NEWBORN SCREENING – CONGENITAL HYPOTHYROIDISM (NBS-CH)

"THE FIRST TEST"

"Let the first test be the best step"

LET'S JUST DO IT…

Because it's NOT a privilege, but a fundamental right of every baby !

Congenital hypothyroidism (CH) is the most common cause of preventable intellectual disability. If identified and treated with in first two weeks of life, these infants will have normal physical and mental development. Any delay in treatment leaves the child with low IQ permanently. However, as CH is often asymptomatic in early infancy, less than a third get diagnosed before 3 months of life. This, in addition to the availability of a simple diagnostic blood test, and low cost & easily available treatment, makes CH the best candidate for any screening program. Forty five years after the development of first screening program in Canada, universal newborn screening for congenital hypothyroidism as a means to prevent mental retardation, by early detection and treatment is a standard of care in all of the developed countries. Even as many developing countries like China and Mexico incorporated NBS into public sector healthcare programs, India is still in the contemplating stage for the last few decades. Screening for CH is one of the few programs in preventive medicine that has an impact on public health with a positive cost to benefit ratio of 10 to 1. The incidence of CH in India varies between 1 in 1172 to as high as 1 in 727 in some southern states. Years after the ICMR prevalence study (results published 2013) which is supposed to pave the way for universal Government sponsored

"CONGENITAL HYPOTHYROIDISM IS THE MOST COMMON CAUSE OF PREVENTABLE INTELLECTUAL DISABILITY"

"IT'S MY RIGHT"

A Simple Blood Test

A low cost & Easily Available Medicine.

NBS, there are no indications of progress, except in Kerala and Goa, where state governments launched newborn screening programs few years ago.
Indian Society for Pediatric and Adolescent Endocrinology (ISPAE) has recently published clinical practice guidelines, giving clear set of guidelines on screening cut offs, diagnosis, management and follow up of CH. The TSH test costs no more than Rs100-200. It can be done at **any dependable local lab in a venous sample, or sent as a dried blood sample (DBS) on filter paper to one of the screening labs**. It is the responsibility of each individual pediatrician and OBGYN, to take initiative and start screening all the babies born under their care, one baby at a time. To their credit, some of us have already been doing NBS-CH screening for some time now. It is time rest of us incorporate this best practice into our clinical care.

….

One in 1000 babies in India are born with Congenital Hypothyroidism.
That is 27,000 children per year with preventable mental disability!

A call to all fellow OBGYNs and Pediatricians…

Let's take a pledge!

