Praying for a Cure

A swollen stomach was the only clue to Gina's son's rare problem

I rushed to the loo — at six weeks pregnant, I was suffering awful sickness. But I couldn't fuss about myself — my son James, two, had begun vomiting as well.

Soon he had a high fever and wouldn't drink, so my husband and I took him to our local hospital. James was admitted with pneumonia, but a doctor noticed his tummy looked swollen. 'His liver and spleen are inflamed,' he said.

James recovered from the pneumonia with intravenous antibiotics, but his tummy was still big. Six months of hospital tests were all inconclusive.

Two months on, I gave birth to William, but we still didn't know what was wrong with James.

Finally, James was referred to Great Ormond Street Hospital in London. Six months later, a specialist diagnosed ASMD Niemann-Pick Disease Type B.

'It's a rare inherited disease that can cause the major organs to malfunction,' he explained.

My husband and I sat terrified. 'There is no treatment and no cure,' I remember hearing.

Later, I surfed the web. There were different types of Niemann-Pick Disease — Types A and C were more serious. James had Type B — the disease wouldn't affect his brain.

It was a comfort, but then came the next shock. My husband and I discovered we were both carriers of the disease. It's how we'd passed it on. We'd simply had no idea.

Heart banging, I wanted more information and rang the support group – the Niemann-Pick Disease Group (UK). Luckily, there was a Niemann-Pick Family Conference being held in Telford, Shropshire, that weekend, so I decided to go along.

I remember walking into the hall. There were children in wheelchairs, some of whom were very seriously affected by Niemann-Pick disease. There were parents of kids who had died of the disease.

I almost ran away. But I forced myself to stay. For James' sake, I had to find out all I could. It was a positive meeting — the parents and families I met were so supportive.
'Lots of research is being done,' I told my husband when I got home, 'and time is on James' side.'

With the help of the NPDG (UK), we gradually found out more and more about the disease. With every year that's passed, my husband and I have grown more hopeful.

Gene therapy could offer a cure in the distant future. And closer than that, enzyme replacement therapy is being trialled in America. It's something the support group has been pushing for and a real breakthrough in combating this awful disease.

James is now a teenager, a football-loving lad who doesn't look or feel ill. The only signs something is wrong are that he's small for his age and his tummy is still big and hard.

It's difficult for him to sit cross-legged and he avoids contact sports in case he gets knocked. Thankfully, his brother William is clear of the disease.

We know James' condition may deteriorate as he grows older and he could develop breathing difficulties. But I refuse to think of that. As I told my husband right back in the beginning, time is on our side. A cure will be found.

GINA, mum to James and William