Constitutional mismatch repair deficiency syndrome Kod Orpha: 252202 Kod OMIM: 619097

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
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.