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