Von Gierke's disease

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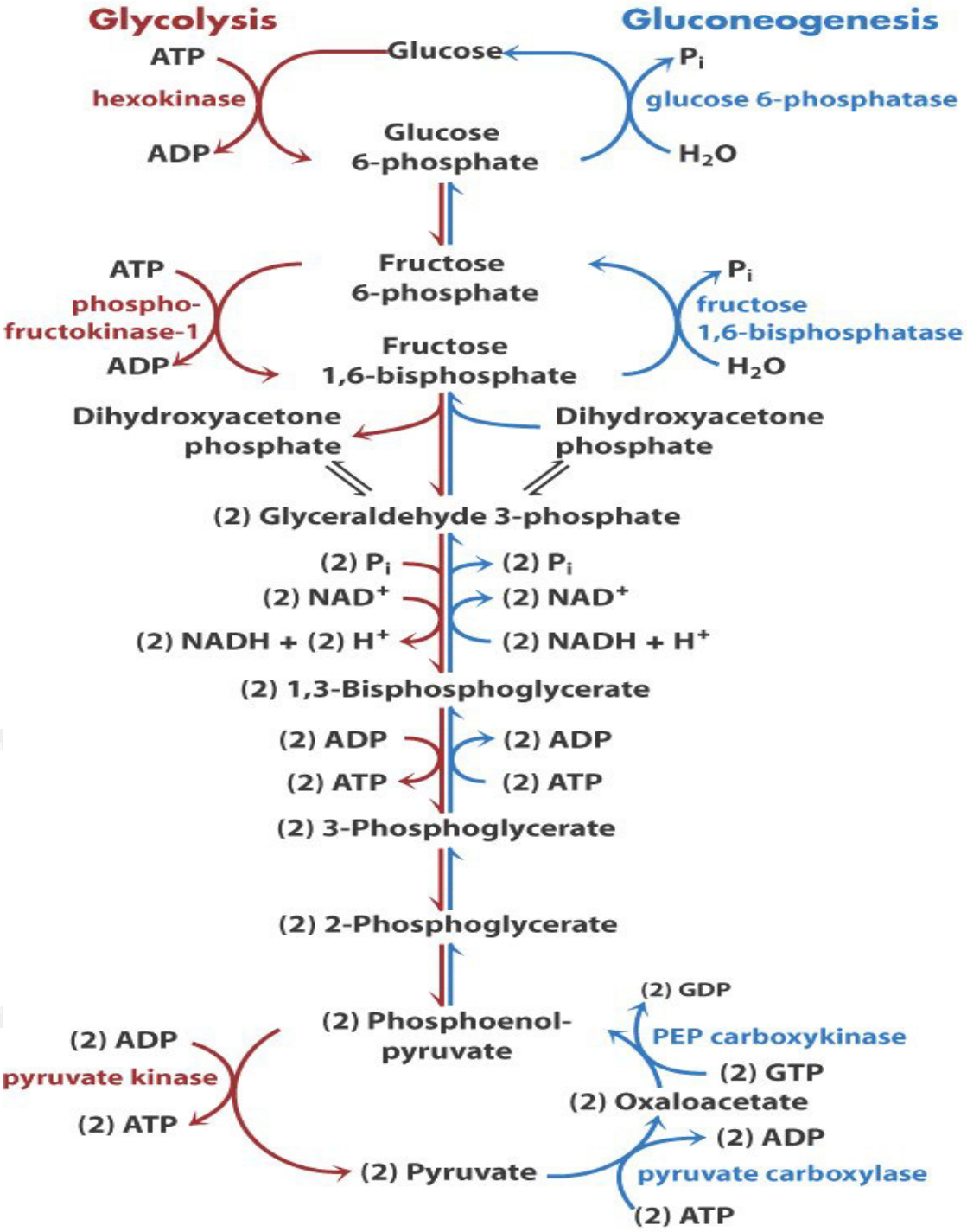
Glucose -6-Phosphatase

- Glucose 6 Phosphatase is require in gluconeogenesis as well as glycogenolysis.
- It convert Glucose-6-phosphate into glucose.
- Physiologically ,Glucose-6-phosphatase is absent in muscle. It is only present in Liver.
- Hense, liver help to provide glucose from glycogen as well as substrate of gluconeogenesis.


Glycolysis
Gluconeogenesis
ATP
hexokinase
ADP
Glucose
Glucose 6-phosphate
P_i
glucose 6-phosphatase
H_2O
ATP
phospho-fructokinase-1
ADP
Fructose 6-phosphate
Fructose 1,6-bisphosphate
P_i
fructose 1,6-bisphosphatase
H_2O
Dihydroxyacetone phosphate
Dihydroxyacetone phosphate
(2) Glyceraldehyde 3-phosphate
(2) P_i
(2) NAD^+
(2) NADH + (2) H^+
(2) P_i
(2) NAD^+
(2) NADH + H^+
(2) 1,3-Bisphosphoglycerate
(2) ADP
(2) ATP
(2) ADP
(2) ATP
(2) 3-Phosphoglycerate
(2) 2-Phosphoglycerate
(2) Phosphoenol-pyruvate
(2) ADP
pyruvate kinase
(2) ATP
(2) Pyruvate
(2) GDP
PEP carboxykinase
(2) GTP
(2) Oxaloacetate
(2) ADP
pyruvate carboxylase
(2) ATP

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