AIMS For a better birth...

Pregnancy, Childbirth and Marfan Syndrome

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Editor's note: In this very interesting article, Victoria Hilton from the Marfan Trust writes about the genetic condition Marfan Syndrome and explains how it may affect someone during their pregnancy. She includes two personal stories from women with this condition, the first of which may be distressing to read.

By Victoria Hilton for the Marfan Trust

The promise of a new life brings new hope but also apprehension and worry. Pregnancy is a life-changing event, driven by shifting emotions and physiological change. Childbirth is a journey like no other, an intense rollercoaster of sensations and feelings, culminating in the most magnificent of physical achievements. All this becomes further heightened when a genetic illness is woven into the mix. Pregnancy with a lifelong disorder - one that can be passed on - adds a new dimension of worry to an already-stressful situation.

Marfan syndrome (MFS) is a genetic condition which carries a 50% chance of transmission to each offspring. It is a disorder of the body's connective tissue with implications for the eyes, skeleton, and, most dangerously, the heart. Pregnancy can exacerbate an underlying weakness caused by MFS and should be carefully planned from beginning to end, together with an obstetrician and a cardiologist.

Connective tissue helps provide structure to our body, binding skin to muscle and muscle to bone. It is made of fine fibres and 'glue' including a protein called fibrillin. This tissue provides the stretchy strength

of tendons and ligaments around joints and in blood vessel walls. It also supports the internal organs. In 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 ramifications and can affect the eyes, lungs, gut, nervous system, skeleton and, potentially fatally, the cardiovascular system.

Our body's fist-sized motor engine, the heart, is where magic happens. It beats approximately 100,000 times a day, pumping blood around the body to keep us alive. Pregnancy adds to its workload, with the cardiovascular system making significant adaptations to nourish and accommodate the growing baby. For instance, blood volume increases between 30% and 50%. The body's largest artery, the aorta, is responsible for carrying the oxygen-rich blood from the heart to the rest of the body and it can become imperilled in Marfan syndrome. The stretchiness that is characteristic of the syndrome can affect the aorta which, if untreated, may dangerously widen and fatally tear. This is known as an aortic dissection and the risk rises with pregnancy as hormonal changes affect vessel walls and the volume of blood circulating increases. Another cardiovascular symptom of Marfan is a leaking heart valve (particularly the mitral valve) which can billow backwards and allow blood to flow in the wrong direction, making the heart work even harder. Again, pregnancy compounds the situation.

Ideally, Marfan syndrome should be diagnosed at an early age whereupon a medical plan is put in place to manage the often complex and multi-faceted manifestations. Any aortic problems are treated with medication - beta-blockers and angiotensin receptor blockers – which slow down the aortic widening. However, as a rare condition in which the signs and symptoms vary greatly from person to person, MFS is sometimes missed and goes dangerously undiagnosed until a crisis occurs. This was Julie's experience, during her 39th week of pregnancy.

Some first hear the words Marfan syndrome across the consulting table. Others grow up knowing them. A select few first encounter the words when recovering from emergency surgery, like Julie.

Julie's Story

> *In July 2010 I returned from a shopping trip, and slumped on the sofa, feeling exhausted. My tiredness was no surprise, I was in my 39th week of pregnancy and I had been out panic-buying baby vests. Just me worrying, in my final trimester, that the 50 tiny little vests, already washed and ironed in the drawer, were not enough!*
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> *After mustering up the energy to put the kettle on, I knew something wasn't right as I stood up. I felt light-headed with a heaviness in my chest that I hadn't had before. Telling my partner I was going to lie down, I reached the bed and as soon my head hit the pillow, I felt the most intense pain in my chest. It was so strong that I sat bolt upright – not easy at 39 weeks pregnant! I knew I had to get to hospital.*
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> *My partner and I walked to the car but during the journey, my condition deteriorated. Just when I thought the pain in my chest couldn't get any worse, it intensified. My back stiffened and stuck in an arched position. I couldn't feel my legs. "Something's not right", I repeated, over and over. I was*

cold all over. The traffic near the hospital was gridlocked. We went to drive around it and a police car appeared. An officer got out to speak to my partner and on seeing something was wrong, he jumped in our car and drove it, following his colleague in the police car ahead, into the Accident & Emergency Department at St George's Hospital in Tooting.

