

Opis choroby *

Definicja

Shprintzen-Goldberg omphalocele syndrome is a very rare inherited malformation syndrome characterized by omphalocele, scoliosis, mild dysmorphic features (downslanted palpebral fissures, s-shaped eyelids and thin upper lip), laryngeal and pharyngeal hypoplasia and learning disabilities.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

3164

Kod OMIM

182210

Kod ICD10

Q79.2

Kod ICD11

LD2F.1Y

*Źródło

orphanet