

When Myelofibrosis is Skin-Deep: A Clinical Clue to a Rare Diagnosis

Nayak A.R. et al.: Cutaneous Clues to Rare Myelofibrosis

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A 24-year-old male presented with an 8-year history of transfusion-dependent anemia. Complete blood counts revealed pancytopenia with hemoglobin 6.9 g/dL, white blood cell count $2.1 \times 10^9/L$, and platelet count $80 \times 10^9/L$. Examination showed grade 2 clubbing, coarse facial features, and skin thickening over face, hands, and soles (Figure 1). Radiographic imaging revealed acro-osteolysis of the digits, cortical thickening of long bones, and symmetrical shaggy periosteal new bone growth (Figure 2). Clinical examination was not suggestive of any pulmonary condition and 2D Echo ruled out cardiac pathologies.

Bone marrow biopsy demonstrated grade 3 reticulin fibrosis (Figure 3). Testing for *JAK2*, *CALR* & *MPL* was negative. Clinical exome sequencing identified a homozygous *SLCO2A1* mutation (exon 11, p.Leu516SerfsTer12), confirming pachydermoperiostosis (PDP). Treatment with prednisolone and etoricoxib leads to increase in hemoglobin up to 9g/dL & he became transfusion independent till the last follow up (18 months from diagnosis). The three hallmarks of PDP are periosteal new bone growth, digital clubbing, and pachydermia [1]. Mutations in *SLCO2A1* impair cellular uptake of prostaglandin E2 (PGE2), preventing its intracellular degradation. The elevated PGE2 causes periostosis, digital clubbing, and skin thickening [2]. Selective COX-2 inhibitor Etoricoxib & prednisolone has shown some efficacy in improving anemia and reducing fibrosis [3,4]. This case emphasises the importance of careful physical examination in young patients with unexplained myelofibrosis & identification of *SLCO2A1* mutation has therapeutic relevance.

Keywords: Pachydermoperiostosis, *SLCO2A1* mutation, Pancytopenia, Hypertrophic osteoarthropathy, Myelofibrosis

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Patient Consent- Written informed consent was obtained from the patient for publication of this case and accompanying images.

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Figure 1. Skin thickening and corrugation noted over face(A), palms(B) and feet(C). Digital clubbing was also noted(D).



Figure 2. Pachydermoperiostosis. (A) AP radiograph of bilateral distal leg and ankle region shows bilateral diffuse shaggy periosteal new bone formation in diaphyseal regions of tibia and fibula with symmetrical cortical thickening (white arrows). (B) AP radiograph of bilateral hands and distal forearm shows similar cortical thickening and periosteal reaction notable along the long bones [radius, ulna and second metacarpals] (white arrows). Partial acroosteolysis is seen in the left third finger (red arrow). Widening of the ends of the long bones are also noted (arrowheads in A and B).

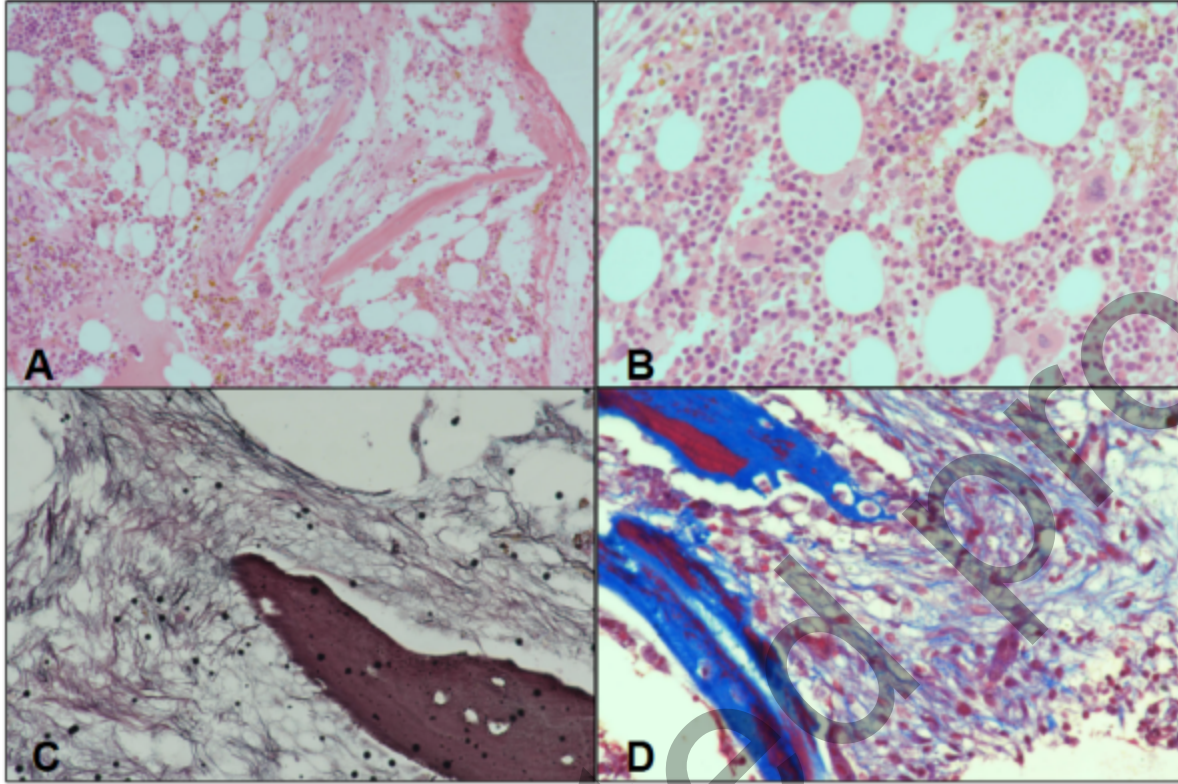


Figure 3: Bone marrow biopsy showing intertrabecular fibrosis (A), megakaryocytic proliferation (B), increased fibrosis as noted on reticulin & Masson's trichrome stain (C & D)