+

Additional Enzymes

+

A bunch of bit players

Esoteric Enzymes

Aldolase

5'NT

Pyruvate Kinase

Glucocerebrosidase Galactocerebrosidase

Sphingomyelinase

Alpha-galactosidase-

Hexosaminidase A

Ceramidase

Aldolase

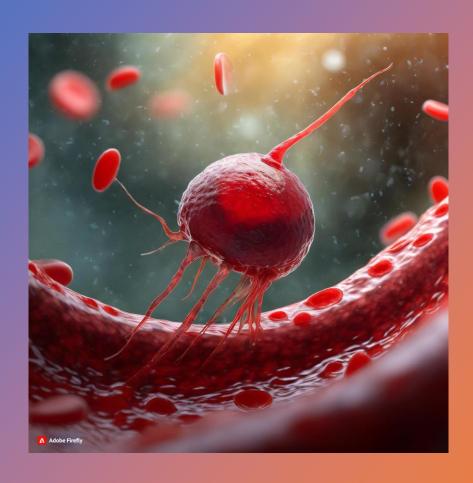
- Enzyme rises in response to skeletal muscle disorders
 - Muscular dystrophy
 - Inflammation like polymyositis, dermatomyositis
 - Rhabdomyolysis
 - Muscle pain, weakness due to infectious diseases
 - Muscle problem caused by medication





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

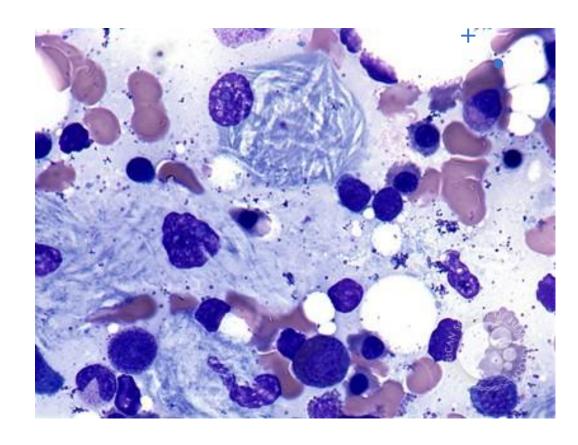


Pyruvate Kinase

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

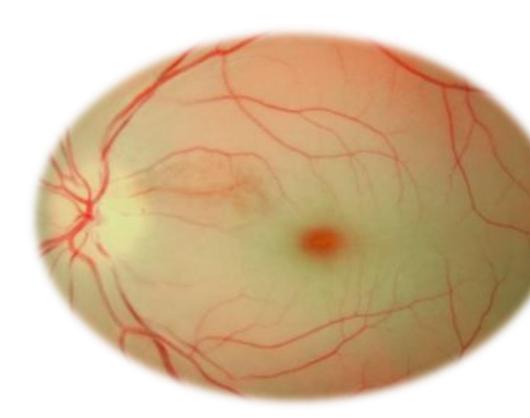
Glucocerebrosidase

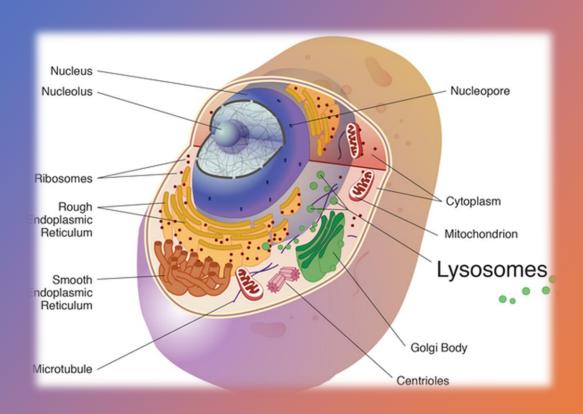
- Enzyme active in lysosomes
- Deficiency causes Gaucher's Disease a lysosomal storage disease
 - Macrophage filled with glucocerebrosides fill organs
 - "Wrinkled tissue paper appearance"
- Recombinant enzyme used for treatment
- Related Enzyme GALACTOcerebrocidase
 - Deficiency causes Krabbe Disease



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

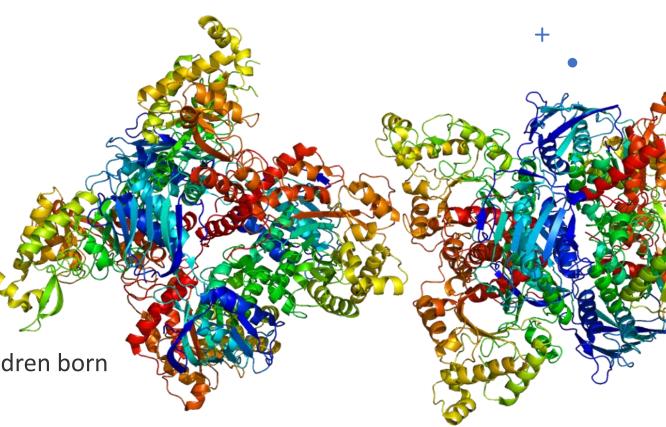
• Those affected usually die between 2-6

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



ogical Symptoms

Id brain atrophy ts and developmental delay gy in both CNS and PNS may increase risk of Schizophrenia sent in FD and SMA-PME

al Triad Symptoms

odules along joints and pressure points ractures is leading to aphonia ften misdiagnosed as JIA

poietic Involvement

matory markers in bloodwork sed of foamy histiocytes and

tion may extend to reticuloendothelial to enlarged spleen and lymph nodes

ogical Manifestations

skin lesions in areas near skin nodules en, hyperkeratosis and large foamy

D showed cross-over with stiff skin

ked to keloid formation.

on & Muscle Weakness

weakness leading to difficulty walking s are initial signs of SMA-PME iron disease

s and wasting may be progressive in

e biopsies may show denervation in

Ophthain

- Formation of Cherry re Storage pathology in re
- Comeal opacities and

Respirato

- Labored breathing lead
- Foamy histiocytes in lu
- Pneumonia and infection

Gastrointest

- Severe persistent diarr
- Gastrointestinal lesions
- Increased apoptosis of

Liver

- Palpable liver and hepa
- Severe cases may dev
- Storage pathology in h Kupffer cells

Bone

- Demineralization and e
- Progressive osteoporos
- In milder forms may sh

Ceramidase

- Farber Disease
 - Develop lipogranulomatosis

Mnemonics to match!

"Gentle Koalas Navigate Furry Tunnels For Fun"

Breaking it down:

Gentle: Gaucher (G)

Koalas: Krabbe (K)

Navigate: Niemann-Pick (N)

Furry: Fabry (F)

Tunnels: Tay-Sachs (T)

Fun: Farber



"Graceful Giraffes Swim Across Huge Canyons, Hunting Zebras"

Breaking it down:

- Graceful: Glucocerebrosidase
- Giraffes: Galactocerebrosidase
- Swim: Sphingomyelinase
- Across: Alpha-galactosidase-A
- Huge: Hexosaminidase A
- Canyons: Ceramidase

