DOWN'S SYNDROME ASSOCIATION

Susannah writes about her daughter Catriona

Parent story

Catriona is 25 and has a dual diagnosis of DS and ASD resulting in severe and complex learning difficulties. She has always displayed mild repetitive movements (stimming) associated with autism and chosen to distance herself from others in social situations, only joining in on her terms.

She attends daycare provision four days per week and whilst choosing to sit separately from her peers, would get up and join in with dancing, music sessions and other activities.

She enjoyed an active social life with frequent theatre, live music and cinema visits. She would sit calmly at venues and loved to chat about the show or film afterwards. She loved swimming and, despite her chronological age, still enjoyed an adventure playground.

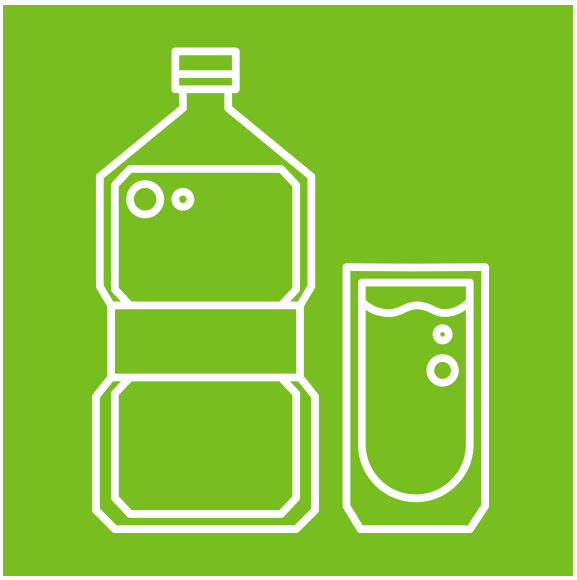
She spent a lot of time on her iPad at home and watched a huge variety of music videos that she loved to sing along to. She also loved her extensive collection of family films on DVD.



Catriona is essentially non-verbal with a limited number of clear single words and a few two word sentences. She communicates with speech, signs and gestures, and a communication book containing pictures and symbols. She has always been keen to communicate with those close to her. She has always been affectionate on her own terms and had a wicked sense of humour. She loved to entertain us with singing and dressing up and found bodily functions endlessly amusing; she laughed readily and often.

Catriona has always preferred the company of non-learning-disabled adults as they would allow interactions to be led by her; she has never sought friendships with her contemporaries. However, family members and close family friends have always been important to her, and she would ask frequently when certain individuals would be visiting. Her adult sister provides respite care and Catriona was always keen to know when her next visit was due.

What happened



Since the night of 27 September Catriona has displayed extreme bodily and facial movements, accompanied by loud, distressed, guttural vocalisations. The intensity of these episodes has resulted in a weight loss of over a stone – she is 4'9" and currently 6st 9lb.

She displays an almost constant need to move – wriggling, hand clenching, gurning, bouncing, rocking. Conversely, on occasions, she will appear to freeze and appear detached from her surroundings; she will lie back and stare at the ceiling. Episodes of manic giggling for no apparent reason also occur on occasions.

Eating and drinking can be challenging. Catriona no longer requests a drink; we make sure she has a plentiful supply of liquids as her almost constant movement causes her to become dehydrated very quickly. It is not unusual for her to take two hours plus to eat a meal, she seems to lose concentration and forget to eat.

Catriona always took herself to the toilet as necessary and would request help with bottom wiping. Since regression, retention of urine has become a problem. She will go up to 8 hours without going to the toilet, despite frequent encouragement and will not seek support with wiping.



She has lost almost all interest in the well-being of family and friends and no longer asks after them or greets them when they visit. She no longer leaves her room to seek out interaction with us.

Catriona displays extreme anxiety and often indicates that she is scared, particularly when she needs to leave the house. She is reluctant to leave her room and will not go to daycare without her iPad, charging plug. She refuses to remove her headphones throughout the day and will only join in with limited activities and then only from her separate table.

It is no longer possible to visit the cinema or theatre. We have tried and had to leave venues as Catriona's huge movements, arm waving and jumping up and down, accompanied by extreme, throaty noises, made it impossible for us to remain.

Her interests have narrowed significantly. She now watches a very limited number of music videos and is particularly obsessed with just one. She has a film on the TV simultaneously and keeps turning the volume up even though she has her headphones connected to her iPad.

The frequency and intensity of some of the Catriona's behaviours have reduced recently (in percentage terms by about 20%) since Catriona was prescribed 1mg Lorazepam TDS a month ago.

Was there anything that happened before September in Catriona's life?

The only change we can think of is that Catriona moved to a new daycare provider in March 2023. However, she seemed to transition well and the facilities and professionalism at the new provider are far better than her old placement.

Investigations and treatments

October 2023

GP sent an urgent referral to Neurology at Maidstone Hospital. This was downgraded by the hospital and given a 10 month wait time.

