Additional Enzymes

A bunch of bit players

Esoteric Enzymes

Aldolase	5’NT	Pyruvate Kinase	Glucocerebrosidase Galactocerebrosidase
Sphingomyelinase	Alpha-galactosidase-A	Hexosaminidase A	Ceramidase

Aldolase

- Enzyme rises in response to skeletal muscle disorders
  - Muscular dystrophy
  - Inflammation like polymyositis, dermatomyositis
  - Rhabdomyolysis
  - Muscle pain, weakness due to infectious diseases
  - Muscle problem caused by medication

5’-Nucleotidase

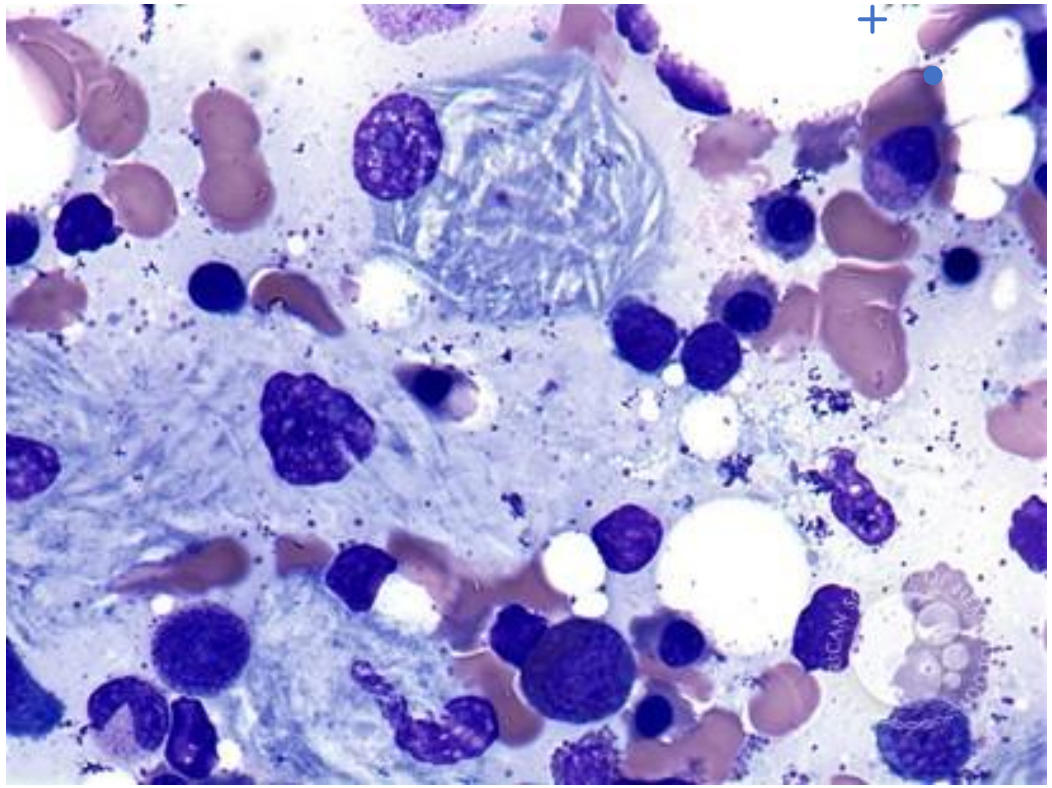
- Originally discovered in snake venom!
- Elevated in hepatobiliary disorders
  - Not elevated in bone disorders like ALKP
  - Used to differentiate source of ALKP
    - Cholestasis, bile duct obstruction, pancreatic tumor, liver flukes etc may cause rise

Pyruvate Kinase

- Enzyme involved in glycolysis AND gluconeogenesis
- Autosomal dominant/recessive inheritance
- RBCs need anaerobic ATP generation
  - Without this Na-K ATPase cells shrivel, die, hemolyze

