Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by moderate to severe developmental delay/intellectual disability with absent or limited speech development, various behavioral problems (including autistic features, hyperactivity, or aggressiveness), and craniofacial anomalies such as long face, high and prominent forehead, bulbous nose with low-hanging columella, thin vermillion of the upper lip, palatal (cleft palate, high-arched palate, and bifid uvula) and dental (abnormal upper incisors) abnormalities, and micrognathia. Hypotonia and feeding difficulties are frequent. Other supportive findings may include skeletal anomalies with low bone density and abnormal brain imaging.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SAS
SAS

Kod ORPHA
576278

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

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*Źródło

orphanet