Catatonia as a Treatable Cause of Late Regression in Rett Syndrome: A Case Report

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Introduction. Rett syndrome is an X-linked neurodevelopmental disorder characterized by loss of speech, motor, and cognitive abilities that typically occurs after a period of normal development. Diagnosis is generally made between six months and two years as developmental delays and regression become noticeable. Catatonia is a neuropsychiatric syndrome with motor and behavioral abnormalities, including disturbances to speech, movement, and responsiveness. Individuals with neurodevelopmental disorders are at elevated risk of catatonia, though diagnosis is often difficult due to variable and overlapping symptomatology.

Case Report. A 10-year-old female diagnosed with Rett syndrome presented with several months of poor oral intake, weight loss, insomnia, mutism, catalepsy, ambitendency, grimacing, and worsening agitation with self-injury. Examination revealed catatonia, and treatment with lorazepam yielded notable improvement in symptoms. Medical work-up including neuroimaging, lumbar puncture, and serum studies was unremarkable, and low-dose lorazepam was continued outpatient with some benefit. A viral illness caused acute worsening of catatonic symptoms, requiring rehospitalization and titration of lorazepam to 24mg daily. There was subsequently marked improvement in catatonia, including reduction of aggression and self-injury, improved sleep and return of some meaningful speech.

Discussion. Very few descriptions of catatonia in Rett Syndrome exist in the medical literature. Regression in Rett Syndrome tends to be prominent in early development, though catatonia should be considered when late motor and behavioral changes occur. This case illustrates catatonia as a treatable cause of regression in Rett Syndrome leading to a return of previously acquired skills and abilities, highlighting the importance of early recognition and treatment.