A Case of Neutropenia and Hypogammaglobulinemia
Patient LK

- **HPI:** 6mo previously healthy Asian boy.
  - Admitted to General Pediatrics for fever and rash consistent with eczema herpeticum with a group A Strep and Staph super-infection x 2 days. The rash was worsening despite treatment with PO acyclovir.
  - Noted to be neutropenic with ANC 100.
- **PMH:** Eczema.
- **Meds/All:** None except recent PO acyclovir.
- **Family Hx:** Mom with history of genital herpes. No childhood cancers or immunodeficiencies.
- **Social Hx:** Healthy older siblings, including a healthy brother.
Exam & Labs

**Physical Exam:**
- General: febrile (38.7), irritable but consolable
- HEENT: no thrush, oral lesions/sores, nor gum hypertrophy
- Lymph Nodes: none
- Abdomen: no organomegaly
- GU: normal descended testes
- Skin: multiple erythematous, crusted lesions and crops of vesicles on face, ears, hands and feet

**Admission labs:**
- Blood counts:
  - WBC 3.6 with ANC 100
  - Hgb 10.3
  - Plts 415
- Metabolic panel: normal
- Blood culture: no growth
- VZV DFA: negative
- HSV DFA: negative
- Wound culture: +group A Strep, +Staph aureus
Hospital Course

- **Super-infected eczema herpeticium**
  - Treated with IV acyclovir and keflex with gradual resolution.

- **Neutropenia**
  - Peds Heme/Onc consulted on hospital day 2
    - Review of CBCPDs over 48hr of his admission showed ANC 0-100 and mild anemia with MCV 73.
    - Peripheral smear – leukopenia with neutropenia, lymphopenia and monocytosis with a mild normocytic, normochromic anemia.
Peripheral Smear & Bone Marrow

- Peripheral smear most consistent with transient viral-induced myelosuppression; however, due to the severity of the super-infection and the extent of his neutropenia quantitative immunoglobulins were requested.

- Flow cytometry negative for leukemia.
- FISH t(15;17) negative for AML M3.
Hospital Course (cont.)

- Quantitative immunoglobulins:
  - IgG 24 G/dL
  - IgA <6 G/dL
  - IgM 5 G/dL

- IVIG (0.6 G/kg) x 1 given.

- Neupogen 5mcg/kg/day given x 2 days until ANC > 1000.
Final Diagnosis

- Primary immunodeficiency panel:
  - T helper 67.8% (absolute: 2104 cells/uL)
  - T cytotoxic 20.8% (absolute: 647 cells/uL)
  - NK cells 3.5% (absolute: 109 cells/uL)
  - B cell 0.6% (absolute: 19 cells/uL)

- BTK gene testing:
  - Positive for novel hemizygous V546D missense mutation in the BTK gene consistent with X-linked agammaglobulinemia.

*Bruton X-linked Agammaglobulinemia*