

MEN2B AND ME: ETHAN (1)

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.

"I hope this film can help somebody in the future get an early diagnosis, so that they won't go through what we've gone through" Ethan's mum, Jane

Ethan's Mum, Jane:

Ethan has MEN2b, that's Multiple Endocrine Neoplasia type 2B. Basically he has full lips and lumps on his tongue and they're nodules or neuromas, tumours that are not cancerous, and unfortunately this disease also comes with a form of Medullary Thyroid Cancer in early infancy, and as Ethan was diagnosed at twelve-and-a-half, obviously his cancer had twelve-and-a-half years to grow.

We'd been waiting so long to get a unifying diagnosis to have a name for what Ethan's problems were. We honestly thought, seeing how many different things Ethan had wrong with him that we by now, knew all the individual parts of him that were wrong, and it would just be a name, a syndrome name. It was a real shock to find out that Ethan had MEN2B, and to find out that he had a disease that meant he'd had cancer since he was a baby was absolutely devastating.

Right from when Ethan was born we realised that something wasn't right. The first signs were immediately at birth he was projectile vomiting really badly and he was diagnosed with severe reflux. Originally, he was terribly constipated. We were told he was lactose intolerant, fructose intolerant. He stopped breathing many times. Sometimes it could be as many times as four times an hour. It became clear as he got older that he had other problems too. His eyesight wasn't great and we were told he had a tethered spinal cord and he had to have surgery on his spinal cord to de-tether it. Ethan also had bumps on his eyelid and lumps in the neck. We were told for a long time that the bumps we could see and feel in Ethan's neck were things like 'waxy nodes' or 'nothing to worry about', but they weren't. They were thyroid cancer, Medullary Thyroid Cancer, spreading into his lymph nodes.

A year ago, nearly, Ethan had major surgery so that has removed an awful lot of the cancer. It's called a thyroidectomy and Ethan is monitored roughly every three months with blood tests, particularly looking at calcitonin and CEA which are markers that are relevant to medullary thyroid cancer and looking to see how the numbers go up and unfortunately for Ethan his numbers are going up. There was a time we were hopeful the numbers were going to go down after the surgery and they did for the first blood test, but since then, sadly, they've been going up which is not a good sign. It means that the cancer is progressing. Ethan has got multiple metastases in his lungs and therefore his cancer is incurable.

"If we had had an early diagnosis life would be very different." Ethan's Dad, Carl

Early diagnosis in MEN2B is crucial. If Ethan had been diagnosed before he was one, he would have been able to have a prophylactic thyroidectomy. Basically it means they'd have taken his thyroid out before the cancer had a chance to grow and spread, so for Ethan, an early diagnosis would have meant a cure from cancer. Being told at twelve years old that you have an incurable cancer is very

difficult. In himself, he's bright, resilient, amazing, but he has been through a really tough time and I'd be lying to say that it wasn't difficult; it really has been quite a challenge.

Ethan's Dad, Carl:

Inside we're absolutely broken but you can't keep living your life like that. It's not fair for us, it's not fair for Ethan, and it's not fair for the rest of the family, so we just want to grab each day now and just go for it.

Ethan:

I don't worry about the future. I just like live life as it comes, so yeah, I just, I'm excited for what's going to happen. I'm not scared about it or worried. I'm excited to see what happens in the future, for me and my family.

End



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.

We are here to break the isolation so often felt by those with these rare diseases.

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