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Wyburn Mason Syndrome

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Synonyms of Wyburn Mason Syndrome

- Bonnet-Dechaumme-Blanc syndrome

Disorder Subdivisions

- No subdivisions found.

General Discussion

Wyburn-Mason syndrome is an extremely rare nonhereditary disorder that is present at birth (congenital). Affected infants have arteriovenous malformations (AVMs), which are developmental abnormalities affecting the blood vessels, specifically the arteries, veins and capillaries. Arteries typically carry oxygen-rich blood from the heart to body cells, while veins transport oxygen-deficient blood to the heart and lungs for the exchange of oxygen and carbon dioxide. The network of very tiny blood vessels (capillaries) that normally connects arteries and veins may be absent and the arteries and veins may be directly linked together. Larger AVMs may consist of a tangled mass of abnormal or malformed blood vessels. AVMs associated with Wyburn-Mason syndrome are usually found in the eyes and midbrain. The exact cause of Wyburn-Mason syndrome is unknown.

Symptoms

The symptoms associated with Wyburn-Mason syndrome vary greatly among affected individuals based upon the specific number and location(s) of associated arteriovenous malformations. Affected infants may have abnormalities affecting the eyes, central nervous system and, in rare cases, the skin.

In Wyburn-Mason syndrome, AVMs may range from absence of the capillaries to the presence of large masses of widened, twisted, tangled blood vessels known as a racemose hemangioma. Absence of capillaries results in the abnormal, direct connection of the arteries to the veins. This abnormal connection can result in excessive blood flow.

AVMs in Wyburn-Masons syndrome often affect the thin layer of nerve cells that lines the back of the eyes (retina). In some cases, an AVM may extend into the eye socket (orbit) or midbrain. The specific symptoms associated with an ocular AVM vary depending upon the exact location and extent the abnormality. Small AVMs affecting tiny blood vessels may not cause any symptoms (asymptomatic) and may be difficult to detect. Large AVMs such as a racemose hemangioma may cause significant loss of vision, usually from lack of blood flow to the retina (retinal ischemia).

Additional eye abnormalities may occur in some individuals with Wyburn-Mason syndrome including forward displacement of the eyeball so that the eyes bulge (proptosis), drooping of the upper eyelid (blepharoptosis), abnormally widened (dilated) blood vessels of the thin membrane that covers the outer surface of the eye (conjunctiva), and nerve paralysis (palsies).

AVMs of the central nervous system may not cause any symptoms (asymptomatic) or can cause severe symptoms. Although AVMs are present at birth, in many cases they may not cause symptoms until the second or third decade of life or even later. Neurological symptoms associated with Wyburn-Mason syndrome include severe headaches, vomiting, seizures, paralysis (palsy) of various cranial nerves and neck stiffness (nuchal rigidity). Spontaneous bleeding (hemorrhaging) of these lesions can lead to the sudden onset of symptoms. If the bleeding is severe, it can cause partial or full paralysis of one side of the body (hemiparesis or hemiplegia) or even death.

In rare cases, the skin may be involved in Wyburn-Mason syndrome including the formation of small bumps or clusters of blood vessels (angiomas) on the face. If the jaw bones are involved, dental procedures can lead to excessive bleeding. In rare cases, other areas of the body have developed AVMs including the lungs or the kidneys or other bones and muscles.

Causes

The exact cause of Wyburn-Mason syndrome is unknown. It is considered a developmental abnormality characterized by arteriovenous malformations. No specific genetic abnormality or hereditary tendencies have been identified. The specific, underlying mechanism(s) that cause the vascular malformations in Wyburn-Mason syndrome are not known. However, they are thought to result from abnormalities of blood vessel development during embryonic or fetal growth.

Affected Populations

Wyburn-Mason syndrome is an extremely rare disorder that appears to affect males and females in equal numbers. The incidence or prevalence rates of Wyburn-Mason syndrome in the general

population are unknown. The disorder is named for the investigator (Dr. R. Wyburn-Mason) who extensively described the disease entity in 1943.

