Walker-Warburg Syndrome: Demonstration of Cerebellar Cysts with CISS Sequence

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Introduction

Walker-Warburg syndrome (WWS) is an autosomal recessive disorder characterized by a triad of congenital muscular dystrophy, brain anomalies, and ocular abnormalities. The brain anomalies mainly include type II lissencephaly (cobblestone cortex), pontine and cerebellar hypogenesis, cerebral or cerebellar hypomyelination, cerebellar polymicrogyria with or without cysts, and variable callosal hypogenesis. Constructive interference in steady state (CISS) sequence, a heavily $T_2$-weighted sequence, is ideal for demonstrating the presence of cerebellar cysts on magnetic resonance (MR) imaging. We report the complete imaging spectrum in a patient with WWS and emphasize the utility of CISS sequence in the imaging of cerebellar cysts.

Case Report

A male infant was born to parents of consanguinous marriage at full term by normal vaginal delivery. No prenatal imaging was performed. At birth, the infant was hypotonic with weak reflexes, macrocrania, and bilateral enophthalmos with right-sided leukocoria. Biochemical investigations and screening for congenital infections were normal.

Computed tomographic (CT) scan of the brain performed on the third day of life revealed hydrocephalus with hypoplastic cerebellum and absent vermis (Fig. 1). Magnetic resonance (MR) imaging of the brain revealed marked asymmetric dilatation of the ventricles and hypomyelination. The cortex was thick with irregular projections into the underlying white matter, giving the appearance of cobblestone cortex (Fig. 2a). The body and splenium of the corpus callosum were not visualized, suggesting partial agenesis. There was pontine hypogenesis with a dorsal kink at the mesencephalo-pontine junction (Fig. 2b). The cerebellum appeared malformed and contained multiple tiny cysts on axial $T_2$-weighted and turbo inversion recovery (TIR) images (Figs. 2c, 3). On CISS sequence (repetition time [TR], 10.76 ms; echo time [TE], 5.38 ms; matrix size, 256×256; voxel size, 0.7×0.7×1 mm), the cysts were much better appreciable as a result of the sequence’s inherent heavy $T_2$ weighting and high contrast (Fig. 3). Ultrasound B scan of the orbits revealed bilateral retinal detachment with right-sided persistent hyaloid artery, suggesting persistent hyperplastic primary vitreous (Fig. 4a, b, c). All these classic findings led to diagnosis of WWS.
Discussion

CMDs are mainly classified into 4 types: pure CMD, Fukuyama CMD, muscle-eye-brain disease (MEBD), and Walker-Warburg syndrome, the latter being the most severe.\(^3\) Cerebellar polymicrogyria and cysts may be present in all but the pure type. MR imaging is the principal imaging modality used to diagnose brain anomalies in WWS, which include severe diffuse cobblestone cortex, complete absence of cerebral and cerebellar myelin, cerebellar polymicrogyria (with or without cysts), pontine and cerebellar vermian hypoplasia, hydrocephalus, variable callosal hypogenesis, and encephalocele.\(^3,4\) Ocular abnormalities are essential features of WWS and include bupthalmos, congenital cataract, coloboma, glaucoma, optic nerve hypoplasia, persistent hyaloid artery, retinal detachment, and microphthalmia.\(^5\) The last three were present in our case.

The presence of polymicrogyria and associated cerebellar cysts is seen in CMDs and in congenital infections.\(^3,6\) The cysts are present within or adjacent to the polymicrogyria and are partially lined by leptomeningeal tissue on pathological studies. They are probably formed from the subarachnoid spaces that were engulfed by the malformed cerebellar cortical folia, particularly at the junction of normal and abnormal cortices.\(^7\)

The CISS sequence is a heavily T2-weighted high resolution steady-state gradient-echo sequence, with intrinsic high contrast between fluid, such as cerebrospinal fluid (CSF), and neuroparenchyma. It is a 3-dimensional sequence that enables high resolution image reconstruction in other planes. It is useful to evaluate the cisternal segments of cranial

![Image](image-url)

**Fig. 1.** Axial computed tomographic (CT) image shows hydrocephalus with hypoplastic cerebellum (thin arrow) and absent vermis (thick arrow).

![Image](image-url)

**Fig. 2.** (a) Axial T2-weighted image shows hyperintense signal in the white matter (thin arrow) with thickened cortex and irregular projections into the underlying white matter (thick arrow), suggestive of cobblestone lissencephaly. (b) Sagittal T1-weighted image shows absent body and splenium of corpus callosum (long thin arrow) and pontine hypogenesis (short thin arrow) with dorsal kink at the mesencephalic pontine junction (thick arrow). (c) Axial turbo inversion recovery (TIR) image shows presence of multiple subtle tiny cerebellar cysts (arrow).
nerves, the ventricular system, cavernous sinuses, cisternal spaces, patients with CSF rhinorrhea, brain tumors and spinal diseases.\textsuperscript{8,9} The sequence is ideal for demonstrating the presence of cerebellar cysts in CMDs on MR imaging.

Lack of consistent chromosomal anomalies in WWS makes its prenatal diagnosis difficult. However, fetal brain MR imaging and transvaginal fetal neurosonography can be used to detect lissencephaly.\textsuperscript{10,11} Thus, parents can be properly counseled regarding the poor prognosis of this disease.

**Conclusion**

Although rare, Walker-Warburg syndrome should be considered in the differential diagnosis of a hypotonic child with severely delayed milestones and ocular abnormalities. Brain MR imaging is a powerful tool to visualize the full spectrum of congenital brain anomalies in CMD. CISS sequence should be performed in every case for good visualization of cerebellar cysts.

**References**