Glycogen Storage Diseases

↓

metabolic defects

↓

concerned with

- glycogen synthesis ←
- → degradation.

* It is due to defect in enzymes which can be generalized or tissue specific.

Types of glycogen storage diseases.

- → Type I (Von Gierke disease)
- → Type II – (Pompe's disease)
- → Type III – (Cori's disease)
- → Type IV – (Anderson's disease)
- → Type V – (McArdle's disease)

(1) Von Gierke disease – Type I

caused due to the deficiency of enzyme

Glucose-6-phosphatase

Glucose-6-phosphate $\xrightarrow{\text{Glucose-6-phosphatase}}$ Glucose

- deficiency of Glucose-6-phosphatase
↓ leads to
Glucose-6-Phosphate ↑↑↑↑
↓
[DEPTH OF BIOLOGY]
accumulated in liver.
↓
increase the size of liver
Hepatomegaly.

* This causes Hypoglycemia or low blood sugar
↓
No free glucose form to ↑ the blood glucose.
↓
- This also causes Hyperlipidemia and Hyperuricemia (fat/protein catabolism) [DEPTH OF BIOLOGY]

(2) Pompe disease (Type II)
↓
genetic disorder → glycogen gets build up in body's cells.

* This results from the deficiency of an enzyme called acid alfa glucosidase (GAA)
↓
This helps in breakdown of glycogen in body.

(3) Cori Disease Type III [DEPTH OF BIOLOGY]
caused due to lack of debranching enzyme
↓
α-1,6-Glucosidase
* Its deficiency causes abnormal glycogen str.

* Normally:

- glycogen is branched
- debranching enzyme cut 1-6 bond and release it!

↓

so that it can become glucose.

→ Due to the deficiency of enzyme

↓

Bond remains uncut

↓

leads to abnormal str. of glycogen

↓

This cause some symptoms like Von Gierke disease

↓

because there too no free glucose is produced

↓

causes hypoglycemia

and abnormal accumulation of super branched glycogen structures in liver

↓

causing Hepatomegaly.

(4) Andersen disease - Type IV

also known as glycogen storage disease (GSD)

- caused by deficiet activity of glycogen branching enzyme.

This results in accumulation of abnormal glycogen in liver, muscle, and/or in other tissues. [DEPTH OF BIOLOGY]

(5) McArdle's disease / Hers's disease - Type V

- caused due to lack of Glycogen Phosphorylase.
  - In Muscles → McArdle's disease
  - In Hepatic → Hers's disease.

* McArdle's disease (muscle)

- muscle cramp on exertion
- Hypoglycemia on exertion
- Myoglobinuria

[DEPTH OF BIOLOGY]

* Hers's disease

- Hepatomegaly
- fasting hypoglycemia.

[DEPTH OF BIOLOGY]