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Ministry of Health

Congenital Hypothyroidism

Procedure for Neonatal Screening and Management

Third Edition

2024

Department of Woman and Child Health

Directorate General of Primary Health Care Ministry of Health

Sultanate of Oman

Contents

List of Figures

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ACRONYMS:

ACTH	Adrenocorticotropic hormone
BBA	Born Before Arrival
BFFP	Birthing Facility Focal Point
BP	Blood pressure
CH	Congenital hypothyroidism
DWCH	Department of Women & Child Health
EPI	Expanded Program of Immunization
FSH	Follicle-Stimulating Hormone
FT4	Free thyroxine
IV	Intravenous
LH	Luteinizing hormone
MD	Medical Doctor
mg	Milligram
ml	Milliliter
mm	Millimeter
MO	Medical Officer
PEAS	Performance Evaluation Assessment Scheme
TBG	Thyroxine-Binding Globulin
TFT	Thyroid function Test(s)
TSH	Thyroid-Stimulating Hormone
TSH-R	TSH Receptor
µg	Microgram

PURPOSE

Congenital hypothyroidism (CH) occurs at an incidence of one case in every 3,000 to 4,000 births in most geographic areas of the world. Late detection in the neonatal period may result in mental retardation and poor physical development of the child. Considering the feasibility of preventing CH through early screening and intervention, many countries have initiated a routine screening program which in found to be cost effective. Following the pilot study conducted in Oman in 2004, the MOH of Oman initiated a National Neonatal Screening Program for CH in 2005. To evaluate the functioning of the systems and services, Performance Evaluation Assessment Scheme (PEAS) was carried out in the year 2008. The objective of the evaluation was to identify deficiencies and rectify them. Although the evaluation indicated an overall efficiency of the system, it also highlighted the need for greater elaboration of the protocols, documentation, data accumulation and summation, in addition to parental counseling, communication and education. Therefore, this document needs to be updated regularly to address the above issues.

The development of this procedure document has taken place in perspective of recent literature review on the latest evidence-based recommendations on CH screening, followed with a review by a panel of national experts including the National Laboratory Medicine Committee, as well as, a feedback from the National Health Service Screening programs, using Newborn Blood Spot for CH Initial Clinical Referral Standards and Guidelines as documented from "Congenital Hypothyroidism: A 2020–2021 Consensus Guidelines Update—An ENDO-European Reference Network Initiative Endorsed by the European Society for Pediatric Endocrinology and the European Society for Endocrinology", particularly with their evidence on screening program based on neonatal blood TSH.

The purpose of the procedure is to provide the most up-to-date and evidence-based clinical framework for the optimal management of congenital hypothyroidism for all health-care providers including general practitioners, family physicians, pediatricians, nurse practitioners, radiologists and laboratories.

SCOPE:

This document covers sample collection procedures; stages of the screening process; follow-up of results and case management; parental counseling; laboratories and related logistics. It also specifies the roles and responsibilities of health care providers at primary, secondary and tertiary health care

levels. Flow charts have been updated in order to address latest changes to the procedures.

DEFINITIONS:

1.

All cases with repeated (venous blood sample) TSH ≥ 40 mIU/L should be diagnosed as hypothyroidism and treatment to be started immediately.

2. **Suspected CH**, If the TSH concentration in the second sample is as follows:

Age	**TSH level**
1-6 days	≥20 - < 40 mIU/L
7-14 days	≥10 - < 20 mIU/L
>14 days	≥5 - < 10 mIU/L

Central hypothyroidism is suspected:

If the cord or neonatal blood TSH concentration is as in the below table, then Central hypothyroidism is suspected and TFT (TSH, FT4) should be requested for confirmation.

Age	**TSH level**
0-6 days	< 2.4 mIU/L
≥ 7 days	< 0.6 mIU/L

DETAILS: PROCEDURE

A. FRAMEWORK

Congenital hypothyroidism (CH) screening of neonates is a health service that should be provided to all newborns in all health care facilities of the Sultanate of Oman, which include Ministry of Health (MOH) institutions, sister governmental institutions and private health institutions throughout Oman. The following should be noted:

1. Therapeutic management and follow up should be provided at the secondary and tertiary health care levels.
2. The components of neonatal CH screening and management service should include:
    I. Screening through measuring cord blood thyroid stimulating hormone (TSH).
    II. Retesting of neonates who have elevated TSH through measuring neonatal blood TFT.
    III. Handling and follow-up of inconclusive or invalid samples.
    IV. Initiating treatment for confirmed CH cases.
    V. Educating and counseling parents of neonates with CH.
3. Cord blood samples should be collected and immediately sent to the laboratory preferably within 5 days.
4. Laboratory focal point and maternity staff nurse in charge are responsible to trace the result of TSH and act immediately when TSH is elevated.
5. Result of TSH should be documented in the white copy of the pink card before dispensing the copies to parent's institutions.
6. TFT (TSH, FT4) should be considered in all late discharge of neonates from Special Care Baby Unit (SCBU).
7. If TSH result is elevated or inconclusive the test should be repeated to confirm the results immediately.
8. All cases with elevated TSH of ≥ 40 mIU/L should be referred to secondary hospitals for treatment, then to tertiary hospital for follow up and further management.
9. Parents of confirmed cases of CH should be counseled on the nature of the disease and its impact on child development by pediatrician.
10. Follow up and management plans should be discussed and agreed upon with parents.
11. Confirmed cases of CH should be immediately notified on the congenital anomaly and genetic disorder notification form (H/P-4) which is available in AL-Shifa 3+.
12. Recall and confirmation of the congenital hypothyroidism cases should be arranged

internally in the hospital.

B. Blood Collection And Processing

1. Umbilical cord blood sampling

I.	Collect 3 ml of cord blood with a 5 ml syringe within 2-3 minutes of birth.
II.	Transfer the blood to a 5 ml plain glass tube with red cap.
III.	Inform parents about the test and that they will be notified if test results are abnormal.
IV.	Ensure that documents accompanying specimen are appropriate, accurate, and complete.
V.	Transport blood to the hospital laboratory (Lab.) at room temperature.
VI.	If the health facility can't process the blood samples, then it should be stored at 4^0c and transport it later to the regional lab.

2. Venous blood sampling

In the case of invalidity of cord blood sample "(e.g. cord necrosis, etc.):

1. A repeated venous blood sample should be drawn from the baby and sent to the lab immediately before discharge.
2. Follow up the case after the TSH result with caution for the difference in TSH cut-off value by age.
3. Follow the same procedures for sample collection and documenting the reason for the invalidity of venous blood sample.

3. Special considerations

3.1 Born Before Arrival (BBA)

Collect 2-3 ml of blood sample by venous-puncture (neonatal blood, not cord blood and not placental blood) while the baby is in hospital before discharge and send it to the hospital laboratory as early as possible.

3.2 Home delivery

Collect blood sample by venous-puncture at the first contact and send it to a laboratory with healthcare facilities for TSH testing as soon as possible. Please note that even if the baby showed up late (later than 6 days), the test should still be done to avoid further damage by

congenital hypothyroidism.

3.3 Pre-term, low birth weight babies, and neonates from multiple births particularly in case of monozygotic twins.

It is recommended that the Thyroid Function Test (TSH, FT4) is performed at birth, at two weeks and at one month in the following cases: -

- All preterm neonates of less than 30 weeks of gestation, and those with a birth weight less than 2000 grams, as there might be a delayed rise in TSH due to immaturity of pituitary –thyroid feedback mechanism.
- Neonates born between 30-37 weeks of gestation as they have almost similar TSH values as compared to 37 weeks. However, in addition to the basal blood for TSH, checking of TFT is required before discharging the baby from the hospital.
- Multiple pregnancies as they have a risk of CH threefold higher than single ones. Furthermore, twins are frequently preterm and/or low birth weight and may have a delayed increase of TSH.

3.4 Very sick Neonates:

For all neonates admitted in SCBU, TFT should be repeated at 2-4 weeks after birth and before discharge, examples are neonates with cardiac disease, neonates underwent cardiac surgeries and those who are on Dopamine administration.

3.5 Neonates with hypothalamic/pituitary disorders (with central hypothyroidism):

Cord TSH is not a good indicator of central hypothyroidism. If central hypothyroidism is suspected clinician should refer the case to pediatric endocrinologist for further evaluation.

3.6 In certain conditions like children with Down Syndrome, TSH should be repeated after one year then annually, along with FT4 and thyroid antibodies. Parents should be informed and a note should be made in the child's health record in the "screening assessment for children" with Down Syndrome section. (refer to Guideline for Medical Management of Children and Young People with Down Syndrome)

3.7 Hypothyroxinemia in low-birth-weight infants should not be treated with levothyroxine sodium (L-T4). Doctor should consult pediatric endocrinologist for further clarification and

management plan.

