ORIGINAL RESEARCH PAPER

Paediatric Medicine

# A CASE REPORT OF JOUBERT SYNDROME

KEY WORDS:

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## INTRODUCTION

Joubert syndrome is an autosomal recessive disorder (ciliopathy) with significant genetic heterogeneity that is associated with cerebellar vermis hypoplasia and the pontomesencephalic molar tooth sign (a deepening of the interpeduncular fossa with thick and straight superior cerebellar peduncles). It is associated with hypotonia, ataxia (as toddler), characteristic breathing abnormalities including episodic apnea and hyperpnea (which improves with age), global developmental delay, nystagmus, strabismus, ptosis, and oculomotor apraxia.

## CLINICAL DIAGNOSIS

The features necessary for a diagnosis of classic JBTS include the following (1) The molar tooth sign on axial views from cranial MRI studies comprised of these 3 findings: cerebellar vermis hypoplasia (CVH), deepened interpeduncular fossa, and thick, elongated superior cerebellar peduncles (2) intellectual impairment/ developmental delay, of variable degree; (3) hypotonia in infancy; (4) one or both of the following (not required but supportive of the diagnosis): irregular breathing pattern in infancy (episodic apnea and/or tachypnea, sometimes alternating) and abnormal eye movements (nystagmus and/or oculomotor apraxia (OMA).

### Molecular Genetics

Mutations in the eight ciliary/basal body genes INPP5E, AHI1, NPHP1, CEP290, TMEM67/MKS3, RPGRIP1L, ARL13B, and CC2D2A have been identified in subjects with JSRD.

### Case Presentation

A 11-month-old male child born at 38 weeks of gestation by normal vaginal delivery out of non consanguineous marriage with birth weight 2.6 kg, appropriate for gestational age, cried immediately after birth to 22 years old primi-gravida mother. On examination patient have hypotonia, breathing abnormalities like episodic apnea, global developmental delay, nystagmus, and oculomotor apraxia.

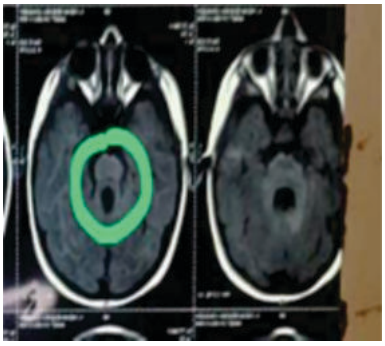
### Management

- Examination of high-quality MRI scan to assess for cerebral malformations, neuronal migration disorders, or cephaloceles that could portend a poorer prognosis or seizures, if not done at the time of diagnosis
- A baseline neurologic evaluation with particular attention to tone, respiratory pattern (tachypnea and apnea), eye movements, development, and cerebellar function
- Sleep history with polysomnogram as baseline evaluation and particularly if symptomatic apnea is present
- Assessment of oromotor function by a speech therapist and/or by fluoroscopic swallowing studies
- Developmental assessment with age-appropriate tools
- Evaluation by a pediatric ophthalmologist via dilated eye examination for colobomas and retinal changes, as well as strabismus and ptosis, with consideration of specialized testing such as visual-evoked potentials, electro-retinogram, and ocular motility testing

### Genetic Counseling

Genetic counseling by a clinical geneticist is imperative to help JS families understand mechanisms of inheritance, repetition risk, reproductive options, and genetic testing. In vitro fertilization with preimplantation testing for known pathogenic variants, heterologous insemination, and/or prenatal testing (including prenatal imaging and invasive diagnostics) are possible options to avoid repetition in a family.

**Pic.1 Hypotonia + nystagmus**



**Pic.2 Molar tooth sign**

## REFERENCES

[1] Nelson essentials of pediatrics 21st international edition, section 26, chapter 609.9, Agenesis of cranial nerve and dysgenesis of posterior fossa, pp 3073.
[2] National center for biotechnology information, National library of medicine, Joubert syndrome.