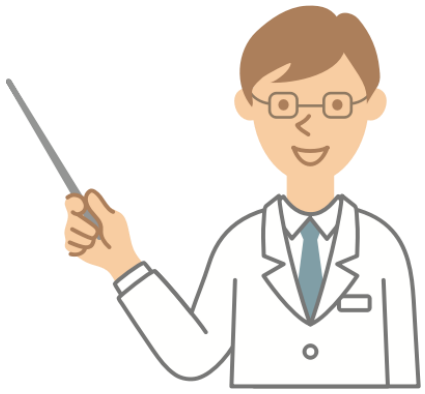
HOUGE-JANSSENS SYNDROME (TYPE 3)

Houge-Janssens Syndrome – Type 3 (HJS-3) is a rare genetic disorder characterized chiefly by neurodevelopmental delay, epilepsy, and various behavioral disorders.

SYMPTOMS

- Intellectual and/or developmental delay (mild to profound)
- Speech delay
- Seizures
- Hypotonia
- Brain abnormalities
- Autism spectrum disorder (ASD)
- Digestive dysfunction
- Other symptoms have been reported.



CAUSE

HJS-3 is caused by a mutation in the gene PPP2CA. To date, most cases are thought to be caused by a *de novo* mutation in this gene. The nature of the genetic variant majorly determines the overall clinical severity, which can range from very mild to very severe.

DIAGNOSIS

Diagnosis is typically made through genetic testing. Modalities of testing include whole exome sequencing. Brain abnormalities may be detected on imaging.

TREATMENT

There is no cure for HJS-3. Treatment involves managing the symptoms of the disorder (i.e. physical therapy, speech therapy, anticonvulsants, etc.).

OUTLOOK

Children may succeed in school given appropriate accommodations in the classroom. Although the natural history of this disorder is still largely unknown, barring complications from conditions such as epilepsy, those who are afflicted with HJS-3 may expect to live a full life commensurate with the severity of neurological disorder present.

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