## Screening newborns for congenital hypothyroidism

Congenital hypothyroidism (CH) is a common preventable cause of mental retardation with a world wide incidence varying from 1:3000 to 1: 4500 live births [1]. It has been estimated that 50,000 newborns each year are at risk of CH in the southeast Asian region [2]. Although screening of newborn for CH has been a routine practice in developed countries for more than 40 years, developing countries have addressed this concern only in the past decade [3]. A study done in August 2005 at Teaching Hospital, Gampola and base hospitals Mawanella and Nikaweratiya showed a high incidence of CH [4]. This stressed the importance of implementing a national screening programme for CH in Sri Lanka. We conducted a pilot study conducted in the Teaching Hospital, Mahamodara to assess the feasibility of a regional screening programme for CH.

This study commenced on 3rd October 2006 after obtaining approval from the Ethical Review Committee of the Faculty of Medicine, University of Ruhuna. Informed written consent was obtained from mothers at the time of drawing blood just before leaving the hospital. The blood spots were collected from 7 hours to 10 days after the delivery. A questionnaire was used to record the demographic data. Heel-prick blood spots were collected on a special filter paper (Schleicher and Schuell No 903) by pre-intern medical officers working in the Nuclear Medicine Unit and Department of Paediatrics of the Faculty of Medicine. They were trained on the heel-prick technique by the paediatrician in the research team. Air dried blood spots were stored in a sealed plastic box and transferred to the laboratory. Samples with poor quality or inadequate spot were rejected. Thyroid stimulating hormone (TSH) measurement was done by the radioimmunoassay technique using assay kits obtained from Netria limited, UK. Neonates with blood spot TSH concentration of 20 mU/L or above in whole blood after 72 hours of birth were considered at risk for CH [2] and were recalled for serum confirmation. The cut-off value for TSH in the blood spots collected prior to 72 hours of life was set at 40 mU/L [2]. A total of 1650 infants were screened by the end of August 2007. Only few blood spots (n=6) were rejected due to poor quality (sample rejection rate was 0.4%); 1348 neonates (81.7%; 95% CI 81.4%-82.0%) were screened within 72 hours of birth.

Mean blood spot TSH value was 11.47 mU/L (95% CI 10.90-12.01). When the blood spot TSH cut off of 20 mU/L was used, 244 neonates (14.8%; 95% CI 14.1-15.3%) were identified as at risk. 216 of them had TSH <40 mU/L and were not called for serum confirmation. The other 28 neonates (either TSH >20mU/L and sample collected after 72 hours of birth or TSH >40mU/L and sample collected before 72 hours of birth) were called for serum confirmation. Only 1 true positive case of hypothyroidism was identified among them so that the incidence of true CH was 1:1650 in our study.

Even though neonatal screening for CH has become a routine practice in most parts of the world for prevention of profound neurological impairment and growth retardation in infants, Sri Lanka still relies on clinical screening due to non availability of a routine screening programme. Clinical screening causes a delay in diagnosis, treatment and results in a poor outcome of the affected children, causing an enormous burden to the family, society and the country. Estimated incidence of CH in this pilot study is relatively higher than the global incidence. However, it is comparable to that of countries like Iran, Turkey, India and Bangladesh [5-6], and the two hospitals studied in Sri Lanka [4].

The total number of live births reported during the study period (19 months) in the Professorial Obstetrics Unit and Teaching Hospital, Mahamodara was 4,476 and 13,601 respectively. In our study we have assessed only 12.1% of live births in this hospital. If we are to implement a regional screening programme sample collection must be handled by the hospital staff concerned. It is hoped that paediatricians of the health institutions in the South will utilize this service.

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**Manjula Hettiarachchi<sup>1</sup>, Chandrani Liyanage<sup>1</sup>** and **Nayana Liyanarachchi<sup>2</sup>**, <sup>1</sup>*Nuclear Medicine Unit* and <sup>2</sup>*Department* of *Paediatrics, Faculty of Medicine, University of Ruhuna, Galle, Sri Lanka.* 

Correspondence: MH, *e-mail:* <*nmu\_galle@yahoo.com* >. Received 14 May and revised version accepted 5 December 2008. Competing interests: none declared.