Opis choroby *

Definicja

Bosley-Salih-Alorainy syndrome (BSAS) is characterized by variable horizontal gaze dysfunction, profound and bilateral sensorineural deafness associated commonly with severe inner ear maldevelopment, cerebrovascular anomalies (ranging from unilateral internal carotid artery hypoplasia to bilateral agenesis), cardiac malformation, developmental delay and occasionally autism. The syndrome is caused by homozygous mutations in the HOXA1 gene (7p15.2) and is transmitted in an autosomal recessive manner. The syndrome overlaps clinically and genetically with Athabaskan brain dysfunction syndrome (ABDS,). However unlike ABDS, BSAS does not manifest central hypoventilation.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
69737

Kod OMIM
601536

Kod ICD10
Q87.8

Kod ICD11
LD2H.Y

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*Źródło

orphanet