

The Role of Psychiatric Consultation in Diagnosis and Early Intervention in an Adolescent with Down Syndrome Disintegrative Disorder

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Introduction

- Down Syndrome Disintegrative Disorder (DSDD) is a rare but severe condition involving regression in adolescents with Down Syndrome (DS)^{1,2,3,4,5,6}.
- Early and accurate diagnosis is crucial to prevent further deterioration and improve outcomes^{1,6}.
- This case highlights the importance of psychiatric consultation in the early diagnosis and management of DSDD^{4,5}.

Case

- Patient: 15-year-old male with Down Syndrome.
- Presenting Symptoms (Over 12 Months):
 - New-onset aggression and self-injurious behavior
 - Agitation and anxiety
 - Hallucinations
 - Increased stereotypy/stimming
 - Anorexia and insomnia
 - Decline in cognitive abilities, language skills, and social engagement
 - Loss of self-care skills (urinary incontinence)
- History:
 - Diagnosed with Down Syndrome at birth
 - Previously stable developmental progress with speech and occupational therapy
 - No prior psychiatric history
 - Supportive family, special education school

Table 1: Diagnostic process

	Details
Initial Concerns	Rapidly progressing aggression and regression
Methods	Clinical interviews with parents
	Mental status examination
	Review of developmental history
	Bush-Francis Catatonia Rating Scale
	Extensive Laboratory and Imaging: CBC, CMP, TSH, T3/T4, Lead, Folate, Vitamin D, Ammonia, Lactic Acid, CRP, serum and CSF autoimmune encephalopathy, celiac screen, CSF analysis with culture, hearing test and imaging (EKG, EEG, MRI)
Challenges	Differentiating DSDD from other potential causes of regression, such as neurological (e.g., seizures, catatonia) or metabolic disorders due to autoimmune/inflammatory etiologies.
Final Diagnosis	Down Syndrome Disintegrative Disorder (DSDD), characterized by regression in cognitive, behavioral, and functional abilities without an identifiable medical cause.

Table 2: Timeline of recovery

Time	Intervention	Outcome
During Admission	2 Doses of IVIG (0.4 g/kg per day x 5 days)	~50% return to baseline
1 Month Follow Up	Quick return to baseline	
Additional 4-month F/U	2 Additional doses of IVIG (1 mg/kg/month) outpatient	~90% return to baseline

Outcomes

- Stabilization of behavioral symptoms
- ~90% return of lost skills
- Reduction in anxiety and repetitive behaviors
- Improved quality of life for the patient and decreased caregiver stress
- Weight gain from antipsychotic was reported but were able to wean off patient in 6 months from it.

Discussion

- This case highlights the critical role of psychiatric consultation in diagnosing and managing DSDD.
- Early intervention, including pharmacological and behavioral therapies, can significantly improve outcomes.
- Interdisciplinary collaboration is essential for managing complex cases of DSDD.
- The limited understanding of DSDD and lack of standardized guidelines pose challenges in diagnosis and treatment.

Conclusion

- Early psychiatric consultation is vital for adolescents with Down Syndrome presenting with unexplained regression.
- Prompt recognition and treatment can stabilize the patient's condition and improve their quality of life.
- Increased awareness of DSDD among consultation-liaison psychiatrists is crucial for timely intervention.

Reference

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