Hollie's NP-C Journey

We knew nothing about Niemann-Pick disease until 2007 when our second child Hollie was born. We already had a son, Joshua who was 3 years old at the time. Thinking back my pregnancy with Hollie had always felt different to when I was carrying Joshua.

I would regularly visit my midwife and question whether my "bump" was growing as it should. I felt extremely small in comparison to when I carried Joshua certainly in the latter stages of pregnancy. At about 32 weeks my midwife measured my stomach with a tape measure and told me I was measuring approximately two weeks behind but reassured me it was unlikely to be anything to worry about however it continued to play on my mind.

By 37 weeks I was measuring 4 weeks behind and my midwife immediately referred me to our local hospital for a scan as she was worried that the baby was failing to grow. This resulted in my pregnancy being induced at 38 weeks and Hollie was safely delivered on the 11 June 2005 weighing a tiny 5lb 12oz. Even when she was born and handed to me I remember saying to the midwife "she is so tiny, something must be wrong with her" but the midwife replied "she is small but perfectly formed".

We returned home with Hollie the following day and Joshua was over the moon with his new baby sister. Straight away we noticed Hollie was quite "tanned" looking but we knew it was not uncommon for new-borns to have jaundice for the first few days so never gave it much more thought….

As the early days passed Hollie's jaundice seemed to subside but her stomach looked a strange shape. Again, we were not overly concerned as we put it down to the fact that Hollie was small but feeding well and her stomach was just swollen from all the milk she was drinking. But then we noticed that Hollie seemed to become more and more unsettled at night, she would scream constantly and if she wasn't screaming she was feeding continuously. Nothing seemed to settle her at night and it began to concern us that this was more than just a simple case of colic. Then I began to notice how jaundice Hollie would look first thing in the morning when I would pick her up from her Moses basket.

The midwife however appeared content with Hollie as she was growing well and her jaundice would appear to fade as the day went on. It wasn't until our Health Visitor did a home visit at 6 weeks that things changed. I undressed Hollie so she could be weighed and straight away my Health Visitor raised concern about the shape of Hollie's tummy and the fact that she still appeared jaundice. We were immediately referred to our GP. I knew

something was wrong when the GP asked me to leave the room whilst she spoke to the hospital and then came out and told me to take Hollie there straight away.

By the time I reached Milton Keynes Hospital I was frantic with worry, numerous doctors were tapping Hollie's stomach and talking amongst themselves. I then had to face the dreaded blood tests for the first time, a procedure which is now second nature to Hollie! I can recall standing in the corridor in floods of tears from hearing Hollie's screams as three doctors tried to get blood from her. From Milton Keynes we were referred to Kings College Hospital in London where we were told that Hollie's liver functions were extremely poor and she was very unwell.

From this point to diagnosis took a further two years. Hollie underwent extremely evasive testing for numerous illnesses and liver related diseases. All tests drew a blank but Hollie's liver functions were extremely poor and doctors were seriously concerned and baffled. Then strangely at around ten weeks old Hollie's liver functions started to return to near normal and we were allowed to take her home. She continued to develop normally apart from her enlarged spleen. Hollie was classed as having "undiagnosed liver disease" and put under six monthly review. Life returned to normal and we felt like we were extremely lucky to have escaped anything more serious.

The only sign that Hollie ever had a problem was her distended tummy. We had expected this to subside with time but it never did. Every time Hollie had a review at Kings College Hospital the ultrasound would show that her spleen was enlarging although her liver seemed to be stable. Every doctor we came across told us not to worry and her spleen was just enlarged due to the early liver disease she had suffered.

It wasn't until we met a doctor on a routine liver review at the beginning of 2007 that had come across Niemann-Pick disease before that we were put on the road to a devastating diagnosis. By this time Hollie's spleen measured a huge 14.5cm on the scan and he felt a fresh investigation was now required. Hollie underwent further invasive testing, a 2nd liver biopsy, and bone marrow and skin biopsy. No one could have prepared me for the news I was to receive over the telephone at work shortly after from a Registrar who appeared to know little about the disease. I was told that tests indicated that Hollie could have Niemann-Pick Type C and that she would start to decline any time after the age of 2 and lose the ability to walk and talk. He said he was extremely sorry especially as Hollie had been doing so well. I was told to go and look on the internet to find out more and come back in six to eight weeks when the results were ready. I went straight to my desk and typed into an internet search "Niemann-Pick Type C".

