

## Azaria's Story

Having completed our Manchester phase of the drug trial, our next hurdle was to get back to our local hospital for treatment. Initially we thought that this would be straight forward, however life's not like that in this household! When our six months was nearly up and big plans for a party back in Plymouth all in place, the carpet was pulled out from beneath our feet again. Plymouth's Derriford Hospital (only 15 minutes drive from home) did not have a Paediatric intensive care, and the drug company insisted that her treatment be carried out at the nearest hospital with this facility, in case she should need it any time. This meant that the process of setting up our transfer was back to square one. Our nearest unit was in Bristol, 2 hours from home, but at least we would be back living at home.



*Azaria enjoying her donut, while receiving her infusion in the hospital.*

In January 2005, eleven months after treatment started, we were transferred to Bristol for Azaria's fortnightly infusions. The staff were lovely and the Sister of the investigations unit, where the ERT was given, remembered Azaria from her previous visit, when our consultant gave us real hope that the treatment could help her live a normal life. She was surprised to see how well Azaria was looking. Ian took us up the first week, so I would get my bearings of where to go. Although I'd driven ever since I was old enough, it was always Ian behind the wheel on any trips and it had been a long time since I had done any motor way driving, and I

having to do the trip each fortnight, but what options did I have. My mum was fantastic and volunteered to come with us, which was handy as I didn't have to deal with Azaria's bored requests for drinks and toys and anything else from the back of the car, at the same time as driving. It also helped boost my confidence and help me relax, as my mum has always believed in me and can always make me feel good about anything!

Just prior to our transfer, we had a special holiday. When I had told my oldest friend about Azaria's diagnosis she decided that she would raise some money for us to have a family holiday in Euro Disney. With moving to Manchester and the drug trial, it was a year before we finally got around to going. Azaria was much stronger now and able to enjoy the rides she was big enough to go on – although Minnie Mouse was quite a different story, just by saying hello she reduced Azaria to tears! We have very happy memories of a fantastic holiday which are all down to friendship.

It was around this time that we had decided that Azaria's sleep pattern had improved enough to finally move her out of our bedroom. Her breathing was no longer laboured at night and more often than not she slept right through – finally!

She was going to share a room with her big sister and we had planned to move Azaria out of her cot into the bottom bunk with Kezzie, now aged 6 in the top. However, whilst at the Hotel at Euro Disney, Kezzie had not wanted to sleep in the top bunk. Ian had jokingly told me as I was looking at the bed options, to have a look at the Estate Agent's web site. Well that was it, the long slow process of moving house had begun!

By now both Kezzie and Isaac (6 & 4) were at the local school. They were both settled and very happy. The area surrounding the school is all 3 bedroom semi detached houses, just like ours. The challenge was to find a 4 bedroom house that was close enough for Azaria to be able eventually to walk to school too. After about 3 or 4 months we accepted an offer on ours which meant that we had to find somewhere now to make the chain complete. We had seen a

property that was a nice size in a nice location. The garden was too steep for Azaria to manage and would have to be levelled for her to play safely and it was about 20 minutes for the children to walk to school – probably half an hour or more for Azaria. Despite my reservations we put in an offer on it and the process was underway. As the weeks went on we overcame all the problems that always crop up whilst trying to move, and on week 12 when everyone had been discussing dates for moving everything came to a halt. Our buyer had pulled out, leaving the chain broken beyond repair as it was another 2 to 3 months before another offer came along. The whole family was disappointed, especially the girls, their friends (also sisters) lived two doors away, and they were all looking forward to being neighbours! I was relieved, I was sure that we could find something closer to school. I do feel walking is good exercise and I love being able to walk to school and activities with the children.

Eventually we found my dream home, with Ian's dream garden! It was in a cul-de-sac, within easy walking distance from school (10 minutes on a bad day for Azaria), in a very quiet road and a big back garden. It was a great place for Azaria to practise on her trike, she had it for her 2<sup>nd</sup> birthday and had been unable to pedal for herself, but this was ideal for her to be able to build up the strength in her thighs to propel herself. We started 2006 by moving to our new home on Friday 13<sup>th</sup> – I've never been superstitious, and there were no hiccups.

