

Introduction

Hi – I'm Vic and I am what doctors consider to be a patient with a severe case of McCune Albright Syndrome and Fibrous Dysplasia. You are Learning about the symptoms of MAS & FD at this conference from some of the world's leading experts in the field. I hope that relating my personal experience will help you understand that as life changing as these conditions can be, that with the right support and positive outlook you can lead a relatively stable life.

This is the first time I've presented to such a large group of people, so sorry if I'm a bit nervous.

My Journey With FD & MAS

I'm 28 years old and have lived with MAS from birth. My mum had a normal pregnancy and uncomplicated birth. I was small but perfectly formed. Within two weeks my parents knew something wasn't right as I had poor muscle tone, was a poor feeder and I developed the classic café o lait mark on my face and left buttock. I developed complications at 6 weeks, being admitted to Cheltenham General Hospital with pneumonia and I was then blue lighted to Bristol Children's Hospital.

The next symptom to show itself was Cushing's Syndrome which affects the pituitary gland which causes the overactive adrenal glands to produce excess cortisol. This spontaneously cleared at 6 months. X-rays identified abnormalities with my bone structure and this turned out to be Fibrous Dysplasia which affected all my bones. Along with the café o lait mark this allowed a senior geneticist from Great Ormond Street to diagnose I had McCune Albright Syndrome. (At about 6 months)

At last my parents had a label for my condition, but because there was very little information published and no Internet as such, we were no further forward on the long-term outlook. We were told at the time there were only 9 known cases in the UK.

I developed a cardio myopathy (a thickening of the heart muscle) which was treated with medicine which again spontaneously resolved, with a paper being written by my heart consultant as they had never seen this before.

I had my first period at 2 ½ years old, being the onset of precocious puberty, which was managed successfully with tamoxifen initially and then the use of the contraceptive pill.

I have had many fractures from about 18 months old, two to my tibia's, femurs and humerus and one to my ulna. I was mobile albeit with crutches and my parents encouraged me to walk wherever possible to maintain bone density. In the course of my treatment I had to have rods and pins inserted into my upper and lower legs, particularly as I started to develop the classic shepherds crook neck of my right femur.

In between fractures I suffered like many people with FD from severe bone pain which affected sleep, concentration and quality of life. I was referred to Dr Shaw at Birmingham Children's Hospital to be part of a trial using Pamidronate infusions which then led onto the use of oral Reisedronate. These bisphosphonates helped with the pain and I underwent regular bone density scans.

At 14 years old I had a torsional twist in my right ovary due to a cyst developing in it, initially diagnosed as an appendicitis. The ovary was removed along with my appendix. I have had scans subsequently to check for further cysts as we have learnt this complication has been seen in other female MAS patients.

At age 10 the onset of scoliosis meant I came under a spinal consultant who advised use of a body brace to try and limit the curvature, however by age 14 it was deemed that I would develop breathing issues unless spinal surgery was considered. In 2006 I underwent surgery to insert rods in my back and unfortunately this resulted in paralysis post operation at C6/C7 level. I was in rehab then for 6 months.

This presented me with a new set of challenges and we did for a period lose focus on my MAS & FD in coming to terms with dealing with paralysis. During this period my thyroid had to be treated with

radioactive iodine when it became overactive, yet another MAS symptom.

I had a period of being unwell two and a half years ago where as part of the diagnosis scans revealed I had a benign cyst in my liver again a condition that can arise with some MAS patients.

What We Have Learnt

In the early days information was limited and not being shared. My dad came across a Foundation in the USA called the Magic Foundation, which focused on rare conditions of which MAS was one. Through contact with them we attended a conference in Chicago in 1997 where we met Dr Stanton, Dr Feullan and Dr Schenker. Through this conference we learnt about the work being carried out at the NIH in Washington, who subsequently were contacted and were able to provide research information to help our doctors in the UK.

This leads me to my list of lessons learnt and survival tips:

The importance of having information regarding your condition and access to experts in the field of FD and MAS. It was only when we requested a change of spinal consultant that we learnt of Prof Kassim Javaid at the Oxford Nuffield Centre. We had no idea up until then of his expertise in the field of FD and MAS, the RUDY study and of course the FDSS UK group and these conferences.

