

## 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 vermilion 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