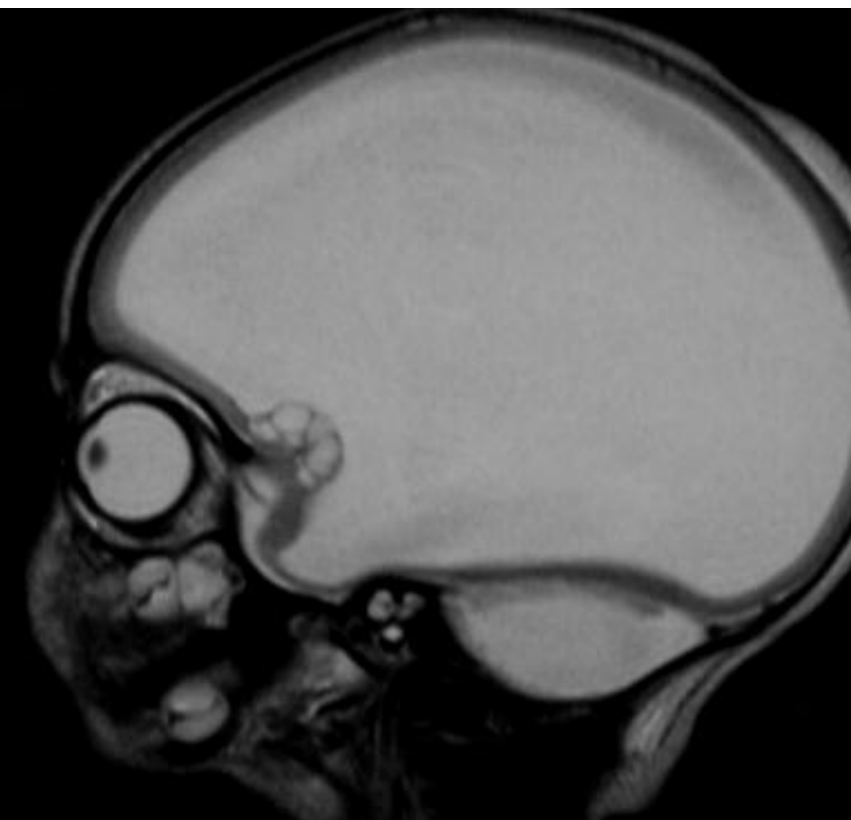


SWISS SOCIETY OF NEONATOLOGY

## Walker-Warburg syndrome

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This male infant was born to a 31-year-old G3/P3 by induced vaginal delivery at 37 4/7 weeks of gestation due to progressive fetal hydrocephaly. A complex cerebral malformation with hydrocephaly, agenesis of the corpus callosum had been diagnosed at 20 weeks of gestation. Cord-pH values were 7.24/7.33 and Apgar scores were 3 at 1, 6 at 5, and 6 at 10 minutes, respectively. The infant was noted to have increased muscle tone with marked hyperreflexia and poor respiratory effort. He was intubated at 20 minutes of age and transferred to the neonatal intensive care unit.

On admission to the NICU, the infant was paralyzed and mechanically ventilated. His birth weight was 2550 g (P10-25), his length was 46 cm (P3) and his head circumference was 35.5 cm (P75-90). There was a median palatal cleft. His cardiorespiratory status was unremarkable. His abdomen was soft without organomegaly. He had a small scrotum and penis, and his testicles were not palpable.

Initial laboratory investigations were non contributory. Chest X-ray was normal. An abdominal ultrasound was only remarkable for bilateral hyperechogenicity of the renal cortex. A cerebral ultrasound showed massive dilatation of the lateral ventricles (Fig. 1). Parenchymal thickness was reduced to 3 mm. There were bilateral hyperechogenic lesions containing multiple small cysts between the frontal and temporal lobes, which protruded into the ventricular lumen (Fig. 2).

