

# Azaria's Story

*October 2011*

Azaria (date of birth 03.12.2002) coped very well during her first year at primary school, spending one day each fortnight at the hospital school whilst having treatment. During a routine visit to Manchester Hospital, we were asked how we felt about home treatment. We would have our own nurse come to the house, make up the drug and administer it all in the comfort of our own home. I couldn't believe how lucky we were, what an opportunity. We would be given a special medication fridge and all the supplies required would be delivered each month. By the end of Azaria's first school summer holidays, we had said farewell to the team of nurses who had looked after us at Derriford Hospital, and met our new home care nurse.



Azaria playing and going over the net

The new routine meant that Azaria would be able to go to school in the morning, her nurse would arrive and make up the medication, in time for her to come home at lunch break and have her treatment in the afternoon. This took a lot of pressure off me too as Kezzie and Isaac were by then in years 5 and 4 respectively, and as school was only 5 minutes up the road, they were old enough to walk home on their own safely.

It was about this time when Azaria first met the cleft palate team from Bristol Frenchay Hospital. After some investigations, it was apparent that Azaria had no movement in her soft palate. The consultant was amazed that she had developed her own way to compensate for this by squeezing her throat muscles to try to replicate the sounds she was unable to make normally.

At first the consultant didn't think they could do anything to help, but after a couple of days, I had a phone call from him to say that he had had an idea of something that may help. It involved an operation to create a flap that would help block the nasal escape. After consulting with our team in Manchester, we followed our initial gut feeling, and decided against the operation. However, from those discussions, came about the idea of fitting a brace to Azaria's teeth, with a metal loop attached to the back to try to stimulate the palate into moving. So in May of 2009 the brace was fitted, initially with a small loop, to allow her to adjust, as starting with a large loop would provoke the gag reflex. As the loop was extended until it reached the point of maximum effect, Azaria had regular visits at school from speech and language therapist, and she worked really hard with us at home.

A recent visit to the cleft team has shown that with the aid of the loop on the brace, there is now movement in the palate, however it is not sealing the nasal passage off completely. The plan is now to make the loop bigger and lift the palate, as this will make the seal. The progress has been good and the brace has definitely improved the clarity of Azaria's speech, even if it is now a permanent feature.

Azaria is now doing private speech and drama lessons once a week, which she really enjoys. She has so far taken 3 LAMDA speech exams, passing two with distinction and one merit. Despite her

speech problems, she has been very confident reciting her poetry and discussing books with her examiner.

This is not the only success we have enjoyed with the 'theatre'. Azaria had been having ballet and tap lessons for quite a while, but was increasingly not so keen on tap, so we dropped that and let her concentrate on ballet. She passes her preliminary exam with a merit – being commended for her points, which I believe is the flip side of lack of muscle strength, pointing is easier to her than most of the other children! In February of this year the dance group put on a show, which they do every other year. This time it was their version of Alice in Wonderland, being staged at the local village hall. Azaria enjoyed going back to tap as well for a couple of months so she could take part in more of the dances. With Big sister Kezzie taking part too – with her first speaking role, we were a very proud family all going along to watch. Their dance instructor asked if there was a charity to support sufferers like Azaria, as it was the first time she had been taking part. They raised £800 from the show for the AGSD, and many mums spoke to me afterwards to find out more about Pompe Disease. Azaria has decided that she wanted to stop ballet for a while, she found the exercises where they lie on the floor and lift their head in modern ballet difficult. Instead she is doing an after school club for gym and dancing, starting after Christmas. I want her to enjoy the clubs that she does and not just keep on with them to try to build strength, so maybe swapping around will keep her interest and strength.



Azaria plays the clarinet

Azaria is getting on very well at school, and I am very glad to say that missing time for treatment and hospital appointments doesn't at present seem to be having an impact academically. She really enjoys reading, and craft and always tries her best. She is now in year 4, and along with all her classmates, she is learning to play the clarinet. The school ordered a special instrument for her, which is easier to blow, which I had been concerned would be a problem. So far she has only had one lesson, so it's too soon to say how that is going, but she can make a noise from it.

Azaria has progressed from Rainbows to Brownie Guides, which is better as they meet on a treatment day, so she was only able to go fortnightly, but as Brownies meet a later she can go after treatment.

Support from family and friends has always been really important to us, and we have been incredibly lucky. They have helped us tremendously through some very difficult times, and also been there at times when we've had excitement to share. Eighteen months ago my mum lost her fight against cancer. She had given so much in the way of support to me and the family, I really don't know how I would have managed without her. I still miss sharing with her all the little triumphs and tribulations, but I know she would be incredibly proud of her youngest granddaughter and all that she is achieving.

One strength in which Azaria isn't lacking is character. She is very strong willed for someone so little! Sometimes I need to remind myself that it is a good thing, I am sure that has seen her make such good progress, but at times as a mum it is very trying and tiring!



Azaria riding her bike without stabilisers

This summer we had a garden make-over, and now have a 'grown up' play area for the children. The trampoline is still popular and we have a scramble net and some log stepping stones. I was a little nervous initially with Azaria going over the net, but she clammers over it, maybe not with such confidence as her brother (who I'm sure is a very close relation to the apes), but having fun none the less. She is also riding her bike without stabilisers, although she does struggle with up hill. Our road is a cul-de-sac and reasonably level, so she can ride along quite happily, but unfortunately it is off a steep hill, which means whilst Azaria likes to ride to local places, it is hard going!

Life is busy, and fun. We try to take everything in our stride and make the most of what we have and can do. Azaria enjoys most of the things that other children of her age do, and maybe she can't do them quite as well as the others she always gives it her best shot.

***Written by: Jo Moyse (Mother of Azaria), United Kingdom***