

## 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 psychoruchowego 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