

Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterised by several dysmorphic features, hypotonia, developmental delay, intellectual disability, behavioral problems, visual and hearing abnormalities, constipation, and feeding difficulties. Common dysmorphic features include coarse facies, broad forehead, synophrys, bushy eyebrows, deep-set eyes, downslanting palpebral fissures, epicanthus, depressed nasal bridge, bulbous nasal tip, posteriorly rotated ears, full cheeks, thin upper lip, inverted nipples, and hirsutism. Behavioral problems tend to be dominated by ADHD, but anxiety, aggressive outbursts and autistic features may also present.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Desanto-Shinawi syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
466943	616708	Q87.0

Kod ICD11

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*Źródło

orphanet