

The Ocular Phenotype of Joubert Syndrome in an Adolescent: High Myopia with Oculomotor Apraxia and Torsional Nystagmus

Running title: Ocular phenotype of Joubert syndrome

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ABSTRACT

Background: Joubert syndrome (JS) is a rare autosomal-recessive ciliopathy defined by a characteristic mid-hindbrain malformation the “molar tooth sign” (MTS) with infantile hypotonia, global developmental delay, episodic neonatal breathing dysregulation and abnormal eye movements. Ocular involvement is frequent and often requires ophthalmic care.

Case presentation: A 15-year-old boy with a previously established diagnosis of Joubert syndrome presented with longstanding poor vision in both eyes. He had had infantile hypotonia with grossly delayed milestones (head control at 3 years, walking at 5 years, first words at 6–7 years), hearing impairment, and a history of episodic rapid breathing in infancy; his parents were non-consanguineous. Examination revealed bilateral high myopia with astigmatism (right $-7.00/-4.00 \times 180$, left $-13.00/-3.50 \times 170$; best-corrected acuity 6/18 and 6/60), ocular motor apraxia with compensatory head thrusts, torsional nystagmus, intermittent exotropia, an anomalous head posture (head tilt to the right, face turn to the left) and left pseudoptosis. The fundus showed myopic changes without retinal dystrophy or coloboma, and corneal topography showed high but regular astigmatism with normal pachymetry. Spectacles were prescribed, contact-lens correction was offered, and continued multidisciplinary care with genetic counselling was reinforced.

Conclusion: High myopia, oculomotor apraxia, nystagmus, strabismus and ptosis are characteristic ocular features of JS. Recognising and managing them refractive correction, amblyopia and low-vision care, and monitoring is an important part of the multidisciplinary care of these children; in an undiagnosed child, such findings should prompt neuroimaging.

Keywords: Joubert syndrome; molar tooth sign; ciliopathy; high myopia; oculomotor apraxia; nystagmus

INTRODUCTION

Joubert syndrome (JS) is a genetically heterogeneous, predominantly autosomal-recessive neurodevelopmental ciliopathy first described in 1969.¹ Its diagnostic hallmark is the molar tooth sign (MTS) on axial brain magnetic resonance imaging (MRI) thickened, horizontally oriented superior cerebellar peduncles with a deep interpeduncular fossa, arising from cerebellar vermis hypoplasia and a maldeveloped ponto-mesencephalic junction.^{2,3} The core clinical triad comprises infantile hypotonia evolving into ataxia, global developmental delay, and the characteristic

neuroradiological malformation, variably accompanied by episodic neonatal breathing dysregulation and abnormal eye movements.⁴

Because more than 40 cilia-related genes are implicated, the phenotype is broad, and additional involvement of the retina, kidneys, liver and skeleton defines recognised sub-groups.^{4,5} Ocular manifestations are among the most consistent features. We describe the ocular phenotype and its management in an adolescent with a previously established diagnosis of Joubert syndrome, in whom high myopia, oculomotor

apraxia, torsional nystagmus and an anomalous head posture were prominent.

CASE REPORT

History: A 15-year-old boy, a previously diagnosed case of Joubert syndrome, was brought to the ophthalmology out-patient department with longstanding diminution of vision in both eyes and spectacles that had become inadequate. He was born at term (40 weeks) by normal vaginal delivery after an uneventful, well-supervised pregnancy with regular antenatal care and supplementation and no maternal illness; the birth weight was 2.8 kg and the Apgar scores were 10 and 10, with no neonatal seizures, jaundice or intensive-care admission. A history of episodic rapid breathing was noted during infancy. His developmental milestones were grossly delayed (below), and he has hearing impairment. There was no significant family history, and the parents were non-consanguineous. The diagnosis of Joubert syndrome had been established in infancy on MRI of the brain.

Developmental history: Milestones were grossly and globally delayed: head control at 3 years (achieved with physiotherapy) and independent walking at 5 years; visually-guided behaviour (fixing and

following) at 3 years; social and cognitive interaction at 5 years; and first words at 6–7 years (following speech therapy). He was normally fed, follows a vegetarian diet, is fully immunised, and currently attends school.

