Questions from the Regression Webinar, 28th of July 2026

Disclaimer,

The questions and answers below are taken directly from the Q&A section of the Regression Webinar held on 28 July 2026.
The questions were copied directly from the webinar Q&A and have not been edited, amended, or altered by Down Syndrome Ireland. The responses were provided directly by Jonathan Santoro and Colm Doherty, who kindly answered the remaining questions in writing following the webinar.
The information provided reflects the speakers' expert knowledge and experience at the time of the webinar and is intended to support understanding and awareness of Down Syndrome Regression Disorder (DSRD). It is provided for general information and educational purposes only and should not be interpreted as individual medical advice, diagnosis, or treatment.
Every person with Down syndrome is unique, and symptoms, investigations, and treatment options will vary depending on individual circumstances. If you have concerns about your own health or that of a family member, you should always discuss these with your GP, consultant, or wider healthcare team, who can provide advice based on your individual needs.
Down Syndrome Ireland is committed to providing families with access to up-to-date, evidence-informed information while continuing to advocate for improved awareness, research, and clinical pathways for people with Down syndrome experiencing regression.

Q. What clinical features do you find most reliable in distinguishing true DSRD from depression, catatonia, or dementia?
A. Encephalopathy and altered mental status are often the best differentiators. That and time course, as depression, catatonia and dementia are much more slow in onset.

Q. Is aggression a symptom?
A. Yes - it is part of the behavioural dysregulation cluster

Q. We have had a number of children presenting with features consistent with DSRD in the DS clinic. Clinical workups are carried out as per guidance from Dr Santoro. They are typically normal and the wait then begins for acceptance to CAMHS ID which is very difficult for the child and the parents.
It would be extremely beneficial to have a dedicated neurology team in the health system to provide a service for these young people who have both the expertise and experience to treat effectively.
It is a topic that needs more studies and discussions between families and professionals to develop a clear pathway
A. Agreed

Q. What dose of Lorazepam should be used for an average sized adult?
A. The dosing is variable but can be as low as 1 mg per day and as high as 16-20 mg per day. Higher doses must be administered under close observations with the treating clinicians.

Q. Have you seen DSRD without catatonia, or do you think catatonia is almost always present if we look carefully?
A. Yes - about 25% of patients with DSRD will not have catatonia.

Q. Jonathan, have you treated regression in patients who have also been treated by lorazepam and/ECT and what were the outcomes.

A. Yes - many of our patients receive multiple therapies. IVIg and lorazepam is probably our most frequently utilized model.

Q. Will the clinic do the blood work up? Or does that need to be done elsewhere?

A. yes, the clinic will do all of the neuroinflammatory work up including EEG, MRI, LP and Bloods

Q. Jonathan - has any patient shown a slow down in the regression by more exercise, healthy eating - and positive reinforcement of self worth?

A. Typically at the time of the regression symptoms, patients are less able to exercise or eat healthy so this is hard to answer. That said, when patients are getting treated and starting to get back to their baseline, it is an excellent opportunity to have plenty of exercise, a balanced diet and positive reinforcement!

Q. How long do you typically continue high-dose Lorazepam in DSRD-associated catatonia before concerns about tolerance arise?

A. We do not really see tolerance as such. The hypnotic effects of Lorazepam are not generally seen unless the dose is escalated very quickly. Once you really a plateau of post effect, it tends to be sustained. That said, you should still decrease the dose slowly afterwards as withdrawal can occur. Never stop lorazepam suddenly.

Q. Are we offering IVIG treatment in Ireland? Or any alternative immunosuppressive treatment in Ireland?

A. I've heard of some families who are accessing IVIG but I think its important to have the full investigation of neuroinflammation done before starting IVIG and I'm pretty sure this full service is not be being offered.

Q. My daughter's regression involved a lot of giggling, laughing, rocking, and a significant loss of speech. She also experienced anxiety, but with the right therapies that became much more manageable. Her main ongoing difficulty is her speech. Given this history, what would be the usual therapeutic approach to help improve or regain speech? Are there specific therapies or interventions you would typically recommend? Speech therapy has not

worked over years. It feels like it is very difficult for her to put the words together.

