Abstract: Congenital hypothyroidism (CH) ranks very high among the most common disorders treated in a paediatric endocrinology clinic. In the Western world, CH is detected in 1:3,500 to 1:4,000 newborns (1) by the neonatal thyroid screening (NTS) programme. The main goal of NTS commenced in the 1970s is an early diagnosis of CH and prompt initiation of ongoing thyroxine replacement therapy, to be administered regularly for the affected infants. This measure is aimed to avoid the inevitable outcome of permanent neurological handicap in untreated children or in whom the onset of treatment was considerably delayed. When treated early, CH is also one of the most common preventable causes of mental retardation. In this regard, while emphasizing the role of NTS in early diagnosis, it is worthy of note that the incidence of CH reported from NTS carried out in various centers in India is much higher, viz., 1:1,200 to 1:1,500 newborns. (2). Naturally, it follows that NTS must be universally available in India to identify all affected infants by prompt diagnosis to begin regular replacement therapy in the initial two weeks of life (3).

Keywords: Congenital Hypothyroidism, Paediatric Endocrinology, Thyroid Screening.
Most of the babies born with CH look entirely normal at birth and may not have any obvious symptoms at all, because maternal thyroxine crosses the placenta and may last in the newborn up to 3-4 weeks of age. Some babies are sleepy and are difficult to feed, but even euthyroid babies may have these symptoms. Hence the futility of clinical findings at birth and the importance of screening all newborns at birth, before the baby shows any definite signs of the condition.

Untreated infants may develop hoarse cry, constipation, low muscle tone (floppiness), cold extremities, prolonged physiological jaundice, delayed development, a large tongue (macroGLOSSIA) but these are seen very infrequently. In the past, prior to TNS, when hypothyroid children did not receive treatment until they were several months old, many of them had learning difficulties, stunted growth, and mental retardation. With the advent of NTS, it has been well established that early commencement of thyroxine within two weeks after birth, normal growth and development can be achieved.

NEWBORN THYROID SCREENING

NTS is performed at birth on all newborns using the cord blood or on the fifth postnatal day with a heel-prick blood test for TSH (thyroid-stimulating hormone). Serum TSH from the anterior pituitary will be high when thyroxine levels are low. If the test is done too early after birth, it may show elevated TSH, which may be a false positive result. The results in preterm infants will need to be assessed as per gestational age. When serum TSH is high indicating hypothyroidism, a confirmatory test is done with a venous blood sample for serum free thyroxine and repeat TSH. If the diagnosis of CH is confirmed now with a low serum free thyroxine and high TSH, then a thyroid nuclear scan is advised to arrive at a definitive aetiological diagnosis of CH. This helps the paediatric nurse and the attending physician to counsel the parents regarding the need for lifelong thyroxine replacement for achieving optimal results.

TREATMENT OF CH

When the diagnosis of CH is confirmed, the baby is best managed by a paediatric endocrinologist or a Paediatrician with a special interest in endocrinology. Treatment with levothyroxine (synthetic thyroid hormone, T4) should be started without delay. The dose of thyroxine is 10-15 micrograms/kg body weight/day and will need regulating regularly as the baby grows. The treatment is monitored by periodic blood tests that measure free thyroxine and TSH levels. These tests are carried out every few months during the first year of life and every three to six months during infancy and childhood. The thyroxine level in the blood should be aimed at keeping it in the upper half of the reference range and keeping TSH in the low range.

