A Case Study on Poirier-Bienvenu neurodevelopmental syndrome (POBINDS)

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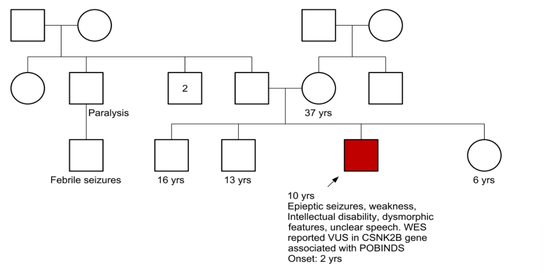
INTRODUCTION

- The Poirier-Bienvenu neurodevelopmental syndrome (POBINDS) is a rare neurological disorder.
- Caused by mutations in the *CSNK2B* gene which encodes for a subunit of casein kinase, *CK2* involved in neuronal growth and synaptic transmission.
- Characterized by early-onset seizures and variably impaired intellectual development (ID).
- Current evidence suggests that POBINDS could manifest with a wide range of symptoms, including developmental disability (DD), and growth abnormalities.
- Severity of neurologic impairment is highly variable.
- Some patients may have refractory seizures and be bedridden with no meaningful speech, others may have treatment-responsive seizures and achieve normal psychomotor development.
- Less than 100cases have been reported in the literature.
- POBINDS has Autosomal Dominant pattern of inheritance.

Family History and Pedigree:

No significant family history was seen. History of febrile seizures was seen in one of his paternal cousin



CASE HISTORY

10year old male child, born to a non-consanguineous couple, 3rd child in birth order presented with

- lethargy
- Weakness
- frequent falls low weight of 22kgs
- lean and thin
- poor academics
- epileptic episodes with uprolling of eyeballs and jerks
- Irritability drooling while sleeping
- Chewing difficulties
- Inability to perform his daily activities by own
- unresponsiveness.

This case was referred to Strand Life Sciences in the month of November'23 from Bangalore Child Neurology and Rehabilitation Center.

GC OBSERVATIONS

- Detailed birth history showed full term, normal delivery and child cried immediately after birth.
- Child had dry scaly skin and skin rashes after birth.
- Normal Gestational history.
- Normal developmental milestones.
- Had noticeable frequent falls, at 2yrs.
- Fatigue
- High arched neck.
- Long fingers, arms, legs
- Thin build.
- Drooping eyelids.
- Long philtrum
- Wide spaced teeth
- Unclear speech
- Suspected scoliosis
- EEG reported generalised epileptiform discharges

RESULTS

Exome Plus test reported a heterozygous 'variant of uncertain significance' (VUS) was detected in exon 4 of the *CSNK2B* gene associated with POBINDS syndrome.

Management Options:

The different management options include a multisystem care involving their referring clinician, a neurologist, a paediatrician for monitoring the child's health condition. Sanger sequencing is recommended for parents. They were advised to consult a therapist for the child's behaviour, speech and behaviour improvement. The child was given contacts for a few support groups based out of Kerela

GC Session:

The pre-test GC session involved a detailed clinical history of the child, the phenotypic features observed were noted. A detailed 3 generation family history was taken.

The case highlights the importance of detail history taking during a Pre-Test GC, for differential diagnosis of a case, followed by a specific test recommendation for arriving at a proper genetic diagnosis.

Majority of POBINDS patients presents with a wide spectrum of phenotypes described has made the diagnosis and clinical follow-up progressively challenging for clinicians.

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