Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders
Loeys Dietz
CONNECTING THE UNCONNECTED AND STRENGTHENING LIVES

Guide to Pregnancy in Marfan & Loeys-Dietz Syndromes

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What Are Marfan & Loeys-Dietz Syndromes?

Marfan and Loeys-Dietz syndromes are inherited genetic disorders of the body's connective tissue, affecting any gender, race or ethnic group. Connective tissue helps provide structure to the body, binding skin to muscle and muscle to bone. It also supports the internal organs.

Connective tissue is made of fine fibres and 'glue'. One fibre is called fibrillin. In Marfan syndrome (MFS), a change in the fibrillin-producing gene, fibrillin-1, means that this protein is deficient in connective tissue throughout the body, creating an unusual stretchiness and weakness of tissues. This has far-reaching implications and can affect the eyes, lungs, gut, nervous system, skeleton and, most dangerously, the cardiovascular system. Symptoms can vary widely from person to person with people experiencing mild to severe manifestations. MFS affects roughly 1 in 3,000 people which means that approximately 18,000 people in the UK have MFS. We estimate that half remain undiagnosed.

75% of patients inherit the condition whilst 25% develop it as a result of a spontaneous (new) gene change. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

Every year, on average in the UK there are over 200 new cases of Marfan Syndrome diagnosed.

Marfan syndrome was first described by a French paediatrician, Dr Antoine Marfan, in 1896. It is caused by a change in the gene for fibrillin-1 on Chromosome 15.

Patients with Loeys-Dietz syndrome (LDS), were once thought to have Marfan syndrome, such are the similarities between the two conditions.

LDS was first described in 2005 by Drs Bart Loeys and Harry Dietz. It is caused by a variant in one of the genes in the transforming growth factor-beta signalling pathway, TGF-β. The genes that cause LDS are: TGFβR1, TGFβR2, SMAD3, TGFβ3 and TGFβ2. TGF-β is a crucial signalling pathway involved in various cellular processes, including cell growth and immune responses. It is a key player in development and tissue repair. When a gene change occurs, it has implications for many systems of the body. Whilst symptoms vary from patient to patient, with some being similar to those of Marfan syndrome, the most characteristic are: arterial tortuosity, aortic enlargement, bifid uvula, curvature of the spine, high-arched palate and over-crowded teeth.

This pamphlet is intended for patients with both Marfan and Loeys-Dietz syndromes.

Loeys-Dietz syndrome is much rarer than MFS and the incidence is currently gauged to be 1 in 100,000 although this is probably an underestimation.

Planning a Pregnancy

Specialists agree that the most important step to a safe and successful pregnancy with MFS/LDS is planning. A person with MFS/LDS has a higher risk of aortic dissection than that of the general population and this is why careful planning and accurate risk assessment is so important: aortic dissection is life threatening.

Hopefully, if you were diagnosed at a young age, you will have had the opportunity to discuss these issues with your specialist doctors over several years and learn about the steps you will need to take prior to any pregnancy. Sometimes, a diagnosis of MFS/LDS can be much more recent, and the implications can come as a great shock. It is important to remember that it is better to be aware of your condition so that doctors can keep you and your baby safe throughout any pregnancy.

Inheritance

Before starting a family, you may wish to think about inheritance. MFS/LDS are genetic conditions, they are ‘autosomal dominant', this means you have a 50% chance of passing the genetic change on to any child you may have. This applies to men and women with Marfan syndrome.

Pre-implantation Genetic Diagnosis (PGD) is now available, embryos can be tested prior to implantation to determine whether they have an alteration to the genes that cause MFS/LDS. Another option is to have amniocentesis in pregnancy to determine whether the baby has MFS/LDS. Other people decide to try for a natural pregnancy and accept the 50% chance of having a child with or without MFS/LDS. You can discuss all these options when you have a pre-pregnancy appointment.

