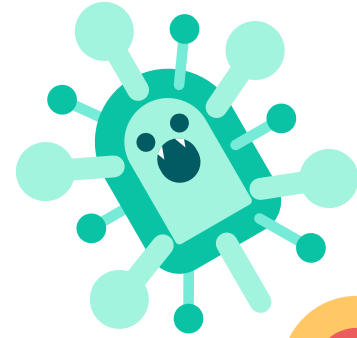
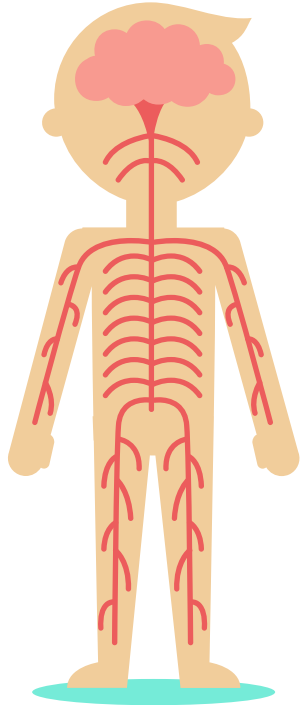


Diagnosis of an Immunodeficiency



Bianca Minter, Virginia Tam,
Victoria Walden

OUR PATIENT: JOHNNY



At 1

Recurring sinus infections (ethmoid sinuses), β - hemolytic streptococci



At 3

Pneumonia from *Pneumocystis jirovecii*

