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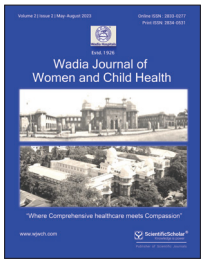

Clinical Image

Café-au-lait spots-what lies underneath

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An 11-year-old girl was diagnosed with high-grade osteosarcoma. She had a history of T acute lymphoblastic leukemia at 6 years of age. On examination, she had multiple café-au-lait spots on the trunk. Due to the presence of café-au-lait spots, 2nd malignancy, and 3rd degree consanguinity, we investigated for cancer predisposition syndromes which revealed a homozygous mutation in PMS2 exon 2 (c.128_130del) suggestive of constitutional mismatch repair deficiency (CMMRD) syndrome.

Family screening revealed multiple café-au-lait spots and the same homozygous mutation in the 5-year-old asymptomatic brother [Figure 1] and heterozygous mutation in the parents.

Differential diagnosis of café-au-lait spots and predisposition to malignancy includes CMMRD, Fanconi anemia, Neurofibromatosis, Bloom syndrome, Noonan syndrome, Tuberous sclerosis, and Ataxia telangiectasia.

CMMRD is a rare autosomal recessive disorder, with an incidence of 1 per million patients, caused by biallelic mutations in MMR genes including MLH1, MSH2, MSH6, or

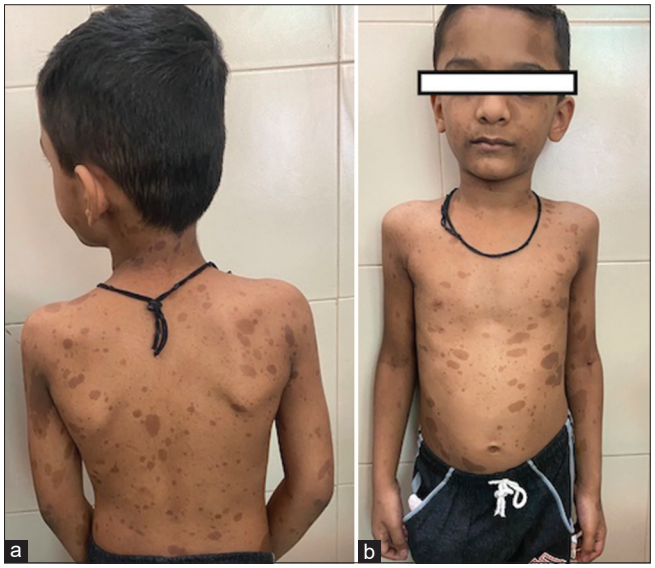

Figure 1: Asymptomatic sibling of index case (a) Multiple café-au-lait spots on the back. (b) Multiple café-au-lait spots on the trunk.

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PMS2, resulting in increased susceptibility to mainly hematological, colorectal, and brain tumors. Heterozygous mutations in these genes lead to autosomal dominant Lynch syndrome predisposing to colorectal and endometrial malignancies, usually not before 40 years of age.

Suspicion and early diagnosis of CMMRD help provide appropriate counseling and design surveillance strategies for further malignancies.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The author(s) confirms that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.