A case of catatonia in White-Sutton Syndrome

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BACKGROUND

White-Sutton Syndrome is a neurodevelopmental disorder caused by a pathogenic variant in the POGZ gene. Little is known about the psychiatric comorbidities of this disorder and even less about pharmacological treatment.

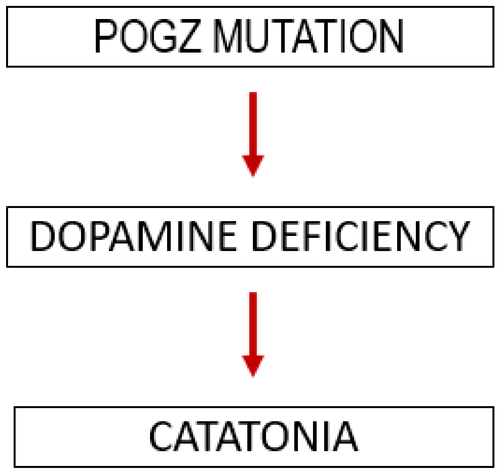
CASE

Patient is a 58 year old female with a history of intellectual disability who was brought to the emergency department for a gradual decline in cognitive function, withdrawal, and incontinence. She had no psychiatric history until several months prior when she became withdrawn and less interested in her hobbies. A local psychiatrist started citalopram and paliperidone. Soon after, she became disheveled and began responding to unseen others. She was admitted to a local hospital where oral paliperidone was transitioned to injectable paliperidone palmitate. Cognitive function worsened after discharge, prompting her presentation to the emergency department. Examination revealed short stature, hypertelorism, retrognathia, and microcephaly. She exhibited immobility, mutism, staring, and posturing. She was given 1mg of intramuscular lorazepam. Catatonia symptoms improved and she was admitted to the psychiatric hospital. Outpatient medications were discontinued and she was discharged after several days without need for further lorazepam.
She was followed in the outpatient clinic and her Catatonia symptoms gradually recurred. Lorazepam was restarted and increased to 3mg TID. It was then cross titrated to clonazepam 1mg TID. After several months of no catatonic symptoms a slow taper was started. The dose got as low as 0.25mg daily. Unfortunately, symptoms recurred and clonazepam was again increased. Given the patient's atypical presentation, she was referred for genetic testing. Exome sequencing revealed findings consistent with White-Sutton syndrome. We contacted Dr. Sutton who confirmed that she is the oldest known patient with White-Sutton Syndrome. She is currently stable on a regimen of escitalopram 5mg daily, clonazepam 0.5mg TID, amantadine 100mg BID, and vitamin b12 1,000mcg daily.

DISCUSSION

Little is known about the natural course of White-Sutton Syndrome. Mutations in the POGZ gene correlate with cognitive dysfunction, developmental delays, autism spectrum disorder, hypotonia, and craniofacial abnormalities (White et al. 2016). POGZ is located on chromosome 1q21.3 and is a transposable element with a zinc finger domain involved in kinetochore assembly and mitotic sister chromatid cohesion and mitotic chromosome segregation (Nozawa et al. 2010). Since this patient's catatonia followed the introduction of an antipsychotic, it is possible that POGZ may play a role in dopamine regulation and catatonia.

PROPOSED MECHANISM



CONCLUSION

The presence of catatonia in a patient with White-Sutton Syndrome without previous psychiatric comorbidities could implicate the role of POGZ in dopamine regulation and catatonia.

ACKNOWLEDGEMENTS

POGZ truncating alleles cause syndromic intellectual disability. White et al. Genome Medicine 2016.
Human POGZ modulates dissociation of HP1alpha from mitotic chromosome arms through Aurora B activation. Nozawa et al. Nature Cell Biology 2010.