C. CLINICAL FEATURES OF CONGENITAL HYPOTHYROIDISM:

Clinical features of CH may not be fully apparent at birth and might take time to become recognized. However, the following features are considered the main clinical features of CH:

- Large tongue
- Hoarse cry
- Facial puffiness
- Umbilical hernia
- Hypotonia
- Mottling
- Cold hands and feet
- Lethargy
- Large anterior or posterior fontanels
- Delayed linear growth
- Goiter

Other nonspecific signs:

- Prolonged, un-conjugated hyper-bilirubinemia (jaundice)
- prolonged gestation (> 42 weeks), feeding difficulties
- delayed passage of stool
- hypothermia or unexplained respiratory distress in full term infants

D. THE SCREENING PROTOCOLS

1. The initial screening sample – TSH analysis is performed on a cord blood sample.

2. Samples with TSH ≥ 40 mIU/L are followed by immediate action and notification of the Maternity Nurse-in-Charge, who will notify the concerned clinician for re-calling the baby and re-sending a new neonatal blood sample for TSH and FT4.

3. It is the responsibility of the laboratory to ensure that a second sample for the newborn is sent to the laboratory.

4. Second sample – It is preferable to send the second neonatal venous blood sample at least 2-

7 days after delivery and must be before 7 days.

5. In certain circumstances, where TSH testing and analysis service is not available within the same governorate, then samples are sent to another governorate for processing. In such cases, the Head of Women and Child Health at both governorates should establish a communication channel to convey the result. A similar communication channel must be established for sharing results regarding neonates born outside their governorates.

E. REPEATED TEST:

1. The baby should be considered to have suspected CH If the TSH concentration in this second sample is as follows:

   ≥20 - < 40 mIU/L (day 1-6) or

   ≥10 - < 20 mIU/L (7-14 days) or

   ≥5 - < 10 mIU/L (> 14 days).

   For suspected CH, repeat TSH and FT4. If TSH remains high and FT4 comes low (according to the lab reference range by age], **start treatment immediately** and refer to the pediatrician/pediatric endocrinologist for further evaluation.

2. In repeated tests,
   1. **If TSH ≥ 40 mIU/L** start treatment immediately and an urgent referral to a pediatric /pediatric endocrinologist has to be done.
   2. **If TSH remains high** ((7-14 days) > 20 miu/L,> 14 days > 10 miu/L), and **if FT4 is low**, start treatment immediately and refer to the pediatrician/pediatric endocrinologist for further evaluation.
   3. **If TSH remains high** ((7-14 days) > 20 miu/L,> 14 days > 10 miu/L)), and **if FT4 is normal**, refer the child urgently to pediatricians in your region for further evaluation and decision of the thyroid status.

F. REFERRAL OF NEONATES WITH POSITIVE SCREENING RESULTS

1. The first clinical appointment with a pediatrician/Pediatric Endocrine is mandatory to start treatment as early as possible (on the same day or the next day, and must be within one week).
2. Parents should be informed of their baby's positive screening result.
3. Refer the case to a center with Pediatric Endocrinologist for further management and

evaluation (Annex 4).

G. COMMUNICATION CHANNELS AND FLOWS:

1. As soon as a positive screening test is detected, laboratories should report the results verbally to the Pediatric /Pediatric Endocrinology team, the Head of Women and Child Health, as well as the health professional responsible for communicating the results to the parents. (Figure 1: screening protocol flow diagram).

2. The health professional at parent institution should arrange an appointment for the neonate with the Pediatrician.

3. The outcome of the first appointment should be reported in the neonates electronic record and documented in the child health record (pink card) under the screening part.

4. All confirmed positive cases of Congenital hypothyroidism should be notified on the Congenital Anomalies and Genetic Disorders Notification Form (H/P-4) (Electronic or hard copy if the electronic notification is inactive), and recorded in the quarterly report for annual national registry of CH (Annex 6), and submitted to the Head of Woman and Child Health or Child Health focal point at hospitals.

H. TREATMENT AND FOLLOW UP OF CASES WITH CONGENITAL HYPOTHYROIDISM:

A baby with confirmed CH must be started on treatment with oral levothyroxine (no later than the first two weeks of life or immediately after confirmatory serum TSH results in infants identified in a second routine screening test).

Note 1:

If the result of the repeated test comes within 7 days of life, the treating physician should wait for the result of the TSH, before initiation of oral levothyroxine.

If the result of the repeated test takes more 7 days, then initiation of levothyroxine should be started until the result of the repeated TSH test comes. If the repeated test for TSH shows normal, then treatment should be stopped.

