# Down Syndrome Regression Disorder (DSRD)

***Sara's decline had been sudden and devastating, "it was like she had died and I didn't have time to mourn her because I had this stranger in my midst who needed a lot of care,"***

Dr Eileen Quinn, mother to Sara and Developmental Paediatrician at the University of Toledo, Ohio.*

Dr Ella Rachamim
Community Paediatrician
Barnet Child Development Team
Royal Free Hospital Trust

*https://www.thestar.com/news/gta/a-mystery-illness-stole-her-son-doctors-in-canada-couldnt-find-the-answer-but-she/article_9dfff7d0-63b3-11ef-b1a2-936fa34e1412.html

# Jude’s story

Jude 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.

In November 2023 Jude became withdrawn, he lost interest and joy.

*"Within a week of him getting ill, I was telling doctors his brain had changed. My instincts were right. He became a different person. He was disappearing physically, mentally and emotionally before our eyes. I was begging doctors to listen."*

# *"Watching my son deteriorate and be a shadow of who he was has affected my whole family profoundly."*

- Jude became mute - no sound was made at all. He usually hummed to himself, laughs, intonates words and sounds but he was silent.
- He refused food and drink and became so dehydrated he was hospitalised.
- He was not able to feed himself and would have to be fed, he lost 9kg in 4 weeks.
- He was sad and would cry and not engage in any activity at school at home.
- He became stiff and catatonic - loosing the ability to balance and walk up stairs.
- If we tried to move him, he would freeze and it would be a real physical task to move him.
- He would stare into space in the same spots for hours at a time.
- He wouldn't sleep in his own bed
- He lost toileting functions and would soil and wet himself frequently
- He then went from delirium to fatigued.
- After a few weeks, it changed again and he would scream, be very aggressive and angry.
- Then barely sleeping at all

# How was Jude with all this?

He became completely mute for a few months too so when putting cannulas in the only way I knew he was in pain was his eyes.  He didn't flinch or make a single sound. That image hasn't left me.

To think of how Jude has been feeling with no means to understand or communicate has been heart wrenching. He has been overcoming challenges for months, being poked and prodded with cannulas, CT scans, hospital admissions, psychiatric changes.

*“No NHS Doctor or GP (or private specialist at the Children’s hospital) knew anything about DSRD……. At times the feeling of isolation and despair was crippling.”*

*“Take him to a health food shop and give him freshly squeezed orange juice and stop treating him like he has Down Syndrome”........ “Not to worry as when he turns 18 years old he will be social care’s problem.”*

- If this person didn’t have Down Syndrome, we’d be rushing them to hospital. We’d be giving them a million-dollar workup”*

*Dr Santoro, Director of neuroimmunology at Children’s hospital, Los Angeles

*Honestly to finally find a consultant that listened made me cry with relief… I felt at times I was going mad! I never ever thought anyone would start listening.*

- Jude was his first case in over 25years.
- ***Impression: "Jude's symptoms fit a neuroregression syndrome associated with Down's syndrome as described by other Doctor's in the USA."***

# Down syndrome regression disorder (DSRD)

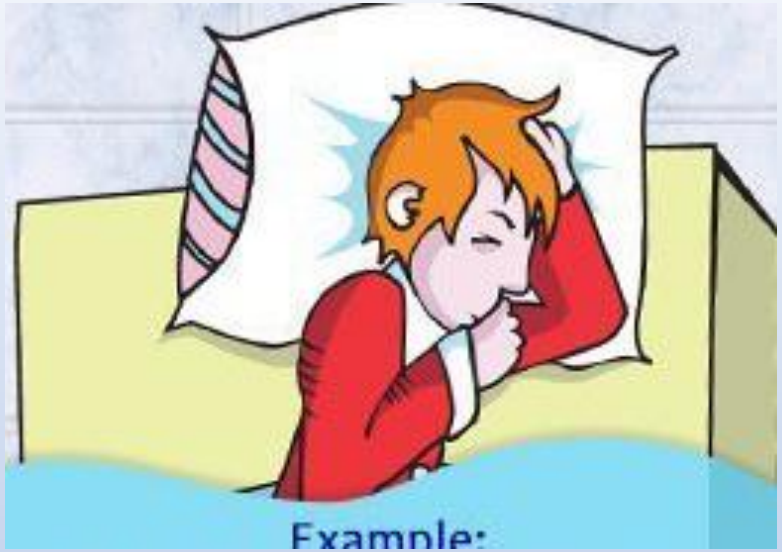
(Down syndrome disintegrative disorder)