Wyburn-Mason syndrome is sometimes grouped with the phakomatoses or neurocutaneous syndromes. This broad group of disorders is characterized by masses or tumors that may grow in the brain, spinal cord and other organs. In children, skin lesions are also prominent. Unlike other so-called phakomatoses, Wyburn-Mason syndrome rarely has skin abnormalities.

Related Disorders

Symptoms of the following disorders can be similar to those of Wyburn-Mason syndrome. Comparisons may be useful for a differential diagnosis.

Sturge-Weber syndrome is a rare inherited disorder characterized by the presence of a port wine colored birthmark (angioma) on the facial area and intracranial abnormalities that are present at birth (congenital). Affected infants may also have an enlarged head (macrocephaly). Generalized seizures and additional neurological symptoms usually occur between one and two years of age. Vascular lesions (telangiectasias and angiomas) in the brain usually involve the occipital or parieto-occipital regions. Glaucoma may be present in the eye located on the same side of the face where the port wine stain occurs. This eye may also be enlarged. The iris may remain blue, even though the other eye may change to another color as the infant matures. Sight in half of the visual field may be defective or absent in the affected eye. (For more information on this disorder, choose "Sturge Weber" as your search term in the Rare Disease Database.)

Von Hippel-Lindau disease is an autosomal dominant condition characterized by multiple localized tissue malformations called hemangioblastomas and angiomas. These growths may be found in the retina, brain, kidneys, adrenal glands, and other organs. Symptoms may include headaches, dizziness and difficulty coordinating muscle movement (ataxia). Chronic high blood pressure (hypertension) can also occur. The disorder may begin during young adulthood or may develop during early childhood. Aneurysms (bulges in blood vessels) may develop and form an angioma (nodule) that resembles a balloon. Benign tumors (pheochromocytomas) of the adrenal glands may be present as well, causing chronic high blood pressure, pounding heartbeat, headache, cold hands and feet, and excessive sweating. This condition is found equally in males and females and all ethnic groups. (For more information on this disorder, choose "von Hippel Lindau" as your search term in the Rare Disease Database.)

Vascular malformations or angiomas are localized collections of blood vessels that are abnormal in structure or number, lead to altered blood flow, and are not cancerous (nonneoplastic). Most vascular malformations are present at birth (congenital) and are suspected to arise between three and eight weeks of gestation, but the specific defect in embryogenesis has not yet been identified for each type of malformation. Some vascular malformations are not congenital, but caused by trauma, radiation, or other injury to the spinal cord. They are typically classified by size, location, and type of change, with the four most common being capillary telangiectasias, cavernous malformations, venous malformations, and arteriovenous malformations. Vascular malformations are sporadic and solitary in the majority of affected persons; however documented cases of autosomal dominant forms exist as well.

Standard Therapies

Diagnosis
A diagnosis of Wyburn-Mason syndrome may be made based upon a thorough clinical evaluation, a detailed patient history, and identification of characteristic findings, especially ocular findings. Imaging studies such as a computed tomography (CT) scan or magnetic resonance imaging (MRI) may be performed to detect potentially dangerous central nervous system (CNS) malformations. During CT scanning, a computer and x-rays are used to create a film showing cross-sectional images of certain tissue structures. An MRI uses a magnetic field and radio waves to produce cross-sectional images of particular organs such as the brain.

Treatment
No specific treatment for Wyburn-Mason syndrome exists. Treatment is directed toward the specific symptoms that are apparent in each individual. Some AVMs may not require treatment, especially retinal lesions which usually remain stable. If lesions in the eyes cause bleeding (hemorrhaging) in the retina or the clear, jelly-like substance that fills the middle of the eye (vitreous), laser treatment or the use of extreme cold to destroy abnormal tissue (cryosurgery) may be performed in an attempt to control the bleeding. Surgical removal of the vitreous (vitrectomy) has been performed in some cases if bleeding is persistent, although surgery is controversial.

