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BILATERAL TOTAL RETINAL DETACHMENT AT BIRTH: A CASE REPORT OF WALKER-WARBURG SYNDROME

Navid Hakim[1], Cristina Soare[2] and **Jamil Hakim[2]**
[1]Princess Royal University Hospital, UK
[2]Queen Mary's Hospital, Sidcup, UK

Walker-Warburg syndrome (WWS) is a disorder characterized by ocular and brain malformations, and congenital muscular dystrophy. Retinal malformations are common in WWS; however bilateral retinal detachment is a rare occurrence. We present a case of a newborn baby delivered at 36+3 weeks, who was the first living child of consanguineous parents of Turkish origin. On antenatal anomaly scans, the fetus had hydrocephalus that had increased throughout pregnancy and a diagnosis of hydrancephaly was made at 36 weeks of gestation. Hypotonia, cleft lip and palate, poor suck and absent gag reflex were noted at birth. Ophthalmic examination at the age of two days revealed bilateral funnel retinal detachment. B-scan ultrasonography confirmed these findings and magnetic resonance imaging (MRI) of the brain was performed at the age of 13 days to establish a diagnosis. The MRI showed lissencephaly, hydrocephalus, thin rim of brain parenchyma, with a cobblestone appearance of the cortex and pontine and cerebellar hypoplasia, consistent with the diagnosis of WWS. The infant deteriorated and died at 39 days of age from complications associated with the brain anomalies. In summary, bilateral retinal detachment is extremely rare and in association with hydrocephalus and posterior fossa anomalies strongly suggests the diagnosis of WWS.

navidhakim@gmail.com