

Dandy Walker Syndrome:

Dandy-Walker Syndrome is a congenital brain malformation involving the cerebellum and the fourth ventricle. Insults of varying severity to the cerebellar hemispheres and fourth ventricle are believed to be the genesis. Nature and cause are unknown.

History:

Described by Dandy and Blackfan in 1914.

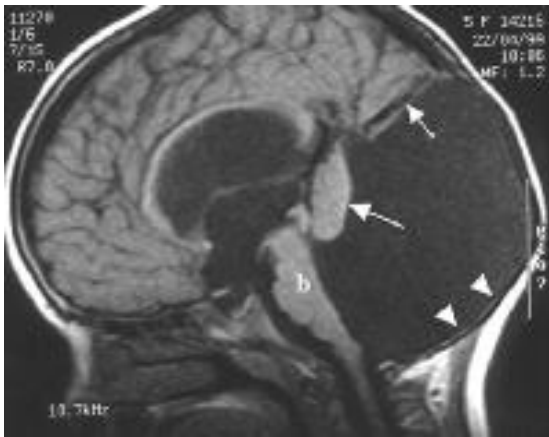
1954 Benda emphasized that atresia of the cerebellar outlet foramina is not an essential feature.

1963 & 1972: D'Agostino and Hart respectively defined the triad as noted below.

Key features of this syndrome are:

- Cystic dilation of the fourth ventricle
 - agenesis or hypoplasia of the *cerebellar vermis*
 - enlargement of the posterior fossa
- * Supratentorial hydrocephalus is considered a complication, not part of the malformation.

- Approximately 70-90% of these patients have hydrocephalus which develops post-natally. The syndrome can appear dramatically or develop unnoticed.



The hypoplastic vermis is everted over the posterior fossa cyst (large arrow). The cerebellar hemispheres and brainstem (b) are hypoplastic.



large cerebrospinal fluid cyst in the posterior fossa and hypoplastic cerebellar hemispheres with a winged appearance (c)

Inheritance:

Etiology is heterogeneous however familial occurrence has been reported.

Predisposing factors:

Rubella, cytomegalovirus, toxoplasmosis, warfarin (Coumadin), alcohol, and isotretinoin.

Frequency:

1 in 25,000 births; F > M

Morbidity/Mortality

12-50% mortality rate – improved now with placement of shunts.

Prognosis is usually only moderately favorable

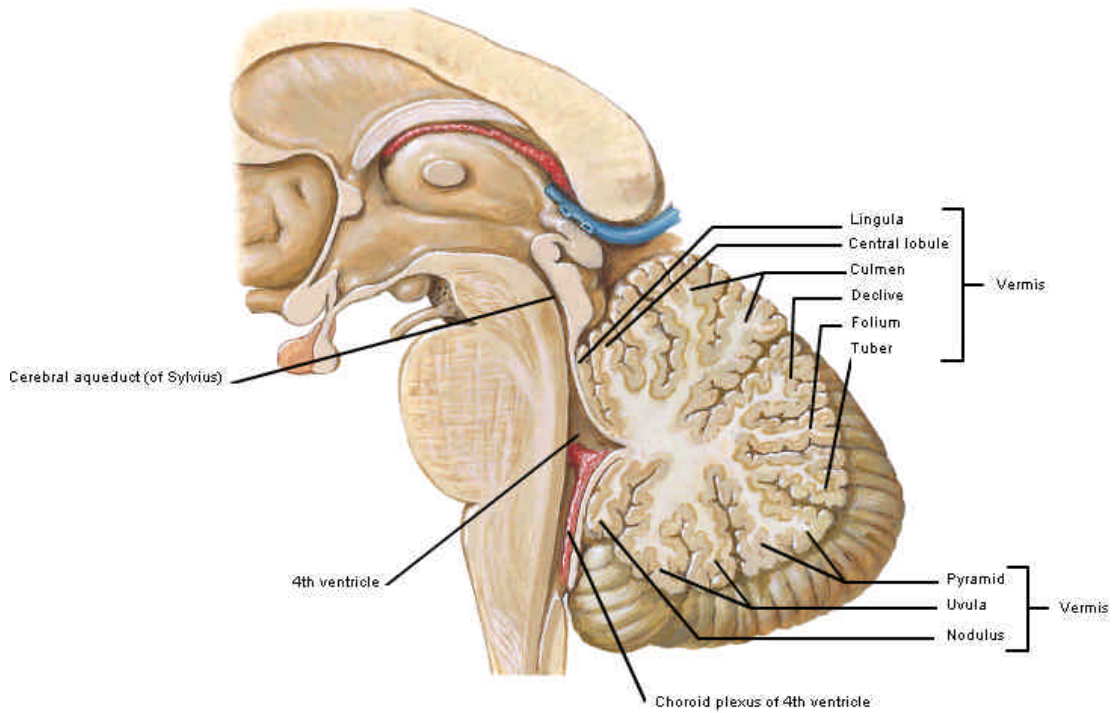
Clinical presentation:

- Usually present with developmental delay, enlarged head circumference, or signs and symptoms of hydrocephalus.
- Difficulty with balance, spasticity, and poor fine motor control are common
- Hearing or visual difficulties, systemic abnormalities, and CNS abnormalities are associated with poor intellectual development.
- An estimated 80% of patients had normal ventricles at birth, and by age 1 year, 80% had ventriculomegaly.

Dx: T1 weighted MRI is best diagnostic study for evaluation. CT is useful to rule out other types of hydrocephalus.

Dandy-Walker Syndrome is frequently associated with disorders of other areas of the central nervous system including absence of the *corpus callosum* (the connecting area between the two cerebral hemispheres, and malformations of the heart, face, limbs, fingers and toes.

- D'Agostino AN, Kernohan J W, Brown JR: The Dandy-Walker syndrome. J Neuropath Exp Neurol 1963; 22: 450-70.
- Bordarier C, Aicardi J: Dandy-Walker syndrome and agenesis of the cerebellar vermis: diagnostic problems and genetic counselling. Dev Med Child Neurol 1990 Apr; 32(4): 285
- Murray JC, Johnson JA, Bird TD: Dandy-Walker malformation: etiologic heterogeneity and empiric recurrence risks. Clin Genet 1985 Oct; 28(4): 272-83
- National Institute of Neurological Disorders and Stroke
National Institutes of Health
Bethesda, MD 20892



Mega cisterna magna

- Normal cerebellar hemispheres and vermis
- Large retrocerebellar CSF collection communicates with fourth ventricle
- Normal fourth ventricle
- Occasionally, quite enlarged posterior fossa