1. **The starting dose of oral levothyroxine should be 10-15 µg/kg/day, with a maximum dose of 50 µg /day**. The target of treatment is to normalize TSH within the first 6 weeks. The dose of levothyroxine may need to be reduced if TSH is suppressed or if the baby is showing signs of overtreatment.

   Babies with significant endogenous thyroid hormone production may need smaller initial doses. *(See note 6)*

Note 2:

Treatment with levothyroxine should lead to normalization of free T4 and a 50% reduction in TSH within days. However, TSH normalization can take weeks and timing does not correlate well with the administered levothyroxine dosage or the severity of the underlying diagnosis. The aim of treatment is therefore to increase free T4 close to the upper half of the age- specific reference range within the first 2 weeks of treatment and to normalize the TSH within the six weeks. However, some cases may take longer, and in those cases detailed history must be taken to view patient's history of medication compliance or it might be due to the type of thyroid like thyroid agenesis. Most of the time TSH will normalize within two weeks, however, few cases will need longer time, especially agenesis. Free T4 concentrations may exceed the normal reference range at the time of TSH normalization but significant elevation should be avoided. Regular dose adjustments may be required.

1. Parents should be shown how to administer and prepare oral solutions of levothyroxine tablets.

Parents should be provided with the hormone in pill form and taught proper administration, as follows:

• The pills can be crushed in a spoon; dissolved with a small amount of breast milk, water, or other liquid immediately before administration; and administered to the child with a syringe or dropper
• The pills should not be mixed in a full bottle of formula
• Toddlers typically chew the tablets without problems or complaints

.

2. All children under treatment should be followed up rigorously as follows:
   - Neonates with congenital hypothyroidism shall be seen by the regional paediatrician on regular basis for routine biochemical markers (TSH, FT4) and clinical examination including developmental assessment.

   - If the child has clinical signs suggestive of congenital hypothyroidism, treatment should be started even if TFT values are normal.

   - If TFT results are not conformity with the expected levels while on treatment suspect noncompliance to treatment.

   - The child should have TSH and FT4 done at all follow-up visits. The child should also be re-evaluated for clinical signs of hypothyroidism.

3. The follow up visits should be as follows:
   a) **The first follow-up visit:** 2 weeks after starting the treatment.
      i. If there is a facility for a thyroid scan, it should be arranged immediately.
      ii. If there is no facility for a thyroid scan, treatment should commence immediately.
   b) **Second follow-up visit**: 4 weeks after the first visit.

c) **Third follow-up visit:** 8 weeks after the second visit.

d) **Fourth follow-up visit:** 3 months after the third visit.

e) **Subsequent visits**: a visit every 3 months until 4 years of age.

f) **Follow-up visits:** at six monthly intervals if the disease is under control.

I. RE-EVALUATION:

- **Re-evaluation of the children with congenital hypothyroidism in whom no etiological diagnostic assessment was carried out during early infancy due to different causes (e.g., preterm, being ill).**
- **Re-evaluation is mandatory when:**
  - Initial evaluation has shown a normally located gland, with or without goiter, in neonates with positive thyroid antibodies,
  - In children who have required no increase in L-T4 dose since infancy,
  - In children in whom no enzyme defect has been identified, either because no molecular genetic investigations have been carried out or because investigations have proved negative for all mutations tested.

- **All children under treatment should be re- evaluated at 3 years as below:**
  - If thyroid scan was not done during the neonatal period, it should be done after completing 3 years of age. Thyroxin should be stopped for one month before the thyroid scan.
  - Perform TSH, FT4 and FT3 (Child should be off treatment for 4 weeks before the test).
  - If thyroid ultrasound and scan, TSH, FT4 and FT3 are normal and child has no clinical signs, discontinue treatment.

- **Desirable additional diagnostic test**

  Appropriate imaging techniques (radioisotope and/or ultrasound scans) may help to establish whether the thyroid gland is:
  - Normally situated and normal in size and shape
  - Normally situated but abnormal in size and shape
  - Ectopic
  - Absent

(See note 5)

J. CLINICAL EVALUATION AND CONFIRMATORY DIAGNOSTIC TESTS

I. The clinician responsible for assessing the baby with a positive screening result shall take a detailed history and perform a comprehensive physical examination. *(See Note 3)*

Note 3:

Babies with CH are more likely to have associated anomalies, particularly congenital heart defects and hearing loss and require careful neonatal examination and follow up. A complete history, including maternal thyroid status (previous history of thyroid dysfunction, maternal anti-thyroid medications), maternal diet (e.g. vegan or other low iodine diet) and family history should be obtained.