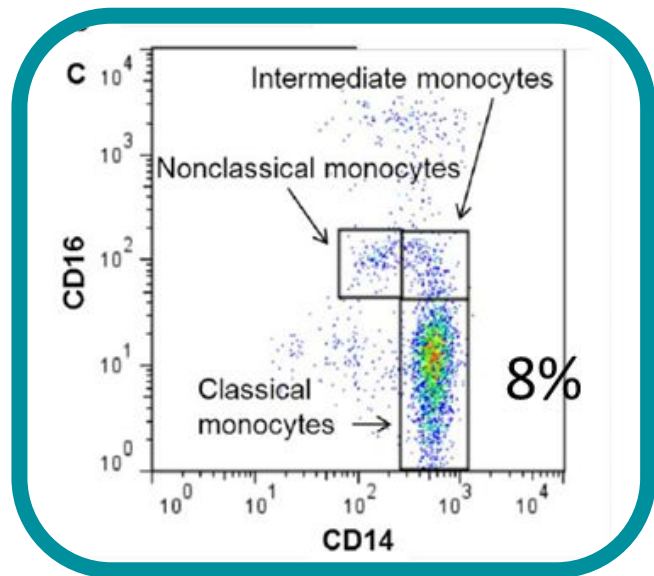


Family

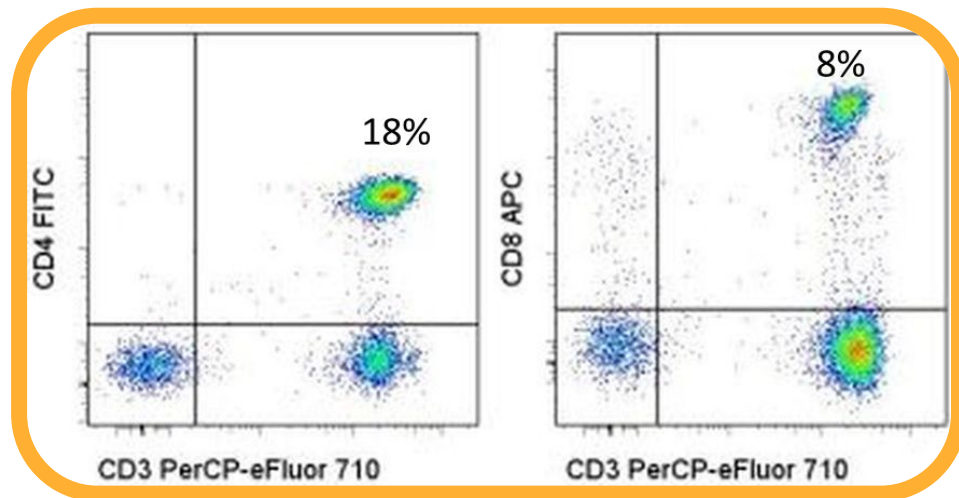
No family history of immune disorders



Flow Cytometry



Normal % Monocytes: 2-8%



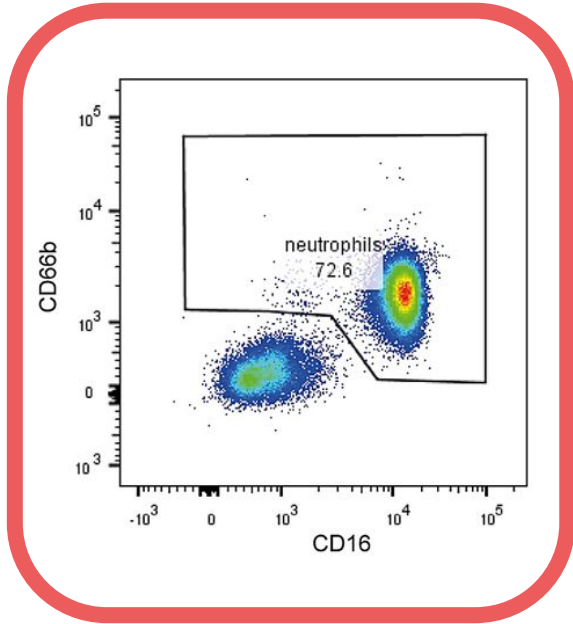
Normal % CD4 T cells:

30-60%

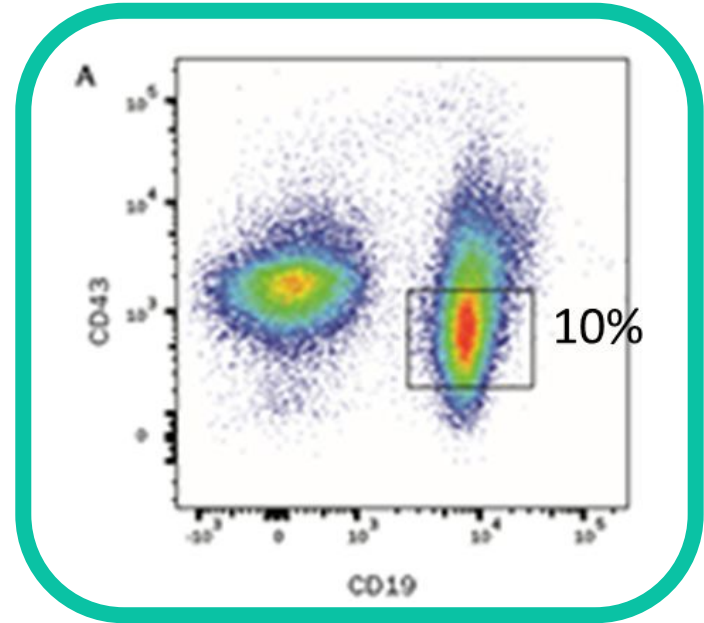
Normal % CD8 T cells:

10-30%

Flow Cytometry

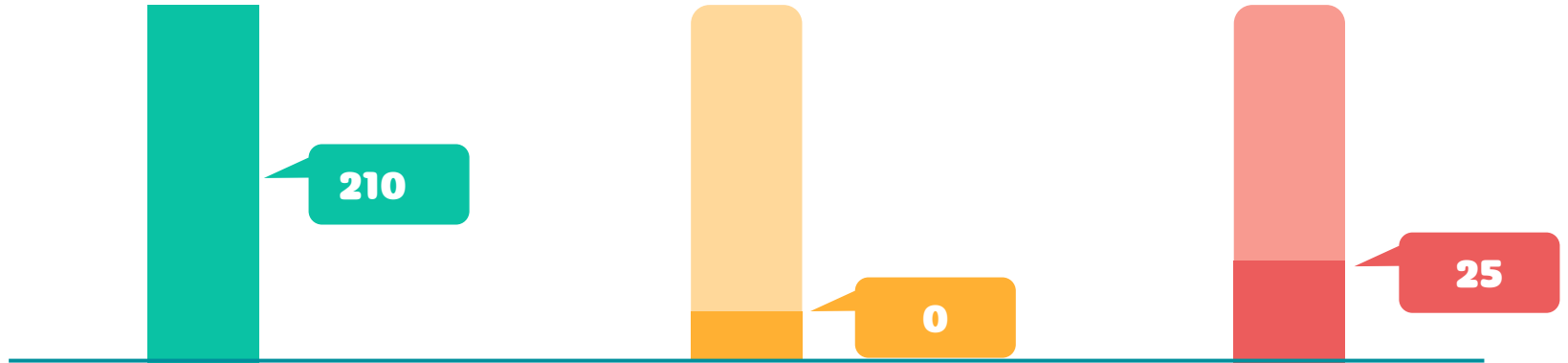


Normal % Neutrophils:
40%-60%



Normal % B cells: 5-10%

Enzyme-Linked Immunosorbent Assay (ELISA)



IgM

Normal serum
antibody levels
(mg/dl): 75-150

IgA

Normal serum
antibody levels
(mg/dl):
150-225

IgG

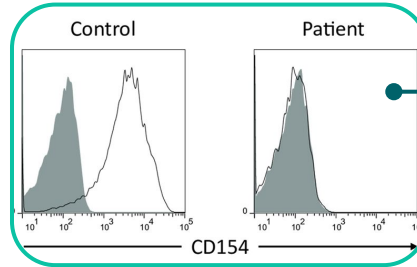
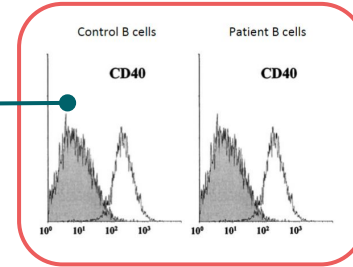
Normal serum
antibody levels
(mg/dl):
600-1500

More Flow Cytometry (And Western Blot Analysis)