Pre-Pregnancy Counselling

Anyone planning a pregnancy will want to make some positive changes to their health: eating well, reducing alcohol intake, giving up smoking, taking folic acid supplements. These are all just as important in individuals with MFS/LDS but there are other things to consider

- Are you taking medication that may be damaging to a baby? Beta blockers such as Bisoprolol have been proven to be safe in pregnancy but angiotensin receptor blockers such as Irbesartan or Losartan cannot be taken and are recommended to be stopped with guidance from your cardiologist following a positive pregnancy test
- Have you had recent imaging of your aorta with MRI, CT, or echo? It is vital that up-to-date measurements of your aorta are available so that during pregnancy this can be watched closely for any signs of further dilatation. ESC (2025) guidelines recommend that imaging of the entire aorta with CT or MRI is performed for all women with known or suspected aortic disease prior to pregnancy.
- Most importantly, if you are planning a pregnancy, tell your cardiologist so they can refer you to a Cardiac-Obstetric clinic. You will be seen in this clinic, usually by a Cardiologist, Obstetrician, Anaesthetist and Midwife who can have a detailed conversation with you about the monitoring you will need during your pregnancy and the ways in which your care may differ to that of a person without MFS/LDS.

A detailed family history is important at this point as a history of aortic dissection in relatives is significant and needs to be considered when assessing your risk

These appointments give your medical team the opportunity to provide you with a more personalised risk assessment based on all your medical information. Any changes to your care are made to keep you and your baby safe during pregnancy and will be based on the best evidence and guidelines available.

Preparing for Appointments

- Take a trusted partner, parent or friend with you, there will be lots of information to take in and it can help to have someone else listening with you
- Think about questions you want to ask before you go along and write them down
- Take a list of all your medications and dosages
- This appointment may be in a different hospital with different specialists to your usual team of doctors, so it is helpful if you bring along your previous letters or scan results
- Feel free to write things down during the appointment, your doctors will send a letter, but it can be helpful for you to take notes in your own words

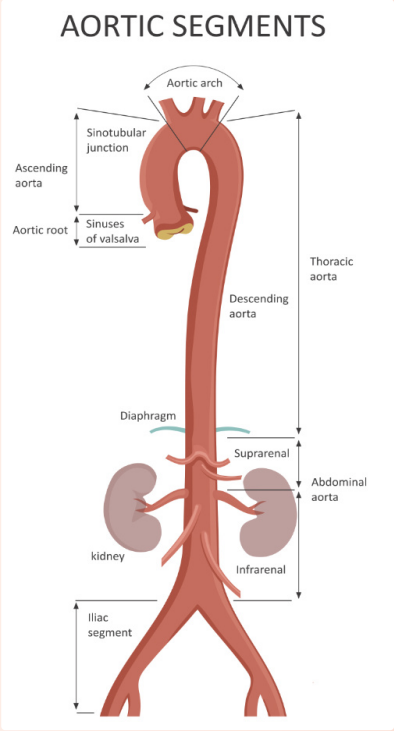
Surgery Prior to Pregnancy

Your doctors will need to review all your medical information in order to consider whether they feel pregnancy is safe for you to consider. There are guidelines that doctors can use to help them decide whether it would be sensible for you to have surgery to your aorta prior to pregnancy. Several factors, including they type of connective tissue disorder you have, the size of your aorta, your specific gene change, family history and other risk factors will be considered when deciding if you would benefit from aortic surgery prior to pregnancy. This is because as the aorta gets bigger there is a higher risk of a tear or dissection of the aorta. In addition to the diameter, the process of pregnancy can also lead to other changes that can increase the risk of dissection:

Increased volume of blood circulating

Hormonal changes affect the structure of blood vessel walls

If you do need surgery before becoming pregnant you will be closely followed up by your medical team in the weeks and months following your operation. You will need time to recover and will need to be seen in the pre-pregnancy clinic again so that the specialists can assess your risks again based on the outcome of your surgery.



Once you are pregnant you will be monitored closely, usually every 4 weeks from around 12 weeks of pregnancy with an echo to check on the size of your aorta and the function of your heart. You may also have an MRI scan which is safe during pregnancy.

Your blood pressure will be closely monitored.

You will have additional growth scans for your baby if you take beta blockers as these can mean your baby has a lighter birth-weight (be small for dates).

Hormonal changes during pregnancy can also affect your joints. Many individuals with MFS/LDS suffer from joint hypermobility and chronic pain and this can be exacerbated during pregnancy or after birth but can be helped with physiotherapy.

Delivery

You will usually be advised to have your baby in a hospital that has cardiac surgical support should you need it during delivery. This may mean that you cannot have your baby in your local hospital, but this is to ensure that you and your baby are in the safest place during and after delivery with access to all the care you could need.

Vaginal delivery is usually possible, unless there have been concerns about a change in your aorta, but an early epidural is recommended to prevent the rises in blood pressure that come with pain and pushing. It will be important to determine prior to pregnancy whether you have dural ectasia (another consequence of MFS/LDS) as this may have implications for the effectiveness of an epidural. Previous scoliosis surgery may also affect administration of an epidural so this needs to be considered in advance by your anaesthetist.

Sometimes a caesarean section will be necessary, for example, if there has been dilatation of your aorta during pregnancy or if your baby is in a position that will make delivery difficult.

Vaginal delivery will often be assisted with either ventouse (vacuum cup applied to the baby's head) or forceps as this reduces the effort of pushing and the increased pressure this puts on the aorta.

In addition to the risk of aortic dissection, individuals with MFS/LDS can also be at higher risk of some other obstetric complications, and these will be monitored too.

- Premature rupture of membranes and premature delivery
- Poor wound healing or tears
- Post-partum haemorrhage

The risk of tears to the aorta is highest during the third trimester of pregnancy and for around 12 weeks post-partum. You will stay in hospital for longer than usual. This is to ensure you are safe and if any problems arise, they can be quickly dealt with.

Going home

Once you go home you should continue to pay attention to any unexpected symptoms:

- Severe chest pain or back pain often described as a ripping or tearing pain
- Severe stomach pain
- Shortness of breath
- Neurological symptoms like those of a stroke such as numbness or loss of movement in your limbs, difficulty speaking, visual disturbances

If you have any of these, you should immediately attend A&E and make sure that the doctors know that you have MFS/LDS, have recently had a baby, and have an increased risk of aortic dissection.

Breastfeeding

You can breastfeed but you will not be able to restart your angiotensin receptor blockers (e.g., Irbesartan/Losartan) whilst doing so. You can continue to take beta blockers like Bisoprolol and will be restarted on other medication once you choose to stop breastfeeding. You will be referred back to your cardiology clinic for ongoing surveillance and will need to have an echo or MRI scan in the months following the birth to ensure there have been no further changes to the aorta.

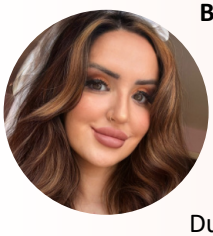
Further Pregnancies

Individuals with MFS/LDS can go on to have further pregnancies and the same precautions and monitoring would be put in place again. These decisions would be influenced by factors including:

- Any complications during your previous pregnancies
- Changes to the aorta during or since your previous pregnancies
- Whether you have required aortic surgery
- If you experienced a dissection during or since your previous pregnancies
- Interval since your last pregnancy

For these reasons planning and pre-pregnancy counselling are again vital before considering further pregnancies.

Pregnancy experience with Marfan syndrome



By Zoe Ridgway

Becoming pregnant with my beautiful daughter was an unexpected blessing but a very scary experience. I was 19 when I learned I was pregnant, and against my cardiologists wishes I decided to go through with the pregnancy,because despite the odds I would still love and cherish her.

During pregnancy I was very frightened about the fact my child might have Marfan syndrome as it was a 50/50 chance. I was offered Non-Invasive Prenatal Testing (NIPT) but declined it due to the risks involved and decided to have my daughter tested after I gave birth.

During pregnancy I had regular echocardiograms (heart scans) and ECGs, I was in hospital every 2/3 months for checks and also scans of the baby.

