Sam's Story:
Our Fight with ASMD Niemann-Pick Type A

Sam was born in September 1997. Not a normal birth, just a little help needed from our friendly midwives and consultants, all 7 of them in total. They were bored and had nothing to do and we were the night's entertainment.

Sam never sat, walked, well he did with help and his second word was bugger, the normal "dad" being his first. I was diagnosed as being a paranoid mother and in need of help because I was always chasing the doctor and health visitor to see why he wasn't doing what he was supposed to be. "All children develop at different rates!" apparently.

We went to Isle of Wight for a holiday and Sam was admitted to St Mary's Hospital with Viral Bronchiolitis. We were only going to be there for 5 days. Sam was in hospital for 5 nights.

Within 2 weeks of that he had Chicken Pox. His spots had spots. He was covered. We went to the doctors because it was so bad. At some point during this day I had noted that I hadn't needed to change him. I told the doctor. His reply was "to the hospital you go and don't come back till Sam has had a full nappy"!!

The Consultant on the children's ward was wonderful and we were asked several questions regarding our family backgrounds. "Are you related?" was a good question to ask "yes, we are married" "no are you blood relations?" "NO!" What a question, never been asked that one before!

Blood tests were done and what was going to be a couple of hours landed up being a couple of days. When we were discharged we were told that we were to return back to the ward within 2 weeks, when they would have the results and would be taking more blood.

Apparently this was to see if Sam's white blood cell count had altered in anyway.

Unfortunately, Sam had other ideas and he landed up back in hospital with Rota virus before this appointment came. During our stay we were told that we would have to go to Birmingham Children's hospital. We were going to go to ward 8, the Liver unit.

Here we met Professor Deidre Kelly. She would be our rock during our stay. More tests were done and the outcome of what we didn't want to hear came with a bang. ASMD Niemann Pick Type A (NP-A). What? Never heard of it; I think they must have made this one up!!! We told our families, and friends. During this time we found out who our friends were and those who weren't.

We soon had follow up appointments to go to both in Northampton and Birmingham. We had been given a place within the Child Development Centre (CDC) for a Tuesday morning and Sam had Room 1 on Paddington Ward and a place at Rainbows Children's hospice in Loughborough.

Things were beginning to happen or not! During this time I was pregnant with my second son, Iwan. I had to go to John Radcliffe, Oxford for a pre-natal test called Chorionic Villus Sampling (CVS) test. Thankfully it came back negative.

Sam continued to progress as much as he could. His second Christmas saw Sam attending a party at the CDC. He sat in his chair eating bread sticks and Wotsits. This was the last time he would feed himself.

In January 1999, Iwan arrived, at this point everything changes. I think this is where Sam had decided to let go. Iwan had arrived and Sam started to go downhill. His head started to flop to one side.

Little things happened to begin with; his head control, then not being able to pick things up. Gradually he flopped completely. Then he lost his ability to feed. A Naso-Gastric tube (NG tube) was added to his good looks. Goodness he hated it and pulled it out at every opportunity. For someone that had little or no control over his movements he did very well to pull it out and I soon had to learn how to put them back.

Sam had grown so much because of his food that he was having through the NG tube. With his arms and legs he was just like a rag doll. But he was still my Sam.

We fought with the local NHS for a chair. We ordered it in the March and August it arrived. August being too late, Sam was too floppy to sit in it. The Occupational Therapist (OT) adapted the head rest so he had two big red things that came out of the side. He looked a bit like Mickey Mouse.

We tried to get Birmingham to put in a PEG feeding tube (Percutaneous endoscopic gastrostomy) instead of the NG tube, as Northampton hadn't dealt with a child with NP-A before and didn't want to risk it, so they say.

Birmingham said that Sam was to go on the waiting list. Waiting list, how long is this? They told us "A couple of months!" We said "But Sam may not last that long". They said "He is on the list!"

In July, we had a letter from a lady called Jackie Imrie; apparently she was our Niemann-Pick Nurse. The letter said if there is anything that I can do!!!!

No more had to be said. On the phone and explained the situation. "Ok leave it with me, let me see what I can do and I will be back to you"

Within the week Jackie was back in contact, I can give you two dates. Both were before Sam's christening, "don't worry leave it till the Monday after and come up to Manchester. After all we don't want Sam to be ill for his christening do we?"

On the Monday we went to Manchester Children's Hospital and met Jackie; a lovely, friendly, warm lady. The surgery was done the next day and everything went well, we were home by the following weekend.

The weeks that followed Sam smiled and laughed. Though a little grumpy. We think Sam's eyesight had gone and he could only see light and dark. But he was the happiest we had seen him for ages, although he did land up back in hospital for his 2nd birthday when his PEG tube got infected but he was a happy chappy.

On the 16th October work colleagues had a disco in honour of Sam. Sam being the guest of honour made his feelings felt. He hated it, loud noises, people making a fuss of him. He complained bitterly. My godson took pity on him and held his hand for the whole evening, not budging from his side. Not until it was way past his bedtime did he leave his side.

At 2.30 the next morning we were woken to silence, nothing unusual you would say. Sam snored loudly, but there was silence. We knew at this point we had lost him.

I phoned Paddington Ward to ask what I should do, the nurse I spoke to just cried and said sorry she couldn't help me, another took over and told me that I should call the out of hours doctor, they would come out and declare his death.

Over the next week all went by in a mist of something, not sure what. Each day we had a house full, what with friends, family, vicars and all. The funeral was lovely a packed house, people from all sorts of life. Each holding a special memory of our little boy.

Oh - I received a letter from Birmingham Children's hospital at the beginning of November, asking Sam to come on the 29th November for his peg. He had passed away on the 17th October!!!

At some point during this period we had been given some money and we also had some money from the disco. A few friends suggested that we should start up a charity and that was what we did, "The Sam White Trust Fund!"

The fund gives financial support up to £1,000 to every family that has a child diagnosed with NP-A. They can do what they want with it, in the past some have used it for funeral payments others have brought a therapy pool, it is for whatever they see fit.

We are still going and have helped a fair few families over the years. I have also been the family contact for NPA and have spoken to people from all over the world and I am still in contact with a couple.

We have received money from all over and held several events over the last 10 years or so.
I am there for any family that wishes to have someone to talk to about NP-A, as someone that has gone through what they are going through. This I never had!

Since then I have had a third son, Luke, whose claim to fame is that he is the youngest delegate to attend a Niemann-Pick Disease Group (UK) Annual Family Conference at just 36 hours old!

Between Iwan and Luke I also had another baby. Unfortunately this one turned out to be a positive for NP-A.

By Kris Burrows, Sam's loving mum

Story told in loving memory of Sam White
14/09/97 – 17/10/1999