RETROSPECTIVE ANALYSIS OF CLINICAL SYMPTOMS USED FOR DETECTION OF CONGENITAL HYPOTHYROIDISM

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Although Neonatal screening is an essential program for early identification of congenital hypothyroidism (CH) many countries including Sri Lanka do not have such a program. Presently investigations are done based on clinical screening. This study reports on a retrospective study of 523 babies aged between 4-12 weeks who were subjected to laboratory investigations based on clinical screening between the period of January 1999 and May 2002.

In these children, the serum thyrotrophin hormone (sTSH) and Total thyroxin (TT4) were assayed. Clinical information was gathered from the request form and the laboratory values were obtained from the Nuclear Medicine database. There were 278 male babies and 245 females babies with median age of 8 weeks. Main clinical features prompting laboratory evaluation were "Clinical hypothyroidism", prolonged jaundice, constipation, delayed bone age, umbilical hernia, failure to thrive, and Down's syndrome.

It was seen that those patients who were referred for laboratory screening yielded a higher than expected proportion of elevated sTSH (>20mu/L with associated low T4 levels). All 22(4.2%) patients who had sTSH > 20 mU/L including 5 (0.95%) patients who had sTSH level more than 60mu/l were reported having prolonged jaundice. Furthermore, all babies who had prolonged jaundice had sTSH values over 5.5mu/l. Those who had sTSH above 0.3mU/l and less than 5.5mU/l (reference ranges for adult female 0.38-4.8mU/l and male 0.45-3.6mU/l), i.e. within euthyroid range had no jaundice. Other clinical symptoms used in clinical screening also had a higher yield of high sTSH levels compared to the incidence of hypothyroidism by routine screening as reported in the literature.

It is suggested that clinical screening for CH is useful for further laboratory evaluation of when routine neonatal screening is not available.