Glycogenosis / Questions and case reports

Questions

1. **Glycogen biosynthesis**
   - A – requires inorganic phosphate as one of the substrates
   - B – involves the formation of α-1 → 6 branches by glucan unit transfer from α-1 → 4 bonds
   - C – includes synthesis of UDP-glucose directly from uridine triphosphate and glucose-6-phosphate
   - D – involves the transfer of glucose residue in UDP-glucose to the reduced end of the "primer" of glycogen
2. **Glucagon acts by:**
   - A – inhibits cAMP-dependent protein kinase in the liver
   - B – stimulates glycolysis in the liver
   - C – stimulates gluconeogenesis in muscles
   - D – stimulates glycogen phosphorylase phosphorylation in the liver
   - E – stimulates glycogen synthase dephosphorylation in the liver
3. **Type I glycogenosis (Gierke´s disease) is caused by:**
   - A – hepatic glucose-6-phosphate dehydrogenase deficiency
   - B – glucose-6-phosphatase deficiency in the liver and kidneys
   - C – an abnormal structure of liver glycogen
   - D – amylo-1 → 6-glucosidase deficiency in the liver and muscles
   - E – hepatic phosphorylase deficiency

Answers

Case reports

Newborn slightly hypotrophic, with cyanosis and marked hypoglycemia

It is a newborn born in the 38th week of gestation, 2,100 g (adequate weight: 3,300 g), length 47 cm, slightly cyanotic (for hypoxia), with tachycardia (35 / min). Glycaemia: 0.8 mmol / l (lower limit for newborns: 2.5 mmol / l). Mother 35 years. In the last trimester, she had mild hypertension and recurrent urinary tract infections, vomiting and eating very little.

Questions:

1. What is the cause of such low levels in the newborn?
2. How is the energy metabolism of a fetus different from a newborn?

Answers

An infant with recurrent hypoglycaemia

An infant with recurrent hypoglycemia from birth was tested with glucagon and 1 hour after a carbohydrate diet. Glycaemia rose from 3.9 mmol / l to 6.1 mmol / l. 3 hours later, the blood glucose dropped to 2.5 mmol / l. However, no increase in blood glucose was observed after further glucagon administration.

Question: What is the cause of this form of glucagon response

- A – Deficiency of hepatic glycogen phosphorylase or glycogen "mediator"
- B – Glucose-6-phosphatase deficiency.
- C – Defect in the glucagon receptor.
- D – Inability to secrete an adequate amount of glucagon.
- E – Problem in gluconeogenesis.

Answers

Patient with type V glycogenosis (McArdle´s disease) and patient with type VI glycogenosis (Hers´disease)

The cause of both conditions is an inherited deficit of a key glycogen degradation enzyme: glycogen phosphorylase.

Question:

1. Which of these types has more severe clinical symptoms and why?

Answers

References

Related articles

Other chapters from the book MASOPUST, J., PRŮŠA, R .: Pathobiochemistry of metabolic pathways

Source

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- MASOPUST, Jaroslav and Richard PRŮŠA. *Pathobiochemistry of metabolic pathways.* 1st edition. Prague: Charles University, 1999. 182 pp. 38- 40. ISBN 80-238-4589-6 .