

SHORT COMMUNICATION

NEW BORN SCREENING IN PREVENTING CONGENITAL  
HYPOTHYROIDISM

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Congenital Hypothyroidism is one of the most common preventable causes of mental retardation which is highly prevalent in our society due to lack of a national neonatal screening program, lack of education of the parents, increased consanguinity, and lack of suspicion from doctor's leads to delayed diagnosis and an increased incidence of congenital hypothyroidism in our society. This Menace can be easily tackled with a mass neonatal screening program and effective legislation which would make sure that serious efforts are being made to eradicate this preventable disease from Pakistan.

**Keywords:** Congenital hypothyroidism, cretin, screening, Pakistan, hypothyroidism, mental retardation

J Ayub Med Coll Abbottabad 2015;27(4):953-4

Congenital Hypothyroidism—one of the most preventable causes of mental retardation which is highly prevalent in our society due to mere fact that Clinicians aren't highly suspicious and this disease is overlooked as a possible cause of early onset retardation in our clinical setup.

Congenital Hypothyroidism is one of the most common preventable diseases that has an incidence of 2% in Pakistan which is 3-4 times more than the west.<sup>1-3</sup> Newborn suffering from a congenital absence of Thyroid hormones have symptoms including feeding difficulties, constipation, enlarged tongue, hypotonia and umbilical Hernias as well.<sup>4</sup> One of the major potential aggravators of this diseases is that this condition is always overlooked and delayed screening results in Mental Retardation.<sup>4</sup>

Screening of Congenital Hypothyroidism is performed via a simple a blood test that is to be performed at the least by the 5<sup>th</sup> day.<sup>5</sup> TSH levels or Free T4 levels are both used for screening for congenital Hypothyroidism which can be confirmed by finding an elevated level of TSH in the serum.<sup>6</sup> Mass neonatal screening was made possible by the development of radioimmunoassay of T4 and TSH levels from blood spots on filter paper.

Recent studies in Pakistan have shown the increased risk of neonates developing congenital Hypothyroidism. A study carried out at the Aga Khan University showed the incidence of congenital Hypothyroidism to be more than 4 times than that of the west.<sup>2</sup> The 20 year Audit of Congenital Hypothyroidism Screening At Aga Khan University showed increased incidence of Congenital Hypothyroidism in infants born between 1987 and 2007 but because of missing data , true incidence wasn't established.<sup>7</sup> However

a detailed analysis of the same screening program in a 10 month period in 2008 which had a compliance rate of 99%, resulted in an incidence of 1 in 1600 cases.<sup>7</sup>

Another study carried out at Pakistan Institute of Medical Sciences in 2013 showed an incidence of 3 cases in the 1337 newborns screened which are astonishingly high numbers despite a small sample size.<sup>8</sup> The reason for this being as around 80% of the deliveries in Pakistan take place in villages and midwifery clinics which have no access to such education or a basic laboratory which can easily screen for the disease.<sup>7</sup> Another important reason is that Pakistan has alarmingly high rates of consanguinity ranging from 46-61% which also contributes to the increased incidence of Congenital hypothyroidism in Pakistan.<sup>7</sup> A study carried out in West Midlands, England showed an incidence of 1 in 781 in Children of Pakistani descent which was attributed to increased Consanguinity in Pakistani population.<sup>10</sup>

Apart from this a low level of suspicion of Physicians even in mothers who have a history of Hypothyroidism has also led to the surge of this preventable illness.<sup>2</sup> Delayed Diagnosis has played a pivotal role in the surge of this preventable disease which was shown by a study in Faisalabad, Punjab which showed that only 14% of cases were diagnosed before the age of 3 months while around 42% of the cases were diagnosed between 1-5 years of age.<sup>9</sup>

The way forward in regard to this problem is education of the Young doctors and midwives and urgent referral of any such case to a tertiary care hospital which can be developed with a high level of suspicion. Legislating the provision of Congenital Hypothyroidism screening made

mandatory for all hospitals/tertiary care providers and specials grants to be provided to hospitals so that they can introduce Mass Neonatal screening for Congenital Hypothyroidism with the help of the government and World Health Organization, which would eventually enable us to eradicate this preventable disease from Pakistan.

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