GUEST COLUMN

What is Down Syndrome Regression Disorder?

Nia Parker, from Down Syndrome WA, urges the local medical community to be aware of this heartbreaking disorder and help find some answers.

Down syndrome is the most common cause of genetic intellectual disability, occurring in approximately 1/800-1000 live births.

The medical profession, and society in general, has come a long way in terms of attitudes to education and opportunity for those living with an intellectual disability and many adults with Down syndrome now live independently – working and integrating fully into the mainstream community.

Imagine then, how devastating it is to see this independence, confidence and skills disappear over a short period of time – sometimes in as little as 6-12 months, with no explanation as to why a 25-year-old, who has a job, catches public transport and plays basketball, has suddenly lost the ability to self care, takes an hour to eat a bowl of cereal and whispers (if they speak at all).

These are just a few of the many symptoms of Down Syndrome Regression Disorder (DSRD).

The first paper discussing this condition was published in the 1940s, but little since until five years ago when the level of interest sharply increased. The condition was described by a variety of names, but an international group of researchers collaborated to reach a consensus on nomenclature, diagnostic criteria and testing (published July 2022, *Frontiers in Neurology*).

The Down Syndrome Association of Western Australia (DSWA) has established a support group for parents who have a teen or adults showing signs of regression. Most have seen a GP, some have been prescribed anti-depression/anti-anxiety medication or referred to a psychiatrist, others to a neurologist, but most are just told that it's "probably early onset dementia"

Perth Children's Hospital has a T21 clinic, but when a child reaches 18 and too old to attend, there is no adult centre for Down syndrome providing specialist care, so each patient sees their own GP who has possibly never heard of this condition and therefore unaware of how to approach the situation.

DSRD is a cluster condition, with diagnosis by elimination. Many people with Down syndrome suffer from autoimmune disorders, so often the first point of call is to rule out any of these conditions. Unfortunately, at this time there are no definitive biomarkers for DSRD. Some will show changes on cerebral MRI, others won't, some will show anomalies in CSF taken by spinal tap, others won't.

Globally, treatments currently being offered include lorazepam (an anti-IVIg (an immune regulating therapy) and tofacitinib (a type of drug which suppresses the immune system). There are documented cases where one or more of these treatments have yielded remarkable improvements in symptoms, even after regression has been present for many years.

A multidisciplinary clinical trial of these three treatments began in July this year in the US.

Research into DSRD is hopefully going to provide some answers as to the cause of this condition, as well as consensus on diagnostic tools and first-line treatment. Until then, the message is that the devastating loss of skills and the onset and development of unusual behaviours such as catatonia and increased OCD may not be the signs of early onset dementia, but

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DSRD has a devastating impact on the person with Down syndrome and their families, carers and immediate community. Watching a person who has created a meaningful place in society, working and contributing, suddenly lose their spark, their skills and the love and energy they bring is heart breaking.

For the young person, you can see their frustration, often manifesting in aggression directed toward parents and carers, they withdraw, lose their jobs, stop leaving the house, and in many cases stop communicating.

For parents and carers who have spent a lifetime supporting their child to have a meaningful life, it is not only heartbreaking but exhausting. Parents are returning to carrying out tasks such as dressing and bathing, being desperately concerned as their family member won't eat and experiences rapid weight loss, and frustrated that everything that has worked so far, is gone.

The impact on families is devastating, parents are physically, mentally and emotionally exhausted, being left with feelings of hopelessness and loneliness. More broadly, siblings, other family and close friends are also affected, struggling to understand, having difficulty now communicating with a person that, to date, they have been deeply connected with.

The needs of the person with Down syndrome increases dramatically, family members may have given up work, some have had to change their lives completely to support their family member, and many have struggled with the impact on their own mental health. It is a crisis for all involved.

At DSWA, we are bringing together families who are experiencing some symptoms of regression to provide support and open discussion, enabling us to collect information from the families that have self-identified these regressive behaviours.

In doing so, DSWA is collating data from each family, and as more families reach out, it is the intention that we will have a big enough cohort to attract funding for research in this area, here in WA.

At present there is a small study being undertaken at the Mater Institute in Queensland but otherwise there is very little activity around DSRD in Australia. The DSWA DSRD support group has recently received requests from families in both Queensland and Tasmania to join the group.

It is DSWA's goal to raise awareness of DSRD and to seek professional and financial support for research into the condition to assist with diagnosis and treatment. To read our case studies, view our small data set or review the documents (including journal articles and webinars), please reach out to chair@downsyndromewa.org.au or view our website, **www.downsyndrome.org.au/wa** MF

ED: Nia Parker is chair of DSWA.