A. This may be part of DSRD as language regression is part of the spectrum, but other causes of language loss need to be investigated and so a neurologic opinion is required.

Q. Is participation in sport a factor in delaying the possible onset of DSRD

A. I am unaware of sport participation being protective

Q. if we as therapy clinicians are finding some of these symptoms and presentations described with our service users...is there a process we follow or how or what should we do?

A. I think we need to work with DSI to come up with a comprehensive pathway from community to specialist care and I commit to working on this with other services.

Q. My 47 yr old DS sister has been diagnosed with early dementia. She has so many of the presentations of DSRD. Can this be diagnosed at her age.

A. This seems very old to be diagnosed with DSRD and early features of Dementia which can be seen in this age group can mimic those with Catatonia; I would recommend the ID dementia Clinic in Tallaght to help with these distinctions.

Q. What percentage of patients who have been diagnosed with DSRD within the 2.5 years respond to IVIG?

A. Roughly 80%. After that, the numbers decline slowly to a low of 50% after 5 years.

Q. Apologies, I have adhd and my brain started working overdrive, you may have answered this already, if I suspect symptoms in my child where do I start investigation? Worth noting he has also DCD and adhd (still waiting for CAMHS but his brothers psychiatrist already noted it when e tagged along to his brothers apt and multidisciplinary team is in agreement)

A. see above answer for creating the pathway

Q. so could a blood test determine this?

A. At the moment there is no single blood test that can make this diagnosis. There is active research occurring to identify a biomarker that is both sensitive and specific for the disease.

Q. My brother is showing signs of DSRD and currently on Sertraline for OCD & anxiety. Would it be worth speaking to his psychologist and querying about lorazepam instead?

A. They should engage with his clinical team to make sure this is being considered.

Q. in developing family advocacy and trying to navigate the very significant systemic issues in the Irish Health service do you think that gathering the lived experiences of people and families would be helpful in lobbying and seeking changes by the HSE and Government to address the needs and to putting in place a pathway and hub bit also the supports that families need.

A. Yes, We've done a similar ethnographic piece of research on lived experience with another rare disease that is under recognised Tuberous Sclerosis Complex and it has been very valuable to have this data in policy discussions. This is time consuming but rewarding work; my PhD student spent 3 years gathering data on TSC.

Q. How long does Colm think it will take the HSE to get the systems in place to allow a compressed diagnostic and multidisciplinary response that can commence timely immuno treatment for people who need it? given that there are huge waiting lists, overshadowing, knowledge gaps in adult services?

A. I think that we will get some sort of referral pathway in place within 6 months under our won steam, we need a coordinated response from the HSE to help sustain and develop the programme but this could take years

Q. being a jak inhibitor tofacitinib for arthritis can this help a child of 13 . I find the aggression has calmed in my son since .

A. JAK inhibitors like tofacitinib can have positive effects on mood and executive function (cognition). At present there is no data to support the use of these medicines to prevent the symptoms of DSRD but these are used very frequently in individuals with Down syndrome and arthropathy.

Q. I think it is more prevalent and has been misdiagnosed as early onset dementia.

A. The age is the key risk factor for dementia in DS. We should not be contemplating a diagnosis of dementia in people aged under 30 years with Down syndrome.

Q. unfortunately the HSE WILL NOT allow adults to have MRIs under GA and if so it can take over a year to get past the waiting list. this is a system failure that is excluding disabled people so it may take approaches to the high court.

A. Accessing brain imaging can be difficult but many hospitals have proper pathways in place now and this should not be a barrier

Q. My son has cutaneous mastocytosis. He's only two. But I presume that's a totally unrelated skin condition rather than having inflammatory links?

A. Skin diseases often have an inflammatory component, particularly in Down syndrome. That said, cutaneous mastocytosis is not one of them and immunotherapy is not currently advised to treat this condition.