

"Wildervanck syndrome, also known as cervicooculoacoustic syndrome, is a rare genetic disorder that primarily affects females. The disorder is characterized by a skeletal condition known as Klippel-Feil syndrome (KFS); abnormalities of certain eye (ocular) movements (i.e., Duane syndrome); and/or hearing impairment that is present at birth (congenital). In individuals with KFS, there is abnormal union or fusion of two or more bones of the spinal column (vertebrae) within the neck (cervical vertebrae). Duane syndrome is characterized by limitation or absence of certain horizontal eye movements; retraction or ""drawing back"" of the eyeball into the eye cavity (orbit) upon attempting to look inward; and, in some cases, abnormal deviation of one eye in relation to the other (strabismus). In some affected individuals, additional physical abnormalities may also be present. In most cases, Wildervanck syndrome appears to occur randomly for unknown reasons (sporadically). As mentioned above, Wildervanck syndrome primarily affects females. Since the disorder was originally described in 1952 (L.S. Wildervanck), almost 90 cases have been reported in the medical literature. According to some reports, approximately one percent of females with hearing impairment may be affected by Wildervanck syndrome. Wildervanck syndrome may be detected at birth or during the first year of life based upon a thorough clinical evaluation, identification of characteristic physical findings, and specialized tests. Diagnostic studies may include advanced imaging techniques, such as computerized tomography (CT) scanning or magnetic resonance imaging (MRI). During CT scanning, a computer and x-rays are used to create a film showing cross-sectional images of internal structures. An MRI uses a magnetic field and radio waves to form detailed cross-sectional images of certain organs and tissues. Such techniques may help to detect and characterize abnormalities of the inner ear, union or fusion of certain bones of the spinal column (e.g., cervical vertebrae), possible impingement of vertebrae on the spinal cord, or other abnormalities potentially associated with the disorder. In addition, in some individuals with Wildervanck syndrome, exploratory surgery may be conducted to detect malformations of the middle ear (exploratory tympanotomy). Additional specialized tests may also be performed to confirm or characterize other abnormalities that may be associated with the disorder (e.g., certain ocular findings, congenital heart defects, renal abnormalities, etc.)."