

Dandy-Walker malformation

Description

Dandy-Walker malformation affects brain development, primarily development of the cerebellum, which is the part of the brain that coordinates movement. In individuals with this condition, various parts of the cerebellum develop abnormally, resulting in malformations that can be observed with medical imaging. The central part of the cerebellum (the vermis) is absent or very small and may be abnormally positioned. The right and left sides of the cerebellum may be small as well. In affected individuals, a fluid-filled cavity between the brainstem and the cerebellum (the fourth ventricle) and the part of the skull that contains the cerebellum and the brainstem (the posterior fossa) are abnormally large. These abnormalities often result in problems with movement, coordination, intellect, mood, and other neurological functions.

In the majority of individuals with Dandy-Walker malformation, signs and symptoms caused by abnormal brain development are present at birth or develop within the first year of life. Some children have a buildup of fluid in the brain (hydrocephalus) that may cause increased head size (macrocephaly). Up to half of affected individuals have intellectual disability that ranges from mild to severe, and those with normal intelligence may have learning disabilities. Children with Dandy-Walker malformation often have delayed development, particularly a delay in motor skills such as crawling, walking, and coordinating movements. People with Dandy-Walker malformation may experience muscle stiffness and partial paralysis of the lower limbs (spastic paraplegia), and they may also have seizures. While rare, hearing and vision problems can be features of this condition.

Less commonly, other brain abnormalities have been reported in people with Dandy-Walker malformation. These abnormalities include an underdeveloped or absent tissue connecting the left and right halves of the brain (agenesis of the corpus callosum), a sac-like protrusion of the brain through an opening at the back of the skull (occipital encephalocele), or a failure of some nerve cells (neurons) to migrate to their proper location in the brain during development. These additional brain malformations are associated with more severe signs and symptoms.

Dandy-Walker malformation typically affects only the brain, but problems in other systems can include heart defects, malformations of the urogenital tract, extra fingers or toes (polydactyly) or fused fingers or toes (syndactyly), or abnormal facial features.

In 10 to 20 percent of people with Dandy-Walker malformation, signs and symptoms of

the condition do not appear until late childhood or into adulthood. These individuals typically have a different range of features than those affected in infancy, including headaches, an unsteady walking gait, paralysis of facial muscles (facial palsy), increased muscle tone, muscle spasms, and mental and behavioral changes. Rarely, people with Dandy-Walker malformation have no health problems related to the condition.

Problems related to hydrocephalus or complications of its treatment are the most common cause of death in people with Dandy-Walker malformation.

Frequency

Dandy-Walker malformation is estimated to affect 1 in 10,000 to 30,000 newborns.

Causes

Researchers have found mutations in a few genes that are thought to cause Dandy-Walker malformation, but these mutations account for only a small number of cases. Dandy-Walker malformation has also been associated with many chromosomal abnormalities. This condition can be a feature of some conditions in which there is an extra copy of one chromosome in each cell (trisomy). Dandy-Walker malformation most often occurs in people with trisomy 18 (an extra copy of chromosome 18), but can also occur in people with trisomy 13, trisomy 21, or trisomy 9. This condition can also be associated with missing (deletions) or copied (duplications) pieces of certain chromosomes. Dandy-Walker malformation can also be a feature of genetic syndromes that are caused by mutations in specific genes. However, the brain malformations associated with Dandy-Walker malformation often occur as an isolated feature (not associated with other health problems), and in these cases the cause is frequently unknown.

Research suggests that Dandy-Walker malformation could be caused by environmental factors that affect early development before birth. For example, exposure of the fetus to substances that cause birth defects (teratogens) may be involved in the development of this condition. In addition, a mother with diabetes is more likely than a healthy mother to have a child with Dandy-Walker malformation.

[Learn more about the gene associated with Dandy-Walker malformation](#)

- FOXC1

Additional Information from NCBI Gene:

- ZIC1
- ZIC4

Inheritance

Most cases of Dandy-Walker malformation are sporadic, which means they occur in people with no history of the disorder in their family. A small percentage of cases seem to run in families; however, Dandy-Walker malformation does not have a clear pattern of inheritance. Multiple genetic and environmental factors likely play a part in determining the risk of developing this disorder. First-degree relatives (such as siblings and children) of people with Dandy-Walker malformation have an increased risk of developing the condition compared with people in the general population.

When Dandy-Walker malformation is a feature of a genetic condition, it is inherited in the pattern of that condition.

Other Names for This Condition

- Dandy-Walker complex
- Dandy-Walker cyst
- Dandy-Walker deformity
- Dandy-Walker syndrome
- DWM
- DWS
- Hydrocephalus, internal, Dandy-Walker type
- Hydrocephalus, noncommunicating, Dandy-Walker type
- Luschka-Magendie foramina atresia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Dandy-Walker syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0010964/>)

Genetic and Rare Diseases Information Center

- Isolated Dandy-Walker malformation (<https://rarediseases.info.nih.gov/diseases/6242/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

- DANDY-WALKER SYNDROME; DWS (<https://omim.org/entry/220200>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28Dandy-Walker+syndrome%5BTIAB%5D%29+OR+%28Dandy-Walker+malformation%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

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