ADNP Related Syndrome

https://pubmed.ncbi.nlm.nih.gov/32275126/

Background:

Autism spectrum disorder (ASD) affects approximately one in 59 children. Variants in the activity-dependent neuroprotector homeobox ADNP (OMIM #611386) gene may be one of the most common single-gene causes of syndromic ASD. Most patients diagnosed with ADNP syndrome have ASD as a comorbidity, and all patients have mild-to-severe intellectual disability.

Methods/case report:

We present a case report of a patient diagnosed with ADNP syndrome at 2.5 years of age. The patient has many of the key features of the syndrome, including ASD, global developmental delay, behavioral problems, congenital heart defect, early tooth eruption, and vision problems. The patient's initial presentation included congenital diaphragmatic hernia (CDH), which has not been previously reported in this condition.

Results:

The patient exhibited frequent behavioral outbursts and was initiated on antipsychotic medication with near-complete resolution of symptoms allowing her to engage more fully in early intervention therapies leading to progress in language acquisition.

Conclusion:

This short report provides guidance for antipsychotic medication dosing to improve early intervention outcomes. This is the first report of CDH in this syndrome.