Dandy-Walker Syndrome [1]

By: Bohnenberger, Alexandra Keywords: Dandy-Walker Syndrome [2] Human development [3]

Dandy-Walker Syndrome is a congenital brain defect in humans [4] characterized by malformations to the cerebellum [5], the part of the brain that controls movement, and to the ventricles, the fluid-filled cavities that surround the cerebellum [5]. The syndrome is named after physicians Walter Dandy and Arthur Walker, who described associated signs and symptoms of the syndrome in the 1900s. The malformations often develop during embryonic stages. In early infancy, symptoms include slow motor development and a progressive enlargement of the skull due to cerebrospinal fluid accumulation called hydrocephalus [6]. The prognosis of Dandy-Walker syndrome is highly variable, ranging from minor or negligible birth defects to profound malformations, disability, or early death.

Walter Dandy was a neurosurgeon in the early 1900s in the US who studied the cause of the enlargement of the head associated with what researchers later called Dandy-Walker syndrome. In 1910, Dandy reported that he had observed a thirteen-month-old female with a fluid-filled sac, called a cyst, causing the enlargement of the fourth ventricle, as well as anomalies in the cerebellum [5]. The infant had a high fever and convulsions, followed by the enlargement of the head three weeks after the onset of symptoms. The infant died two months after Dandy's observations.

During the infant's autopsy, Dandy found severe fluid build-up in the brain, called hydrocephalus [6]. Dandy noticed a fluid-filled sac, called a cyst, within the fourth ventricle surrounding the cerebellum [5]. The cyst, he observed, obstructed the flow of the cerebral spinal fluid that fills the ventricles, causing the swelling of the brain. Dandy noted that the medial part of the cerebellum [5], the cerebellar vermis [7], was also small. He found that two normal anatomical channels (called foramina of Magendie and Luschka) were absent, preventing cerebrospinal fluid from exiting the fourth ventricle. Based on those observations, Dandy concluded that the build-up of cerebrospinal fluid was caused by the absence of the foramina of the fourth ventricle, not allowing for cerebral spinal fluid to drain from the brain.

Arthur Walker was a neurosurgeon at Johns Hopkins Hospital [8] in Baltimore, Maryland, who studied cases of congenital defects that mirrored Dandy's descriptions. Walker, like Dandy, noted the partial absence of the cerebellar vermis [7] in addition to the absence of the foramina of Magendie and Luschka. However, in 1942, Walker added the partial absence of the cerebellar vermis [7] as a symptom of the congenital condition.

In 1954, Clemens Ernst Brenda designated the term Dandy-Walker Syndrome to describe the type of hydrocephalus [6] caused by an absence of the foramina of Magendie and Luschka which Dandy and Walker had independently identified. However, Brenda noted that although the obstructions of the foramina of Magendie and Luschka characterized Dandy-Walker Syndrome, the blockage may not have been the main cause of the syndrome. He hypothesized that the syndrome consists of a developmental anomaly of the fourth ventricle, not necessarily the foraminal blockage.

Researchers and physicians of the 1960s and 70s continued investigating the symptoms associated with Dandy-Walker Syndrome. They expanded the defining characteristics of Dandy-Walker syndrome to include not only the complete or partial absence of the cerebellar vermis [7], but also the dilation of the fourth ventricle and the enlargement of the cavity holding the cerebellum [5] and the brainstem, called the posterior fossa. Into the twenty-first century, medical experts continued to revise the characteristics of Dandy-Walker Syndrome.

Doctors used tools to diagnose Dandy-Walker syndrome. Technicians sometimes detected the syndrome during pregnancy [9] using an ultrasound [10], a tool that used sound waves to create an image of something inside a body. If a doctor identified a fetus [11] with possible Dandy-Walker syndrome, she often ordered further tests such as amniocentesis or a fetal echocardiogram [12]. Amniocentesis [13] is a prenatal medical procedure that doctors use to identify genetic birth defects [14] that tests for chromosomal abnormalities and infections. An echocardiogram was a form of ultrasound [10] that picked up sound waves of a beating heart to identify possible rhythmic abnormalities.

After birth, Dandy-Walker Syndrome can cause other malformations and developmental delays. Within the first year, symptoms include the enlargement of the skull, poor motor development, and impaired development of speech and language. The syndrome is often accompanied by other birth deformities of the brain, heart, face, and limbs, as well as additional central nervous system [15] anomalies. A common central nervous system [15] anomaly is the absence of the corpus callosum [16], a bundle of nerve fibers connecting the two hemispheres of the brain.

Diagnostic tools after the infant is born differed from prenatal diagnostic tools for detecting Dandy-Walker syndrome. Those tools included computerized tomography (CT) scans, and magnetic resonance imaging (MRI) scans. CT scans used x-ray [17] radiation [18] to produce images of cross-sections of the brain. Doctors used CT scans to assess the size and shape of ventricles, and they inferred the degree of obstruction to the cerebrospinal fluid flow. CT scans also showed acute bleeding and detected obstructions of the ventricles within the brain. Unlike CT scans, MRI scans used magnetic fields and radio waves rather than possibly damaging x-rays to create images of internal structures. Those anatomical pictures allowed doctors to detect brain malformations and the quality of the cerebellar vermis [7]. The images from any scanning technology were used to reveal the type of malformation causing the obstruction of cerebrospinal fluid or delays in the development of the infant.

Doctors treated Dandy-Walker Syndrome mainly by relieving the associated symptoms, such as reducing pressure in the cranial cavity created by the cerebrospinal fluid build-up from hydrocephalus [6] and managing brain abnormalities. Physicians sometimes drained the cerebrospinal fluid from hydrocephalus [6] using several methods. A common method was the ventriculoperitoneal shunt, which used a catheter that surgeons pass through a hole made through the skull into the brain. Surgeons connected the catheter to a valve and tunneled a second catheter to reach the peritoneal cavity within the abdomen where the cerebrospinal fluid is diverted and reabsorbed. Alternatively, the ventricles were drained to other cavities such as those in the heart and the lungs. Another procedure was endoscopic third ventriculostomy, a procedure in which a doctor made a small perforation in the third ventricle, allowing cerebrospinal fluid to escape from the blocked ventricular system and normalized pressure on the brain without using a shunt.

In the early twenty-first century, the outcome for infants with Dandy-Walker syndrome varied. Overall mortality for patients with the syndrome was about twenty-seven percent with most deaths attributable to associated malformations, uncontrolled hydrocephalus [6], shunt malfunction, or infection. Physicians also reported sudden death in multiple cases. Some studies suggest that the sudden death may be due to an inadequate blood supply to the brain caused by obstructing malformations. Many surviving patients were developmentally delayed and mentally disabled. However, those without additional birth defects [14] often developed normally and did not experience the more severe symptoms of Dandy-Walker Syndrome.

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Subject

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