



Bulbar Palsy, Progressive, with Sensorineural Deafness

Alternative Names

BVL Syndrome
Pontobulbar Palsy with Deafness
Brown-Vialetto-Van Laere Syndrome

WHO International Classification of Diseases

Diseases of the nervous system

OMIM Number

211530

Mode of Inheritance

Autosomal recessive; ?heterogeneous

Description

Brown-Vialetto-Van Laere syndrome is a rare disease, typified by progressive bilateral sensorineural deafness, and neurological disorders, usually involving the lower cranial nerves. Rarely, the upper cranial and spinal nerves may also be involved. Typical symptoms presented by patients include neurosensory deafness, weakness of facial and neck muscles, shortness of breath, swallowing difficulties, atrophic tongue with fasciculations, and occasional mental retardation and vocal cord paralysis.

Only a few more than 30 cases of this disease have been reported worldwide. It has been reported that the sex ratio for the disease is 1 male:5 females, with an even greater bias towards females in familial cases. Differential diagnosis includes amyotrophic lateral sclerosis, progressive bulbar paralysis of Fazio-Londe, Nathalie syndrome, Boltshauser syndrome, and Madras disease.

Molecular Genetics

BVL syndrome has been known to show familial, as well as sporadic occurrence. Majority of the familial cases show an autosomal recessive mode of inheritance. However, at least one report each points to an

autosomal dominant or X-linked mode of inheritance for the syndrome.

Epidemiology in the Arab World

Lebanon

Megarbane et al. (2000) reported for the first time a consanguineous Lebanese family, with at least three affected children presenting with the same severe features with rapid deterioration. The first case was of an 8-year old boy, whose hearing and walking had begun to get progressively impaired since 2-1/2 years of age. On physical examination, he was weak and slender with weight in the 5th centile, totally deaf, and hypotonic. Neurological examination showed axial and appendicular hypotonia; dorsal scoliosis; tongue fasciculation, paucity of spontaneous movements; important muscular weakness involving muscles of neck, shoulders, and upper arms with major muscular wasting; clawed hands with thenar and hypothenar amyotrophy; absent deep tendon reflexes; and no Babinski response. A left curved scoliosis, and thin diaphyses of the long bones were visible in the radiological examination. The patient died three years later, probably due to acute respiratory failure. The patient's younger brother too presented with a similar clinical pattern. By the age of 7-years, his speech was limited to minimal vocalization. He also presented with diffuse muscular wasting, mild kyphoscoliosis with a right gibbosity, complete impairment of manual dexterity, distended neck veins, limited chest expansion with paradoxical respiration, and a protruberant abdomen, among other features. Neurological symptoms were similar to his brother's, with bilateral facial paresis. Incomplete right branch block and a tachycardia of 115 beats/min was recorded in the ECG. Needle electromyography was neurogenic and showed fibrillation activity particularly at the upper arms. This patient also died suddenly after four months. The parents



and other siblings of these patients were normal, except for a sister, who apparently, also presented with similar symptoms, but died at the age of 4 years. Another affected member of the kindred, a third cousin to these affected patients, also presented with the same symptoms. His neurological features were identical to his cousins. Nerve conduction and electromyogram studies revealed an absence of sural and sensory median potentials, but normal motor velocities, suggesting that this disease be classified as a severe sensorimotor neuropathy. The clinical features of all the patients were indicative of the BVL syndrome. The family reported clearly showed the autosomal recessive inheritance of the disorder. Megarbane et al. (2000) were of the opinion that the sporadic cases reported earlier, could be explained by the mutated allele

being present in one parent, and a de novo mutation coming from the second parent. The authors concluded that identification of the gene responsible for the disorder in this family would be a major step towards understanding the etiology and pathogenesis of the disease.

References

Megarbane A, Desguerres I, Rizkallah E, Delague V, Nabbout R, Barois A, Urtizbera A. Brown-Vialetto-Van Laere syndrome in a large inbred Lebanese family: confirmation of autosomal recessive inheritance? *Am J Med Genet.* 2000; 92(2):117-21.

Contributors

Pratibha Nair: 18.7.2006

