An Unusual Presentation of Gilbert's Syndrome in a Neonate

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Introduction

Prolonged jaundice is a common clinical presentation, of which the vast majority of children will be found to have a benign unconjugated hyperbilirubinemia. However, a small group of children will be found to have underlying conditions which may need further investigation and management.[1]

Gilbert's syndrome is generally considered to be a benign condition associated with intermittent and asymptomatic episodes of jaundice secondary to a mild unconjugated hyperbilirubinaemia commonly noted in times of stress, dehydration, or illness. It is not usually noted to cause significant hyperbilirubinaemia, and does not routinely require treatment.[2]

History and Case

A term 29-day old baby boy presented to the Prolonged Jaundice Clinic and was found to be significantly jaundiced with a bilirubin of 617mmol/litre on a blood gas. He was immediately referred to Paediatric A&E where this was confirmed on laboratory serum bloods and established to be mostly unconjugated.

Despite his age, he was started on intensive phototherapy and treated for sepsis. On examination, he was clinically well despite the generalised jaundice, with no evidence of encephalopathy or dysmorphic features, and was noted to have pigmented stools. There were no concerns regarding his feeding or growth at the time, nor was there any family history of any medical issues and his parents were non-consanguineous.

Treatment and Investigations

The bilirubin levels were responsive to phototherapy, and so the lights were weaned down relatively quickly, although the levels would rapidly rise each time the phototherapy was stopped, requiring intermittent treatment with phototherapy.

A trial of exclusive formula feeds was started to rule out breast-milk induced jaundice, but his bilirubin levels continued to rise. After discussion with the local tertiary liver centre, he was also started on a regular dose of phenobarbitone and ursodeoxycholic acid.

An ultrasound scan of his abdomen showed no abnormality. His other liver function tests were normal, and a screen for G6PD, hepatitis, infections, and other first line tests for unconjugated hyperbilirubinaemia were all normal. Genetic tests were also sent for Crigler-Najjar syndrome and Gilbert's syndrome, for which he was found to be homozygous for the c.-41_-40TA[7] variant in the UGT1A1 promoter which is associated with Gilbert's syndrome.

He was discharged from the Neonatal Unit and continued on intermittent home phototherapy with the assistance of the PATCH Nursing Team in order to maintain low bilirubin levels whilst long-term management plans could be instigated.

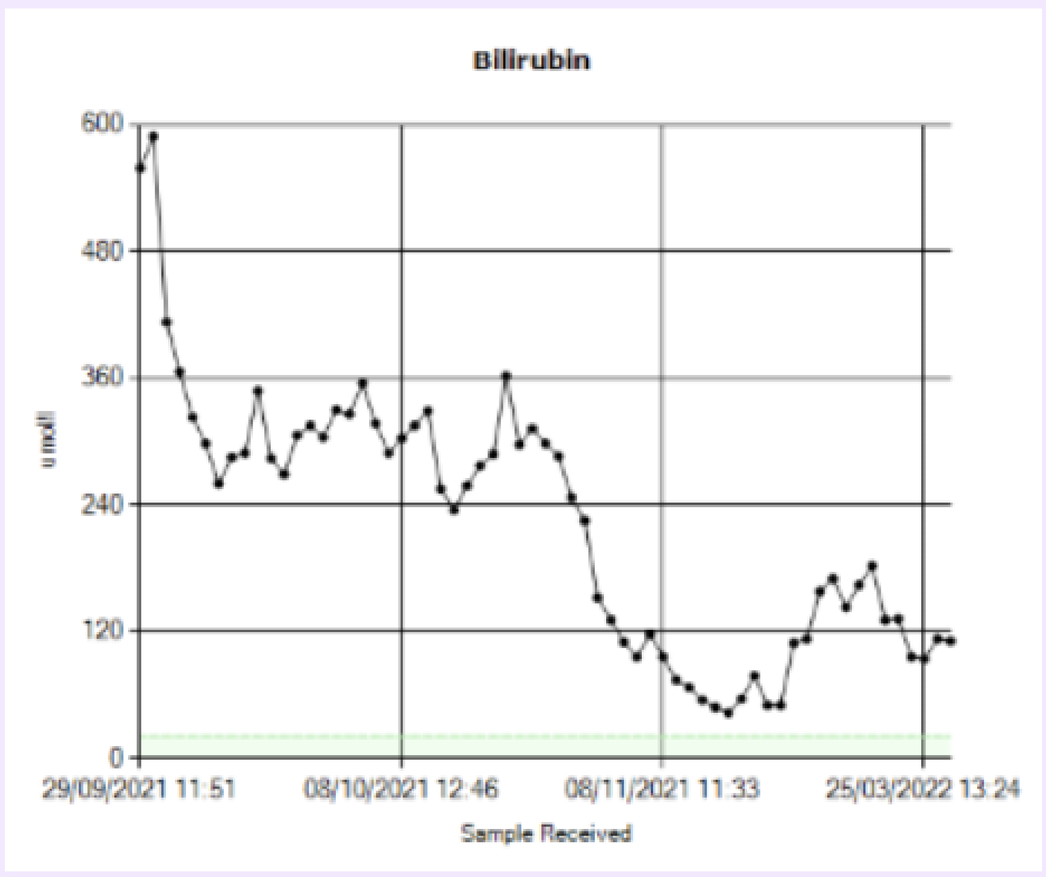

Fig.1: Graph to show the fluctuating levels of bilirubin.

Service Provision

This case allowed us to consider how we could provide care for the patient and family within the home setting and whether other babies requiring phototherapy may benefit from treatment at home. We are very grateful for the PATCH team for regularly attending their home for reviews and blood tests.

Conclusion

Despite prolonged jaundice being a common and usually benign presentation, it is important to always consider the causes which may need further investigation and management.

Gilbert's syndrome is not usually considered as a cause of prolonged and significant jaundice needing home phototherapy, but we present an unusual cause where no other cause for the persistent hyperbilirubinaemia could be identified.

The diagnosis of Gilbert's syndrome must therefore always be suspected when persistent jaundice and raised unconjugated hyperbilirubinemia occur in the absence of other diagnoses.

References

1. Gilmour, S. "Prolonged neonatal jaundice: When to worry and what to do." *Paediatrics & Child Health* vol. 9,10 (2004): 700-704.
2. Bosma, P.J. "Inherited disorders of bilirubin metabolism." *Journal of Hepatology*, vol. *38* (2003): 107–117.