Questionnaire constitutive mismatch-repair-deficiency (CMMRD) syndrome
Assessment of criteria for testing

Patient-no. : CMMRD-20___-_____ ☐ male ☐ female _____
DOB

Referring Center:_____ Clinician_____

Tel.-No.: _____ Email:_____

Tumor entity: _____ Age at diagnosis: _____
(list all if there are more than one tumor in the patient)

Questions:

1) Consanguinity of parents ☐ yes ___ _____ ☐ no
specify relationship

2) Café au lait spots, hyperpigmented skin areas ☐ yes ☐ no

number of macules > 1cm [ ] maximal size (cm) [ ]

body distribution ☐ segmental ☐ generalized

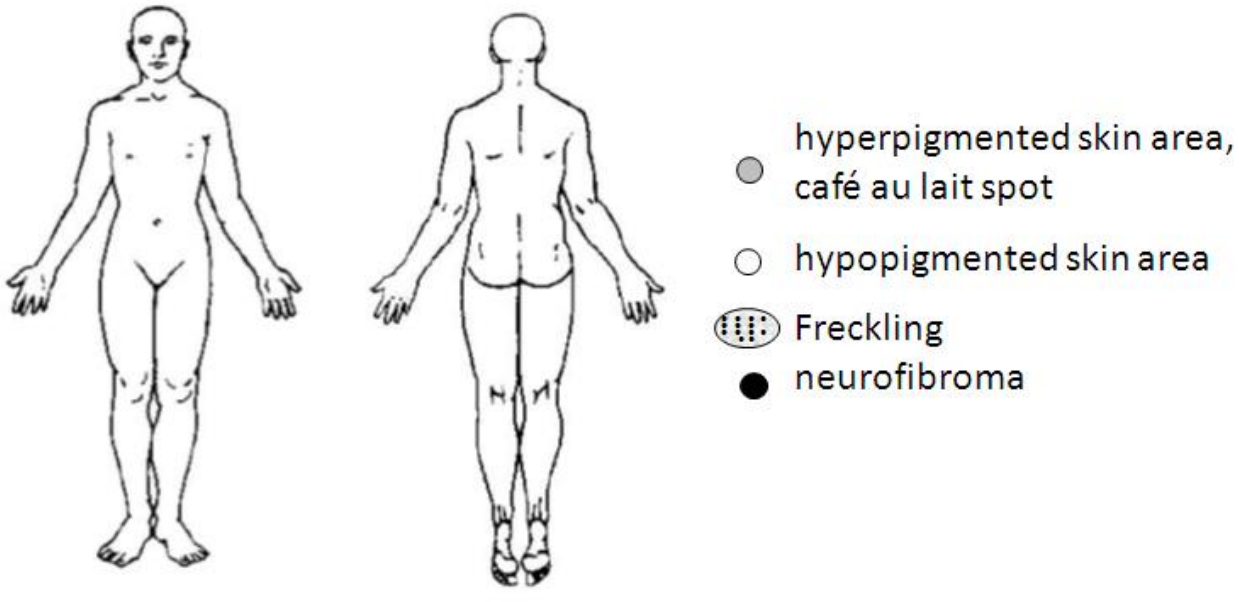
3) Other signs of NF1 ☐ yes ☐ no _____
Specify type & number

4) Hypopigmented skin areas (ash-leave spots) ☐ yes ☐ no

number of macules > 1cm [ ] maximal size (cm) [ ]

body distribution ☐ segmental ☐ generalized

Ad 2-4) Indicate localization and type of cutaneous and pigmentary alterations in this body scheme:



5) Pilomatricoma (=clacifying epithelioma of Malherbe) ☐ yes ☐ no

number [ ] age at diagnosis in years [; ;] localisation:_____

6) Other cutaneous alterations (e.g. hemangioma, hairy naevi, Lupus erythematosus etc.)

☐ yes _____ ☐ no

specify

7) Congenital malformations ☐ yes _____ ☐ no ☐ not known

(specify*)

(*e.g. brain: developmental vascular anomalities (DVAs), brain cavernoma, agenesis of corpus callosum +/- gray matter heterotopia; bone:e.g. syndactyly; urinary tract or genital parts; heart and vessel; dysmorphic features; other)

8) Defect of immunoglobulin production: repeated infections ☐ yes ☐ no

_____

specify (e.g. > 8 otitis per year before 4y-o; >4 otitis per year after 4 y-o; 2 pneumonitis or sinusitis per year; other)

reduced IgG2, IgG4 and/or IgA levels ☐ yes _____ ☐ no ☐ not known

specify

9) Lynch syndrome- associated carcinomas (colon cancer, endometrial cancer, rarer: stomach, ovarian, pancreatic, urothel, bile duct, small intestine) in the family

☐ yes ☐ no ☐ not known

_____

specify (relationship to patient, tumor entity, age at diagnosis),
if more than one relative is affected mention all)

10) Sibling with café au lait spots and/or other pigmentation alterations and/or other signs of NF1

[ ]no. of siblings ☐ yes _____ ☐ no

specify signs

11) Sibling (1.-grade cousin) with malignancy

☐ yes _____ ☐ no

specify (tumor entity, age at diagnosis)

Ad 9-11) Draw a pedigree on a separate sheet and indicate family members with malignancies

Send a copy of the completed form to study center: email: katharina.wimmer@i-med.ac.at
FAX: 0043-(0)512-9003-73510
Division Humangenetik der Medizinischen Universität Innsbruck, Peter-Mayr-Str. 1, 6020 Innsbruck, Austria

Evaluation

Patient fulfills criteria for CMMRD testing ☐ yes ☐ no

Criteria for CMMRD testing in a cancer patient (≥3 points)		
Malignancies/pre-malignancies:	Points assigned	Points in patient
carcinoma from the LS spectrum[a] at age <25 years	3 pts	
multiple bowel adenomas at age <25 years and absence of *APC* /*MUTYH* mutation(s) or a single high grade dysplasic adenoma at age <25 years	3 pts	
WHO grade III or IV glioma at age <25 years	2 pts	
NHL of T-cell lineage or sPNET at age <18 years	2 pts	
any malignancy at age <18 years	1 pt	
Additional features:	Points assigned	
clinical sign of NF1 and/or ≥2 hyper- and/or hypo-pigmented skin alterations Ø>1 cm in the patient	2 pts	
diagnosis of LS in a 1st or 2nd degree relative	2 pts	
carcinoma from LS spectrum[a] before the age of 60 in 1st-, 2nd- or 3rd-degree relative	1 pt	
a sibling with carcinoma from the LS spectrum[a], high grade glioma, sPNET, or NHL	2 pts	
a sibling with any type of childhood malignancy	1 pt	
multiple pilomatricomas in the patient	2 pts	
one pilomatricoma in the patient	1 pt	
brain MRI in the patient: multiple developmental vascular abnormalities (DVAs) or agenesis of the corpus callosum or non-therapy-induced cavernoma	1 pt	
consanguineous parents	1 pt	
deficiency/reduced levels of IgG2/4 and/or IgA	1 pt	
Total points in patient	//////////////	

[a] colorectal, endometrial, small bowel, ureter, renal pelvis, biliary tract, stomach, bladder carcinoma

If the patient fulfills criteria for CMMRD testing

Patient agrees with gMSI testing: ☐ yes ☐ no

IHC of MMR genes initiated: ☐ yes _____ ☐ no
indicate department performing the analysis

MSI tesing in tumor tissue initiated: ☐ yes _____ ____ ☐ no
indicate department performing the analysis

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