Opis choroby *

Definicja

A rare genetic neurological disorder characterized by the presence of two or more of the main criteria for classic Rett syndrome (loss of acquired purposeful hand skills, loss of acquired spoken language, gait abnormalities, stereotypic hand movements), a period of regression followed by recovery or stabilization, and five out of eleven supportive criteria (breathing difficulties, bruxism, impaired sleep pattern, abnormal muscle tone, peripheral vasomotor disturbances, scoliosis/kyphosis, delayed growth, small cold hands and feet, inappropriate laughter or screaming spells, decreased pain sensation, and intense eye communication). Like classic Rett syndrome, it almost exclusively affects girls, while the disease course may be either milder or more severe.

Dane

Klasyfikacja
Choroba

Synonimy
Atypical RTT
Atypowy RTT
Wariant zespołu Retta
Rett syndrome variant

Kod ORPHA
3095

Kod OMIM
613454

Kod ICD10
F84.2

Kod ICD11
LD90.Y

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*Źródło

orphanet