

Opis choroby *

Definicja

X-linked intellectual disability-acromegaly-hyperactivity syndrome is characterised by severe intellectual deficit, acromegaly and hyperactivity. The syndrome has been described in two half-brothers. Dysarthria, aggressive behaviour, a characteristic facies (an acromegalic and triangular face with a long nose) and macroorchidism were also present. The mother displayed moderate intellectual deficit and milder facial anomalies. Central nervous system anomalies were identified in the two boys: subarachnoid cysts and hyperdensity in the pontine region.

Dane

Klasyfikacja

Choroba

Kod ORPHA

85327

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet