Table 3. Scoring system for aiding CMMRD diagnosis from C4CMMRD (adapted from Aronson et al 2022; PMID: 33622763)

Malignancies/premalignancies: one is mandatory; if more than one is present in the patient, add the points:	**>=3 points = CMMRD features meets PM3 criteria after excluding the diagnosis of NF1 or LFS as individuals with those disorders could easily get to 3 points.**
Carcinoma from the LS spectrum* at age <25 years.	3 points
Multiple bowel adenomas at age <25 years and absence of APC/MUTYH mutation(s) or a single high-grade dysplasia adenoma at age <25 years.	3 points
WHO grade III or IV glioma at age <25 years.	2 points
NHL of T cell lineage or sPNET at age <18 years	2 points
Any malignancy at age <18 years.	1 point
Additional features: optional; if more than one of the following is present, add the points:	
Clinical sign of NF1 and/or ≥2 hyperpigmented and/or hypopigmented skin alterations Ø>1 cm.	2 points
Diagnosis of LS in a first-degree or second-degree relative.	2 points
Carcinoma from LS spectrum* before the age of 60 in a firstdegree, second-degree or third-degree relative.	1 point
A sibling with carcinoma from the LS spectrum*, high-grade glioma, sPNET or NHL.	2 points
A sibling with any type of childhood malignancy.	1 point
Multiple pilomatricomas in the patient.	2 points
One pilomatricoma in the patient.	1 point
Agenesis of the corpus callosum or non-therapy-induced cavernoma in the patient.	1 point
Consanguineous parents	1 point
Deficiency/reduced levels of IgG2/4 and/or IgA.	1 point

*Colorectal, endometrial, small bowel, ureter, renal pelvis, biliary tract, stomach, bladder carcinoma

CMMRD, constitutional mismatch repair deficiency; LS, Lynch syndrome; NF1, neurofibromatosis type 1; NHL, non-Hodgkin's lymphoma; sPNET, supratentorial primitive neuroectodermal tumours.