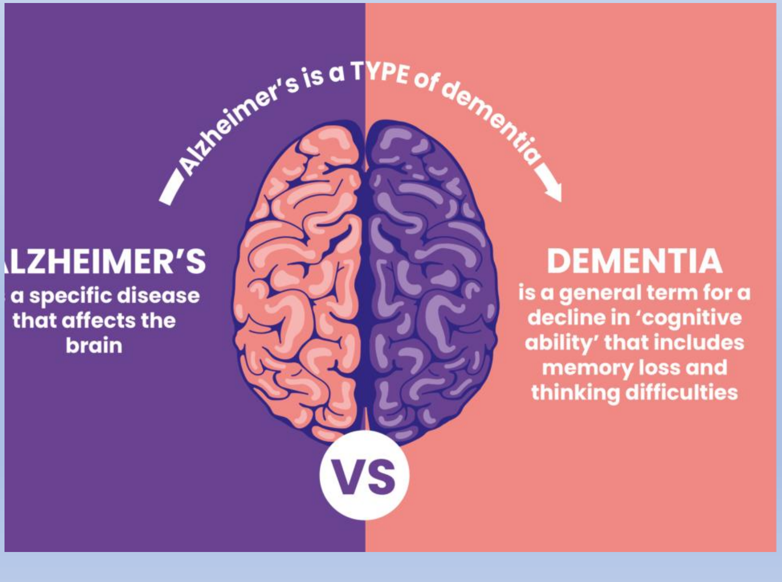
- Young people with DS
- Previously higher functioning
- Suddenly lose their ability to
  - communicate
  - dress or feed themselves
  - use the bathroom or even sleep

***Down syndrome regression disorder (DSRD) was first described in 1946 in a paper about 26 patients with catatonic psychosis.***

***However, there was little research on the condition for over 70 years.***

***DSRD was often misdiagnosed as early-onset Alzheimer's disease, late-onset autism, or schizophrenia.***



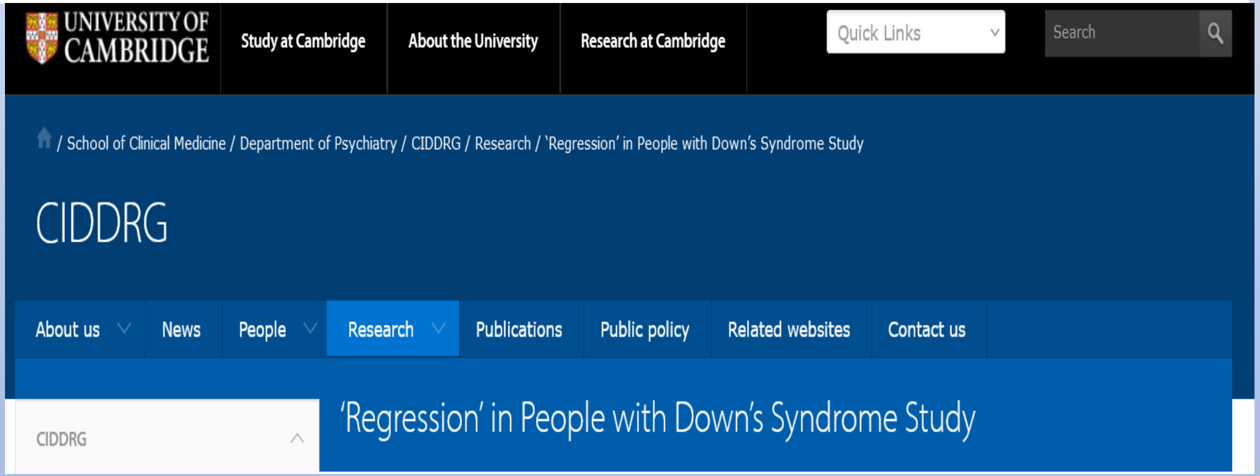


- Mood disorders (30%)
- apathy, extreme slowness or catatonia (37%)
- stereotypies (27%)
- aggressive behaviour (40%)
- Speech impairment (94%)
- MRI (mostly normal – 11/15)
- brain atrophy (n = 2) and
- hippocampal abnormalities (n = 2)

