Opis choroby *

Definicja

Constitutional mismatch repair deficiency syndrome is a rare, inherited cancer-predisposing syndrome characterized by the development of a broad spectrum of malignancies during childhood, including mainly brain, hematological and gastrointestinal cancers, although embryonic and other tumors have also been occasionally reported. Non-neoplastic features, in particular manifestations reminiscent of neurofibromatosis type 1 (e.g., café-au-lait spots, freckling, neurofibromas), as well as premalignant and non-malignant lesions (such as adenomas/polpyps) are frequently present before malignancy development.

Dane

Klasyfikacja
Choroba

Synonimy
CMMR-D syndrome
Zespół CMMR-D

Kod ORPHA
252202

Kod OMIM
619097

Kod ICD10
D80.8

Kod ICD11
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*Źródło

orphanet