Investigational Therapies

AVMs inside the cranial compartment of the skull (intracranial AVMs) may be treated in one of three ways. Many intracranial AVMs may be completely removed with a surgical procedure. Some intracranial AVMs, because of their location within the brain, are not considered good candidates for surgical resection. For these AVMs that are not good candidates for surgical removal, treatment with radiation should be considered. Radiation is delivered to the precise location of the AVM by a procedure called radiosurgery. Several different forms of stereotactic radiosurgery may be used including Linac, Gamma Knife, Cyberknife, or others. Finally, embolization or occlusion of the abnormal arteries by a catheter technique can be considered in certain cases. Referral to a neurosurgeon is essential in order to best define the most appropriate treatment for any given AVM.

Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov. All studies receiving U.S. Government funding, and some supported by private industry, are posted on this government web site.

For information about clinical trials being conducted at the NIH Clinical Center in Bethesda, MD, contact the NIH Patient Recruitment Office:

Tollfree: (800) 411-1222
TTY: (866) 411-1010
Email: prpl@cc.nih.gov

For information about clinical trials sponsored by private sources, contact:
www.centerwatch.com

Wyburn Mason Syndrome Resources

Organizations:

Genetic and Rare Diseases (GARD) Information Center
PO Box 8126
Gaithersburg, MD 20898-8126
Phone #: 301-251-4925
800 #: 888-205-2311
e-mail: N/A
Home page: http://rarediseases.info.nih.gov/GARD/

March of Dimes Birth Defects Foundation
1275 Mamaroneck Avenue
White Plains, NY 10605
Phone #: 914-997-4488
800 #: N/A
e-mail: N/A
Home page: http://www.marchofdimes.com

National Organization of Vascular Anomalies
PO Box 38216
Greensboro, NC 27438-8216
Phone #: N/A
800 #: N/A
e-mail: admin@mail.novanews.org
Home page: http://www.novanews.org

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References

TEXTBOOKS
Traboulsi EI. Ed. A Compendium of Inherited Disorders and the Eye. Oxford University Press. New York, NY. 2006:125-126

Shields JA. NORD Guide to Rare Disorders. Lippincott Williams & Wilkins. Philadelphia, PA. 2003:667.

Magalini SI, et al., eds. Dictionary of Medical Syndromes. 4th ed.New York, NY: Lippincott-Raven Publishers; 1997:855.

Champion RH, Burton JL, Ebling FJG. Eds. Textbook of Dermatology. 5th ed. Blackwell Scientific Publications. London, UK; 1992:493.

JOURNAL ARTICLES
Skorin L, Jr., Simmons DK. Wyburn-Mason syndrome. Mayo Clin Proc. 2008;83:135.

Dayani PN, Sadun AA. A case report of Wyburn-Mason syndrome and review of the literature. Neuroradiology. 2007;49:445-56.

Reck SD, Zacks DN, Eibschitz-Tsimhoni M. Retinal and intracranial arteriovenous malformations: Wyburn-Mason syndrome. J Neuroophthalmol. 2005;25:205-8.

Ponce FA, Han PP, Spetzler RF, Canady A, Feiz-Erfan I. Associated arteriovenous malformation of the orbit and brain: a case of Wyburn-Mason syndrome without retinal involvement. Case Report. J Neurosurg. 2001;95:346-9.

Patel U, Gupta SC. Wyburn-Mason syndrome. A case report and review of the literature. Neuroradiology. 1990;31:544-46.

Wyburn-Mason R. Arteriovenous aneurysm of mid-brain and retina, facial naevi and mental changes. Brain. 1943;66:163-203.

FROM THE INTERNET
Bidwell AE. Wyburn-Mason syndrome. Emedicine Journal, July 24, 2007. Available at: http://www.emedicine.com/oph/topic357.htm Accessed on: October 18, 2007.

National Institute of Neurological Disorders and Stoke. Arteriovenous Malformations Information Page. February 7, 2006. Available at: http://www.ninds.nih.gov/disorders/avms/avms.htm Accessed On: October 18, 2007.

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