Frequently Asked Questions

1. **If no members of my family are affected by Marfan Syndrome, can I still have a child with the condition?**
   Yes. The majority of cases of Marfan Syndrome occur in families with a history of the condition previously reported. However, spontaneous (new) mutations account for up to 30% of all Marfan Syndrome cases. The genetic change occurs randomly and the cause of it is unknown.

2. **Since Marfan Syndrome is a genetic condition, is it possible to have more than one child with the condition?**
   Yes. If either or both parents have the condition, the associated risks apply to each pregnancy. Recurrence of Marfan Syndrome in a family where there is no family history of the condition is low.

3. **Do people with Marfan Syndrome have a shortened lifespan?**
   No. In most cases, people with the condition have a normal lifespan with proper treatment and management.

4. **What are the main medical issues faced by people with Marfan Syndrome?**
   The most prevalent problem faced by Marfan Syndrome patients is complications involving the cardiovascular system. This requires patients to undergo regular cardiac monitoring and refraining from pursuing strenuous activities.

5. **I have Marfan Syndrome and have a young son who was recently diagnosed with the same condition. Can I expect him to exhibit exactly the same symptoms that I do?**
   No. Marfan Syndrome is one of several disorders said to show variable expressivity. This means that symptoms and severity vary from patient to patient, even among those from the same family. Your son may be more or less severely affected than you are.

References

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MALAYSIAN RARE DISORDERS SOCIETY

Rare Disorders Series

Marfan Syndrome

What is Marfan Syndrome?

Marfan Syndrome is an inherited disorder affecting connective tissues in the body.

Connective tissues strengthen the body's structure. Disorders involving them affect the skeletal and cardiovascular systems, eyes and skin.

It is caused by changes (mutations) in the *FBN1* gene, located on the fifteenth chromosome pair.

Genes are located on chromosomes and contain 'recipes' to make proteins.

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.

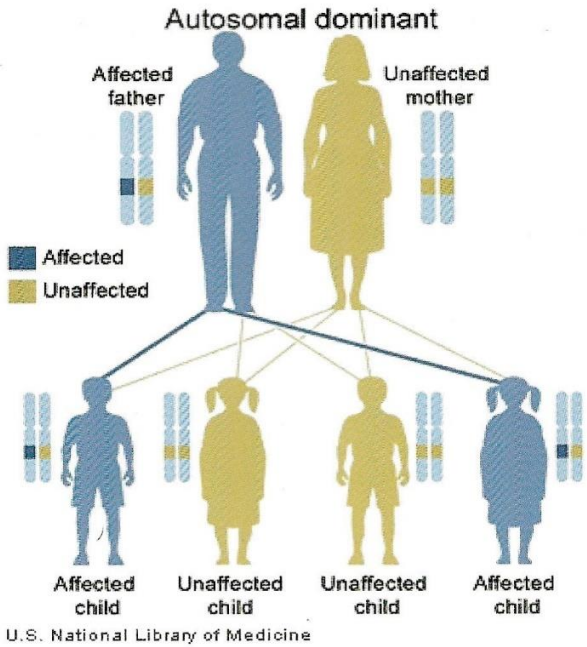
Marfan Syndrome follows an autosomal dominant pattern of inheritance. This means only one copy of the changed gene is needed to result in the condition.

When one parent is affected, there is a 50% chance of passing on the condition to each child.
If both parents have the condition, there is a 75% chance of having a child with Marfan Syndrome.

Inheritance of the mutation happens by chance and is not due to anything either parent did or did not do before or during pregnancy.

Symptoms and severity vary from patient to patient. In most cases, symptoms progress as a person ages.

Patients are advised to consult a geneticist to obtain further information on disease progression.



Inheritance pattern of Marfan Syndrome when one parent has 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 due to arched roof of mouth
- Heart and blood vessel abnormalities

Other ways Marfan Syndrome may affect the body

- Swelling of the covering of the spinal cord
- Increased risk for lung damage
- Pregnancy may give stress to the body

Note that symptoms and severity vary from patient to patient and not all of the symptoms stated above will be seen in an individual with Marfan Syndrome.

Testing

Investigations to make a diagnosis comprise of a thorough physical examination and taking a complete medical as well as family history. An eye doctor may review the patient's eyesight.

Non invasive imaging studies such as radiography, echocardiography and MRI help in evaluating the heart and determining the appropriate intervention needed.

Specific lab tests are only available to diagnose Marfan Syndrome on a research basis. Genetic counselling is usually performed before any genetic testing.

Treatment and management

There is currently no known cure for 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.