

Opis choroby *

Definicja

A rare DNA repair defect other than combined T-cell and B-cell immunodeficiencies characterized by intrauterine and postnatal growth retardation resulting in short stature, microcephaly, glucocorticoid deficiency, natural killer cell deficiency, and recurrent viral infections. Patients may also have increased susceptibility to cancer.

Dane

Klasyfikacja

Choroba

Synonimy

Primary immunodeficiency due to MCM4

deficiency

Pierwotny Niedobór odporności spowodowany deficytem MCM4

Kod ORPHA

75391

Kod OMIM

609981

Kod ICD10

D84.8

Kod ICD11

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*Źródło

orphanet