ADNP Related Syndrome

https://pubmed.ncbi.nlm.nih.gov/27054228/

Clinical characteristics:

ADNP

-related disorder is characterized by hypotonia, severe speech and motor delay, mild-to-severe intellectual disability, and characteristic facial features (prominent forehead, high anterior hairline, wide and depressed nasal bridge, and short nose with full, upturned nasal tip) based on a cohort of 78 individuals. Features of autism spectrum disorder are common (stereotypic behavior, impaired social interaction). Other common findings include additional behavioral problems, sleep disturbance, brain abnormalities, seizures, feeding issues, gastrointestinal problems, visual dysfunction (hypermetropia, strabismus, cortical visual impairment), musculoskeletal anomalies, endocrine issues including short stature and hormonal deficiencies, cardiac and urinary tract anomalies, and hearing loss.

Diagnosis/testing:

The diagnosis of

ADNP

-related disorder is established by identification of a heterozygous

ADNP

pathogenic variant on molecular genetic testing.

Management:

Treatment of manifestations:

Treatment is symptomatic and can include: speech, occupational, and physical therapy; specialized learning programs depending on individual needs; treatment of neuropsychiatric features; nutritional support as needed; standard treatment of gastrointestinal, ophthalmologic, musculoskeletal, endocrine, and cardiac findings; standard treatments for hearing loss, seizures, urinary tract anomalies, and recurrent infections.

Surveillance:

At each visit monitor growth and nutrition, occupational and physical therapy needs; assess for seizures, developmental progress, behavioral issues, gastrointestinal issues, and family needs; annual vision assessment.

Genetic counseling:

ADNP

-related disorder is expressed in an autosomal dominant manner and typically caused by a de novo ADNP

pathogenic variant, the risk to other family members is presumed to be low. Once an ADNP

pathogenic variant has been identified in an affected family member, prenatal testing and preimplantation genetic testing are possible.