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Reply:

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REPLY:

We appreciate that Guerrini-Rousseau and colleagues are raising awareness for constitutional mismatch repair deficiency (CMMRD) syndrome and its phenotypical overlap with neurofibromatosis type 1 (NF-1). All 5 patients described in our article met clinical criteria for NF-1 (Table). None of the patients reported had confirmatory genetic testing; for children who meet the clinical criteria, genetic testing is not typically obtained.

CMMRD is rare (200 patients reported), and our patients had none of the other diagnostic criteria of CMMRD.[1] Café au lait spots are common (up to 97%) in CMMRD, but the other clinical manifestations of NF-1 are much less frequent.[2] Focal abnormal signal intensities have also been reported in CMMRD.

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For a diagnosis of NF-1, at least 2 of the diagnostic criteria should be present[3]

NF-1 Diagnostic Criteria	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5
Six or more café au lait macules >5 mm in greatest diameter in prepubertal individuals and >15 mm in greatest diameter in postpubertal individuals	Yes	Yes	Yes	Yes	Yes
Two or more neurofibromas of any type or 1 plexiform neurofibroma		Yes			
Freckling in the axillary or inguinal regions	Yes	Yes	Yes	Yes	Yes
Optic glioma			Yes		Yes
Two or more Lisch nodules (iris hamartomas)	Yes	Yes			Yes
A distinctive osseous lesion such as sphenoid dysplasia or tibial pseudarthrosis					
A first-degree relative (parent, sibling, or offspring) with NF-1 as defined by the above criteria	Yes	Yes	Unknown (adopted)		
MRI findings: focal abnormal signal intensities	Yes	Yes	Yes	Yes	Yes