Dandy-Walker Syndrome

Dandy-Walker syndrome (DWS) is a condition affecting brain development, primarily involving the cerebellum. Classic anatomical hallmarks of DWS are hypoplasia of the cerebellar vermis, antero-posterior enlargement of the posterior fossa, upward displacement of the torcula and transverse sinuses, and cystic dilatation of the fourth ventricle. It may also form part of the PHACES syndrome (Posterior fossa anomalies as Dandy-Walker malformation (DWM); Haemangioma; Arterial lesions of the head and neck; Cardiac abnormalities as aortic coarctation; Eye abnormalities and Sternal defect).

A large number of associated problems may also be present, such as hydrocephalus (often develops postnatally), atresia of the foramen of Magendie and atresia of the foramen of Luschka.

Epidemiology

- Incidence is approximately 1 in 25-35,000 live births. It occurs slightly more frequently in females than in males.
- Marked variation has been shown in the genetics and aetiology. Most cases of DWS are sporadic, although familial occurrence has been reported and several individuals with de novo interstitial deletions of 3q have been identified.
- DWS may result from chromosomal anomalies or environmental factors. Recognised chromosomal abnormalities associated with DWS include trisomies (trisomy 18, 13, 21 or 9) and triploidy. Recognised single gene defects associated with DWS include Walker-Warburg syndrome or Meckel's syndrome and in these cases the recurrence risk for siblings may be high.
- Associated environmental factors include first-trimester exposure to rubella, cytomegalovirus, toxoplasmosis or warfarin. Maternal diabetes during pregnancy is also associated with increased risk. When the evidence suggests that there is no association with a Mendelian or chromosomal disorder then the recurrence risk is relatively low at between 1% and 5%.

Presentation

- Symptoms often occur in early infancy with 80-90% of individuals becoming symptomatic in the first year of life. Most children have hydrocephalus with abnormally rapid increase in head circumference with bulging at the back of the skull. Development is often affected, particularly with delay in motor skills such as crawling and walking. Spastic paraplegia and seizures may be present.
- In 10-20% of individuals with DWS, signs and symptoms do not appear until late childhood or adulthood. The presentation is different and may be with signs of increased intracranial pressure, such as irritability, vomiting, headaches and convulsions. There may also be signs of cerebellar dysfunction, such as unsteadiness, lack of muscle co-ordination, or jerky movements of the eyes.
- One case report describes conduct disorder, hyperactivity, stereotypical movements and nocturnal enuresis in a 14 year-old.
- Some individuals with malformations characteristic of DWS have no symptoms and the diagnosis may be made incidentally because of neuroimaging done for another reason.
Investigations

- The diagnosis of DWM can be made by antenatal ultrasound.\[10, 1\]
- MRI scan allows a detailed evaluation of DWM lesions and complications. MRI evaluation can then be used antenatally to confirm the diagnosis and to gain more detailed information.\[11\]

Associated abnormalities

DWS is frequently associated with other central nervous system abnormalities including dysgenesis of corpus callosum, ectopic brain tissue, holoprosencephaly and neural tube defects.\[12\]

Other associated abnormalities include heart defects, urogenital malformations, polydactyly or syndactyly and abnormal facial features.

Management

- Treatment involves managing the associated problems - eg, seizure management.
- Hydrocephalus plays an important role in development of symptoms and neurological outcome. Surgical interventions to control the hydrocephalus are the most important part of management. This involves inserting a shunt. This may be to shunt the cyst (cystoperitoneal), the ventricles (ventriculoperitoneal), or both. Endoscopic procedures are also considered an acceptable alternative.\[13\]
- Parents of children with DWS will benefit from genetic counselling if they intend to have more children.

Prognosis

- The effect of DWS on intellectual development is variable. Some children have normal cognitive development while others may never have normal intellectual development, even when the hydrocephalus is treated early.
- Prognosis otherwise depends on the severity of the syndrome and associated malformations.
- A population-based study looking at natural history of DWS showed that mortality is significantly high (ten-fold) in children with DWS compared to controls. However, surgical CSF drainage procedures are beneficial and halve the mortality.\[14\]

Further reading & references

4. Dandy-Walker Syndrome, DWS; Online Mendelian Inheritance in Man (OMIM)
6. What is Dandy-Walker syndrome?; Genetics Home Reference

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Document ID: 1356 (v23)
Last Checked: 29/09/2015
Next Review: 27/09/2020

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