Von Gierke's disease

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Surat
Glucose -6-Phosphatase

• Glucose 6 Phosphatase is required in gluconeogenesis as well as glycogenolysis.
• It converts Glucose-6-phosphate into glucose.
• Physiologically, Glucose-6-phosphatase is absent in muscle. It is only present in Liver.
• Hence, liver helps to provide glucose from glycogen as well as substrate of gluconeogenesis.
Von Gierke's disease

- Deficiency of Glucose-6-phosphatase enzyme
- Inability of liver to provide glucose from glycogen as well as gluconeogenesis.
Von Gierke's disease

• Substrate Accumulated
  – Increase Glucose 6 Phosphate
    • More HMP
    • More Ribose 5 Phosphate
    • More PRPP
    • More Nucleic acid
    • More Break down of Nucleic acid
    • More Uric acid
Von Gierke's disease

- Increase Pyruvic acid
  - More Acetyl CoA
  - More TCA cycle
  - More formation of Cholesterol, Fatty Acid, Ketone body
- Increase Lactic acid
  - Metabolic acidosis
  - What happen to uric acid???
    - Increase Uric acid precipitation
    - Increase formation Sodium Urate Crystal
- Decrease Glycogen utilization
  - Increase un-utilized glycogen storage
  - Glycogen Storage Disease
Clinical Feature of Von Gierke's disease

• Clinical Feature
  – Hypoglycemia
  – Retard growth
  – Lactic acidosis
  – Ketosis
  – Hyperlipidemia
  – Hyperuricemia
  – Gouty arthritis
  – Cirrhosis