Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by Hirschsprung disease, facial dysmorphism (sloping forehead, high arched eyebrows, long eyelashes, telecanthus/hypertelorism, ptosis, prominent ears, thick earlobes, prominent nasal bridge, thick philtrum, everted lower lip vermillion and pointed chin), global developmental delay, intellectual disability and variable cerebral abnormalities (focal or generalized polymicrogyria, or hypoplastic corpus callosum).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
GOSHS
GOSHS
Okrężnica olbrzymia - mikrocefalia
Megacolon-microcephaly syndrome

Kod ORPHA
66629

Kod OMIM
609460

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet