Crigler-Najjar syndrome

Template:Infobox - Genetic disease

Crigler-Najjar syndrome type I

- AR hereditary complete defect of bilirubin-uridine diphosphate-glucuronyl-transferase (UDPG-transferase, UGT1A1 (http://omim.org/entry/191740)) hepatocytes.
- The most severe indirect (unconjugated) hyperbilirubinemia with early complications (nuclear jaundice).

Etiology

- Complete absence of bilirubin-uridine diphosphate glucuronyl transferase → bilirubin conjugation disorder → bilirubin cannot be excreted in the bile

Clinical course

- Icterus with a sharp rise in unconjugated bilirubin during the first hours of life.
- Bile is colorless.
- Stool is brown - the transfer of unconjugated bilirubin through the intestinal mucosa.
- Urine is light, no bilirubin can be detected in it (unconjugated does not pass into the urine)
- Without therapy, nuclear icterus soon develops.

Diagnosis

- Lack of UDPG-trasferase activity in the liver

Therapy

- Intensive phototherapy and exchange blood transfusion, cholestyramine administration.
- Liver transplantation[1].

Crigler-Najjar syndrom type II

- AD hereditary partial defect in bilirubin uridine diphosphate glucuronyl transferase (UDPG transferase) hepatocytes.
- Mild indirect (unconjugated) hyperbilirubinemia.

Clinical course

- Icterus with unconjucated hyperbilirubinemia.
- Bile and urine are stained, conjugated bilirubin can be detected.

Diagnosis

- Lower activity of UDPG-transferase in the liver.

Therapy

- Enzyme induction by phenobarbital[1].

Links

Related articles

- Hyperbilirubinemia in newborns and infants
- Juvenile hyperbilirubinemia

References

1. MUNTAU, Ania Carolina. *Pediatrisc.* 4. edition. Praha : Grada, 2009. pp. 386-388. ISBN 978-80-247-2525-3.

Source

- ws:https://www.wikiskripta.eu/index.php?curid=11500