Opis choroby *

Definicja

A rare autosomal ichthyosis syndrome with prominent neurologic signs characterized by the association of congenital ichthyosis with severe developmental delay, microcephaly, spastic tetraplegia, sensorineural hearing impairment, athetosis, and myoclonus. Marked epileptic discharges with occurrence of tonic spasms have also been reported. Cerebral MRI shows diffuse cortical atrophy. There have been no further descriptions in the literature since 1995.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital ichthyosis-microcephalus-quadriplegia syndrome
Rybia łuska wrodzona - małogłowie - kwadriplegia

Kod ORPHA
2271

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet