

MEN2 and Me: Asher

Multiple endocrine neoplasia type 2B (MEN2B) is a genetic disease in which more than one endocrine gland forms a tumour that can make too much hormone.

"We're very lucky that Asher was diagnosed at only 7 weeks old. Many people with MEN2B don't get diagnosed until later on in their childhood by which point the Medullary Thyroid Cancer has spread which obviously makes the prognosis that much more difficult"

Tara, Asher's mum

Asher has MEN2B which is a very rare variant of Multiple Endocrine Neoplasia. His condition causes Medullary Thyroid Cancer, it also causes growths throughout the gut, it causes tumours, adrenal tumours later on in life as well.

With Asher it presents in a lot of vomiting and also constipation because his bowels don't work properly.

The tumours that you tend to get with MEN2B, particularly the Medullary Thyroid Cancer is very aggressive so usually you start to get cancerous growths within the thyroid within the first year of life.

Asher also has a second rare condition, a chromosomal condition called Cat Eye Syndrome. He is very likely to be the only person in the world with both MEN2B and Cat Eye Syndrome as they are both so rare.

Paul, Asher's Dad

Asher had surgery when he was just five months old to have his thyroid removed. When the thyroid was removed they found Asher already had Medullary Thyroid Cancer growing within it so having this really early diagnosis is so important.

I think the things that the medical professionals to look out for obviously the issues we have with feeding, the issues with the gut and problems around that

One other thing though that's a really good marker is the fact that he doesn't produce tears so if a pediatrician notices that you've got a child that doesn't produce tears and is having problems with the digestive system that is a really good indication that as rare as it could be it could be MEN2B.

"We do live with a great deal of uncertainty around Asher's future."

Tara, Asher's mum

Since Asher's had his diagnosis life's been a real rollercoaster really. Asher is now NG tube fed all the time which means he has a tube which goes down his nose into his stomach. That will be replaced very soon by a permanent gastrostomy which is a feeding tube into the stomach.

Great Ormond Street is our second home. We live there. We see so many different specialists and consultants, all of whom have been amazingly helpful, but life has been very difficult.

Asher's conditions are so all encompassing that we find ourselves being carers more than parents at times, but you know he does all the things that little boys do, you know he crawls around as best he can and you know he does interact with his two sisters but it is a very

different ball game particularly because he has Cat eye syndrome as well, no-one is too sure how the two conditions can interact with one another so we're all on a big learning journey including the Hospitals as well.

When Asher was diagnosed, we obviously, like the majority of people had no idea what MEN2B was so I took to the websites and tried to find information out there, but it was all very medicalized and then I happened to stumble across AMEND's website. That was the first thing that I read which was able to give me some concrete answers.

To have the resource of AMEND there is so valuable on so many levels and it set me off on my own personal journey as I have now become a regional volunteer for AMEND for people with MEN syndromes and their families.

It's important to raise awareness about MEN2B because it is so rare and there is so little information out there on it, you can save lives by raising awareness.

AMEND provides support to patients and their families and friends. Providing information and promoting a wider knowledge of MEN among the medical profession to assist in early and accurate diagnosis and assisting in future MEN research.

Amend.org.uk