From the conference last year, I learnt of a research study at the NIH in Washington regarding MAS & FD patients. I applied to be part of the study and after careful consideration of my condition they invited me to attend. Whilst I had to fund getting to the USA myself, I was given a full MOT at the leading research centre working with Dr Collins and Dr Alison Boyce. They confirmed what I needed to be aware of and to ensure my UK specialists know what is needed to be carried out to ensure my condition is managed.

The NHS is not as well joined up as it could be in pointing FD and MAS patients to the right experts, although we believe from experience this may be partly down to arranging funding between trusts. This highlights the importance of

conferences such as this bringing together specialists and patients to promote information sharing.

Attend regular checkups for each of the areas affected by MAS and FD and take time to keep those appointments.

I learnt from the NIH that MAS patients can suffer from torrential bleeding when undergoing major operations and that measures need to be put in place to manage this. Also, I am not allowed to take Asprin or Long-Term Ibuprofen as this can affect platelet function and affect clotting.

Be proactive – the NHS as good as it is, is under pressure so do keep calling / contacting them – don't rely on the system to always remember you.

I felt the impact of transferring from children to adult services. I expected to receive the same levels of care in terms of priority in outpatient appointments when in adult services. It is important to be aware that there may be such changes. I became ill from my thyroid issues as my Endocrinology follow ups failed once in Adult services and issues were not picked up early on.

Keeping a diary and filing your own medical appointment letters, results and consultant feedback – it's amazing how time passes and you can't remember when some key dates have occurred.

Don't be frightened to get a second opinion in respect of your diagnosis and treatment.

Have your family GP on board, keeping them abreast of what's happening with your condition. I've been fortunate to have two GP's who have always actioned what I've requested, when backed up with the medical advice from both UK and US doctors.

Keeping fit by doing the right exercise that is not too strenuous which may cause fractures. I attend Hydrotherapy and wheelchair dance.

Listen to your body and rest when needed.

Drink lots of prosecco – I'm joking of course but a little does help !

My Goals and Achievements

I have always been encouraged by my parents to be as normal as possible and not let my syndrome define me. As a child I went to main stream school, joined in swimming lessons and participated in PE lessons knowing my limitations. I was taken to France on a school trip albeit I had to have one to one TA's with me. I did what any other young child would do, go to parties, sleep-overs and surround myself with friends

When I was paralysed things dramatically changed as I had to adjust to a whole new set of problems. In rehab I was encouraged to set myself goals to achieve, the first being to leave hospital and pass my GCSEs and carry on to sixth form. Second was to find employment, this proved a bit more difficult as I believe employers are still unsure about employing a disabled person. I was accepted on an apprentice scheme with a local transport firm who on completion took me on full time. I work within a small team who are all fully aware of my disabilities and support me when I have been unwell and hospitalised. Access to work ensured the workplace was set up to meet my needs. My job gives me purpose and I really enjoy it.

Next came learning to drive. I think that was my biggest achievement. I'm sure my Dad has a few extra grey hairs from taking me out when I first started learning in my adapted vehicle, that has no steering wheel for him to grab!

Finally, my next and final goal was to live independently albeit with a full-time personal assistant. I moved into my own bungalow last December. It was a big step as my parents have been there

for me throughout my journey. It hasn't been as difficult as I thought it would be and I'm loving being in my own space.

THE FUTURE

Knowing what I have learnt from my visit to the NIH and all my specialist doctors at the NOC I am more aware of my condition. I understand the need to look after myself, not to panic when I experience unexplained episodes. Although I cannot feel pain below my spinal injury level anymore due to my paralysis, It can manifest itself in other ways.

I don't expect my condition to stay stable for the rest of my life. Armed with the information we have obtained and support from doctors and this group I feel confident I can deal with these changes.

As a family we know how important the FDSS charity is and the need to spread the work being carried out to as wide an audience as possible. We plan to continue to be involved as much as we can. We as a group need to support the research wherever we can and I'd like to encourage you all to join in this.

Thank you for listening.

Any Questions?

