



Branchiogenic-Deafness Syndrome

Alternative Names

Megarbane-Loiselet Syndrome

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

609166

Mode of Inheritance

Autosomal dominant

Description

The branchiogenic-deafness syndrome is an extremely rare condition, having been identified in only a single Lebanese family, so far. Characteristic clinical features of the syndrome include branchial arch anomalies, asymmetrical ear malformations, conductive, sensorineural, and mixed non-progressive hearing loss, internal auditory canal hypoplasia, strabismus, trismus, abnormal 5th fingers, short stature, and learning disability. Many of the clinical features overlap with those of brachio-oto-renal syndrome, and branchi-otic syndrome.

Molecular Genetics

Nothing is definitely known about the molecular genetics of the disorder. However, the mode of transmission resembles an autosomal dominant pattern of inheritance.

Epidemiology in the Arab World

Lebanon

Megarbane et al. (2003) described a brother and sister in a Sunni Lebanese family with a constellation of clinical features that had never been reported previously. The elder brother had been noted to have a bilateral meatal atresia of the external ears and a malformed lobe of the left ear at birth. Hearing impairment was noticed at 2-years of age, and surgery to create a

neo-external auditory canal at the right ear was not successful. Upon examination, at the 24-years of age, his height was below the 3rd centile, and he could speak intelligibly, with some distortions. His right pinna measured in the 50th centile, and the ear had an over folded helix, and hypoplastic tragus and lobe. The left ear had an atretic auditory canal with preauricular tag. Other noteworthy features in the patient included divergent strabismus, difficulty in opening the mouth wide enough, submucosal cleft palate, branchial cleft sinus on both sides of the lower neck, some astigmatism, patchy depigmentation of the skin at hair root, on both eyelids, around the mouth, on the chin and both hands, short terminal phalanx of both 5th fingers, and short terminal 2-4 toes of the right feet. Radiological examination revealed the short phalanges to be due to mild cortical erosions of the terminal phalanges. CT scan revealed temporal bone abnormalities, including bilateral external auditory canal atresia, small middle ear cavity with malformed ossicles, right internal auditory canal hypoplasia, and enlargement of the left vestibular aqueduct. His 20-year old younger sister presented with similar features. Her strabismus was severe, and required surgical correction at 3-years of age. Upon examination, her left ear had an atretic auditory canal with a preauricular tag, while the right ear had a preauricular pit. The terminal phalanges of her 5th fingers were more pronouncedly shortened. But, there was no depigmentation of the skin, or submucosal cleft palate as he brother's. Radiological and CT results were similar to her brothers's. However, she also showed mild erosion of the phalanges of the other fingers, along with atrophy of the soft tissue. In addition, the upper phalanges of her toes showed a pointed aspect. The other siblings of the patients, as well as their mother showed no abnormalities. Their father, was short in stature, and had an ear pit at the margin of his left helix. However, he had normal hearing, as well as



clinically normal hands. His sister and his half brother apparently, had short stature, and branchial sinuses without any hearing problems. The EYA1 gene did not show any mutations in one of the patients examined. Megarbane et al. (2003) compared the clinical features with other known syndromes, including branchio-oto-renal syndrome, brancio-oto syndrome, branchio-oto-facial syndrome, and branchio-oto-uretral syndrome. Although the patients shared many features with these diseases, the disorder could not be defined by any one of these diseases, indicating that it was a new disorder. Regarding the inheritance of the disorder, Megarbane et al. (2003) concluded that the most likely possibility

was that a dominant mutation in a single gene with pleiotropic effects, and intra-familial variability in its expression, was responsible for the genetic defect.

References

Megarbane A, Rassi S, Chouery E, Delague V, Perez de Nanclares Leal G, Tabet M, Castano L, Loiselet J. A new dominant branchiogenic-deafness syndrome with internal auditory canal hypoplasia and abnormal extremities. *Am J Med Genet A*. 2003; 120(2):276-82.

Contributors

Pratibha Nair: 7.8.2006

