DISEASE:
Constitutional mismatch repair deficiency syndrome

NAME:	Constitutional mismatch repair deficiency syndrome
DESCRIPTION:	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.
ORPHACODE:	252202
SYNONYMS:	CMMR-D syndrome
XREF(S):	Orphanet OMIM OMIM ICD-10 OMIM OMIM

ANALYTE(S):	PMS2 MLH1 MSH2 MSH6
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RELATED CONTENT

Related Genetic Tests

- Colorectal cancer / Polyposis (gene panel)
- Constitutional Mismatch Repair Deficiency Syndrome (4 genes)
- Constitutional Mismatch Repair Deficiency Syndrome + Bloom syndrome (5 genes)
- Hereditary cancer (Breast, ovary, colon) (26 genes)
- Hereditary cancer (gene panel)
- Hereditary nonpolyposis colorectal cancer (gene panel)
- Hereditary nonpolyposis colorectal cancer / Lynch syndrome (8 genes)
- Microsatellites instability analysis- MMR genes

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- mutL homolog 1
- mutS homolog 2
- mutS homolog 6
- PMS1 homolog 2, mismatch repair system component

Related Gene Panels

- Cancer (Breast, ovary, colon,…) (26 genes) - ULG
- Colorectal cancer/polyposis (18 genes) - KUL
- Constitutional Mismatch Repair Deficiency Syndrome (4 genes) - KUL
- Constitutional Mismatch Repair Deficiency Syndrome / Bloom syndrome - KUL
- Hereditary predisposition to cancer (47 genes) - IPG
- Lynch syndrome/hereditary nonpolyposis colorectal cancer (5 genes) - UCL - UGent
- Lynch-like panel