What faced me was horrific, I remember my eyes just skipping from one sentence to the next looking for a positive but I just couldn't see anything. All I could see were the words, neurological decline, dementia, and no treatment or cure.

The next ten weeks were the worst of our life. I took it upon myself to search out more information and found the contact details for the Niemann-Pick Disease Group (UK). Toni Mathieson, development manager for the charity also had a daughter with NP-C and she kindly provided me with the support I needed and referred me to the Clinical Nurse Specialist based in Manchester who gave us a clearer picture of the disease and testing process.

The definitive results came through in July but I think we had already accepted the fact it was NP-C long before that. We knew that the liver biopsy and bone marrow biopsy all pointed strongly in that direction and although we tried to remain positive deep down we knew it was now just a case of rubber stamping the results.

People constantly ask us how we cope and comment about how brave we are but I don't think we are brave. We have no choice but to accept that NP-C is part of our lives and the fact that we may never know for sure when the disease will start to show itself fully. The uncertainly that this carries is extremely hard. Each day Hollie learns something new and she is the most happy little girl, bubby and full of fun. She is a very active child and is never happier than when she is dancing to music in the lounge or playing on the slide and climbing frame in the garden.

The thought of Hollie losing the ability to do these things is still unbearable. The immense fear that sometimes engulfs me about the future is terrible but we are determined to try and keep this hidden from Hollie and Josh and want to give Hollie as normal a life for as long as we possibly can, not just for Hollie, but also for her older brother Joshua and her younger brother Lucas who is too young to understand what the future holds for his sister and for the rest of our family - grandparents, aunts and uncles who all find the diagnosis difficult to come to terms with.

Hollie is now 6 and settled in school with a close group of friends. At the moment her symptoms still remain mild although changes are beginning to appear. Hollie has high frequency hearing loss, a neurological symptom of the disease. The high frequency hearing in both ears is now deteriorating and she wears a hearing aid in one ear. By early next year we have been told she will need a hearing aid on her other ear as well. She has some balance problems, sleep disturbances and mild difficulty with upper eye movement. She hates people spinning her around or pushing her back suddenly or the motion of a swing as he makes her feel extremely dizzy and distressed. Her spleen is still very enlarged so she continues to wear a spleen guard during school time play.

Our biggest concerns recently have been the apparent gap appearing between her and her friends in school with writing skills. Her writing is very poor and she now has a laptop to help her complete homework tasks. We are however extremely lucky that we have Hollie in a highly supportive school where the teachers and staff go out of their way to ensure Hollie has the very best care and support to help her continue to progress for as long as possible.

This is a huge relief to us because Hollie absolutely loves going to school and learning new things.

Hollie is on a drug called Zavesca that has been shown to stabilise symptoms of the disease in some patients although it is not without side effects. The hope is that this drug may buy us some time until a better treatment or therapy can be found.

Like every parent with an affected child, we find the progress towards a better treatment or ultimately a cure painfully slow and at times frustrating. We are in no doubt that this frustration and impatience will only get worse as the disease starts to show itself more and time begins to run out for Hollie. We are therefore extremely encouraged by the fact that a clinical trial is starting this year at the National Institute of Health ('NIH') in Maryland into a potential new treatment for NP-C called Cyclodextrin. This will be a Phase 1 study; if the trial reaches Phase 3 it is at this point that it will hopefully become a multi-centre trial giving the opportunity for patients in the UK to enrol on it within a UK based centre. To find out more about this and other research news please visit www.niemann-pick.org.uk

As for the future, we will take each day as it comes, pray for a cure or treatment soon and if that times doesn't come soon enough for Hollie we hope that when the disease starts to progress we will be strong enough to cope with what we will have to face.

Helen Carter
Mum to Hollie aged 6 (NP-C) and Joshua, aged 10 and Lucas, aged 2
www.hopeforhollie.co.uk