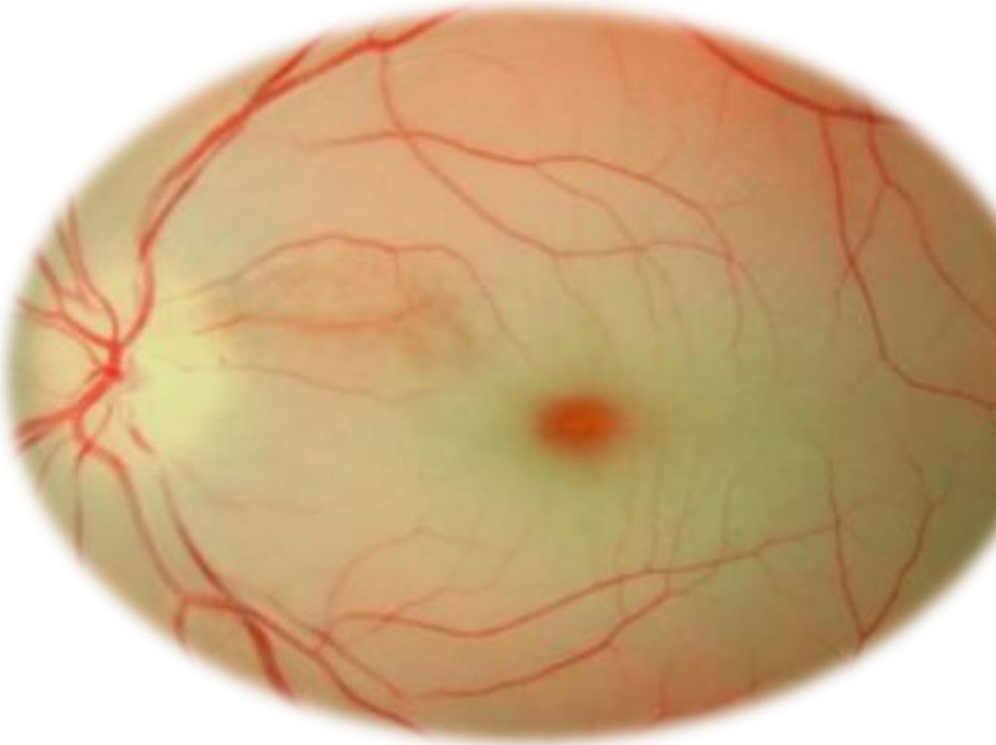
Glucocerebrosidase

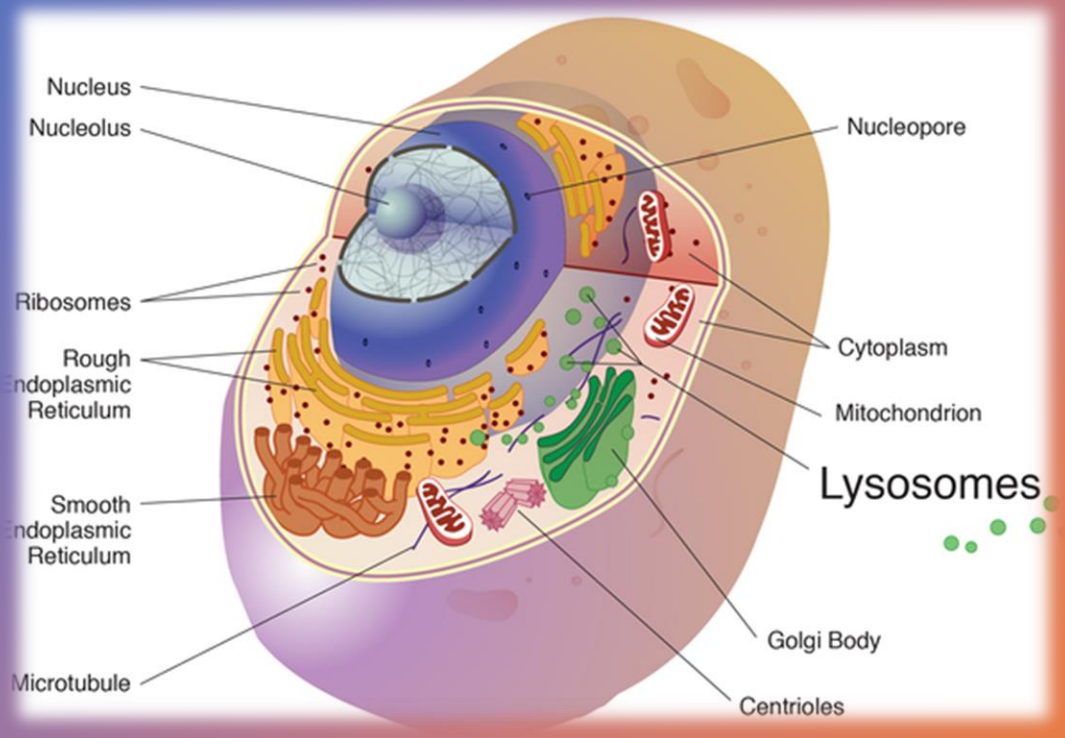
- Enzyme active in lysosomes
- Deficiency causes Gaucher's Disease a lysosomal storage disease
  - Macrophage filled with glucocerebrosides fill organs
  - "Wrinkled tissue paper appearance"
- Recombinant enzyme used for treatment
- Related Enzyme GALACTOcerebrocidase
  - Deficiency causes Krabbe Disease