Azaria was attending Pre-school and was quite happy mixing with the other children, although her speech was delayed, the staff always made every effort to understand what she said and would ask me at the end of the session if there was something they couldn't. I had fairly good understanding of her language, and would always repeat what she said, to clarify that I had understood and also aid others in the conversation so that they too knew what she was saying. Her mouth had always been a problem, firstly leaking, both milk and saliva, chocking on food and she had never been a babbling baby. Her nasal tone didn't help the poor control she had over the muscles which enable speech. We had seen a Food and Drink Speech therapist to help sort out the eating difficulties and we had been given exercises to help enable Azaria to strengthen her muscles. The hanging upside down to move obstructions was becoming less frequent, much to my relief, watching her turn blue and praying for whatever had got lodged to land on the floor was very upsetting for all the family at mealtimes. Her speech wasn't really making much progress and we were referred to the local clinic. After a half an hour appointment, at which Azaria's speech was assessed, I was told we would be seen again in 3 months. That time came and went, and after about 6 months when I tried to chase up the appointment, I was told that they were undergoing a re-structure and they would be in contact in about 9 – 12 months time.

I contacted our Paediatrician who managed to get us back to see the original therapist who had been helping with her eating. She did an assessment and arranged to see her for weekly appointments for the next six weeks. She was quite concerned by her poor speech and suggested that we try to get a place for her in the ICAN nursery, where they specialise in speech and language development. We applied for a place in November 2006, but as only 3 or 4 spaces were available and 12 children had been referred for them, we were not allocated a place. I was sorry that she didn't get a place, but pleased in some ways, as the nursery was on the other side of town and I would not have been able to take all three children to school and collect them myself, and I was relieved that I wasn't forced to decide between them!

During 2006 treatment wasn't our only reason for hospital visits. Isaac had been playing with his sisters on the small trampoline which Azaria had for Christmas, and he managed to break his arm – to think when we bought it we were worried what damage Azaria would do to herself on it! Anyway within 2 weeks of him having his plaster taken off, I was back at Accident and Emergency again. This time it was Azaria's turn. Isaac had been crawling on all fours with Azaria on his back and she had slipped off and managed to fracture her collar bone! It was refreshing to be visiting hospital with something 'normal'!

The children were enjoying being in their new home and making the most of the safe outdoor playing. Azaria wanted to be like her big sister and for her 4<sup>th</sup> birthday had a pair of roller skates – complete with safety helmet knee and elbow pads! Kezzie the dutiful big sister holds her hands as they shuffle down the road and back, and who knows maybe one day they'll be having skating races! After much debate we decided to buy a play house on stilts for the new

back garden. We were concerned about Azaria's ability to climb safely up and down the ladder, but Kezzie and Isaac were always there with her to help, but they don't need to anymore!

In July 2006 with Myozyme now licensed, we managed to transfer to Derriford Hospital in Plymouth for treatment. This was wonderful for family life, as I was now able to take Kezzie and Isaac to school and pick them up, whilst still being there for the treatment with Azaria. It also meant that post infusion observation time was cut to half an hour which shorted our day too.

Isaac had out grown his bike, and so we bought him a new one for his birthday, so we put the stabilizers back on his old one and it was passed on to Azaria. It was a few weeks before she managed to get anywhere on the road, but gradually managed to start cycling for herself. Now she's fairly good on the flat, but still can't quite manage the hills, but hopefully will soon master the art of braking and then we'll be able to go out together without me having to run to stop her landing in a heap and the bottom of the hill!

At the end of her first year at Pre-school, there was a sports day, which I went to watch along with three very proud grandparents. Unfortunately that was all Azaria went along to do as well! The sense of occasion obviously too much for our little girl, no matter how much we tried there was no persuading her to join in.

Azaria started back at swimming lessons in the September, this time with her best friend Jenny. This was much more successful, and she quickly gained confidence in the water and under water too. Within 2 terms she had her 5 metre badge, and then by the end of the summer term had her 10 metres. She now has her whole swimming lesson without a back float, and really enjoys splashing about!

Isaac had started wearing glasses earlier in the year, and following a conversation with the optician, I realised that muscles are also involved with sight and I hadn't ever thought about any possibilities of Azaria's vision being affected. Although she is long sighted (like Isaac) there is no evidence that her muscle weakness is causing any vision problems, which was good news. She looks very cute in her little pink glasses and has adjusted to wearing them very well, but does need reminding most of the time to put them on!

We ended 2006 very emotionally with Azaria playing Mary in the Christmas Pre-School nativity, Kezzie and Isaac were allowed the morning off school to come and watch along with the rest of the family. She looked very angelic – even when she dropped Baby Jesus on his head – good job it was just a doll!

In January we encountered our first infusion complication – Azaria's port could not be accessed. After conversations with our medical 'team', we decided that Manchester was the best place to sort this out. When we arrived, they decided to try themselves to access, which was about 8 weeks or so since her last infusion into it. She had been cannulated for all the treatments meanwhile, which was much more difficult, with scarring making access to the veins a problem. Much to everyone's surprise, although very stiff initially, they were able to flush her port. It was decided to do a lineogram to check under the X-Ray for any problems, which again was stiff, but that was only to be expected having not been used for so long. Azaria had another flush to try to clean out the line, which appeared to work.

Back in Plymouth for her next infusion, everything went well, however this was short lived, by May we were waiting for a replacement port. We had our 6 monthly check up in Manchester and whilst we were there they tried to get the port replaced, but emergencies pushed us back on the operations list and after two days just waiting, we returned home unsuccessful. It was only a few more weeks before we got a confirmed slot (not an emergency, which they had tried to fit us in as before). Azaria was the first patient on the afternoon's list and by 6 pm we were heading back to the hotel, with Kezzie and Isaac too, as it was the school holidays. We stayed overnight and then travelled back home the following morning. Her port was used the following day in Plymouth and has been fine ever since (touch wood!).

Just prior to Azaria's port being replaced, the children had pooled all their money and bought themselves a 12 ft trampoline. We decided it would be safer for Azaria with fresh wounds from the operation, if we left it all in a box for a week or two! They were all very excited, and when it was finally put up in the garden with the full enclosure to keep them from falling off, and the ladder to enable Azaria to climb up to it, the weather seemed to know and suddenly every single day seemed to rain! They didn't care though and I'm sure that Azaria has built up a lot of muscle by all the bouncing.

Azaria's old buggy, which has covered some miles in it's time, has now been retired finally. I think it's been out once or twice all year. That isn't to say she has some days where she doesn't really want to walk far, but most of the time she can keep going, with the odd piggy back here and there. In fact we went on holiday to Austria this year, and didn't even think about packing it. We didn't exactly 'climb every mountain' but we got around quite well, using the cable cars to cheat our way to the top! It was a fantastic experience for us all, with Ian opting to drive the 900 miles each way, we saw the changing scenery down through Europe and the children gained the experience of another language, even Azaria had a go at a few German words!

Her speech is still making good progress, and she is now able to say many letters that previously she couldn't, although her tone is still very nasal. Her speech therapy assistant was visiting her each week at Pre-school, and with the help and encouragement, she managed to get her 'b and d' sounds and now her 'c' too. When Azaria finished at Pre-school, she was signed off by that Speech therapist and will have to be referred again by the school – all a bit mad and just a paperwork exercise!



*Azaria showing her school uniform just before she goes to school*

It was a very sad day for me when Azaria left Pre-school. Both Kezzie and Isaac had been with them when Azaria was born, and just Isaac when she was diagnosed. They were delighted to get the opportunity to have Azaria, having watched and prayed through those tough times. They had been so supportive over the years I do miss them, and I know that they miss Azaria too, but in such a positive way! Our last sports day was quite different from our first. Azaria took part in all the races, mostly coming last, glad to have the staff there to help in the sack race and the three legged to keep her upright! It was a wonderful day!

Azaria is now at our local main stream primary school, leaving me finally time to get around to this update and catch up with all the years of household neglect! I do miss having her around all day, but I am so proud of all her achievements, and her strength of character. She has settled in remarkably well, and takes part in all school activities. She misses one day a fortnight to have her treatment, but does attend the hospital school on those days, where she is a real hit with all the staff. She is learning to read quite quickly and loves to write and draw.



*Azaria practising for her ballet lesson*

Since starting ballet and tap dancing lessons, a couple of months ago, Azaria has learnt to skip and hop, and she's having great fun developing in this way. In fact she is enjoying life and all the opportunities that it is giving her, and we are all enjoying watching the fantastic progress she is making – thanks to Myozyme.

Jo,  
mother of Azaria, Isaac and Kezzie