Opis choroby *

Definicja

Hypotonia-speech impairment-severe cognitive delay syndrome is a rare, genetic neurodegenerative disorder characterized by severe, persistent hypotonia (presenting at birth or in early infancy), severe global developmental delay (with poor or absent speech, difficulty or inability to roll, sit or walk), profound intellectual disability, and failure to thrive. Additional manifestations include microcephaly, progressive peripheral spasticity, bilateral strabismus and nystagmus, constipation, and variable dysmorphic facial features (including plagiocephaly, broad forehead, small nose, low-set ears, micrognathia and open mouth with tented upper lip).

Dane

Klasyfikacja
Choroba

Synonimy
IHPRF syndrome
Zespół IHPRF
Zespół hipotonii niemowlęcej, opóźnienia psychruchowego i charakterystycznego wyglądu twarzy
Infantile hypotonia-psychomotor retardation-characteristic facies syndrome

Kod ORPHA
371364

Kod OMIM
615419

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet