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AJNR

Reply:

N. Kadom, R.C. Castellino and D.S. Wolf

AJNR Am J Neuroradiol 2019, 40 (6) E32

doi: <https://doi.org/10.3174/ajnr.A6062>

<http://www.ajnr.org/content/40/6/E32>

This information is current as
of May 14, 2025.

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.

<http://dx.doi.org/10.3174/ajnr.A6062>

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 **N. Kadom**

Departments of Radiology and Pediatrics

 **R.C. Castellino**

Department of Pediatrics and Aflac Cancer & Blood Disorders Center
Children's Healthcare of Atlanta
Atlanta, Georgia

 **D.S. Wolf**

Department of Pediatrics, Division of Child Neurology
Emory University School of Medicine
Atlanta, Georgia

For a diagnosis of NF-1, at least 2 of the diagnostic criteria should be present³

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