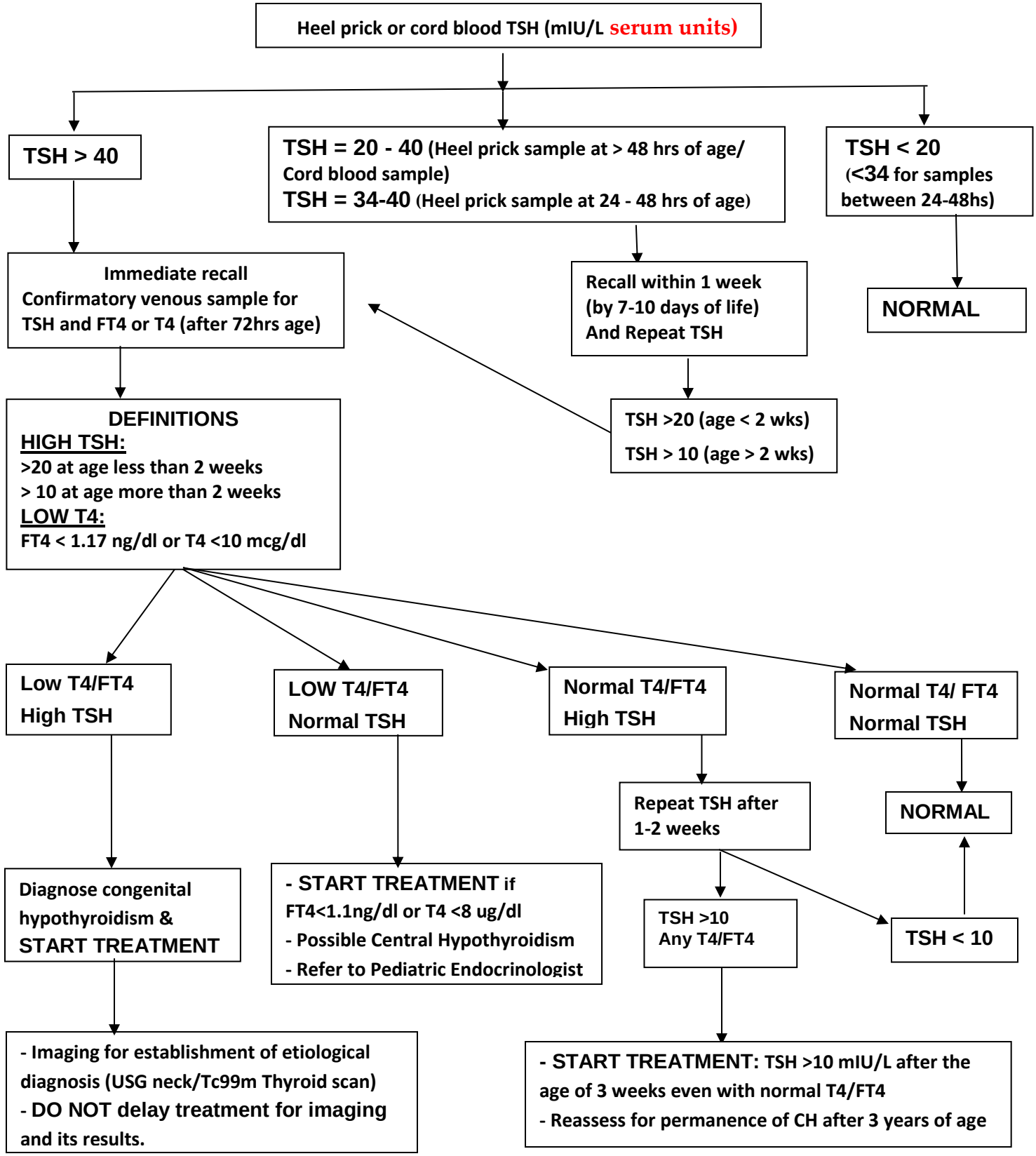
Let's do the NewBorn Screening for CH

Let our first step be the best one by doing

"THE FIRST TEST"

Clinical practice guidelines algorithms ***(based on ISPAE guidelines on CH-2018 with permission)*** **attached for use in your clinics. (Charts A & B)**
This is a public health awareness message. No conflicts of interest declared.

Screening and case detection of Congenital Hypothyroidism in India (Chart A)



Preterm, Sick term babies, IUGR, Twins, and Down syndrome:

If screening test normal - Repeat T4/FT4 and TSH after 2-4 weeks or at the time of discharge whichever is earlier

Treatment and follow up of a baby diagnosed with Congenital Hypothyroidism (Chart B)

Whom to start treatment for:

- **Low FT4 (<1.17 ng/dl) or T4 (<10ug/dl) AND High TSH (>20 at less than 2 weeks, > 10 at more than 2 weeks)**
- **Very Low FT4 (1.1 ng/dl) or t4 (<8 ug/dl) even with normal TSH**
- **Normal FT4/T4 with high TSH (>10 beyond 3 weeks)**

When:

As soon as diagnosis is made (On the same day). Preferably with in first 2 weeks of life.

What treatment:

- LEVOTHYROXINE at 10-15 mcg/kg/day.
- As a single morning dose at the same time every day, on empty stomach (If possible maintain 20-30 minutes gap with next feed).
- Crushed with a spoon and mixed with few ml of breast milk (water after 6 months age).
- Avoid concurrent administration of Iron, Calcium, Soy, Laxatives and antacids.

Follow up:

- First follow up at 2 weeks from start of Thyroxine with FT4/T4 and TSH. Goal is to normalize FT4/T4. TSH takes 2 more weeks to normalize.

Further follow ups:

- Once every 2 months until 6 months age, and once every 3 months until 3 years age.
- After 3 years age (in permanent CH), every 3-6 months till completion of growth and puberty.
- Any major dose adjustment need to be followed by a repeat test in 4-6 weeks.
- Titrate the dose to keep FT4/T4 in upper half of normal range for age (see page.5) and TSH in adult normal range for the lab.
- In case of financial constraints, TSH alone can be used for follow up testing after first year.
- Document developmental milestones and anthropometrical measurements in each visit.

- **Hearing assessment** and **clinical evaluation for congenital anomalies** (e.g. congenital heart disease) in all babies with CH.
- **Genetic counselling**.

Age wise reference ranges for Thyroid function tests

Table 3 Age-wise reference range for thyroid function tests in post neonatal period

Age group	T4	FT4	TSH
1 – 3 mo	82– 235 nmol/L 6.4 – 18.3 μg/dL	13.4 – 44 pmol/L 1.04 – 3.4 ng/dL	0.58–5.57 mIU/L
3 –12 mo	91 – 219 nmol/L 7 – 17 μg/dL	14 – 31 pmol/L 1.1 – 2.4 ng/dL	0.57–5.54 mIU/L
1 – 5 y	91 – 192 nmol/L 7– 15 μg/dL	14 – 26 pmol/L 1.1 – 2 ng/dL	0.56–5.41 mIU/L
5 – 8 y	74 – 166 nmol/L 5.7 – 13 μg/dL	13.4 – 25 pmol/L 1.04 – 1.94 ng/dL	0.55–5.31 mIU/L
8 –12 y	65 – 150 nmol/L 5 – 11.6 μg/dL	12.7 – 24 pmol/L 0.99 – 1.86 ng/dL	0.53 – 5.16 mIU/L
12 –18 y	62 – 136 nmol/L 4.8 – 10.5 μg/dL	12 – 23 pmol/L 0.93 – 1.78 ng/dL	0.51 – 4.93 mIU/L

Lem AJ, de Rijke YB, Toor HV, et al. Serum thyroid hormone levels in healthy children from birth to adulthood and in short children born small for gestational age. J Clin Endocrinol Metab. 2012;97:3170-8

Assay method: Chemiluminescence

TESTING FOR ETIOLOGY

1. **Ultrasonography (USG) and Tc-99m thyroid scan are complementary to each other** in reaching the etiological diagnosis. Scintigraphy can be done up to 7 days after starting treatment, and USG can be done even months later.

2. **Absent, small and dysgenetic gland, hemiagenesis or ectopic thyroid means** hypothyroidism is most likely permanent and **treatment is lifelong**.

3. **If the gland is eutopic (in place) or thyroid scan is normal**, it indicates the possibility of temporary CH. A **reassessment should be done in such cases after the age of 3 y** after stopping treatment for 4 week and repeating the thyroid function test.

4. **If imaging could not be done at the time of diagnosis of CH, then it should be done after age 3 y** (after stopping treatment for 4 week) to establish the etiological diagnosis.