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Continuing Medical Education (CME) Questions

NIH natural history study of FPDMM patients

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Cunningham L, Merguerian M, Calvo KR, Davis J, Deuitch NT, Dulau-Florea A, Patel N, Yu K, Sacco K, Bhattacharya S, Passi M, Ozkaya N, De Leon S, Chong S, Craft K, Diemer J, Bresciani E, O'Brien K, Andrews EJ, Park N, Hathaway L, Cowen EW, Heller T, Ryan K, Barochia A, Nghiem K, Niemela J, Rosenzweig S, Young DJ, Frischmeyer-Guerrerio PA, Braylan R, Liu PP. Natural history study of patients with familial platelet disorder with associated myeloid malignancy. *Blood*. 2023;142(25):2146-2158.

1. **Your patient is a 22-year-old man with familial platelet disorder with associated myeloid malignancy (FPDMM). Based on the ongoing prospective, longitudinal National Institutes of Health (NIH) study from early 2019 to December 2021 by Cunningham and colleagues, which of the following statements about genetic, hematological, and bone marrow findings of patients with FPDMM is correct?**
   - ☐ Half of patients had thrombocytopenia, and nearly all had abnormal bleeding scores
   - ☐ On histologic evaluation of nonmalignant bone marrow samples, the most common finding was an increased number of megakaryocytes
   - ☐ One-third of patients were diagnosed with hematologic malignancies (HMs)
   - ☐ 62% (28 of 45) of families had ≥1 member who developed HMs

2. **Based on the ongoing prospective, longitudinal NIH study from early 2019 to December 2021 by Cunningham and colleagues, which of the following statements about other clinical findings of patients with FPDMM who were evaluated by multidisciplinary teams is correct?**
   - ☐ 93% (42 of 45) of patients had allergic and 80% (24 of 30) had gastrointestinal (GI) symptoms
   - ☐ Among 45 patients seen by the allergy/immunology team, asthma was the most common doctor-diagnosed allergic disorder
   - ☐ Among 30 patients with GI evaluation, diarrhea was the most common patient-reported GI disorder
   - ☐ 29 of 30 patients evaluated by the dermatology team had a history of eczema

3. **Based on the ongoing prospective, longitudinal NIH study from early 2019 to December 2021 by Cunningham and colleagues, which of the following statements about clinical implications of preliminary findings of patients with FPDMM is correct?**
   - ☐ FPDMM is best managed solely by hematologists
   - ☐ Most patients seen for thrombocytopenia, bleeding/bruising, or cancer had prompt detection of *RUNX1* variants and diagnosis of FPDMM
   - ☐ To detect *RUNX1* variants, all exons, introns, and copy number should be analyzed, as called for in diagnostic guidelines and previous literature
   - ☐ The study identified particular *RUNX1* variants linked to cancer risk

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