Images in...

Walker–Warburg syndrome

Sanwar Agrawal

Department of Pediatrics, Ekta Institute of Child Health, Raipur, India

Correspondence to Dr Sanwar Agrawal, drsanwar50@gmail.com

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, hydrocephalus² 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.

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.

Figure 4 Kinking of brain stem.

REFERENCES

1. **Vajsar J**, Schachter H. Walker-Warburg syndrome. *Orphanet J Rare Dis* 2006;**1**:29.
2. **Warburg M**. Hydrocephaly, congenital retinal nonattachment, and congenital falciform fold. *Am J Ophthalmol* 1978;**85**:88–94.
3. **Dobyns WB**, Pagon RA, Armstrong D, *et al*. Diagnostic criteria for Walker-Warburg syndrome. *Am J Med Genet* 1989;**32**:195–210.

This pdf has been created automatically from the final edited text and images.

Copyright 2011 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit

<http://group.bmj.com/group/rights-licensing/permissions>.

BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Agrawal S. Walker–Warburg syndrome. *BMJ Case Reports* 2011;10.1136/bcr.04.2011.4102, date of publication

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow