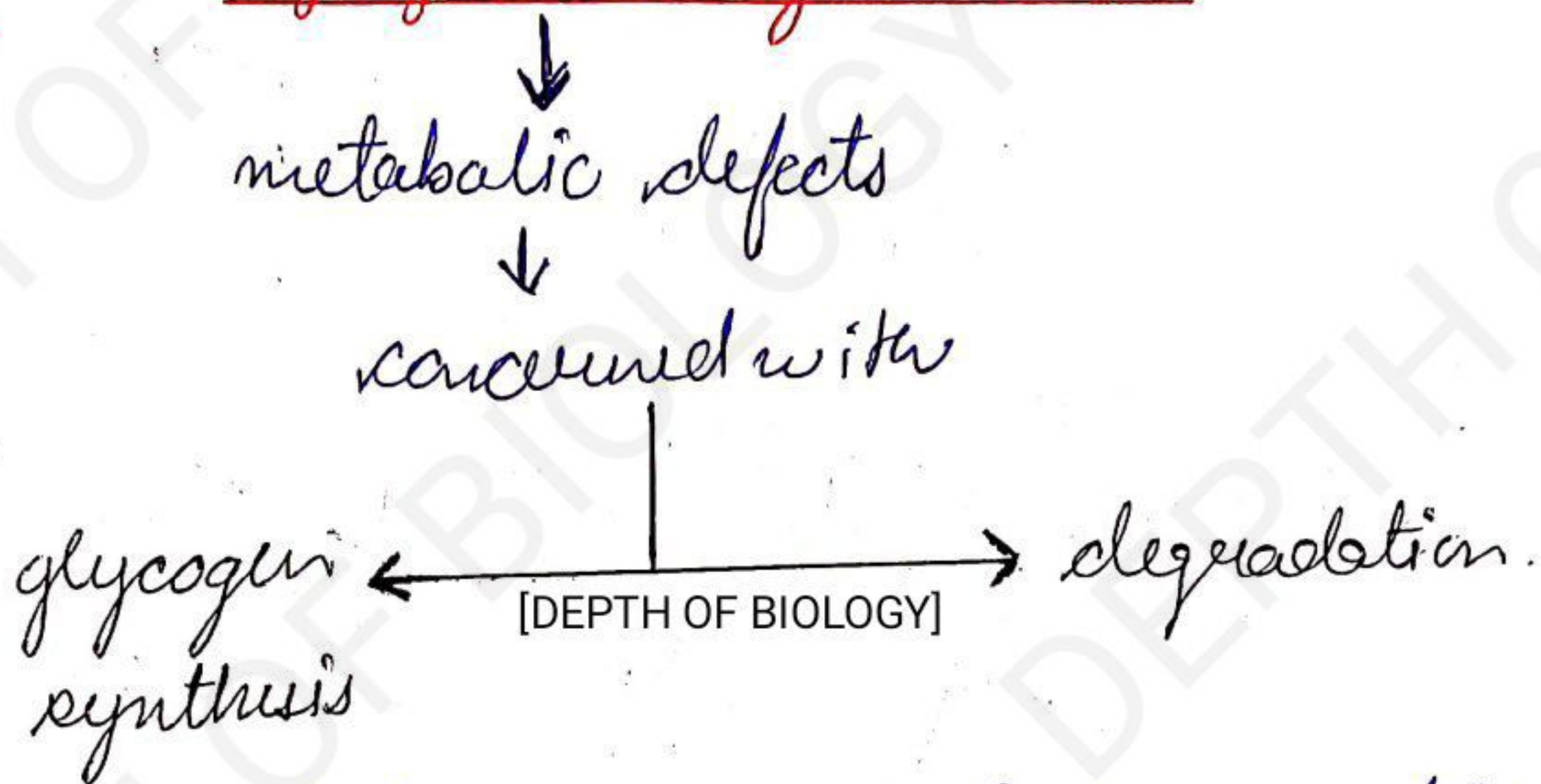


Glycogen Storage Diseases



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

Types of glycogen storage diseases.

[DEPTH OF BIOLOGY]

- Type I (von Gierke disease)
- Type II - (Pompe's disease)
- Type III - (Cori's disease)
- Type IV - (Andersen's disease)
- Type V - (McArdle's disease)

① Von Gierke disease - Type I [DEPTH OF BIOLOGY]

caused due to the deficiency of enzyme

Glucose-6-phosphatase



• 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 from 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 alpha 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!



[DEPTH OF BIOLOGY]

so that it can become glucose.

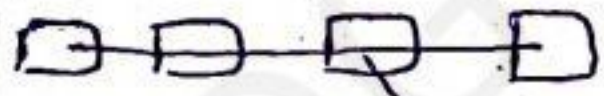
→ Due to the deficiency of enzyme



Band remains uncut



leads to abnormal str.
of glycogen



This cause some symptoms like
Van Gierke disease



[DEPTH OF BIOLOGY]

because here too no free
glucose is produced



causes hypoglycemia
and abnormal accumulation of super branched
glycogen structures in liver



causing Hepatomegaly.

(4) Anderson disease - Type IV

also known as glycogen storage disease (GSD)

- caused by deficient activity of glycogen branching enzyme.

[DEPTH OF BIOLOGY]

↓

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

⑤ McArdle's disease / Her's disease - Type V

• caused due to lack of Glycogen Phosphorylase.

• In Muscles → McArdle's disease

• In hepatic → Her's disease.

* McArdle's disease (muscle)

- muscle cramp on exertion
 - Hypoglycemia on exertion
 - Myoglobinuria
- [DEPTH OF BIOLOGY]

* Her's disease

- Hepatomegaly
- fasting hypoglycemia.

[DEPTH OF BIOLOGY]