Sphingomyelinase

- Another lysosomal enzyme deficient in storage disease
- Niemann-Pick Disease
  - Cherry red spot on retina



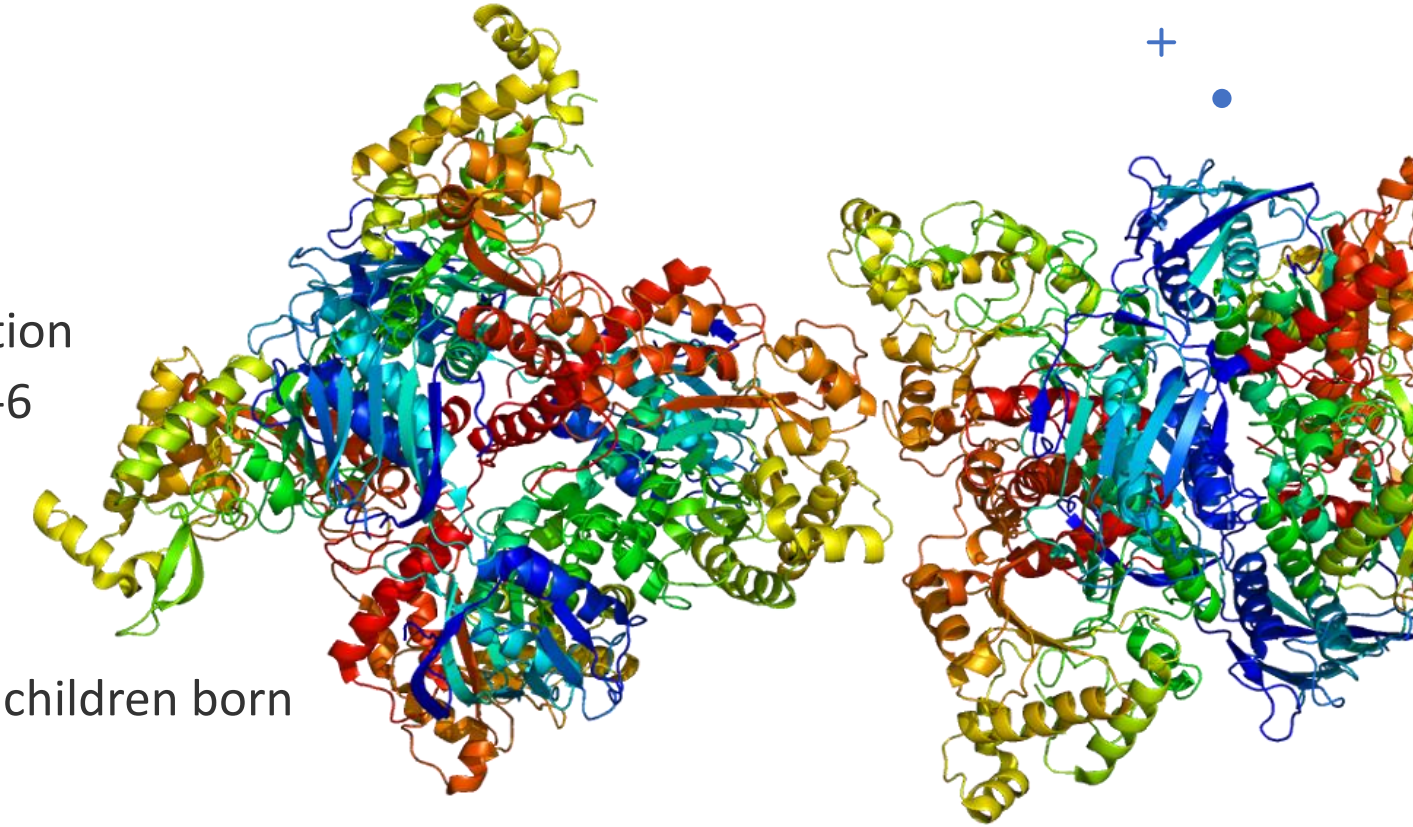


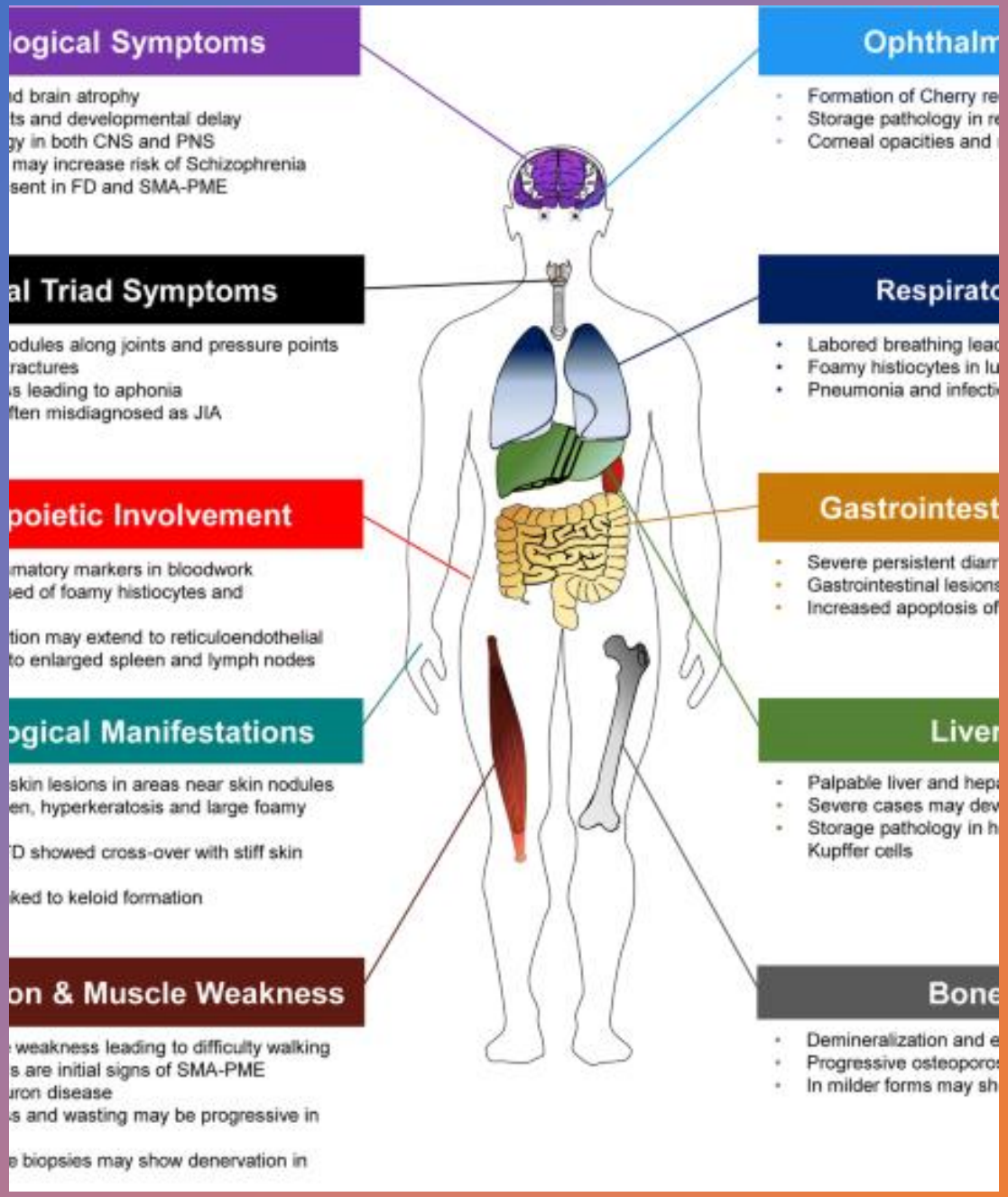
Alpha-galactosidase-A

- X-linked deficiency results in autonomic system infiltration
  - Fabry Disease
  - Mostly affects those male or assigned male at birth (AMAB)

Hexosaminidase A

- Deficiency results in Tay-Sachs
  - Prevalent in Ashkenazi Jewish population
  - Those affected usually die between 2-6
- Genetic counseling success
  - Screening in NY began in 1970s
    - Carrier frequency 1 in 28
    - In 2 years 1000 Ashkenazi Jewish children born 0 cases of Tay-Sachs or Gaucher





Ceramidase

- Farber Disease
  - Develop lipogranulomatosis

Mnemonics to match!

"Gentle Koalas Navigate Furry Tunnels For Fun"

Breaking it down:

- **G**entle: Gaucher (G)
- **K**oalas: Krabbe (K)
- **N**avigate: Niemann-Pick (N)
- **F**urry: Fabry (F)
- **T**unnels: Tay-Sachs (T)
- **F**un: Farber

"Graceful Giraffes Swim Across Huge Canyons, Hunting Zebras"

Breaking it down:

- **G**raceful: Glucocerebrosidase
- **G**iraffes: Galactocerebrosidase
- **S**wim: Sphingomyelinase
- **A**cross: Alpha-galactosidase-A
- **H**uge: Hexosaminidase A
- **C**anyons: Ceramidase