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Radiological Imaging Findings of Walker-Warburg Syndrome

Walker-Warburg Sendromunun Radyolojik Görüntüleme Bulguları

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To the Editor

Walker-Warburg Syndrome (WWS); it is an autosomal recessive, fatal congenital muscular dystrophy with elevated creatinine phosphokinase values, accompanied by eye and brain anomalies (1). Most patients with WWS, the most severe form of congenital muscular dystrophies, are symptomatic at birth or infancy, and patients usually die before the age of 3 years. In this syndrome; a number of eye anomalies such as cataracts, microcornea, microphthalmia, lens defects, optic nerve atrophy or hypoplasia, glaucoma and retinal dysplasia can be seen. In addition, intracranial malformations such as type 2 (cobblestone) lissencephaly, hydrocephalus, hypomyelination in white matter, corpus callosum dysplasia, cerebellar dysplasia, presence of abnormal brain stem and encephalocele may be seen (2). Among the MRI findings, the most typical findings for WWS are; angulation at the level of brainstem-spinal cord junction, type 2 lissencephaly, cerebellar hypoplasia, corpus callosum hypoplasia and cerebellar cysts (3). It is defined as the Z-shape in the literature, different from what we have seen (4).

Microphthalmia and persistent hyperplastic primary vitreous (PHPV) were detected in the right eye in the MRI performed when the patient was 3 days old. There was dilatation in the third and fourth ventricles in both lateral ventricles, and the cerebellar vermis was found to be hypoplasic. In addition, with Type 2 (cobblestone) lissencephaly and interventricular septum agenesis, the brain stem was Z-shaped and the pons was hypoplasic (Figure 1).

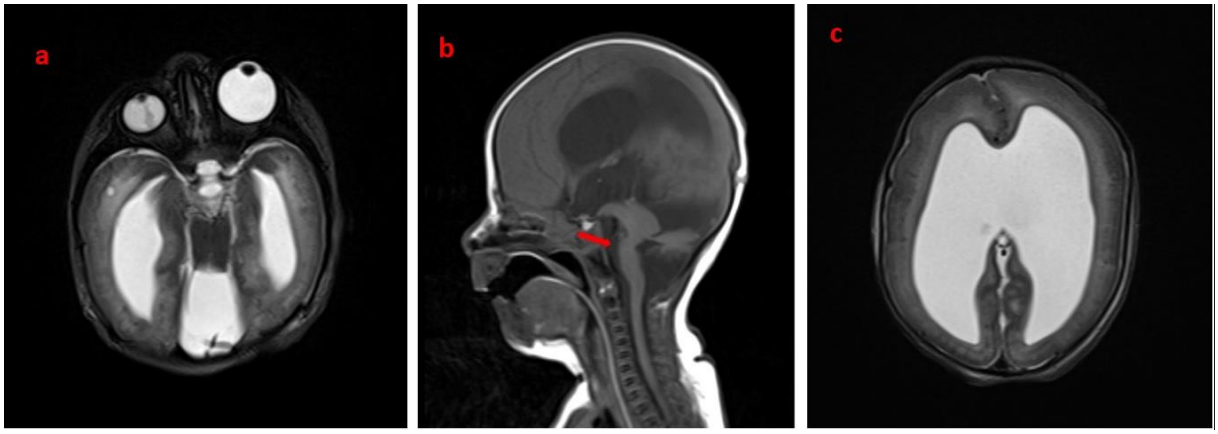

Figure 1 a. Axial T2-weighted image. Microphthalmia and PPHV in the right eye, both lateral ventricles dilated, infratentorial cystic structure behind the mesencephalon. **b.** Sagittal T1-weighted image. Angle (W-shaped) at the level of brain stem-spinal cord, hypoplasic cerebellum, dilated ventricles, infratentorial cystic structure behind the mesencephalon(Red arrow). **c.** Axial T2-weighted image. Both lateral ventricles dilated, septum pellucidum agenesis, type 2 (cobblestone) lissencephaly

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We submitted this letter to highlight the Z-shaped brainstem appearance, an imaging finding that is under-reported in the literature. In our opinion, this image should be called a "W-shape" rather than a Z shape.

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