Ocular examination: Uncorrected and corrected visual acuities are shown in Table 1. Cycloplegic refraction was $-7.00/-4.00 \times 180$ in the right eye and $-13.00/-3.50 \times 170$ in the left, giving a best-corrected acuity of 6/18 (logMAR 0.48) and 6/60 (logMAR 1.00) respectively — bilateral high myopia with high astigmatism, marked anisomyopia and left amblyopia. Ocular motor apraxia was evident, with impaired initiation of horizontal saccades and compensatory head thrusts. There was, in addition, an anomalous head posture (head tilt to the right, face turn to the left) and a torsional nystagmus in both eyes. Ocular alignment showed an intermittent exotropia, and there was a left pseudoptosis. Intraocular pressure was 14 and 15 mmHg with normal central corneal thickness; the anterior segments and lenses were clear. Fundus examination of both eyes showed a healthy optic disc with peripapillary atrophy, a dull foveal reflex, a tessellated fundus and white-without-pressure at the periphery changes attributable to high myopia, without pigmentary retinal dystrophy or coloboma.

Table 1. Visual acuity (Snellen and logMAR).

Measurement	Right eye	Left eye
Uncorrected	3/60 (1.30)	2/60 (1.48)
With previous spectacles	5/60 (1.08)	4/60 (1.18)
Best corrected (new refraction)	6/18 (0.48)	6/60 (1.00)

logMAR equivalents in parentheses.

Systemic Examination

The head circumference was normal. He had a history of infantile hypotonia with grossly delayed milestones and hearing impairment. A detailed neurological examination was not available at our centre.

Investigations

Corneal topography (Pentacam) confirmed high, regular corneal astigmatism corresponding to the refractive cylinder anterior corneal astigmatism 4.2 D (K1 41.4 / K2 45.6) in the right eye and 4.6 D (K1 41.1 / K2 45.7) in the left, with maximum keratometry 46.4 D in both eyes and normal corneal thickness (thinnest pachymetry 517 μ m right, 543 μ m left). Corneal ectasia screening (Belin/Ambrósio) was normal in the left eye (final D 1.05; topographic keratoconus classification negative), while in the right eye some indices were borderline (final D 2.37; classification

“possible”), consistent with the effect of the high regular astigmatism; the right eye was therefore regarded as a keratoconus suspect for follow-up rather than established keratoconus. MRI of the brain, performed in infancy, demonstrated the molar tooth sign thickened, elongated superior cerebellar peduncles with a deepened interpeduncular fossa and a dysplastic pontomesencephalic junction (Figure 1) together with cerebellar vermian hypoplasia (predominantly inferior), widened separation of the cerebellar hemispheres, a prominent fourth ventricle and mild communicating ventricular dilatation; no focal parenchymal lesion or abnormal diffusion restriction was seen. These appearances are diagnostic of a Joubert syndrome spectrum malformation. Electroretinography, audiological assessment, renal and hepatic screening and molecular genetic testing were advised but could not be performed.

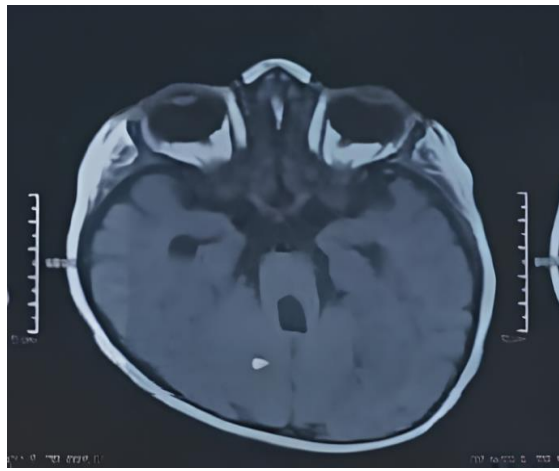


Figure 1. Axial MRI of the brain at the ponto-mesencephalic junction (performed in infancy) showing the molar tooth sign: a deepened interpeduncular fossa with thickened, elongated superior cerebellar peduncles.

Table 2. Ocular features of Joubert syndrome and their status in the present case

Ocular feature	Reported frequency in JS ⁶	Present case
Ocular motor apraxia	~80%	Present
Strabismus (exotropia commonest)	~74%	Intermittent exotropia
Nystagmus	~72%	Present (torsional)
Refractive error (myopia / astigmatism)	Common ⁷	High myopia + astigmatism
Ptosis	~43%	Left pseudoptosis
Retinal degeneration	~38%	Absent (myopic fundus)
Chorioretinal coloboma	~30%	Absent
Optic-nerve atrophy	~22%	Absent

Reported frequencies are from a published review of the ocular phenotype of Joubert syndrome.

