Walker–Warburg syndrome

Renu Suthar, Suresh K Angurana, Usha Singh, Paramjeet Singh

1 Department of Pediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh, India
2 Department of Ophthalmology, Postgraduate Institute of Medical Education and Research, Chandigarh, India
3 Department of Radiodiagnosis, Postgraduate Institute of Medical Education and Research, Chandigarh, India

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Correspondence Address:
Dr. Suresh K Angurana
Department of Pediatrics, Advanced Pediatric Centre, Postgraduate Institute of Medical Education and Research, Chandigarh India

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A 7-day-old female baby, born at term to a non-consanguineous couple with an uncomplicated antenatal period, presented with decreased movements since birth. Examination revealed a weight of 2.2 kg (<3\textsuperscript{rd} centile), a length of 48 cm (15\textsuperscript{th} centile), and an occipitofrontal circumference (OFC) of 38.5 cm (>97\textsuperscript{th} centile). She had dysmorphism, microcornea, micro-ophthalmia \[\text{Figure 1}\], white pupillary reflex, corneal opacity, persistent hypertrophic primary vitreous (PHPV) on the right side, buphthalmos on the left side, and generalized hypotonia. The creatine kinase (CK) level was 1967 U/mL (normal: <190 U/mL). Magnetic resonance imaging of the brain shows characteristic features \[\text{Figure 1}\]b, \[\text{Figure 1}\]c, \[\text{Figure 1}\]d. Renal ultrasonography was normal. The patient was referred to a neurosurgeon and an ophthalmologist for further management, but the parents choose not to undergo any active treatment.

Figure 1: (a) Facial profile shows a large head, depressed nasal bridge, long philtrum, and micro-ophthalmia on the right side. (b) MRI brain T1 sagittal weighted images; (c and d) T2 axial sections showing the abnormal “Z” shaped and hypoplastic brainstem with kinking at the junction of midbrain and pons as well as at the cervico-medullary junction. Cerebellar hemispheres and vermis are atrophic with prominent posterior fossa cerebrospinal fluid (CSF) spaces. A large cyst measuring $5.9 \times 4.5$ cm is seen in the posterior fossa communicating with the fourth ventricle. A small bony defect in the occipital region (5 mm in size) in the midline, along with external herniation of the meninges and CSF into the subcutaneous tissue, causes a mild contour bulge. The supratentorial ventricular system is dilated. The
corpus callosum is hypogenetic with an absent septum pellucidum. The brain parenchyma of bilateral cerebral hemispheres is thinned out. Bilateral frontal lobes show pachygyria and bilateral parieto-occipital regions show cobblestone polymicrogyria. The right globe is smaller in size with a distorted contour that is suggestive of microphthalmia.

Characteristic clinico-radiological features were suggestive of Walker–Warburg syndrome (WWS), which is a rare congenital muscular dystrophy (CMD) with autosomal recessive inheritance associated with brain and eye abnormalities. WWS is the most severe form of CMD with most children dying before 3 years of age with the reported incidence of 1–2/1,00,000 live births.\cite{1,2,3} WWS presents at birth and early infancy with generalized hypotonia, developmental delay, mental retardation, occasional seizures, and facial dysmorphism. Central nervous system abnormalities include type II cobblestone lissencephaly, hydrocephalus, vermian and generalized cerebellar malformation, a flat brain stem with small pyramids, hypomyelination, hypoplasia or agenesis of the corpus callosum, occipital encephalocele, and Dandy–Walker malformation. The eye abnormalities include the presence of cataract, a shallow anterior chamber, microcornea, micro-ophthalmia, lens defects, retinal detachment or dysplasia, hypoplasia, atrophy of the optic nerve and macula, PHPV, and coloboma. Glaucoma or buphthalmos may be present.\cite{1,2,3,4}

In any infant presenting with hypotonia, elevated CK, hydrocephalus, brain and eye malformation, and characteristic imaging, the diagnosis of WWS should be considered.

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References


Figures

[Figure 1]