

# Disorders associated with metabolism pathways

1) Oxphos Diseases (Genetic) mutations in mtDNA  
 mutations in nDNA  
 Leigh Syndrome → Complex I, II, IV

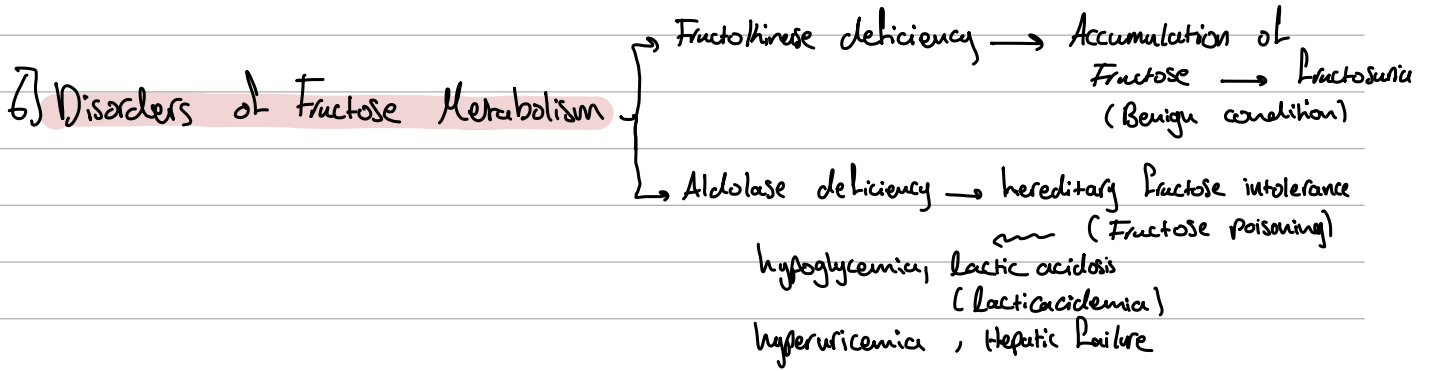
Leber's hereditary optic neuropathy  
 Complex I → LHON  
 Complex III → Sporadic myopathy  
 Complex IV → Sporadic myopathy  
 → Encephalomyopathy  
 Complex V → NARP  
 → Neuropathy, ataxia, Retinitis pigmentosa

2) Carbohydrate digestion  
 → Sucrase-isomaltase deficiency → Gases, organic acids, other osmotically active molecules  
 → Lactase deficiency → Lactose Permentation by bacteria  
 → Permentation of Sucrose  
 → Carbon metabolites → bloating  
 →  $\text{CO}_2$   
 →  $\text{H}_2$  → lost → diarrhea  
 → Can be measured in breath

3) Glycolysis → Pyruvate kinase deficiency → RBCs are affected → Mild to Chronic hemolytic anemia

4) Pyruvate dehydrogenase deficiency → Affect brain → Dependent on TCA cycle  
 → sensitive to acidosis  
 → neurodegeneration, muscle spasticity, early death in neonatal onset

5) Glycogen storage diseases  
 → Von Gierke disease → Glucose-6 phosphatase  
 → Liver, kidney and intestine → Prolonged hypoglycemia  
 → renal disease  
 → Hepatomegaly fatty liver  
 → Growth retardation  
 → McArdle syndrome → Muscle glycogen phosphorylase  
 → ATP shortage → weakness after exercise  
 → Pompe disease → Lysosomes  $\alpha$  (1-4) glucosidase → liver, heart muscle  
 → Massive Cardiomegaly → early death from heart failure  
 → Normal Blood sugar and glycogen storage



Similar consequences that in fructose intolerance