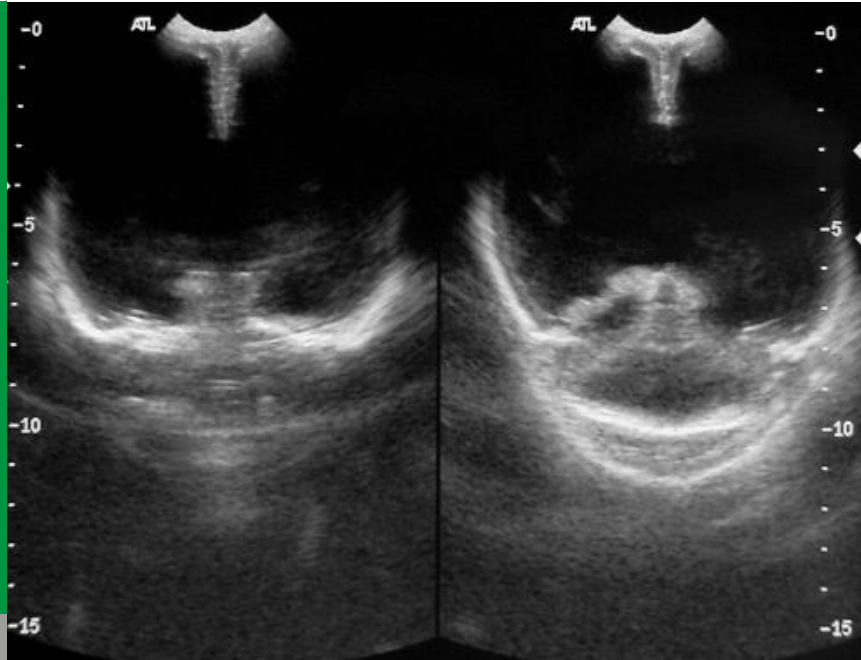


Fig. 1

*Cerebral ultrasound (coronal view): massive hydrocephalus.*

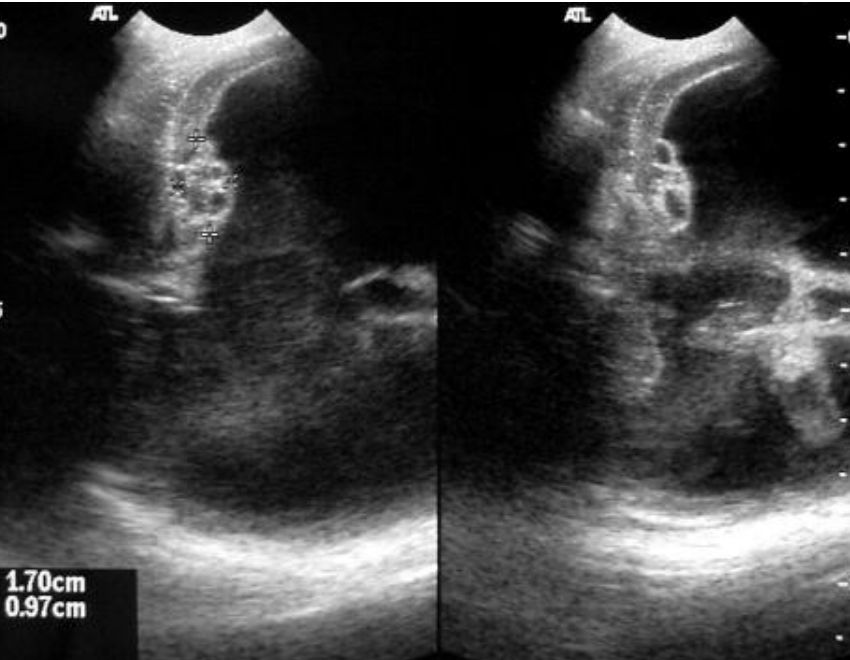


Fig. 2

*Cerebral ultrasound (parasagittal view): massive hydrocephalus.*

An MRI on the second day of life confirmed the US findings with massive hydrocephalus (Fig. 3), absence of gyri (Fig 4, 5), agenesis of corpus callosum, cerebellum and pons. The same cystic lesions seen on ultrasonography were again noted posteriorly to the orbitae (Fig. 3-5).

In the posterior cerebral fossa, there was a large cystic lesion posterior to a very hypoplastic and anteriorly displaced cerebellum (Fig. 6). There was a z-shaped deformation of the medulla oblongata and pontine hypoplasia (Fig. 6).

The suspected diagnosis of Walker-Warburg syndrome was further supported by the observation of bilateral retinal dystrophy and an elevated creatine kinase (882 U/l, normal < 195 U/l).

In view of the infant's undoubtedly poor prognosis, life support was withdrawn on the 2nd day of life following careful discussion with the parents. The infant died 4 hours later in the presence of his parents. Permission for autopsy was not given.



Fig. 3

*MRI (DOL 2): massive hydrocephalus.*



Fig. 4





Fig. 5

*MRI (DOL 2, T2-weighted image): massive hydrocephalus, absence of gyri, cystic retroorbital lesions.*



**Fig. 5**

*MRI (DOL 2, T1-weighted image): z-shaped deformation of the medulla oblongata.*

Initially described by Walker in 1942, the disorder was first suggested as a distinct disease entity by Warburg in 1971 (1). The full spectrum of associated defects was outlined by Pagon et al. (2) and Whitley et al. (3). All patients have type II lissencephaly with widespread agyria and scattered areas of macrogyria and/or polymicrogyria, absent or hypoplastic septum pellucidum and corpus callosum.

Approximately 50% of patients have a Dandy-Walker malformation, and hydrocephalus frequently develops due to mechanical obstruction in the posterior fossa. Retinal malformations have been described in all patients, including microphthalmia, retrolental masses, coloboma, and retinal detachment secondary to retinal dysplasia. In addition, all patients have congenital muscular dystrophy and genital anomalies in males are common. The cleft palate and possible mild renal dysplasia seen in our patient have also been described occasionally in patients with Walker-Warburg syndrome (4). This disorder has an autosomal recessive inheritance and the prognosis is very poor with the majority of affected children dying within the first year of life (5).

## REFERENCES

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