Neonatal Marfan Syndrome

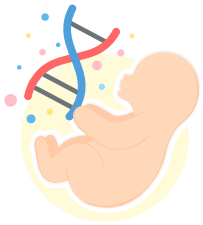
Marfan Syndrome:
An Overview

Marfan syndrome is a genetic condition that affects the body's connective tissue. It is caused by a defect in fibrillin-1, the protein that gives structure to our body.
Marfan syndrome typically affects the heart, blood vessels, eyes and skeletal system, however this varies from patient to patient.

Neonatal Marfan syndrome:

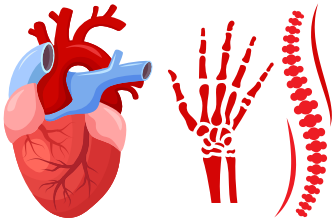
Neonatal Marfan syndrome is the term used to describe the most serious form of Marfan syndrome.

New mutation

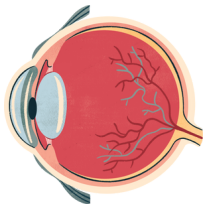


Whilst Marfan Syndrome is classically inherited from a parent, neonatal Marfan syndrome has emerged from a spontaneous gene change. Those affected have different genetic mutations from the more common type of Marfan syndrome. The condition is apparent in early infancy and advances very quickly. It can even be diagnosed before birth.

What are the medical signs?



Neonatal Marfan syndrome can affect the Heart, the Eyes and the Skeletal System. Some common signs that the doctor may look out for include:



- Leaky Heart Valves: which may present with difficulty breathing at birth or a heart murmur
- Aortic Enlargement: this is the main blood vessel leaving the heart, and sometimes the connective tissue deficiency causes it to widen and become enlarged
- Detachment or dislocation of the lens of the eye
- Difficulty seeing and following objects

Neonatal Marfan Syndrome

What are the physical signs?

Neonatal Marfan syndrome can manifest in the following physical features:

- Long and thin stature
- Joint contractures and long fingers
- Chest wall abnormalities
- Downward Slanting Eyes
- Crumpled Ears
- Small Chin

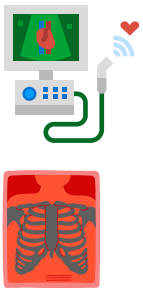
How is it diagnosed?

Clinical Examination

A doctor will examine every baby at birth. By listening to their heart, moving their joints, and looking into their eyes, this may lead to a suspected diagnosis of neonatal Marfan syndrome.

Scans and Images

If a doctor suspects neonatal Marfan syndrome from the clinical examination, further tests will be required, including scans of the heart.



Genetic Testing

The diagnosis will be confirmed by genetic testing which usually involves taking a small sample of blood which is sent to the laboratory to look for a specific fibrillin-1 mutation.

How do we treat it?

Medical

The majority of medication is prescribed to optimise heart function:

- Angiotensin Converting Enzyme ACE) Inhibitor: Lisinopril, Captopril
- Angiotensin Receptor Blockers (ARBs): losartan
- Beta-Blockers: bisoprolol, atenolol
- Diuretics: furosemide
- Others: digoxin, aspirin, milrinone, sildenafil

Surgical

Surgery is sometimes required to correct serious problems with the Heart. Operations may include valve repair or replacement and strengthening of the aorta.

With surgery in nMFS come considerable risk, including:

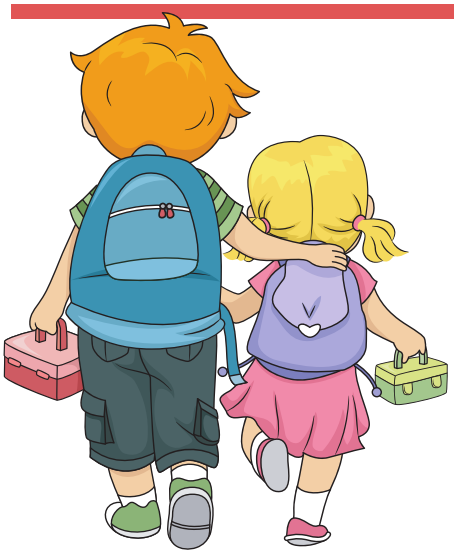
- bleeding or blood clots
- heart rhythm disturbances

Neonatal Marfan Syndrome

Financial & Practical Considerations

Financial Element

Marfan syndrome presents significant financial and practical challenges for affected individuals and their families. Ongoing medical expenses, including regular check-ups, may make it challenging for a parent to maintain a full-time job. The management of associated complications and lifestyle adjustments further adds to the financial burden. Additionally, the need for adaptive equipment, assistive devices, and accessible living spaces may arise to ensure the safety and well-being of the child. Conducting risk assessments for everyday activities, like using a swing, becomes necessary but can be exhausting. Despite these challenges, prioritizing the child's safety and well-being remains paramount for families coping with Marfan Syndrome.



Effect on Siblings

There is a huge impact on siblings of those who are affected with Marfan syndrome. Siblings may experience a mixture of emotions, including worry and concern for their affected brother or sister, and sometimes feelings of jealousy or a need for additional attention from parents. Navigating these complex emotions while fostering positive sibling interactions requires open communication and support from parents.

Long-Term Ventilation

Some children may require long-term assisted mechanical ventilation. This places a high demand on parents. The technology, attendant complexities and parental responsibilities can feel overwhelming and isolating.

Neonatal Marfan Syndrome

Multi-Disciplinary Team

The management of nMFS requires a team of specialists. Amongst this team will be genetic doctors, cardiologists, ophthalmologists, and orthopaedic surgeons. This is in addition to physiotherapists, occupational therapists and specialist nurses - all of whom will work together to ensure the baby's long-term treatment is optimised for their individual needs.

Conclusion

Neonatal Marfan syndrome is a severe, life-limiting condition. Published medical reports suggest average survival is less than 2 years. Palliative care may be recommended by the multidisciplinary team caring for the baby. Those who survive into later childhood have mostly required major cardiac surgery (often more than one heart operation. Initial cardiac operations are to deal with leaky valves and heart failure . If the child survives there is a risk of tearing of the aorta (dissection - and more surgery may be needed to try to prevent this). Surgery on the eyes and joints are also typically required.

Expert care from the earliest age can improve quality and length of life.

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

References:

- Anne Child BH, Antonio Aragon-Martin J, Sage K. Genetic Testing in Marfan Syndrome. Br J Hosp Med (Lond). 2016 Jan;77(1):38-41. doi: 10.12968/hmed.2016.77.1.38.
- Child AH. Diagnosis and management (summary). Diagnosis and Management of Marfan Syndrome. 2016; 3–12. https://doi.org/10.1007/978-1-4471-5442-6_2
- Milewicz DM, Braverman AC, De Backer J, Morris SA, Boileau C, Maumenee IH, et al. Marfan syndrome. Nature Reviews Disease Primers 2021 7:1. 2021;7(1): 1–24. https://doi.org/10.1038/s41572-021-00298-7.
- Child AH. Infantile and neonatal Marfan syndrome. Diagnosis and Management of Marfan Syndrome. 2016; 21–26. https://doi.org/10.1007/978-1-4471-5442-6_4
- S. Strigl, J. M. Quagebeur, and W. M. Gersony, “Quadrivalvar replacement in infantile Marfan syndrome,” Pediatric Cardiology, vol. 28, no. 5, pp. 403–405, 2007.

Got Questions? Get in Touch
info@marfantrust.org
+ 44 (0)333 011 5256
https://www.marfantrust.org
Registered Charity number 328070