Dandy-Walker malformation (DWM) is a brain malformation that occurs during embryonic development of the cerebellum and 4th ventricle. The cerebellum is the area of the brain that helps coordinate movement, and is also involved with cognition and behavior. The 4th ventricle is a space around the cerebellum that channels fluid from inside to around the outside of the brain. DWM is characterized by underdevelopment (small size and abnormal position) of the middle part of the cerebellum known as the cerebellar vermis, cystic enlargement of the 4th ventricle and enlargement of the base of the skull (posterior fossa). DWM is sometimes (20-80%) associated with hydrocephalus, in which blockage of the normal flow of spinal fluid leads to excessive amounts of fluid accumulating in and around the brain. This leads to abnormally high pressure within the skull and swelling of the head, and can lead to neurological impairment. DWM results from defects in early embryonic development of the cerebellum and surrounding structures. A few patients have chromosome abnormalities including deletion of chromosome 3q24.3 (the location of the first DWM genes, known as ZIC1 and ZIC4), 6p25 or 13q32.2-q33.2, or duplication of 9p. In the remainder, it is probably due to other more complex genetic and perhaps environmental factors (teratogens) as the recurrence risk in siblings less than 5%. A few examples of affected siblings with isolated Dandy-Walker malformation have been reported, suggesting autosomal recessive or X-linked inheritance, but most of these are probably CVH and not typical DWM. In these families, the recurrence risk is higher, up to 25%. DWM may also occur as part of a genetic syndrome that includes multiple birth defects, such as the PHACES syndrome of facial hemangioma, heart and sternal defects and DWM. Many other syndromes and chromosome abnormalities have been reported with DWM, but most of these appear to have CVH rather than typical DWM. The frequency of Dandy Walker malformation in the US is approximately 1 per 25,000 -35,000 live births and affects more females than males. Dandy Walker malformation is diagnosed with the use of ultrasound, CT and MRI. Prenatal diagnosis of Dandy-Walker malformation is sometimes made by ultrasound or fetal MRI.