I contacted the DSA for advice. They referred Catriona to their doctor at the DSMIG, and they, very kindly, provided a report. The DSA doctor did not give a diagnosis but suggested that Regression in Down's Syndrome should be considered.



November 2023

Catriona passed out in the bathroom after getting out of bed. She was admitted to A&E. After an ECG and blood tests, dehydration was given as the cause due to the intensity and frequency of her involuntary movements.

GP prescribed Promethazine Hydrochloride 10mg after I begged for help – this had no effect on Catriona's symptoms.

December 2023

I emailed Dr Santoro's team in Los Angeles and asked for their advice, I included videos of Catriona's movement behaviours. Dr Santoro replied personally and said that he felt her symptoms were compatible with a diagnosis of DSRD. He offered to consult on Zoom with UK specialists; this offer has not been taken up to date.

January 2024

After I met with the GP Practice Manager, a neurology appointment was received for the 22 January with Dr Sengupta at Maidstone Hospital.

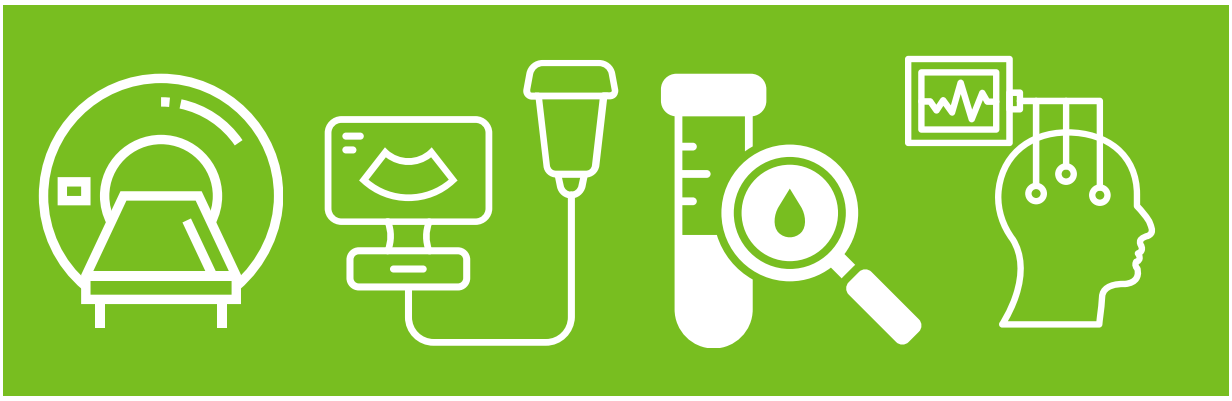
Dr Sengupta is familiar with DSRD and would not rule it out. She mentioned autoimmune Encephalitis and Limbic Encephalitis as possible causes and ordered blood tests, an EEG, a pelvic ultrasound and a CT scan. Lamotrigine was prescribed to, hopefully, alleviate Catriona's anxiety.

March to April 2024

Catriona has series of tests.

- CT scan with mild sedation (diazepam)
- Pelvic ultrasound
- Extensive specialised blood tests
- EEG at King's College Hospital, London

No abnormalities found in any of the investigations.



May 2024

Consultation with Dr Sengupta at Maidstone Hospital. Probable diagnosis given of DSRD. Lamotrigen dose increased to 125mg per day and Clonazepam 1mg TDS added to the prescription. Dr Sengupta said she was arranging for a colleague, Dr Jonathon Chan to see Catriona as he was a more experienced neurologist. His speciality is early onset dementia, and she felt this was a diagnosis that needed to be considered.

An appointment was received with Dr Chan for the end of July.

I arranged for the Learning Disability Community nurse to complete a dementia screening, and Catriona scored well below the level at which further investigation was recommended. Where she did score, all the behaviours were compatible with the symptoms of DSRD.

July 2024

Dr Chan was emphatic that Catriona does not have Alzheimer's Disease. He was very interested in DSRD and in the treatments that had been successful in the US.

He said he would be recommending to Dr Sengupta that an admission should be arranged to Queen's in London for a lumbar puncture and MRI to be carried out, under a general anaesthetic.

He also agreed that a change of medication to Lorazepam, as used by Dr Santoro, could be beneficial as Catriona's current medication had had minimal effect on her symptoms. He felt that both immunological and psychiatric neurological input would be of benefit to Catriona.

August 2024

Copy received of Dr Sengupta's letter to Dr Zandi, Consultant Neurologist at the National Hospital for Neurology and Neurosurgery, in London. She requests in-patient admission for review by UCL autoimmune neurology team. Dr Sengupta gives details of the work of Dr Santoro and references IVIG as a possible treatment.

September 2024

With the agreement of Dr Sengupta, Lamotrigen has been phased out and Lorazepam has replaced Clonazepam. She is currently taking 1mg Lorazepam TDS and we have observed a definite reduction in the severity of her symptoms and a return of some of her baseline personality and behaviours.



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