Management

The clinical and ocular findings were consistent with the established diagnosis of Joubert syndrome. Spectacle correction was prescribed and contact-lens correction was offered for the anisomyopia, with low-vision support for the left eye. The intermittent exotropia and nystagmus were documented for monitoring, and continued multidisciplinary care (paediatric neurology, nephrology, hepatology, audiology, developmental paediatrics and clinical genetics) with genetic counselling regarding the autosomal-recessive inheritance and the approximately 25% recurrence risk was reinforced. A review after one month with spectacles was advised.

DISCUSSION

The diagnosis of JS rests on the MTS in a child with the appropriate neurodevelopmental phenotype; isolated vermian hypoplasia is not sufficient.^{2,4} Ocular features are common but variable. In a review of the ocular phenotype, ocular motor apraxia was the most frequent finding (~80%), followed by strabismus (~74%) and nystagmus (~72%); ptosis (~43%),

chorioretinal coloboma (~30%), optic-nerve atrophy (~22%) and retinal degeneration (~38%) were less common, and exotropia was the commonest strabismus.⁶ Refractive errors, particularly astigmatism and myopia, are frequently reported, with spherical errors spanning approximately +4 to -15 dioptres in one series.⁷

Our patient displayed a characteristic ocular phenotype ocular motor apraxia, the single most frequent ocular sign, together with nystagmus, intermittent exotropia, ptosis and high myopia with astigmatism while retinal dystrophy and coloboma were absent. This is instructive for three reasons. First, although our patient had already been diagnosed, these same features ocular motor apraxia with compensatory head thrusts, an anomalous head posture and a central-type (torsional) nystagmus are, in an undiagnosed child, important clues that should prompt neuroimaging; and in a known case the ophthalmologist carries the ongoing burden of refractive, amblyopia and low-vision care. Second, the fundus changes here were myopic (tessellation, peripapillary atrophy) rather than dystrophic; because retinal degeneration occurs in a

substantial minority of patients and carries prognostic and genotype implications, electroretinography is worthwhile even when the visible fundus is attributable to myopia.⁶ Third, the recognition of JS mandates a multidisciplinary search for renal, hepatic and other systemic involvement, and genetic counselling for the family.

The differential diagnosis includes other MTS-related disorders COACH, Senior-Løken, Dekaban-Arima and oral-facial-digital type VI syndromes distinguished by their additional systemic features, as well as Poretti-Boltshauser syndrome (LAMA1-related cerebellar dysplasia with cysts, typically without a true MTS), Dandy-Walker malformation and rhombencephalosynapsis.^{8,9} Careful correlation of clinical and neuroradiological findings, supported where possible by molecular testing, resolves these.

Strengths

This case report highlights the combination of high myopia with astigmatism, ocular motor apraxia, torsional nystagmus, intermittent exotropia, and anomalous head posture in an adolescent with Joubert syndrome. Detailed refractive assessment, fundus examination, corneal topography, and documentation of previous neuroimaging provide a useful clinical description of the ocular phenotype and emphasize the importance of comprehensive ophthalmic evaluation in affected children.

Future Directions

Further evaluation should include electroretinography, periodic retinal monitoring, audiological assessment, renal and hepatic screening, and molecular genetic testing where feasible. Longitudinal follow-up may help document visual outcomes, refractive changes, ocular alignment, and the effectiveness of spectacle correction and low-vision support. Multidisciplinary care and genetic counselling remain important for long-term management.

Limitations

This is a single case; a detailed neurological examination, electroretinography, systemic (renal and hepatic) screening and molecular genetic confirmation were not available. Nonetheless, it documents the ocular phenotype and the ophthalmic management of a child with Joubert syndrome.

CONCLUSION

Joubert syndrome shows characteristic ocular features high myopia, oculomotor apraxia, nystagmus, strabismus and ptosis. Recognising and managing these features, and monitoring for retinal and other involvement, is an important part of the coordinated, lifelong care of these children; in an undiagnosed child, an anomalous head posture with a central-type nystagmus and a suggestive developmental history should prompt neuroimaging.

Declarations

Consent to publish: Written informed consent for publication of the clinical details and accompanying images was obtained from the patient's parent/legal guardian, as the patient is a minor. A copy is available with the corresponding author.

Author contributions: Both authors were involved in the clinical care of the patient, the conception of the report, and the drafting and critical revision of the manuscript, and both approved the final version.

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