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The Significance of the Newborn Screening Test: A Case Report Detecting Congenital Hypothyroidism and Indirect Hyperbilirubinemia

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The first incidence of phenylketonuria in Ankara in 1983 with a pilot study to investigate the screening program began in Turkey, has been expanded upon determination that high. Diseases such as phenylketonuria, congenital hypothyroidism, cystic fibrosis and biotinase deficiency can be detected with the Heel Blood Test, which is a simple and inexpensive method with a few drops of blood in the next 5-10 days after birth. Jaundice is the most common problem in the newborn and should be carefully monitored with the reason for the toxic effects of bilirubin. Hypothyroidism, one of the causes of indirect hyperbilirubinemia, is one of the most important endocrine diseases of childhood. Congenital hypothyroidism Symptoms and signs are very rare during the neonatal period. It is very important to detect congenital hypothyroidism in the early period and to eliminate the cause of the effect of thyroid hormone on the child's motormental development.

Case Report: A 23-year-old mother was born with a caesarean section weighing 4070 grams. She was referred to our Neonatal Polyclinic on the 11th day of the 11th day because of TSH: 61 IU / mL (0,34-5,6). There were no attributes in his CV and Family History. On physical examination, skin was slightly icteric, pulse rate was 120 / min, respiratory rate was 48 / min and other systemic examinations were evaluated naturally. TSH:> 49,500IU / mL, sT4: 0,26ng / mL, Hemoglobin: 18,1g / dL, Leukocyte: 12200 / uL, Trombosit: 381000 / uL, Total bilirubin: 17,89mg / dL, Direct bilirubin : 0,84 mg / dL, AST: 51,2 U / L, ALT: 31 U / L and Urea, BUN, Urine and Electrolyte values were evaluated as normal. Patient was admitted to Yenidoğan Service for follow-up and treatment in terms of congenital hypothyroidism and indirect hyperbilirubinemia. Treatment started with 10μg / kg L-thyroxine and phototherapy. On the 3rd day of the hospitalization, total bilirubin was discharged upon examination of 13,5mg / dL. The thyroid was evaluated agenesis in ultrasound and scintigraphy. The patient is still being monitored and treated with systemic findings and motor-mental development normally.

Results: Hypothyroidism is a very important disease that requires early diagnosis, follow-up and treatment, indirect hyperbilirubinemia in the short term if not diagnosed, and delay in motor development in the long term. If the patient was not diagnosed, indirect hyperbilirubinemia could reach the upper limit, resulting in the development of a posterior kernicterus or long-term retardation in motor development. As seen in our case, Congenital Hypothyroidism may not give symptoms, so the screening program with Heel Blood Test is of vital importance.

Biography

Muhammet is currently working in the Department of child health and Diseases in Duzce University School of Medicine, Turkey. His areas of interest are Pediatric Health Care.

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