of GERD diagnosis was 11.6% in type II, and 6.3% in type III. Pulmonary complication such as recurrent pneumonia occurred in a half of patients with both GERD and insufficient weight gain.

Swallowing difficulties are very common in type II SMA. GERD and esophageal complaints also occurs in a significant portion of type II and III SMA. Early recognition and intervention of such gastrointestinal dysfunction is mandatory.

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CRIGLER-NAJJAR SYNDROME TYPE II CAUSED BY HOMOZYGOUS DOUBLE MUTATION [T1456G, G211A] OF *UGT1A1* GENE IN A TAIWANESE SIBLING

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Objective: This study aimed to analyze the mutations of UDP-glucuronosyltransferase 1A1 (*UGT1A1*) gene in a Taiwanese family.

Patients and methods: Two brothers aged 24 and 2 months, respectively were born to a Chinese mother who married to a Taiwanese man. They had profound unconjugated hyperbilirubinemia in the newborn period (30.7 and 25.2 mg/dL, respectively). The liver function, hemoglobin and glucose-6-phosphate dehydrogenase levels were within normal limit. They both received intensive phototherapy and the serum bilirubin level gradually decreased to 6-10 mg/dL. The blood samples of the brothers and their parents were sent for DNA analysis including the promoter area of the UGT1A1 gene, exons 1-4, the coding region of exon 5, and their flanking intronic regions. The polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) and direct DNA sequencing were used.

Results: The PCR results for the *UGT1A1* gene showed homozygous G > A substitution at nucleotide 211 in exon 1 and homozygous T > G substitution at nucleotide 1456 in exon 5 ($Gly^{71}Arg$ plus $Tyr^{486}Asp$) in both patients. No other variation was detected in the *UGT1A1* gene. Analyzing the *UGT1A1* gene of the parents showed a homozygous 211G > A plus a heterozygous 1456 T > G variations in their father and a heterozygous 211G > A plus a heterozygous 1456 T > G variations in their mother. All of the tests were confirmed by the direct gene sequence.

Conclusion: Double homozygous mutation [T1456G, G211A] of *UGT1A1* gene is rare in CN-2 and is a novel mutation in Taiwanese children.

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PROLONGED JAUNDICE SCREEN AUDIT-RATIONAL APPROACH TO INVESTIGATIONS

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Background: Extensive laboratory investigations are routinely performed to exclude rare but potentially catastrophic causes of prolonged jaundice.

Aims: This study aims to review the investigations and outcomes of Prolonged Jaundice Screening (PJS) and considers whether it is possible to safely reduce the number of unnecessary blood tests.

Methods: Retrospective analysis of all babies for PJS, between June 2008 and March 2010. Data from the Prolonged Jaundice Proforma was collated in Excel and was analysed using simple statistical methods and percentage calculations.

Results: 100 babies reviewed, age of presentation were between 2 to >8weeks. 97% were fully or partially breast fed and 97.3% showed adequate growth. 90.4% term and 9.6% pre-term. Stool colour and clinical examination was normal in all cases.

8% had conjugated bilirubin above 20% of the total, 19% were noted to have >200mmol/L unconjugated bilirubin levels. All were subsequently found to have normal second line investigations. 55% of babies had abnormal blood results requiring repeat testing. All results normalised on subsequent testing (up to five repeats).

Urine was positive in 25 cases with possible contamination in 20 cases and E.coli in five cases. 2 babies had positive reducing substances, however, Galactosaemia was ruled out in both cases. No cases of cholestasis were identified.

Conclusions: Good clinical history and examination along with split bilirubin is sufficient as first line investigation for PJ screen. Second line investigations are indicated only if there are any concerns on initial screen.