*Article*

**Acute Regression in Down Syndrome**

Benjamin Handen [1,*], Isabel Clare [2], Charles Laymon [1], Melissa Petersen [3], Shahid Zaman [2], Sid O'Bryant [3], Davneet Minhas [1], Dana Tudorascu [1], Stephanie Brown [2], and Bradley Christian [4] on behalf of the Alzheimer's Biomarker Consortium-Down Syndrome (ABC-DS)



/ School of Clinical Medicine / Department of Psychiatry / CIDDRG / Research / 'Regression' in People with Down's Syndrome Study

CIDDRG

'Regression' in People with Down's Syndrome Study

A First Clinical Trial for Down Syndrome Regression Disorder Earns $5.3 Million NIH Grant

*Children's Hospital Los Angeles is leading a pioneering study to find new treatments for a devastating condition in Down syndrome. The study is funded by a five-year, $5.3 million grant from the National Institutes of Health (NIH) Eunice Kennedy Shriver National Institute for Child Health and Human Development.*

# Common pattern

- between the teenage years and mid to late 20s
- sudden, and often unexplained, reduction in expressive language, decreased functional living skills and reduced psychomotor activity
- significant change in long-term needs and independence

# Potential triggers

- Medical (surgery, thyroid disease, coeliac disease, sleep disruption, hormonal cycles, maybe puberty)
- Psychosocial stressors (transition from school, change in living arrangement)
- Something important to the person that might seem insignificant to those around them

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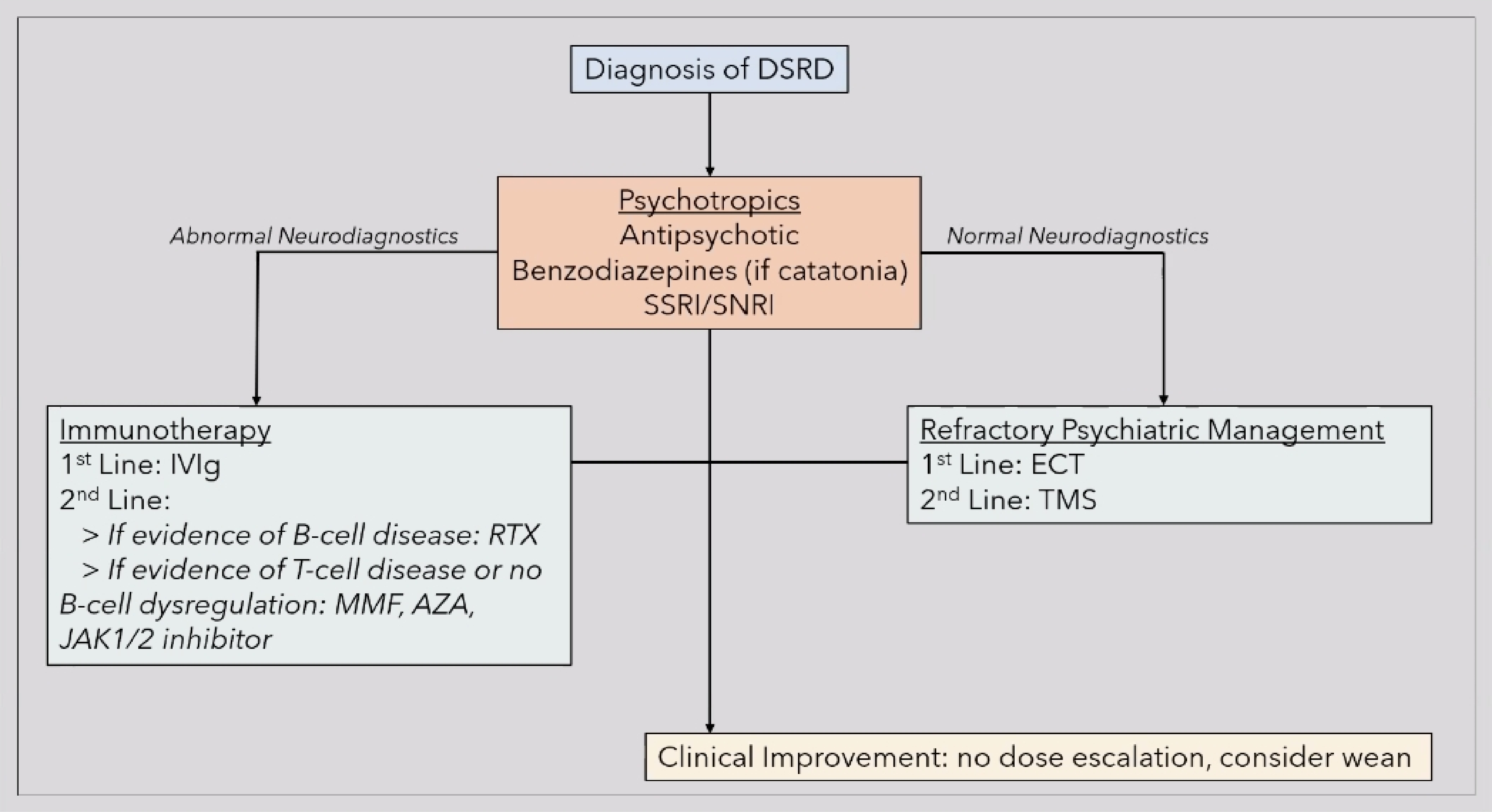
Lorazepam- catatonia and other symptoms

