

Cutaneous Vasculitis as the Initial Presentation of Juvenile Myelomonocytic Leukemia: A Case Report

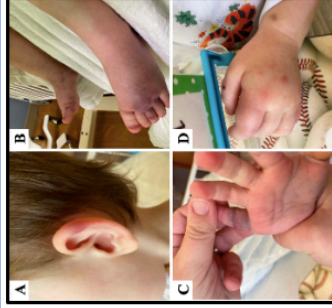
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Background

- Juvenile myelomonocytic leukemia (JMML) is a myeloproliferative neoplasia that targets the Ras signal transduction pathway.¹
- Clinically, JMML is associated with fever, respiratory symptoms, café-au-lait spots, as well as splenomegaly, hepatomegaly, lymphadenopathy, and skin rashes.^{2,3}
- Vasculitis has been reported to occur with a mutation to the CBL gene, however, ours is the first reported case of JMML presenting with cutaneous vasculitis associated with the PTPN11 mutation.⁴

Clinical Presentation



Clinical photographs showing (a) erythematous lesion on left external ear, (b) left foot edema with purpura, (c) edema and purpura proximally on his fifth digit on his right hand, and (d) multiple erythematous papules on the dorsal aspect of left hand and edema of his second digit.

- Skin biopsy was suggestive of cutaneous vasculitis. Next Gen Sequencing with Foundation One panel was done and revealed a mutation in *PTPN11* in his bone marrow.
- He was diagnosed with JMML with a somatic marrow mutation in *PTPN11*. Although vasculitis has been reported to occur with the CBL gene, ours is the first reported case of JMML presenting with cutaneous vasculitis associated with the *PTPN11* mutation.

Histological Findings

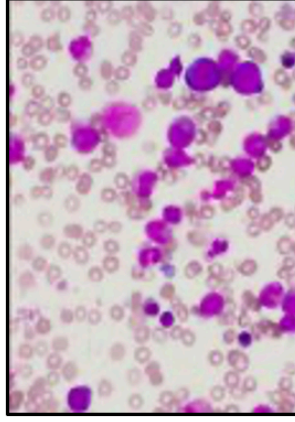


FIGURE 1. Blood smear of bone marrow showing neutrophils with dysplastic features.

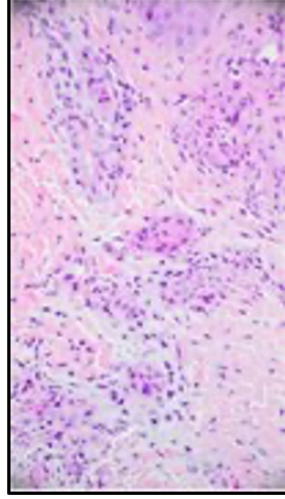


FIGURE 2. Blood vessels within the dermis showing lymphocytic perivascular inflammation with fibrin deposition and nuclear debris.

Discussion

Clinical Findings	Typical JMML Presentation	Our Patient's Presentation with JMML
Organomegaly (splenomegaly/hepatomegaly)	Yes	Yes
Mutation in <i>PTPN11</i> *	Yes	Yes
Fever, Respiratory symptoms, Lymphadenopathy	Yes	Yes
Café-au-lait spot	Yes	Yes
Vasculitis as the initial presentation	No	Yes

Conclusions

Pediatricians should consider JMML when young patients present with the triad of leukocytosis, anemia, and thrombocytopenia, along with dysplastic changes on bone marrow biopsy. In this case, cutaneous vasculitis confounded his clinical picture, but it should be kept in mind that vasculitis can be a paraneoplastic finding. Hematology and rheumatology should work together closely in similar cases to ensure more accurate timely diagnosis and better outcomes, given the high risk this disease carries.

References

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