condition.



SIGNS AND SYMPTOMS

- Tall and thin with disproportionately long limbs
- Sunken or protruding chest
- Flat feet
- Dislocation of the eye lens
- Crowded teeth and high arch palate
- Heart and blood vessel abnormalities

OTHER SIGNS AND SYMPTOMS

- Pregnancy can be dangerous for women with Marfan syndrome, especially if the aortic root exceeds 4.0 cm
- Lung damage

MANAGING MARFAN SYNDROME

Treatments are given to manage symptoms and vary between patients depending on the organ systems affected.

Regular monitoring of the heart, eyes and bones are important.

Some patients may need medication or surgery to manage heart complications.

Restriction of vigorous physical activities is usually necessary as strenuous activities may be dangerous to the heart.

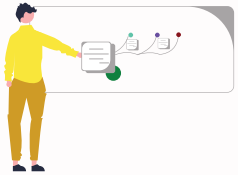
Smoking should be avoided as Marfan Disease patients are at increased risk of lung damage.

Well planned treatment and care programmes are able to reduce complications and improve quality of living.

BENEFITS OF GENETIC TESTING

- Confirmation of a diagnosis

- Provide insights on underlying cause of heart disease



- To begin with risk reduction interventions by making informed medical decisions

- Identify at-risk family members for cascade screening