Down Syndrome Regression Disorder and Catatonia: A Case Report

Background

Catatonia is a psychomotor syndrome occurring in primary psychiatric conditions, medical conditions, and neurodevelopmental disorders such as Down Syndrome (DS) and autism spectrum Disorder (1).

Down Syndrome regression disorder (DSRD) is a commonly noted though poorly understood phenomenon in Down Syndrome, wherein developmental milestones are abruptly lost (2). There is phenomenological and treatment overlap with catatonia, though the syndrome remains poorly characterized (3).

Herein, we present a case of DSRD that was later recognized and treated as catatonia to good effect, emphasizing the importance of careful screening in vulnerable populations.

Table 1: Testing for DSRD (Adapted from Rosso et al, 2020)

Endocrine Studies (Thyroid)	Further genetic testing
Electrolytes and liver function tests	Other inborn errors of metabolism
Nutritional Deficiencies (B12, B9, D)	
STI Testing	
Autoimmune studies (Lyme, PANDAS, ANA)	
EEG	

Case Presentation

The patient was a generally healthy man with DS diagnosed on prenatal screening, well-controlled hypothyroidism, and well-controlled type 1 diabetes on insulin pump. There was no prior reported psychiatric history.

The patient had subacute onset of isolative behaviors, staring, loss of multiple milestones, and idiosyncratic and bizarre use of language. Extensive neurological and medical workup across two medical admissions, including studies for acute intermittent porphyria, were negative.

On psychiatric consultation, the patient presented as bizarre with multiple mannerisms and posturing. Mental status exam also remarkable to reported hallucinations, but there was concern this was better understood as magical thinking. He was discharged on a regimen of fluoxetine 20 mg, risperidone 0.5 mg at night, and lorazepam 0.5 mg daily. Over 2 weeks, this was escalated to 9 mg daily on an outpatient basis with minimal improvement in symptoms. Direct admission from outpatient office was done for ECT consultation due to decreased oral intake and self-care.

After initial treatment, the patient's presentation was dramatically improved, and he received 4 more treatments before discharge on regimen of lorazepam 1.5 mg twice daily.

One month at the outpatient clinic, the patient still has residual abnormalities of speech and difficulty returning to work due to anxiety but was significantly closer to baseline functioning. He reported a qualia of fear while catatonic that had since resolved. Lorazepam continues to be tapered and escitalopram was started for unspecified anxiety.

Disclaimer: The authors have nothing to disclose.

Discussion

Catatonia is a complex syndrome that occurs either independently or because of complex psycho-neuro-immunological dysregulation, but it can still be effectively treated with safe modalities such as benzodiazepines or ECT (4-5).

Rosso et al identify DSRD as a syndrome most notable for behavioral finding compared to prior individual's norms and achieved milestones. There is significant definitional overlap with catatonia, namely of the stuporous type. Their paper describes tiers of medical testing for DSRD, but overt screening and treatment of catatonia is omitted. Furthermore, literature exists showing that catatonia can occur in multiple medical conditions described in their algorithm (6). Though it may be appropriate to consider catatonia an epiphenomenon of general medical conditions in certain context, it is vital that catatonia still be accurately diagnosed in the absence of clear primary medical condition, as in this case.

Learning points

Novel presentations of common psychiatric phenomenon may distract or deter less experienced clinicians.

Different literatures and nosologies may be referring to same clinical phenomenon, even if underlying causes are different.

Even bizarre or complex behaviors can be understood using principles of descriptive psychiatry, and organization or symptoms at the syndromal level provides diagnostic clarity.

References

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