

# Down Syndrome Regression Disorder Self Assessment (Parent/Child) Diagnostic Workup (GP's and Specialists)

## 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.

**Regression is the loss of previously acquired skills.**

Through a national survey, parents identified the list of symptoms and behaviours on page 1 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 reduce quality of life, independence and social engagement.

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

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

This list has been prepared from the lived experience of parents in Western Australia.

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

### 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

### 4. Cognitive and psychological changes

- Incapable of reasoning
- No sense of time / urgency
- Always running late
- Difficult 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 school/work





## 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”

Reference Link: <https://www.dsrf.org/resources/blog/u-turn-down-syndrome-regression-disorder/>

## For the General Practitioner or Specialist Doctor *Questions for parent/carer*

Questions provided by Dr Cathy Franklin - Mater 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 usual behaviours eg walking, stepping sideways (mannerisms)
- unusual facial expressions eg grimacing (grimacing)
- abnormal staring (staring)
- changing handed-ness (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  | Celiac 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

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#### Gastroenterologist

- Colonoscopy + 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.”

Refer to Research Paper: Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus 2022 <https://www.frontiersin.org/journals/neurology/articles/10.3389/fneur.2022.940175/full>

