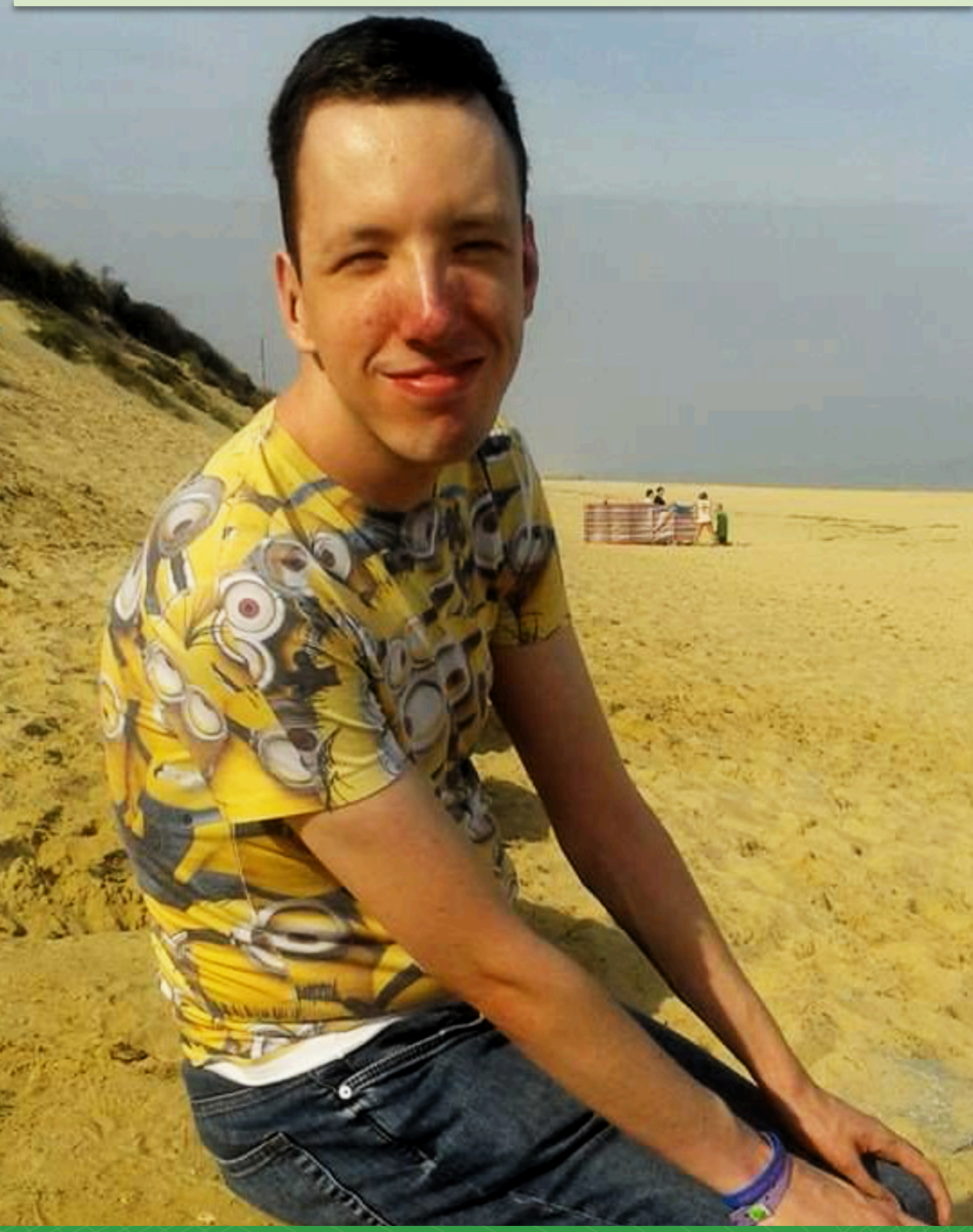
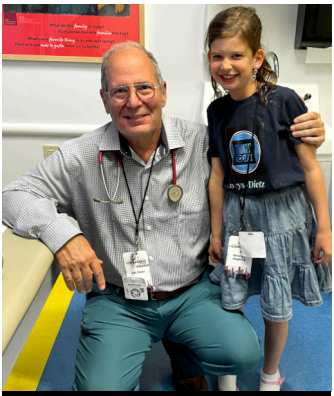
Loeys-Dietz Syndrome



What is Loeys-Dietz Syndrome?



Georgie with Professor Harry C Dietz

Loeys-Dietz Syndrome (LDS) is a connective tissue disorder first described in 2005 by Professors Bart Loeys and Harry C. Dietz.

It is caused by a variant in one of the genes in the TGF-beta pathway-the genes that can cause LDS are TGFBR1, TGFBR2, SMAD3, TGFB3 and TGFB2.

A variant in these genes causes the connective tissue in the body to be weakened, which causes the signs and symptoms associated with LDS.

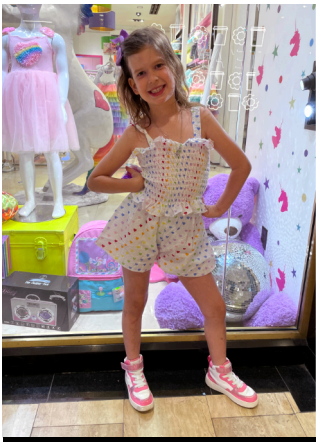
A spontaneous gene change (mutation) could mean someone is the first in their family to have LDS. LDS is inherited in a heterozygous dominant pattern meaning if one parent has LDS, their children each have a 50% chance of inheriting the condition.

What are the signs of Loeys-Dietz Syndrome?

Connective tissue is found throughout the entire body, so LDS can affect many areas of the body.

LDS shares some similarities with other connective tissue disorders, such as Marfan Syndrome. These include aortic dissection, scoliosis (spinal curvature), and deformities of the chest wall. The following is not exhaustive but gives an idea of some of the signs & symptoms your medical team will be looking for:

- Aneurysms–an aneurysm is a widening or dilation of an artery. This is most often seen in the aortic root (where the aorta meets the heart)
- Arterial tortuosity-twisting or spiralling arteries
- Hypertelorism-widely spaced eyes
- Bifid (split) or broad uvula- the uvula is the small piece of flesh that hangs down from the back of the mouth



Georgie has Loeys-Dietz Syndrome type 1

Biographies

Darran & Mum Debbie

Meet our cover star, Darran Jones, an avid fan of Elvis Presley and Disney. He and his mother, Debbie Wood (pictured together, left) both have Loeys-Dietz syndrome. Darran loves to impersonate Elvis, as this image reveals!

Meanwhile, lovely Georgie (pictured, right), has LDS1. She has met one half of her condition's namesake as you can see from the picture above.

Thank you, Darran, Debbie and Georgie for so generously giving a face to the condition of Loeys-Dietz Syndrome.

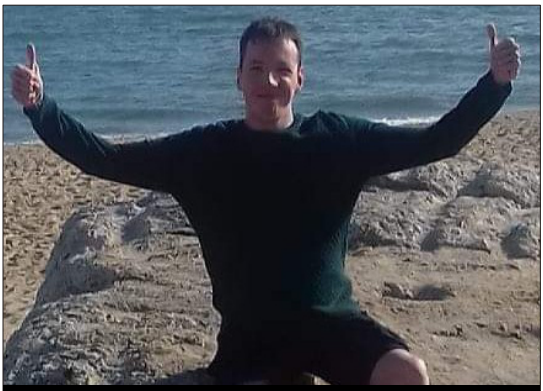
Georgie

How will my diagnosis impact me?

Being diagnosed with a condition such as LDS can be both physically and emotionally challenging.

You are likely to see a variety of specialist doctors and other medical professionals, including cardiologists, geneticists, and cardio-thoracic surgeons.

There is no cure for LDS, and therefore your treatment will try to help with the symptoms and complications of LDS. This will depend on how your LDS impacts you specifically, but it could involve medicines, physiotherapy and potentially surgery.



Darran Jones

How will my LDS be managed?

Whilst LDS affects patients in different ways, and therefore management will be specific to you, there are some general management steps such as:

- Monitoring of the aorta/blood vessels:
    - Yearly echocardiograms (a scan of the heart)
    - Regular CT/MRI angiograms of the head, neck, chest, abdomen, and pelvis to look for aneurysms and dissections
- Medication:
    - Medicines to reduce stress on the heart, such as beta blockers and angiotensin receptor blockers (ARBs)
- Safe exercise:
    - Gentle regular exercise such as walking, cycling, and swimming are beneficial for patients with LDS
    - Contact sports such as football and rugby should be avoided, as should strenuous activity such as weightlifting or long-distance running
- **Genetic testing might be recommended for other members of your family**
- **Pregnancy can pose additional risks and requires careful planning and specialist management**

Cardiac Emergencies and Aortic Dissection

Cardiac Monitor

Aortic dissections can occur at a smaller diameter and at a younger age in LDS than in other connective tissue disorders, like Marfan syndrome. This is why regular surveillance is so important. It's vital that younger people with LDS are listened to if they are describing severe symptoms that could be a sign of aortic dissection. It is important to be able to recognise the signs of a potential aortic dissection and seek immediate help by calling 999 and asking for an ambulance.

Signs of an aortic dissection include:

- Sudden, severe onset chest or interscapular (between the shoulder blades) pain. Can be described as a 'ripping' or 'tearing' pain
- Pale skin
- Weak pulse
- Numbness or tingling
- Paralysis (sudden-onset, temporary paralysis of both legs is a rare sign of aortic dissection)

It is important to tell both the paramedics and doctors looking after you that you have Loeys-Dietz syndrome, which means you have a higher risk of aortic dissection.

References

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