LABORATORY SCREENING FOR CONGENITAL HYPOTHYROIDISM IN EARLY INFANCY- A HOSPITAL BASED STUDY

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Neonatal screening measuring serum thyroid stimulating hormone (sTSH) and free thyroxin (FT4) is an essential program for the early detection and prevention of Congenital Hypothyroidism (CH) and its consequences on physical, mental, and sexual development of infants and children. Although national screening for CH is available in many other countries, such a screening programme is not available in Sri Lanka where laboratory testing is done only if there is a suspicion of CH after clinical screening. This situation leads to a delay in diagnosis and treatment with its consequences. This study aims to determine thyroid function and dysfunction in babies born at the Teaching Hospital Peradeniya in Sri Lanka, between January 2002 - June 2003, by estimation of sTSH and FT4 in early infancy.

Seven Hundred and twenty four (724) infants aged between 4-14 weeks with a mean age of 8 weeks were screened for CH using sTSH and FT4 estimation on venous blood samples. There were 365 females and 359 males. Four hundred and ninety four babies had normal sTSH and FT4. Three (3) who had no clinical evidence of hypothyroidism showed biochemical evidence of CH with TSH values more than 60 and low FT4. Of them, one infant had a lingual thyroid and the other showed thyroid agenesis on Tc^{99m} thyroid scan. The other was lost to follow up. Two hundred and twenty six (226) had mild to moderate increase in TSH (5-40mIU/L) with normal T4 levels. Seven of these who had a moderate increase in TSH (20-40mIU/L) had thyroid Tc ^{99m} scans which were found to be normal. One infant who repeatedly showed a normal sTSH and subnormal FT4 had severe constipation, a small umbilical hernia and an absent thyroid on Tc ^{99m}scan. This child and the three children with high TSH and low FT4 were referred to a Pediatrician for thyroxin replacement.

This study detected four babies with CH by laboratory tests although three had no clinical evidence of hypothyroidism. This sample even though it is relatively small shows the importance of laboratory screening for congenital hypothyroidism and the poor sensitivity of clinical screening for CH prompting the need for a national neonatal screening program for CH in Sri Lanka. This study also showed 226 infants with elevated sTSH(5-40 mIU/L) and normal FT4 who subsequently showed normal thyroid function without treatment. It is recommended that babies with mild to moderate increase in TSH (5-40mIU/L) be closely followed up with reassessment of the assays within a few weeks. A study indicates iodine status will be necessary in this group to differentiate iodine deficiency from other forms of transient hypothyroidism.