Levothyroxine is given in tablet form, crushed and mixed with a small quantity of water or breast milk and administered with a spoon or with a medicine dispenser pipette, 30 minutes prior to a feed. It should never be added to the baby’s feeding bottle in bottle-fed infants. Older children can take the tablet with a drink of water. Concurrent administration of calcium or iron supplements should be avoided and must be done after an interval of at least 2 hours. When given in the right dose, there are no side effects from treatment, as thyroxine only replaces the hormone the thyroid is unable to produce. Significant periods of over-treatment or under-treatment should not happen if blood tests are carried out at regular intervals. Thyroxine is inexpensive but highly effective and needs to be given only once a day. The regularity of treatment needs to be ensured. The vast majority of children who have been screened at birth and diagnosed and treated from an early age will grow up normally, going to a normal school and living a normal independent life as an adult. Screening for CH at birth and initiating and continuing regular treatment in proved cases of CH means that a low IQ and other development problems can be avoided. The parents will need to be educated in detail regarding CH and their fears and anxiety allayed.

CASE PRESENTATION

VRS, who had never been shown to a physician, presented for the first time at the age of 8 years 9 months in the paediatric endocrinology clinic for poor growth and constipation. Her parents did not recollect any neonatal problems. All her early developmental milestones in infancy were delayed. She had very clear speech. Her handwriting was slow but she was reported to have average school performance. She was clinically a picture of full-fledged hypothyroidism with the characteristic facies (depressed bridge of the nose, frontal prominence, blepharophimosis, and puffiness of eyes), short limbs, intact primary dentition, cold clammy dry skin, bradycardia and severe short stature (Fig. 5). Her height age was 3 years 8 months and her weight age was 4 years 4 months. A small lingual thyroid was clearly seen on examination of the neck. The parents will need to be educated in detail regarding CH and their fears and anxiety allayed.
DISCUSSION
Once the diagnosis of CH is confirmed, parent education regarding the condition is absolutely essential. Many parents will feel guilty that they produced a child who is not perfect and as one who requires daily medication. They are often misled by others in the community that short term indigenous medications will hasten recovery and obviate the need for lifelong therapy. This happens even after the parents have been concrete evidence of CH. Poor growth is a common manifestation of CH although it may be very severe as in our Index case, still, parents heed to the advice of elders in the family that the child “will grow over the years” and no medical intervention is necessary. This approach results in neurodevelopmental delay which is irreversible. Our patient was fortunate that she did not have major neurologiological problems because it is likely that thyroxine from her ectopic thyroid, though adequate, spared her intellectual functions. It is also fortunate that she attained a normal height and pubertal status as a young woman. This was possible only by constant counselling at every session of her visit.

The benefits of NTS for CH are well known for several decades now but it is unfortunate that this is not routinely practiced or available in many parts of India, the second most populated country in the World. Even in well-equipped hospitals, NTS is offered only as a choice and not as an established norm. It is estimated that in our country, nearly 10,000 babies are born with CH every year, which emphasizes the need for organizing NTS as a priority and on a country-wide basis.

Even in the developed world, where NTS has been practiced regularly from the 1970s, as many as 45% of children diagnosed and treated for CH are lost for follow-up by the age of 3 years. The situation in India is probably better among diagnosed cases, with a long term regular follow-up at least in the private setting, because the parents will need to pay for the initial detection and subsequent follow-up and hence they are keen to continue to monitor their children’s growth and development, especially when they have been initially counselled in detail regarding the importance of thyroxine in the child’s life and metabolism.

NURSING IMPLICATIONS
It will be good to have a team of specially trained paediatric nurses to educate the parents of children with CH and to stress the aetiological details to them by which they will understand the need for continuing the treatment for life. For example, a child proven to have absent thyroid will obviously need replacement thyroxine throughout his or her life. Paediatric nurses will also ensure that children with CH are regularly monitored for their growth and development as well as advice from a paediatric endocrinologist or paediatrician for thyroxine dosage regulation. Rebellious toddlers and adolescents who refuse to take regular medication will also need to be taken care of by friendly persuasive counselling.

REFERENCES
Fig 3. Thyroid Gland in Normal location

Fig 4. Enlarged Thyroid Gland in Dysshormono-Genesis

Fig 5: VRS at Initial Presentation

Fig 6: Lingual Thyroid in Index Case
Fig 7. Growth Chart of VRS