IVIg- an immune-regulating therapy

Tofacitinib (JAK inhibitor) - suppresses immune system

It was like patients were waking up from a dream



Diagnosis of DSRD

<u>Psychotropics</u>
Antipsychotic
Benzodiazepines (if catatonia)
SSRI/SNRI

*Abnormal Neurodiagnostics*

*Normal Neurodiagnostics*

<u>Immunotherapy</u>
1st Line: IVIg
2nd Line:
  > *If evidence of B-cell disease: RTX*
  > *If evidence of T-cell disease or no B-cell dysregulation: MMF, AZA, JAK1/2 inhibitor*

<u>Refractory Psychiatric Management</u>
1st Line: ECT
2nd Line: TMS

Clinical Improvement: no dose escalation, consider wean

Courtesy: Dr Stephanie L Santoro MD, Division of Genetics and Metabolism, , Harvard

- *“In the UK there is currently no workup and treatment plan in place and several doctors including Dr Ella Rachamim are advocating for this condition and for trials/diagnostic and treatment options. Further investigation is required to confirm any underlying brain inflammation or other causes.*
- *There are no specific treatment or trials available in the UK currently but data collection is ongoing currently for children with Down's Syndrome presenting with symptoms.”*

# *So it isn't normal but we are in a better place.*

- Currently Jude is still not the Jude he once was. He isn't in 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 over and over and over again the same scene to the exact moment.
- He can still be very aggressive and less tolerant, so he cannot be left alone with his twin little sisters as he can thrash out for no obvious reason.

# It was truly a relief to hear a Doctor confirm our initial suspicions and we can now try and seek help

- To create more awareness to professionals that are first point of contact for families experiencing the sudden onset of symptoms
- Getting a faster diagnosis for our children
- Emotional and well being support is essential

- https://www.downs-syndrome.org.uk/about-downs-syndrome/health-and-wellbeing/regression-in-downs-syndrome/

REGRESSION SUPPORT GROUP
Tuesday 17 June 2025 | 10.30 – 11.45am | FREE
This is a new online support group for parents of those who have experienced a rapid regression in the last few years.

