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February 2013, United Kingdom

Sam and her daughter Poppy

My name is Sam, I'm 34 and was diagnosed with Pompe Disease in February 2010. I live in the UK with my boyfriend Stuart and our beautiful little girl Poppy who was born in January 2012.

Like many people with Pompe Disease I experienced symptoms for several years before I was diagnosed. These started mildly and gradually worsened over 10 years or so. The main things I noticed were difficulties walking, climbing the stairs, getting up out of a chair and picking things up from the floor. I always felt weak and exhausted and was in constant pain. I had breathing difficulties that were causing sleep disruption, excruciating headaches and lethargy, and together these symptoms really impacted on my day to day life. Simple tasks that other people took for granted were becoming a real challenge. I visited several doctors over the years who felt my problems were due to being unfit or stressed. I was someone that had always been healthy, motivated and had exercised regularly so I felt frustrated that no-one seemed to believe my problems were real. I was initially misdiagnosed with Limb Girdle Muscular Dystrophy, but then following further tests it was discovered I actually had Pompe Disease. When I was finally diagnosed I was relieved to understand what was wrong with me, but I was also terrified about my future and felt very depressed in the first few months after learning the diagnosis.

One of the symptoms that caused me most problems was the respiratory issues. I was waking at night feeling panicky and unable to catch my breath. When my breathing difficulties were recognised I was given a bipap to use at night. Although this was a terrible shock, after a few weeks of use I noticed I felt much better. I started ERT soon after diagnosis and built up a supportive network through the AGSD-UK and with other Pompe patients around the world. This helped me to feel more positive and I learnt that Pompe didn't have to ruin my life. I was inspired by many of the people that I contacted and started to see that although my life was going to be different to what I had envisaged, it didn't mean it would all be bad.

From the start, one of the most important questions for Stuart and I, was about how this disease would impact on our ability to start a family. The doctors involved in my care had various opinions, some more optimistic, others very concerned about the impact it would have on my respiratory function and general health. We had many questions related to how pregnancy would affect my body and breathing, how I would manage to care for a child, and most importantly what were the chances of passing of the disease on to our child. We found it really hard to decide whether or not to have a baby. Stuart and I spent a long time discussing the potential problems with the various health professionals involved in my care, but we eventually decided that we wanted to try.

The doctors explained that the likelihood of Stuart being a carrier was extremely low, and because of this they were reluctant to test him. However I did not find this very reassuring as the likelihood of both my parents being carriers would have been 'low' in theory, but they were carriers, and I therefore inherited the disease. It was explained to us that whilst the test can sometimes identify carriers, there are times when it can identify a sequence variant in the gene that has not been previously described. In this case the result would be difficult to interpret and it would not be known if the variant would cause the disease or not. However we were very anxious, and decided that if there was a test available, then we would like to have this done. We fully accepted that we may end up with an

inconclusive result, but felt we needed to do everything possible to prevent us passing on this disease to our child.

Stuart was tested in May 2011 which very fortunately showed he was not a carrier of Pompe Disease. Ironically fate also intervened and I discovered I was pregnant, 3 days before he had the test! This was a big surprise and not part of the original plan but the outcome of the test meant a lot to us and made my pregnancy much easier to deal with, knowing that the baby would not inherit the disease.

PREGNANCY……

Sam being pregnant

I was classed as a high risk pregnancy so was under the care of an obstetrician as well as the midwife from the start. My respiratory consultant and GP also monitored me closely and I was referred to other specialists in London for advice. I stopped Myozyme for the first trimester (on my doctors' advice whilst the baby was developing) but noticed a deterioration in my strength, stability and breathing. I noticed more weakness in my upper body and kept waking up in the night with poor breathing feeling panicky, similar to the breathing problems I experienced before diagnosis. However this resolved once I restarted ERT in the second trimester.

Other than this I had a very straightforward and easy pregnancy. It went very smoothly and was probably no different to someone without Pompe. In fact it was probably much easier than some of my friend's pregnancies. I felt well, energetic, healthy and experienced less pain than before. I had visited Dr Slonim in New York shortly before I found out I was pregnant so I followed his diet and exercise recommendations throughout. I did antenatal yoga and carried this on until I was 41 weeks pregnant. My breathing remained stable - it was slightly more difficult but this is usual for anyone who is pregnant. My bipap settings remained the same throughout. I had monthly overnight oxymetry for the last 3 months and 2 sleep studies to monitor (halfway and towards the end). I continued working full time until 34 weeks. Walking, moving, getting dressed etc. became more difficult towards the end, but this was related to being heavier and larger and I feel it was again normal for pregnancy, not necessarily related to Pompe. Looking back I wish I had enjoyed my pregnancy more but I was so terrified that something would go wrong, that even up until when I went into labour, I didn't allow myself to believe I might have a baby at the end of it.

THE BIRTH……..

Poppy, just a few hours old

My respiratory doctor was keen for me to have a caesarean but all the other doctors suggested I should have a vaginal delivery with an early epidural for pain relief, so this is what I did. I spoke to several other women with Pompe who'd had children and the general opinion seemed to be that a vaginal delivery was preferable if possible.

I was induced at 41 weeks and 5 days. I used gas and air for pain relief then had the epidural after about 7 hours of contractions. Unfortunately the epidural didn't work for me so I still experienced a great deal of pain but I coped ok with this by using my yoga breathing techniques. The doctors had agreed in advance that I would only push for 30 minutes maximum so that I didn't get too tired or compromise my breathing. I pushed for about 45 minutes in the end then they used a ventouse to help the baby out. Again this was

very straightforward and we were discharged home from hospital the following day.

COPING AFTERWARDS……

Back at home

The first few weeks after the birth were definitely the hardest. I was blissfully happy and loved motherhood, but really struggled physically. I found it very difficult to walk or even stand due to very poor balance and pain. I needed help to get washed and dressed and it was difficult looking after Poppy because I couldn't stand up holding her as I felt so unsteady. I couldn't lift her out of the crib, couldn't carry her upstairs and couldn't put her down or pick her up from the floor so really relied on Stuart. I spent most of my days sitting on the sofa, breastfeeding her every 2 hours. I found breastfeeding extremely painful and this pain was worse than the birth! However this improved as time went by. I initially found it difficult to have visitors in the house for too long as it was very overwhelming and tiring. I was scared to leave the house as my mobility was so poor, so I had to build up my confidence gradually with short, easy trips to familiar places. I had to practice doing everything such as using the buggy and lifting Poppy in and out of the car, so that I could trust myself to cope on my own.

Enjoying the Summer together

By the time she was 4 weeks old Stuart was back at work and I have looked after her on my own during the day since then. Whilst there are many things I can't do, I have adapted well and developed different techniques as Poppy grows. I can do most things myself and have an adapted cot and other equipment to help me. I can't pick her up from the floor or stand from a chair with her, but I have found ways of managing. I can't give her a bath as I can't bend and lift her and have to rely on Stuart to do this. I do feel that Poppy and I are in tune with each other and she adapts well. For example from the age of 4 months she knew to put her arms up in the air to help me lift her. Now she can crawl, walk with a walker and pull herself to standing I am finding it much easier. I'm still breastfeeding a year on, and just give her expressed milk during and for 24 hours after an infusion. We go out most days, but I just have to make sure that we go somewhere I am familiar with where I know I can cope, or I make sure I'm with other people who can help me with the things I can't do. My family and friends have been great, and don't make me feel awkward when I need them to lift Poppy. I go to a couple of baby groups and again, the teachers are brilliant in helping me – I feel it's important to do lots of activities so that Poppy doesn't miss out because of my disability.

I wouldn't say it is easy coping with Pompe disease and a baby, but it is definitely easier than I thought it would be. Of course I have bad days – I recently fell for the first time when I was carrying Poppy which was awful and really knocked my confidence – but the good days outweigh the bad days no end. Overall, I don't think that it has worsened my Pompe. In fact I feel the healthiest I have in years. My walking is worse but I feel this is more due to me not exercising as regularly and having become reliant on leaning on the buggy when walking. I feel that ERT, diet and exercise has helped me a great deal as there is no way I could have looked after a baby full time prior to starting ERT. I've also learned to listen to my body and pace myself, and to stop and rest when I need to. I've recently gone back to work, but I'm only doing 3 days a week as I want to save my time and energy for enjoying my days with Poppy. I'm sure more challenges will crop up as the years go by, but I feel proud of how I have managed so far and feel equipped to face whatever happens next. I can honestly say that I am the happiest I've ever been with my little family.

Our family

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