

SHORT COMMUNICATION



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Neonatal Thyroid screening for congenital hypothyroidism

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Thyroid hormone plays an important role in the development of central nervous system.¹ Thyroid hormone deficiency present at birth is called Congenital Hypothyroidism(CH).² The incidence of Congenital hypothyroidism is 1 in 4000.³ A pilot study newborn screening program involving 5000 babies in Nepal detected 1 baby with CH.⁴ Girls are more affected than boys by the condition.⁵ Studies have shown that it is extremely mentally stressful and physically exhaustive to raise a mentally retarded special child for a family.⁶ There is lack of sufficient data regarding prevalence of CH in Nepal and large scale studies are required to see the magnitude of the condition.

Newborn screening (NBS) is testing every neonate for potentially fatal or harmful disorders that are not otherwise apparent at birth. Laboratory investigations for this screening program include testing for tetraiodothyronine (T4) and Thyroid stimulating hormone (TSH).⁷ Congenital Hypothyroidism can be permanent or temporary in nature and can be due to thyroid dysgenesis, defects in thyroid hormone synthesis, due to genetic mutation or thyroid hormone resistance.⁸

Neonatal thyroid screening has been undergoing in many developed countries however many third world countries do not have such programs. So the majority of neonates with congenital hypothyroidism worldwide are not detected and treated early. Thus, congenital hypothyroidism remains a significant public health challenge.^{2,9,10}

The problem with congenital hypothyroidism is that most infants are asymptomatic during early stages of the disease.¹¹ A study from Kathmandu has shown that most children with hypothyroidism

were detected at the age 6 to 36 months when parents are most likely to notice delay in developmental milestones.¹² Timely intervention with thyroxine treatment can prevent severe adverse effects on brain development that may occur due to lack of thyroid hormones.¹³

The trend of thyroid disorders being diagnosed is rapidly increasing in our population because of increased awareness in the medical fraternity and general population as well as easy availability of testing facilities. Many patients with subfertility issues are found to have thyroid function abnormalities.¹⁴ Some patients are first found to have thyroid dysfunction during pregnancy itself with more preterm deliveries.¹⁵ Those who have been found to have thyroid dysfunction during such occasions maybe more concerned about whether their offsprings will also be similarly affected. Current practices of thyroid dysfunction testing in neonates may be restricted to neonates with maternal thyroid issues.

Universal Screening of neonates may help for early detection of thyroid disease and timely treatment to prevent unwanted health complications. Whether to combine screening for other disorders is largely dependent upon multiple issues like disease prevalence, costs of tests etc. Large scale multicentre studies will further help to provide evidence regarding prevalence of this relatively easily amenable condition. However, it should not unduly delay the process of implementing such neonatal screening program.

A joint team effort consisting of obstetricians, paediatricians, physicians, endocrinologists etc. can develop a consensus statement in order to implement such screening program. Some retrospective studies have also advocated that

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screening with TSH from umbilical cord blood samples will provide an alternative screening where a delayed TSH screening is not possible due to various reasons.¹⁶ The challenge lies in conducting such tests in rural areas where facilities for testing as well as management may be a major hurdle. The modality of testing, parameters to be tested, timing of testing, a basic treatment and follow up algorithm should be developed so as to increase uniformity and practicality in case detection and management as well as minimise undue stress and over referrals.

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