Opis choroby *

Definicja

A rare stiff person syndrome spectrum disorder characterized by limb and truncal rigidity, stimulus-sensitive spasms, myoclonus, hyperekplexia, autonomic disturbance, and brainstem involvement or other neurological defects. The condition is progressive and potentially life-threatening, especially due to respiratory failure. It may be associated with the presence of glycine receptor or glutamic acid decarboxylase antibodies, as well as thymomas or lymphomas.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
PERM
PERM

Kod ORPHA
438266

Kod OMIM
184850

Kod ICD10
G04.8

Kod ICD11
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*Źródło

orphanet