A porter arrived and opened my door, getting me onto a wheelchair as I still couldn't feel my legs. I looked up and saw the ceiling lights whizzing past me one by one. That is the last thing I remember until I woke up in the Cardiothoracic Intensive Care Unit at St Georges Hospital several days later.

On arriving at the hospital, I had been taken to the maternity ward and treated as a 'woman in labour'. I was monitored overnight but no one seemed to know what was wrong. A chest X-ray was ordered, and I was given pain relief. The following morning, one of the nurses struggled to get a blood sample from me, and the Senior Midwife arrived on shift. ***I credit her with saving my life****. She now says that she took one look at me and knew my problem was not baby-related. She humbly puts her efforts down to a fresh pair of eyes arriving on shift. An alarm was sounded, and staff rushed to the unit to assist.*

Initially, I was treated as septic. I was put to sleep and taken for an emergency C-Section as the doctors were worried that I was infecting my baby, or vice versa. When my condition deteriorated after the C-Section, a chest scan was ordered. It was then that the issue was highlighted. I had suffered a Type A Aortic dissection. I was prepared for theatre and underwent 9 hours of open-heart surgery to replace my aortic valve and aortic root. My family sat in the family room waiting for updates. They were told I was the sickest person in the hospital with one doctor suggesting they say their goodbyes, as there was not much hope. Thankfully, I had other ideas!

Waking up in intensive care with no idea what had happened to me, was the strangest feeling in the world. Lying there with a tube helping me breathe, unable to talk and vials of drugs at each side of my bed, I felt like a pair of eyes. My partner and my siblings stood at the foot of my bed, giving me thumbs up, telling me I was doing well. I didn't know what I was doing well, but they seemed pleased enough!

My ICU Nurse showed me a photo of a beautiful baby girl, with knowing brown eyes, olive skin and lovely brown hair. My eyes darted around as memories of my pregnancy came back to me. My sister began telling me how my baby was in the Neonatal Unit being cared for by the staff. It all felt like so much to take in and I still didn't understand why I was there. As the days went by, I recovered well and was moved on to a ward, however, a week after my heart surgery, I suffered an ischemic bowel and was rushed to theatre for an ileostomy.[1] *Thankfully it was reversed a year later.*

I spent three weeks in hospital and during that time I was visited by a member of the Cardiology Team. He discussed my progress and mentioned something that would feature in the rest of my life, Marfan Syndrome. No one in my family had ever heard of it, let alone been diagnosed. We had no one tall in our family and the only clue was that my brother had scoliosis when he was a

teenager.

As time went on, the rest of my family were tested. We discovered that my dad, my brother, my new baby, and my brother's two children, all carried the gene. I thought my own diagnosis was hard to take but hearing that I had passed the gene on to my baby, knocked me sideways. I felt so guilty every time I looked at her and I worried constantly about things she was doing, and things she may never get to do. Over time I came to realise that there is a huge benefit to her being diagnosed early on. She is already taking Losartan and this will help her immensely. Medicine moves at such a fast pace and the Marfan Trust do such great research work – who knows what they will discover in years to come!

Julie suffered a Type A dissection, meaning a tear developed in the ascending section of her aorta, just as it emerges from the heart. This is a major symptom of Marfan syndrome, and yet Julie had remained dangerously undiagnosed because she didn't display the visible hallmarks which characterise the condition such as excessive height and elongated limbs.

As Julie's story testifies, Marfan syndrome is a family affair. With Julie's diagnosis came that of other family members including her new-born daughter. Should Julie's daughter choose to have children of her own, she will be offered careful monitoring with pre-conception counselling and, perhaps, a discussion of pre-implantation diagnosis (PGD). Any child of a Marfan parent has a 50% chance of inheriting the gene and so prospective parents have the opportunity to undergo IVF and test their embryos prior to implantation to determine whether they have the gene change that causes MFS. An unaffected pre-embryo can be selected,and implanted in the mother's womb. The parents then know their baby should not have MFS.

As a happy side-note, Julie and her family remain in close contact with our Marfan Trust, with her son recently volunteering at one of our conferences.

In contrast to Julie, Claudia was diagnosed with Marfan syndrome as a small child and entered pregnancy with knowledge of her condition, albeit with worries about how it would all unfold. Claudia approached the Marfan Trust, newly arrived in England from Italy and in her 32nd week of pregnancy. She shares her story, below:

By Claudia

I worried when I fell pregnant because I didn't know enough about my condition. I didn't know my Marfan gene wasn't the most serious manifestation and I didn't know that I could transfer only that gene to my child. I worried that my baby would develop a more severe manifestation of Marfan.

In Italy, my doctor was the one I'd had since I was a child and so she has followed me since I was little. Arriving in London I felt lost. What helped me most was speaking to my Italian doctor who put me in touch with Dr Anne Child, Medical Director of the Marfan Trust

. Speaking to Anne was so reassuring, especially when she explained to me that my gene was not the most severe but a milder variant of Marfan syndrome. The NHS only seemed to know of the most severe cases of Marfan syndrome and they treated me accordingly, as though I was very ill, which I found very stressful.

During my pregnancy, my NHS doctor monitored me very closely and it all went well. In fact, it felt like private treatment. I was given regular echocardiograms as the doctors kept an eye on my aorta. But I would have preferred to be referred to a Marfan pregnancy specialist, and see someone who was an expert in the syndrome. Ideally none of my questions would have been left lingering.

Throughout my pregnancy I called the Marfan Trust for advice and someone was always there to answer my questions. This was very comforting for me.

I hoped for a natural childbirth and my NHS doctor was happy for me to have one. In Italy, it is normal for Marfan patients to have C-Sections. If there is aortic dilatation during pregnancy, then this is what is advised. I was relieved that, in England, I was allowed a vaginal delivery. My aortic measurements were all compatible with trying for a natural childbirth.[2] I was told that I was allowed a maximum of 30 minutes pushing so as not to stress my heart.

Childbirth went well and, as I was considered a high-risk patient, I was kept in hospital for longer than usual and the maternity care was very good. The obstetrician even called me after I was sent home which I thought was very considerate.

In retrospect and although everything went well, I wish my doctor had been an expert in Marfan. I would also have liked to have seen all the medical reports relating to my pregnancy. Not everything was given to me by the doctor. I would also have liked a 24-hour helpline to speak to about my aorta. The Marfan Trust were there for me always and never failed to pick up the phone. I think this is what every pregnant Marfan woman needs.

Claudia has since delivered two more beautiful babies into the world and loves being a mother.

Marfan syndrome is a complex condition that can complicate pregnancy, but with medical guidance and careful planning from pre-conception to post-birth and breastfeeding, a successful outcome can be assured.

For further information on pregnancy and childbirth with Marfan syndrome, pleaseclick here. Meanwhile, the Marfan Trust can be reached on 0333 011 5256 / info@marfantrust.org / www.marfantrust.org

Author Bio: Victoria Hilton is the helpline and communications officer at the Marfan Trust, the only charity in the United Kingdom dedicated to improving and saving the lives of those with the condition.

1 Editor's note: An ileostomy is a surgical procedure that brings the end or loop of the small intestine to an opening created in the surface skin of the abdomen. Bowel ischaemia occurs when blood flow to the bowel is blocked or severely reduced.

2 Editor's note: AIMS reminds the reader that it is the mother who makes the decision (or does the allowing), even when there are strong reasons (as in Claudia's case) to have a medical care plan in place.