B cells

Has CD40 on B cells



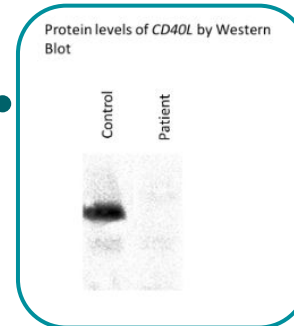
T cells

Has no CD40 ligand on CD3+ T cells

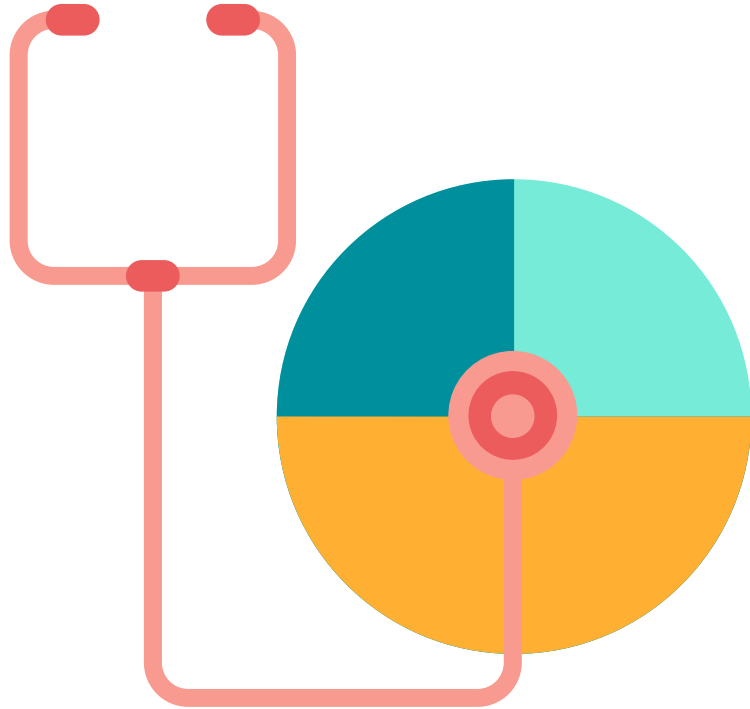


CD40L

Confirmed absence of CD40L with western blot analysis



Diagnosis of Immunodeficiency



Immunoglobulin

Low levels of IgG and
IgA

1

CD40L

Complete absence of
CD40 ligand

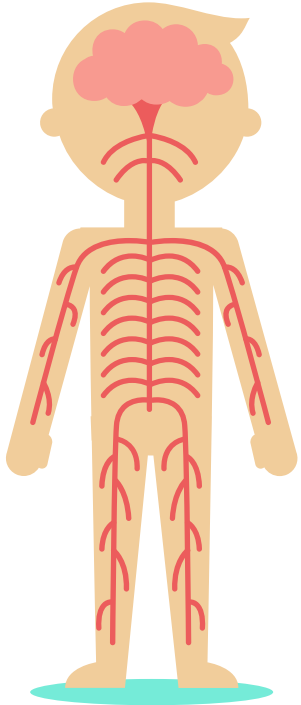
2

Mutation

Mutation in
TNFSF5/CD40L gene

3

Johnny has X-linked Hyper IgM syndrome, Type 1



What is it?

Abnormal B and T cell function; T cells can't communicate with B lymphocytes

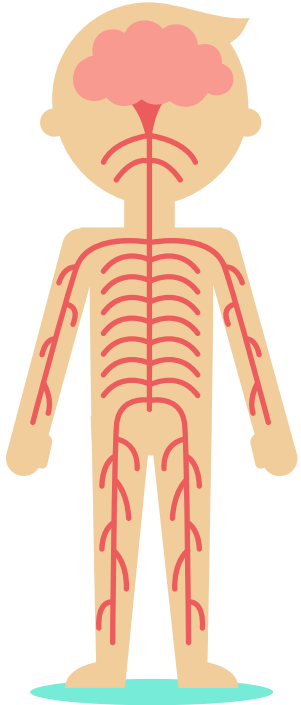
Role of CD40L

Causes recurrent respiratory tract bacterial infections and fungal infections

Prevalence and Similar Conditions

Hematologic, neurologic, liver, oncologic, and gastrointestinal complications

Johnny has X-linked Hyper IgM syndrome, Type 1



Symptoms

Abnormal B and T cell function; T cells can't communicate with B lymphocytes

Complications

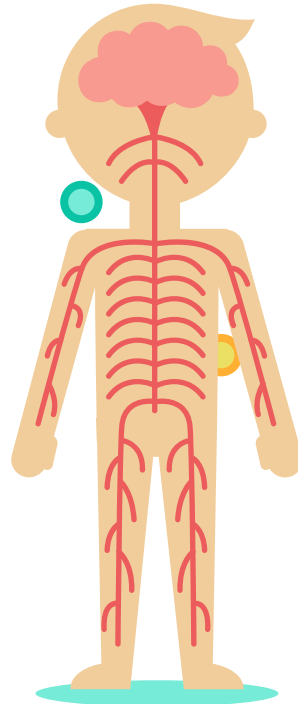
Causes recurrent respiratory tract bacterial infections and fungal infections

Treatments for Hyper IgM Syndrome Type 1



Immunoglobulin Replacement Therapy

Blood-based infusion at regular intervals to raise antibody levels



Antibiotic prophylaxis

Low doses of antibiotics taken everyday to prevent infections



Bone marrow transplant

Healthy blood stem cells from a matched donor



Summary



Tests Ran

- Flow Cytometry
 - Tests to see white blood counts are normal
- ELISA
 - See if antibody levels are normal
- Another Flow Cytometry
 - Look at CD40 and CD40L on T cells
- DNA Mutation Analysis
 - Look at where the DNA mutation is
- Western Blot
 - Determine if the patient has CD40L proteins

Diagnosis

- X Linked Hyper IgM Syndrome Type 1
 - T cells and B cells cannot communicate with each other

Treatments

- Immunoglobulin Replacement Therapy
- Antibiotic prophylaxis
- Bone marrow transplant

