A 10-year-old girl from Northampton is helping scientists in America with world-leading research into a rare genetic condition. Hermione Heseltine, from Abington, was flown out to Washington DC with her family on August 1 to help researchers shed light on a disorder with only 35 known cases in the world.
The condition, known as PPP2R5D, was only discovered in 2015, and is so rare that researchers have almost no data on what causes it or if it can be treated.

Now, Hermione, who was the fourth ever person to be diagnosed with the disorder, could help scientists discover new ways to repair the affected gene for the future.

Hermione's mum Jane, 50, from Abington, said: "Because it's so rare, we want to get it into the public eye and help other families who might not realise their child has the condition get a diagnosis."

Growing up, Hermione could not walk until she was five years old. She cannot speak or read or write, and she suffers from muscle tiredness.
Researchers in America one step closer to cracking rare genetic condition with help of little girl from Northampton - Northampton Chronicle and Echo

Hermione with her dad Robin on their trip to Washington DC.

But despite extensive testing, doctors could not find what was causing Hermione's disability.

Jane said: "Back in 2011, Hermione had some tests taken at John Radcliffe Hospital, in Oxford, on the off-chance it could turn up something.

"Four years later, we got a letter saying they had found something. Hermione had a condition called PPP2R5D. But it was so newly discovered, they couldn't tell us anything about what it was. We couldn't find anything about it on the internet either."

Since then, Jane has connected with other families across the world through Facebook whose children have also been diagnosed with the disorder.
Researchers in America one step closer to cracking rare genetic cond...lp of little girl from Northampton - Northampton Chronicle and Echo

Jane said: "Hermione was able to meet other children like her out in America. It was jaw-dropping. They all had the same mannerisms and made the same sounds. There was another little girl with the same laugh as her. I got to compare notes with the other families too. It was amazing."

Hermione and her family's genes will be used in stem cell research to find a way to repair or treat the affected gene that causes PPP2R5D.

Jane said Hermione, who enjoys horse riding, swimming and trains, was a "superstar" on her trip to America. They flew back to England on August 6.