GLUCOSE 6 PHOSPHATE DEHYDROGENASE AS A CAUSE OF NEONATAL JAUNDICE

A Preliminary Case Report

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Neonatal jaundice has always been one of the major problems for pediatricians. It is well known that blood group incompatibilities are the most frequent cause of neonatal hyperbilirubinemia. However, it has been proven that other, less frequent conditions, such as septicemia, toxoplasmosis, cytoplasmic inclusion body disease, hepatitis, galactosemia, congenital spherocytosis, infantile picnocytosis, could also result in jaundice. But, in spite of the considerable progress that has been made, some cases of hyperbilirubinemia in the newborn still remain unexplained.

Further investigations on the etiology of neonatal hyperbilirubinemia have shown that certain drugs, such as vitamin K, gantrisin, novobiocin, cause hyperbilirubinemia in the newborn.

Beutler and his colleagues (1) were able to demonstrate that an intrinsic defect of erythrocytes is the cause of hemolytic anemias in some patients taking primaquine and similar drugs. Carson (2) et al., have shown that this intrinsic defect is due to an enzyme deficiency —namely, glucose 6 phosphate dehydrogenase (G-6-PD). Further work carried out on the genetic aspects of this enzymatic defect disclosed that it is transmitted as a sex-linked trait affecting males. (3, 4) American negroes, non-Ashkenazic Jews, Greeks, Italians, and some Chinese people are reported to be carriers of this gene. (3) Not long after this basic work, reports began to appear in pediatric literature on G-6-PD deficiency as a cause of neonatal jaundice. (5, 6) Greek and Israeli authors found a rather high percentage of enzyme deficient infants among the newborns who were searched routinely for G-6-PD activity.

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G-6-PD deficiency may very well be of some importance as a cause of neonatal jaundice in Turkey, for two reasons. First, as mentioned above, our close neighbors Israel, Greece, and the Mediterranean Islands have an appreciably high incidence of this deficiency. Second, there are many cases of favism encountered in the western and southwestern parts of Turkey.

In this paper a case of hemolytic disease of the newborn is reported. It is, to our knowledge, the first case of G-6-PD deficiency causing neonatal jaundice reported in this country.

Case Report

A 40 hours old white male was admitted to Hacettepe Children's Hospital in December, 1962, with symptoms of jaundice which had started at the age of 24 hours. Pregnancy and delivery were uneventful. The patient was the fifth child of the family. His first sibling was stillborn. The second developed severe jaundice requiring an exchange transfusion, but died afterwards. The third pregnancy ended with abortion. The fourth child, who did not develop jaundice, is now two years old and healthy.

Physical examination: The pertinent findings were as follows: skin and scleras were deeply icteric; liver and spleen were not palpable; Moro and sucking reflexes were good. The patient's weight was 2800 grams.

Laboratory findings: Hemoglobin was twelve grams per 100 milliliters. The patient's blood group was A Rh positive (E+, C+, c+) and his mother's blood group was A Rh positive (E+, C—, c+). The direct and indirect Coombs tests, as well as the Witebsky test, were negative. Reticulocytes were seven per cent, and many Heinz bodies were seen in the erythrocytes during the reticulocyte count. A peripheral smear showed marked polychromatophilia; no spherocytes were seen. The urine had no reducing agents or inclusion bodies. VDRL was negative. At 41 hours of age, the total bilirubin was 20 mg. per 100 milliliters, with an indirect component of 19.1 mg.

At the age of 43 hours the patient had an exchange transfusion with 500 cc. of A positive compatible blood. He required two more exchange transfusions at 56 hours and 66 hours of age. Prior to the second and third transfusions, the indirect bilirubin levels were 21.1 mg. per 100 milliliters and 17.7 mg. per 100 milliliters, respectively.

Investigation of the etiology of hyperbilirubinemia disclosed no blood group incompatibility. However, examination of the patient's blood by the spot test of Fairbanks and Beutler (7), showed no G-6-PD activity, and our conclusion was that this G-6-PD deficiency was the cause of the patient's jaundice. A review of his second sibling's chart, who died following an exchange transfusion in this hospital in 1958, showed the diagnosis of hyperbilirubinemia of un-

known origin, with possible enzymatic deficiency. This could not be verified at that time due to lack of technical laboratory facilities.

The patient was discharged home at the age of nine days, in good condition. He has been seen twice in the hemotology clinic and is doing well. The entire family is now being investigated for G-6-PD deficiency.

The problem of G-6-PD deficiency in Turkey has not been investigated so far. It is thought, however, that many undiagnosed cases exist. A screening among the newborn babies, as well as a large scale survey in different parts of Anatolia, are being undertaken by our group. We hope to report the results of our findings in the near future.

Summary

A case of neonatal hyperbilirubinemia is reported. Tests indicated a glucose 6 phosphate dehydrogenase deficiency as the cause of the hemolytic disease. The authors have undertaken an intensive study of the incidence of G-6-PD deficiency in Turkey.

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