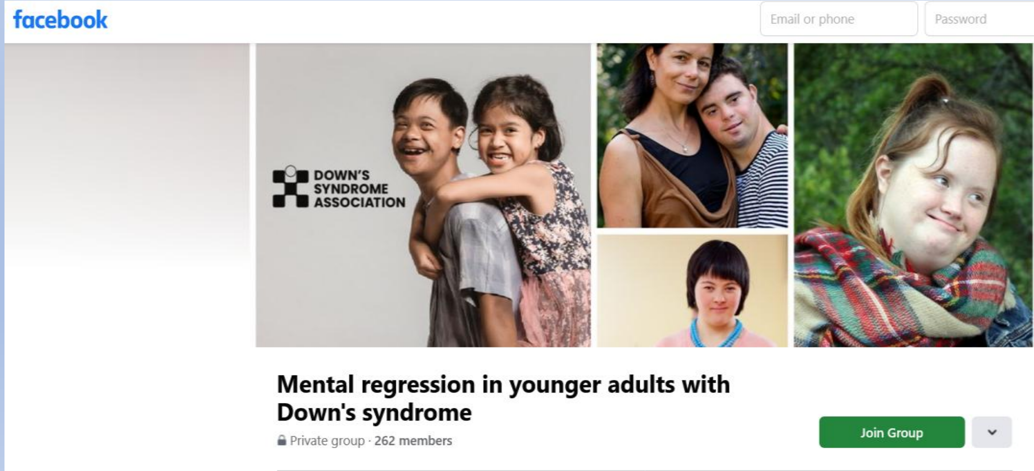
**A mystery illness stole her son. Doctors in Canada couldn't find the answer, but she wouldn't give up**

Mackenzie Wright joined a clinical trial that is bringing hope to Down syndrome families who have struggled with sudden, mysterious regressions.

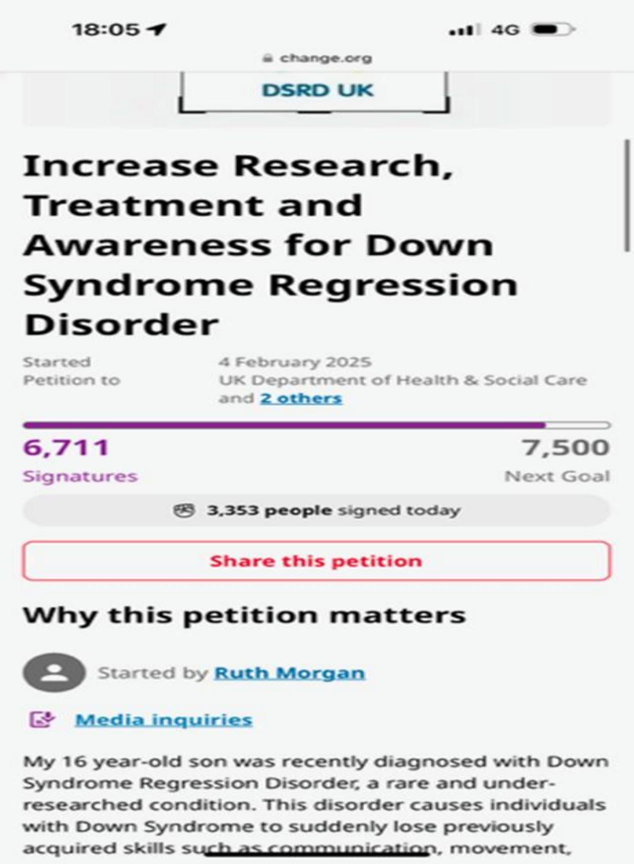
Updated Jan. 5, 2025 at 2:38 p.m.
Jan. 5, 2025

YouTube GB

http://youtu.be/wbvURUte6Zw



**Mental regression in younger adults with Down's syndrome**
Private group · 262 members



**Increase Research, Treatment and Awareness for Down Syndrome Regression Disorder**

| | |
|---|---|
| Started | 4 February 2025 |
| Petition to | UK Department of Health & Social Care and 2 others |

6,711 Signatures
7,500 Next Goal

3,353 people signed today

**Why this petition matters**

Started by Ruth Morgan

Media inquiries

My 16 year-old son was recently diagnosed with Down Syndrome Regression Disorder, a rare and under-researched condition. This disorder causes individuals with Down Syndrome to suddenly lose previously acquired skills such as communication, movement,