Editorial

Newborn screening

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We carry in this issue the Picture Story of a hypothyroid child, diagnosed at the age of 5 months. It is unfortunate that the child was not diagnosed earlier, at a younger age, because the younger the age at which the diagnosis is made and thyroid replacement therapy initiated, the better the prognosis regarding mental development. This child had the classical picture of congenital hypothyroidism i.e. coarse facies, hoarse voice, lethargy, hypotonia, umbilical hernia with constipation. The clinical identification of the newborn with hypothyroidism is often difficult. Retrospective analysis may reveal feeding difficulties, delayed passage of stools, prolonged unexplained hyperbilirubinaemia, large tongue, respiratory distress. One has to resort to the laboratory tests to confirm the diagnosis. The appropriate initial laboratory test is measurement of thyroxine (T4) followed by TSH; a low T4 and an elevated TSH establishes the diagnosis of primary hypothyroidism. If treatment is started before 3 months of age, and life time thyroid replacement is maintained without significant interruption, many have a chance of average intellectual development; of those treated after the age of 3 months or inadequately treated only a small number will attain normal intelligence[1,2].

Congenital hypothyroidirm (CH) represents one of the most common preventable causes of mental retardation[3]. Most infants with CH appear normal at birth as the hypothyroid fetus is protected by placental transfer of maternal thyroid hormones. CH can be detected early only by a screening programme, a T4 and TSH. Screening programmes have been introduced in most developed countries. In North America it is estimated that more than 5 million newborns are screened with approximately 1400 infants with hypothyroidism detected annually.

There are well established criteria for selecting disorders for neonatal screening. Whole population screening for phenylketonuria (PKU) and congenital hypothyroidism (CH) exist throughout the United Kingdom. Babies are screened between the ages of 6 and 14 days by taking a small capillary sample of blood from a heel prick stab. How feasible is it for us to introduce a screening programme for CH? Can this be done with the assistance of the biochemistry department forming a central screening library? It is not practical to do screening on an island wide basis; can this be done on a district basis knowing where CH is more prevalent? We could get the expertise of the nutritional department of the MRI, Family Health Bureau and Epidemiology Department. Studies may be carried out to establish the incidence of the conditions. Screening methods should be suitable, efficient and cost effective. Once the decision is made to introduce a screening programme the practical aspects of the neonatal screening should be studied.

There are neonatal screening programmes in the developed countries for many diseases. The first introduced, the Guthrie test for PKU is unnecessary for us since its incidence is very rare. The neonatal screening will be useful in other inherited diseases with a metabolic basis e.g. disorders of carbohydrate metabolism, galactosaemia and glycogen storage disease.

References

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