Pregnancy was not an easy experience for me, I struggled with low blood pressure and exhaustion. Despite being advised that I could have a natural birth I opted for an elective caesarean due to my medical and family history. I was given lots of support during the operation with cardiologists being present alongside the midwives. I had to have 2 epidurals and a spinal block which finally kicked in, and they safely delivered my beautiful daughter.

Zoe Ridgway has Marfan syndrome

Unfortunately, the care I received after the birth was lacking and I felt very alone at times. Despite this, we have now decided we would like to have another baby and are on a wonderful journey of Preimplantation Genetic Testing IVF, in my case, this would be to ensure that the Marfan gene is not present in any embryos that are reimplanted.

I went to the cardiologist at the end of 2024 and discussed my plans for another pregnancy, they told me how to obtain an IVF referral and recommended that I have surgery to my aorta prior to any further pregnancy to reduce the risk of problems I had Valve-Sparing Aortic Root Replacement (VSRR) and am now recovering. My cardiologist is keeping a close eye on my recovery and will advise when it is safe to try for another pregnancy.

In the meantime, we have been to the fertility clinic where they tested

For families affected by Marfan syndrome (MFS) or Loeys-Dietz syndrome (LDS), Preimplantation Genetic Testing for Monogenic disorders (PGT-M) is a highly effective way to prevent the transmission of these conditions.

Because both syndromes are typically autosomal dominant, an affected parent has a 50% chance of passing the condition to each child. PGT-M allows for the selection of embryos that do not carry the specific family mutation.

How PGT-M Applies Specifically to MFS and LDS

- Targeting the Specific Gene: For Marfan, the test targets the FBN1 gene and for Loeys-Dietz, the test targets genes such as TGFBR1, TGFBR2, SMAD3, TGFB2, or TGFB3.
- The "Probe" Development: Before IVF begins, a lab must build a "custom probe" unique to your family's specific mutation. This usually requires DNA samples from the affected parent and sometimes their parents or other affected relatives to "map" the gene accurately.
- De Novo Mutations: About 25% of Marfan and LDS cases are de novo (spontaneous), meaning the parent is the first in their family to have it. PGT-M is still possible for these individuals, but the lab setup may be slightly more complex since there are no previous generations to use for genetic mapping.

References

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About the Marfan Trust

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Loeys Dietz

CONNECTING THE UNCONNECTED AND STRENGTHENING LIVES

Co-founded in 1988, the Marfan Trust is the sole charity in the United Kingdom dedicated to improving and saving the lives of those with Marfan syndrome. It is estimated that approximately 18,000 people are living with Marfan syndrome in the United Kingdom and around half of these remain dangerously undiagnosed.

The Marfan Trust's three main objectives are to:

- Provide personalised support and medical guidance through its helpline;
- Conduct cutting-edge medical research through its self-funded Sonalee Laboratory, named after a young doctor who tragically died of complications from MFS during her ward round;
- Continue to provide educational information and raise awareness of the condition.

How you can help

The Marfan Trust relies solely on the goodwill of its unstoppable supporters who tirelessly raise funds and awareness, allowing the charity to continue its good work and lift the shadow from this condition.

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You can help to secure the Marfan Trust's future by becoming a member today for just £3 per month:

www.marfantrust.org/pages/10-membership

JustGiving®

Just Giving – **http://bit.ly/3Scj51w**

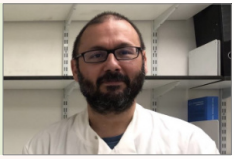
PayPal

PayPal Giving – **https://bit.ly/45NCuwQ**

- BANK: Charities Aid Foundation (CAF) ACCOUNT NAME: Marfan Trust
- SORT CODE: 40-52-40
- ACCOUNT NUMBER: 00017677
- REFERENCE: Your Name (plus campaign name if relevant)

By donating to the Marfan Trust, you are contributing to an ever-growing body of knowledge on the condition, allowing more doctors and medical specialists to deliver the best possible treatment to patients affected by Marfan syndrome.

You can also help to fund a piece of equipment in Dr José Aragon-Martin's Sonalee Laboratory. Email **info@marfantrust.org** to find out more.



The Hospital Saturday Fund
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