

TRANSCRIPT: Emma's diagnosis

Ruth, Emma's mother

Emma is now 18 years old and she was born with congenital Angelman Syndrome.

We had no idea Emma was going to be disabled in any way. She had a developmental delay, which became much more obvious the older she got and we didn't know what was wrong with her and she was tested for all the usual things, like dyspraxia, Fragile X Syndrome, things like that and it all came back negative.

She wasn't diagnosed with Angelman Syndrome until she was three, mainly by the absence of fits. We were told that she would have been diagnosed round about 18 months had she had epilepsy and it wasn't till she started walking in the typical Angelman's fashion that the consultant neurologist said, 'Oh, I know what she's got!'

When we got Emma's diagnosis, I suppose, we were relieved that we knew what it was. We were able to then go on the internet and look up what we were dealing with because I did want to know how the future would pan out.

After we'd had the diagnosis we did go to an Angelman Syndrome conference and that changed my perception of Emma quite a lot because before then, she was just a very cute, blonde haired, gorgeous little girl, with a severe disability and I came home from that weekend feeling I was the mother of a disabled child and one that could be labelled, and that was quite upsetting.

I did have some counselling and that helped a lot, just to get things off your chest about how you feel. I didn't know where to start with counselling and the counsellor had said to me, 'Do you feel Emma has died, and you've got a different child in her place?' and I didn't - and I sort of said, 'Yes' but I didn't - I said, 'Yes' but my husband was horrified. He said, 'Emma's upstairs in her bed, how could you possibly think...' and it was like - the only way I can describe it is - I'm not a religious person - but it's almost like going to God when they're about eighteen months old. It's, 'Oh yes, little Tommy you're going to be a fireman. Oh John, you're going to be a lawyer. Emma, oh we've got nothing written down for you, you're not going to be anything' and you begin to feel as a parent, 'Well I'm gonna have to put something on this bit of paper, you know. She's a person and she will do something and it's up to me to equip her as well as possible to live a productive life.'