Currently 11 yo Hispanic female initially dx with thrombocytopenia at 1 month of age (plts 12K) during hospitalization for fever/aseptic meningitis

Rx with platelet transfusions and IgG but thrombocytopenia recurred within 2 weeks

Bone marrow bx (increased # of mature megakaryocytes; 90% cellular trilineage hematopoiesis); peripheral smear with thrombocytopenia and large platelets

Additional diagnostic testing for vascular malformation, WAS, ALPS, vWD2b, congenital TTP, PNH, multiple infectious etiologies and immune function all normal. Negative FH for thrombocytopenia

PFA, platelet aggregation unreliable due to thrombocytopenia; platelet flow normal
RL

• PE unremarkable except for bruising/petechiae
• Over ensuing 8 years, multiple ED visits for bleeding
• Dx as presumed ITP. Treatment included:
  • Steroids
  • IVIG/WinRho
  • Rituximab, mycophenolate, sirolimus
  • Splenectomy
Also had mild microcytic anemia – alpha thal DNA analysis revealed duplication of alpha chain gene on one allele.

What other test would you consider?
What is the diagnosis?
MEDICH SYNDROME


• Lifelong macrothrombocytopenia, easy bruising/bleeding

• Hypogranular giant platelets with decreased alpha granules and membranous inclusions resembling cigars or scrolls