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Images in...

Walker–Warburg syndrome

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DESCRIPTION

A full-term male neonate born of a non-consanguineous marriage presented to us on the first day of life with large head (figure 1) and generalised hypotonia. He had microphthalmia and white reflex in pupillary areas. There was a family history of three neonatal deaths, the parents did not recall any of these babies having had large head.

His creatine kinase (CK) level was very high (3012 IU/l). A plain and contrast CT brain showed:

1. Lissencephaly and marked dilatation of lateral ventricle and 3rd ventricle with absent septum pellucidum and hypoplasia of corpus callosum (figure 2).
2. Fusion of colliculi with kinking of brain stem, hypoplasia of cerebellum and inferior vermis (figure 3).
3. A large cyst in superior cerebellar cistern communicating with 4th ventricle (figure 4).

Incidentally bilateral eyes showed hyperdense lens with hyperdense vitreous. These imaging features suggested a diagnosis of Walker–Warburg syndrome (WWS). WWS is a rare form of autosomal recessive congenital muscular dystrophy associated with brain and eye abnormalities. WWS has a worldwide distribution. The overall incidence is unknown but a survey in North-eastern Italy has reported an incidence rate of 1.2 per 100 000 live births.[1] It presents with congenital muscular dystrophy, type II lissencephaly, hydrocephalus[2] and eye abnormalities which include anterior eye anomalies (cataracts, shallow anterior chamber, microcornea and microphthalmia and lens defects) and a spectrum of posterior eye anomalies (retinal detachment or dysplasia, hypoplasia or atrophy of the optic nerve and macula and coloboma). Glaucoma or buphthalmos may be present.

A related autosomal recessive disorder, Fukuyama congenital muscular dystrophy, consists of similar but less severe brain changes and congenital muscular dystrophy.

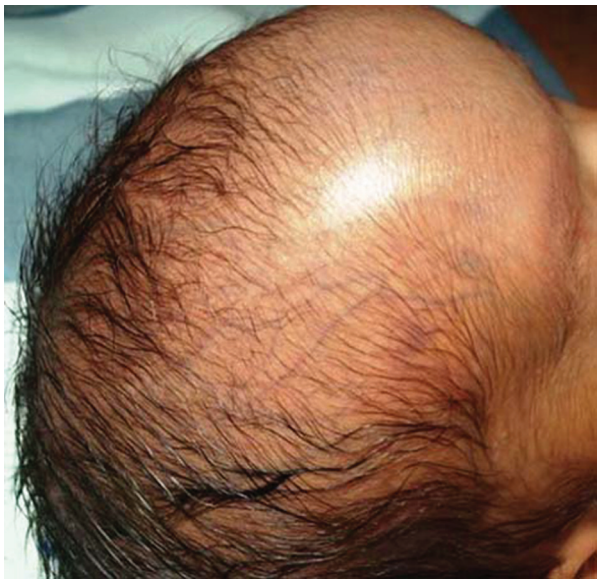

Figure 1 Large head.

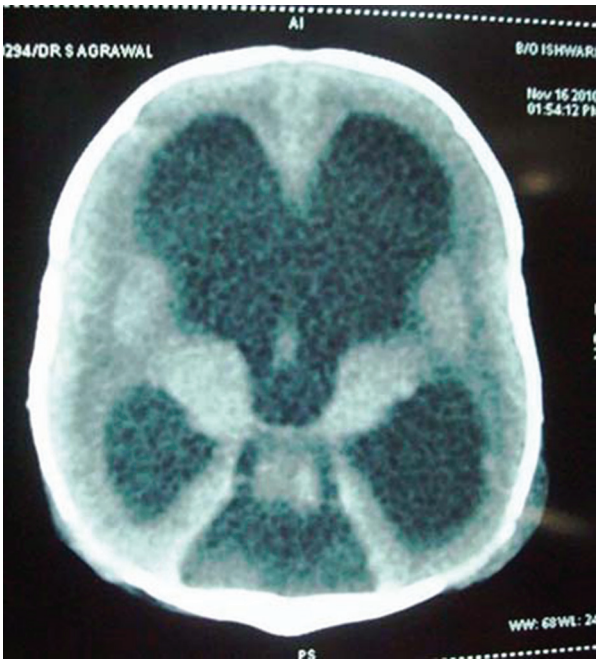

Figure 2 Lissencephaly, absent septum pellucidum, huge dilatation of lateral and 3rd ventricle.

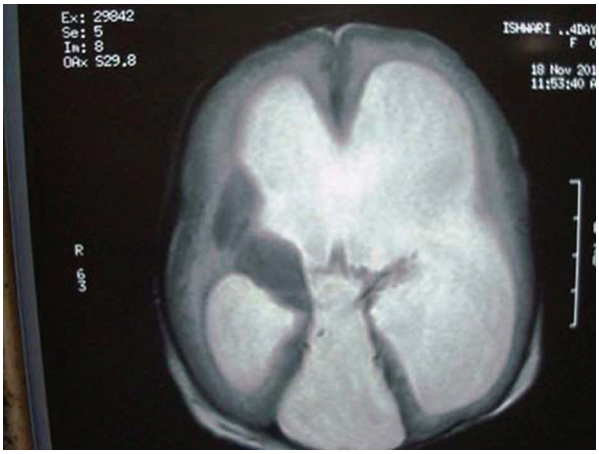

Figure 3 Huge cyst in the cerebellar cistern.

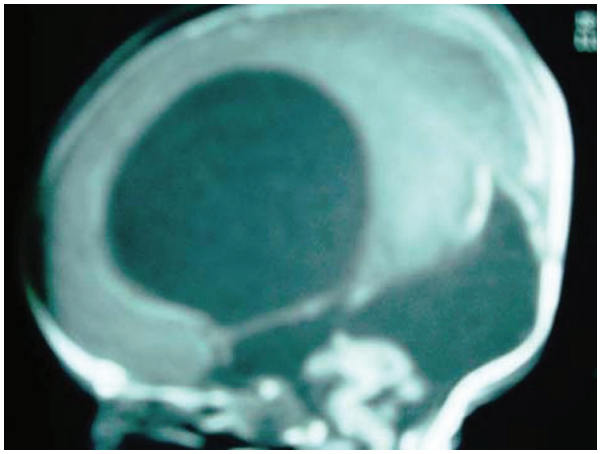

Figure 4 Kinking of brain stem.

It differs from WWS because of consistently less frequent and severe cerebellar and retinal abnormalities.[3]

Hydrocephalus, hypotonia, eye abnormalities, raised CK and the imaging characteristics described above should alert to the possibility of WWS.

Competing interests None.

Patient consent Obtained.

REFERENCES

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