A Patient with DiGeorge Syndrome
now with Massive Splenomegaly
and Hypogammaglobulinemia

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History

- JD: 20 y/o male diagnosed with DiGeorge syndrome in infancy (FISH+)

- PMHx:
  - Cardiac repair in infancy
  - Started on Ca for hypocalcemia early on
  - Age 5: Hypogammaglobulinemia (IgG=17 mg/dL, undetectable IgA) with poor Ab response
    - Started on IVIg with good response

- No family history of immunodeficiency
Clinical Course

- Age 10: Splenomegaly with marked increase in size over the past 2-3 years
  - refractory to multiple doses of steroid and Remicade
  - currently spleen tip reaching RLQ

- Thrombocytopenia: Platelet 40-50,000

- Generalized LAD (1.5-2.0cm)
  - Bx: non-caseating granuloma (other causes of granuloma ruled out)
Clinical Course

- Past 2 years:
  1. Hypercalcemia (iCa 1.78 mmol/L) off supplement
     --responsive to steroid, but →
     --persistent Ca dysregulation: frequent episodes of hypocalcemia (iCa 0.93 mmol/L) after steroid and bisphosphonate administration

  2. ↑↑IgM (3220 mg/dL)
     --normal expression of CD40L by flow cyt.
     --no evidence of clonal B or T cells in peripheral blood
Clinical Summary

- A case of DiGeorge syndrome with unusual clinical features supports the highly variable phenotypic expression and immunologic defects of this syndrome.

- Outstanding issues requiring a better understanding and management plan are:
  - Hypogammaglobulinemia with ↑↑IgM
  - Massive splenomegaly
  - Granuloma
  - Calcium dysregulation
Discussion/Questions

- How do you respond to family who had been repeatedly told that JD has CVID?

- Does JD need splenectomy? What are its potential benefits and risks in this case?

- How do you explain his calcium dysregulation? Is it all from granuloma?

- How do we further work up his elevated IgM level from this point on?