Ruth writes about her son Jude

Parent story

Up to November 2023 my son Jude was independent in many areas.

He could use the toilet, have a shower, put his coat on, get changed for PE, go to bed, eat independently, and move around with ease. He was interactive and would regularly smile, intonate sounds and words despite having verbal dyspraxia. He enjoyed being with family and friends, going swimming and playing basketball. He liked to play with his cars, listen to music and be on his iPad.

What happened



In November 2023 Jude became withdrawn, he lost interest and joy. He refused food and drink and became so dehydrated he was hospitalised. He was sad and would cry and not engage in any activity at school at home. He became stiff and catatonic. He would stare into space in the same spots for hours at a time. If we tried to move him, he would freeze, and it would be a real physical task to move him. He went from becoming fatigued and passive in the early weeks to then screaming and becoming very aggressive and angry.

He then lost toileting functions and would soil and wet himself frequently. He was not able to feed himself and would have to be fed, he lost 9kg in 4 weeks. His toes turned purple, and it took 6 months to get a diagnosis of chilblains.

Jude stopped talking and he didn't utter any sounds. He would not swallow and would drool saliva out of his mouth. His body would tremble. He began laughing for no reason and then a few minutes later thrash around, this is still happening (Summer 2024).

He didn't use his iPad for about five months, he just sat and stared or screamed at us. After a few months he started to repeat scenes of his favourite shows on a loop over and repeatedly watching the same scene again and again to the exact second.

He began collecting objects and hiding them in his bag such as hairbrushes and toilet rolls. He would take photos of his classmates off the wall and bring them home every day.

He would not look at us in the eyes. He would continuously look at his hands and touch the side of his head.

He refused to sleep alone or in his own bed and even now only sleeps for two to four hours a night. The insomnia to this day is extreme.

Very quickly Jude was disappearing emotionally, mentally and physically before our eyes.

Investigations and treatment

At first, I thought Jude had a bug and after nearly a week of being sent home from school I took him to the GP who performed several checks including looking at his ears, throat, temperature, SATS and heart and chest.



She could not find anything wrong with him to explain his symptoms. I informed her that he was refusing food and drink and was becoming dehydrated (evident by the cracks in his lips and his grey appearance).

After a week there was no improvement, and the GP sent him for a precautionary x-ray. It showed that he had a chest infection although there was no cough, cold or other symptoms.

After seven days of antibiotics, I went back to the GP as he was getting worse, and he went for a second chest x-ray even though it was under the three-week mark. At his second x-ray the radiographer remembered Jude, at this point he had lost about 5 kg in weight, and he was very dehydrated. She suggested I take him straight to A&E where he was admitted for dehydration. His ketones were 5.8 and blood sugar 3.4. He was also drooling and refusing to swallow even his own saliva.

This was on the 12 December 2023, nearly a month since he first appeared unwell/different. After a few days in hospital on a drip he was discharged with no other treatment given aside from IV fluids.

Christmas came and we all thought he would get better, but things got dramatically worse. Jude couldn't manage going back to school in January, we staggered his school weeks, two days, three days, half days tried everything, but he was not 'present' mentally or physically. He presented with a new symptom of purple toes, I took him to a GP and eventually he went for blood tests to check for pernicious anaemia however the results were normal. I felt defeated at this point. It was about six to eight weeks of Jude being ill now.



His consultant Paediatrician made contact after I left her a message about his ketones still being elevated. She wanted Jude checked for Type 1 Diabetes which he did not have.

In January 2024, he had an appointment with another consultant paediatrician at our local hospital. His school had written a report, and I had written my observations and findings. This appointment proved to be unhelpful and the Paediatrician didn't look at any of the information.

At the end of February his paediatrician at Queen Elizabeth hospital saw the report from Jude's school and he was admitted for further investigations to rule out clinical possibilities i.e. a brain tumour.

He had a cannula put in and was sent to a ward for observations and had bloods taken. Nothing abnormal came back. It was the decided that a CT scan would be booked for a weeks' time with more specific genetic bloods taken and a test for Addison's disease as at this point this was being suspected.

At the beginning of May Jude had the synacthen test and further tests. He then had a cannula administered and was taken for a CT scan. The Synacthen test was negative, and the CT scan showed Jude had a build-up of calcification in the basal ganglia area of his brain.

We then had an appointment with a neurologist from Addenbrookes Hospital in Cambridge who diagnosed Jude with Down's Syndrome Regression Disorder. He told us the rarity of the condition and Jude was his first case in over 25 years.

Has Jude been offered any medication or treatment?

He was given melatonin for sleep; it didn't work and was difficult to administer.

He has been prescribed Phenergan which helps him drift off to sleep but doesn't necessarily keep him asleep. It took at least 7 days to kick in too.

He has been referred to The Maudsley Hospital in South London and we are waiting for an appointment.

Was there anything that happened before November in his life?

Prior to the changes Jude was not sleeping at all. I had an annual review with his paediatrician in 23 October and I described him as delirious through lack of sleep, he would go without sleep at night and then struggle to stay awake during the day.

Then shortly after symptoms he showed signs of the onset of puberty.

Where are we now?

Currently Jude is still not the Jude he once was. He isn't in the acute phase but remains in a chronic phase with some functioning being normal again such as walking up the stairs, toileting. He is still having moments of delirium and sleeps for only 2-4 hours per night. He also becomes fixated on objects and hides them in his bag or luggage such as hairbrushes or toilet rolls. He constantly repeats what he is watching on tv like parts of his favourite programmes and rewinds repeatedly the same scene to the exact moment.

Although things aren’t back to his normal we are in a better place.



CONTACT US

Down's Syndrome Association

Langdon Down Centre,
2a Langdon Park, Teddington,
Middlesex, TW11 9PS

t. 0333 1212 3007

e. info@downs-syndrome.org.uk
e. training@downs-syndrome.org.uk
w.downs-syndrome.org.uk

Wales
e. wales@downs-syndrome.org.uk

Northern Ireland
e. enquiriesni@downs-syndrome.org.uk



© Down's Syndrome Association 2024