Pediatric Oncall Journal (January-March 2020)
Volume 17, Issue 1: 29
DOI: https://doi.org/10.7199/ped.oncall.2020.3

TEACHING FILES (GRAND ROUNDS)

INDIRECT HYPERBILIRUBINEMIA IN A TODDLER

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ARTICLE HISTORY
Received 22 Mar 2019
Accepted 12 Apr 2019

Clinical Problem

A 1½ years old boy born of non-consanguineous marriage presented with jaundice and clay-colored stools since day 3 of life. There was no itching but there were clay stools and pale urine. The child also had pica. His milestones are normal. He had a 5 years old brother who was asymptomatic and a sister who died at 5 months of age due to indirect jaundice. On examination, weight was 11.6 kg, he had jaundice and systemic examination was normal. There was no organomegaly. He had been investigated for his jaundice previously (table 1). HIV ELISA, HBsAg and Hepatitis C antibody were negative. His liver function test results are shown in Table 1.

Table 1 : Serial Liver Function tests of the child

Age of the child	$2^1/_2$ months	$3^1/_2$ months	6 months	9 months	10 months	16 months
Bilirubin (direct) (mg/dL)	11.1 (0.7)	14.8 (0.9)	19.5 (1.09)	21.8 (1.4)	28.9 (1.6)	22.2 (1.7)
SGOT/SGPT (IU/L)		44/57				
Total proteins/albumin (g/dL)		5.4/3.5				
Alkaline phosphatase (IU/L)		448				

Note : SGOT - Serum glutamic oxaloacetic transaminase, SGPT - Serum glutarric pyruvic transaminase

What is the likely diagnosis and how to manage this child?

Expert Opinion :

Crigler-Najjar syndrome (CNS) most probably type 2. CNS is a rare disease characterized by a complete or partial deficiency of the hepatic enzyme uridine diphosphate-glucuronosyl-transferase (UDP-GT). CNS is inherited in an autosomal recessive manner. Complete deficiency of UDP-GT leads to CNS type 1. It is a severe form of the disease characterized by the accumulation of indirect bilirubin in the basal ganglia and cerebellum leading to kernicterus.[1] Serum bilirubin is usually above 20 mg/dL.[2] CNS type 2 is a due to partial deficiency (decrease by more than 30%) of UDP-GT, thus, it is not as severe as CNS type 1 and generally does not lead to kernicterus.[1] Serum bilirubin levels are typically between 10 and 20 mg/dL but can go up to 40 mg/dL during stressful conditions like pregnancy, infection, intake of some drugs and pregnancy.[3] Diagnosis of CNS can be established by genomic DNA analysis or liver biopsy denoting UDP-GT level.[2] This boy is icteric with no neurological deficits and serum bilirubin level below 20 mg/dL with occasional rise above 20 mg/dL (likely due to an acute exacerbation), thus, the most likely diagnosis in this boy is CNS type 2. Management of CNS type 2 is conservative. Lifelong administration of phenobarbitone is recommended in these patients. Phenobarbitone induces hepatic enzymes and causes a significant reduction in serum bilirubin levels. Additional calcium supplementation can aid in excretion of bilirubin from the gut. Hyperbilirubinemic crisis is managed by plasmapheresis or phototherapy.[4]

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Compliance with Ethical Standards

Funding: None

Conflict of Interest: None

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