



Down Syndrome Regression Disorder Self Assessment (Parent/Child)

Diagnostic Workup (GPs and Specialists)



Down Syndrome
Australia



Down Syndrome
Western Australia



What is Down Syndrome Regression Disorder?

In 2022 a global research paper was produced in which 25 medical professionals from around the world, together agreed on the Nomenclature (Name), Assessment and Diagnostic testing for Down Syndrome Regression Disorder (DSRD).

Regression is the loss of previously acquired skills.

Through a national survey, parents identified the list of symptoms and behaviours on page 3 reflecting the loss of skills and behavioural changes.

DSRD can seriously affect a person's ability to take care of themselves, wash, dress, eat and undertake personal hygiene. Significant regression is also reported in language and communication, social skills, motor skills, memory, and behaviour. Each of these reduces quality of life, independence and social engagement.

The condition is also deeply impactful on the person's family.



Checklist for Parents/Carers of loved ones with Down syndrome experiencing regressive changes

This list has been prepared from the lived experience of parents. Designed to support parents to compile a list of observed changes to support a visit to your General Practitioner.

1. Speech and communication

- Speech deterioration
- Whispered speech
- Stopped talking completely
- Extremely quiet
- Dysphasia (difficulty speaking)
- Changes in handwriting
- Minimal verbal participation
- Has a slower learning pace

2. Physical symptoms

- Catatonia
- Seizures
- Fainting
- Severe weight loss
- Choking
- Hair loss (Alopecia)
- Incontinence
- Spending a lot of time sleeping
- Skin patches or discolouration
- Decline in motor skills
- Very slow/awkward movements
- Unusual facial expressions (eg grimacing)

3. Behavioural and emotional changes

- Extremely slow eating
- Uncontrollable crying
- Aggression
- OCD tendencies rapidly increased
- Can't step over thresholds and cracks
- Loss of interest in life and work
- Sits in the dark for long periods
- Loss of interest in music and hobbies
- Inappropriate laughing and crying (hallucinations)

4. Cognitive and psychological changes

- Incapable of reasoning
- No sense of time/urgency
- Always running late
- Difficulty managing menstrual cycles
- High degree of forgetfulness



5. Daily living skills

Needs assistance with personal care tasks

Requires help to dress

Requires help to shower

Daily wears the same outfits/underwear

Constantly late

Personal hygiene dramatically reduced

6. Social and interpersonal changes

Reduced social engagement

Reduced emotional responsiveness

Falls silent

Not interested in schoolwork

Parents – when to seek help

Dr Eileen Quinn (Paediatrician):

“Any child can have good days and bad days. You think they’ve mastered something, and then suddenly they forget all the words that they used to be able to read. That kind of ebb and flow is pretty normal.

I think the time to really be concerned is when there’s a substantial change that doesn’t get better in a week or two, especially if they start losing more skills and it really starts to affect their daily life and interfere with their ability to do the things that they used to do.”

 www.dsrf.org/resources/blog/u-turn-down-syndrome-regression-disorder/

Parent note:

It is highly beneficial to provide your GP with video footage and a timeline of events as best as you can prepare it.

We encourage you to connect with your local State/Territory association.

 www.downsyndrome.org.au/about-us/contact-us/



For the General Practitioner or Specialist Doctor

Questions to the parent/carer

Questions provided by Dr Cathy Franklin – Master Intellectual Disability and Autism Service (MIDAS), Brisbane Queensland.

Does the person ever:

- get stuck in one position
if so how long for (posturing)
- get stuck in the middle of a
movement – (ambitendency)
- need help getting started with a
movement eg feeding self with
cutlery, taking a step (perseveration)
- have trouble crossing thresholds
eg through doorways or up or
down stairs
- repeat other's speech or movements
(echolalia/echopraxia)

Have you noticed:

- decreased speech/communication
(mutism)
- increased slowness – eg how long
does it take to eat breakfast now
compared to previously
- how long does it take to get dressed
(bradykinesia)
- unusual movements – eye rolling
back, repetitive frequent movements
(stereotypy) or odd ways of doing
unusual behaviours eg walking,
stepping sideways (mannerisms)
- unusual facial expressions
eg grimacing (grimacing)
- abnormal staring (staring)
- changing handedness
(eg was left and now right)
- periods of rushing around, agitation
without obvious trigger (agitation)

NB Inappropriate laughing and crying can also occur and be mistaken for hallucinations.



Diagnostic Workup – General Practitioner and Specialists

General Practitioner

Pathology

FBC	CRP	HbA1c
UEC	Vitamin B12	Coeliac screen
LFT	Vitamin D	HIV, HBV, HCV screen
Ca	TFT	ANA
BSL (random)	Iron studies	Urine MSU M/C/S

**Keep in mind that if there has been a HbA1c, Vit B12 testing in the last year, this is acceptable given that the patient may be out of pocket for these tests if done frequently.*

ECG

Consider:

- ECHO (can request to have an ECG done at the same time)
- Imaging of the brain (CT/MRI – although can be done via neurologist)
- Audiometry

Refer onward at this point if no obvious cause is found.

Specialist Referral

Neurologist

- EEG
- Cognitive assessments
- Lumbar puncture

Geneticist

- Genetic testing

Gastroenterologist

- Colonoscopy and gastroscopy

Psychiatrist

- Autism screening

Respiratory physician

- OSA evaluation

Further Information

Refer to Research Paper: *Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus 2022*. Dr. Santoro says, "Down syndrome regression disorder (DSRD) is really a diagnosis of exclusion. Our workup is not to diagnose DSRD; our workup is to rule out other explanations for it."



www.frontiersin.org/journals/neurology/articles/10.3389/fneur.2022.940175/full

Disclaimer

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