II. Diagnostic tests considered essential in the baby are:

A) Free T4 (plasma or serum)

B) TSH (plasma or serum) *(See Note 4)*

Note 4:

Diagnosis using FT4 and TSH should be performed on plasma or serum sample using the appropriate age-related reference range as defined by the laboratory in relation to the equipment used.

Note 5:

A radioisotope scan and an ultrasound examination may establish the cause of the child's CH and indicate whether the condition is likely to be permanent. Initiation of treatment should not be delayed whilst waiting for thyroid uptake scan. An ultrasound scan can be performed at any stage and investigation need not be confined to the neonatal period.

The thyroid uptake scan has to be done for patients with repeated neonatal venous TSH concentration ≥20 mIU/L(day 1-6). For hospitals with Al-Shifa-3 plus electronic system to send an urgent E-referral to the Nuclear Medicine Department for thyroid uptake scan. Annex 4). In addition, the referring team must call and notify the Nuclear Medicine Department.

Note 6:

Thyroid scans are done during working hours only. During long national holidays, the treatment to be started as per this policy and the thyroid uptake scan will be performed after the child has reached the age of three years and has been off thyroxine for a month.

K. RECALL AND FOLLOW UP OF THE RESULTS:

1. All tests should be recorded in the maternity register by the maternity ward staff nurse.

2. The maternity ward staff nurse should also fill the 'Neonatal screening for congenital hypothyroidism and follow up form, and send it on daily basis to the hospital laboratory focal person (Annex 1).

3. The laboratory focal person should ensure that the numbers of samples received are as per the form and that all samples received are suitable for processing. If the sample is invalid, the maternity ward staff nurse must be informed to recollect the blood sample and send it for repeated test, while the neonate is still admitted in the hospital.

4. In the case of discharge, the hospital maternity staff nurse should trace the case and direct the care giver to attend nearest health institution for repeated test.

5. If the case was unable to be traced and recalled at the parent institutions by the end of one weeks, then Head of Woman and Child Health Section at the governorates should be notified.

6. Result of the screening test should be documented in the Child Health Record (pink card).

7. Report the final feedback to the head of woman and child health at governorates through (Annex 1).

8. Notify all confirmed cases of CH through the Congenital Anomalies and Genetic Disorders Notification Form (H/P-4) (manual/ electronic), electronic notification is available in Al-Shifa system.
9. Head of Woman and Child Health at governorates shall send quarterly and annual reports on the numbers and details of the neonates screened to the Department of Women and Child Health, MOH (Annex 6).

10. It is important to follow the instructions given in Annex 5 regarding the role and

responsibilities of focal points at respective locations.

L. COMMUNICATION AND COUNSELLING

1. Communication with parents:

Communication with parents is essential for ensuring proper follow up and management of cases.

- Parents should be informed/explained of the following:
  - About the test performed and the results obtained, if available.
  - Explain the reasons for repeating the test to the parent.
  - If the results aren't ready at the time of discharge, inform the parents that it is important that they track the results and that they update their personal contact details at the hospital's medical record section as soon as possible.
- It is important that parents are contacted if a test needs to be repeated using the standard script that is provided below.
- Please ensure that you document the details of follow up communication, including the date, the name of the individual who communicated the information and the name of the individual who received the communication (Annex 1)

The standard script for a telephonic conversation with parents of neonates who have tested positive"

- Introduce yourself, greet mother and ask about her health and the baby.
- Ask, "Do you remember that your baby's blood was tested for thyroid function test, hormone levels (TFT).
- Wait for response and say, "We would like to retest his/her blood because the result is doubtful".
- Ask, "how soon you can come and bring your baby for the re-test".
- If she is not proposing to come soon, tell her "it is important to come soon because your baby may require treatment after the test result".
- Request her to come with the baby to perform the test and proceed with the needed management.

2. Parent's counseling and support:

- All parents of cases confirmed CH cases should be counseled by a specialist (e.g. Pediatrician) at the initiation or soon after initiation of treatment, as follows:
  - Issue the parent brochure and ask parents to read it before counseling.
  - Provide counseling as per standard procedures (see below),
  - Let the parents ask any other questions if they wish.
  - Check the understanding of parents at the end of counseling.
  - Refer the parents to a family who is successfully managing a child with CH (if available and agreed by both families).

3. Counseling in hypothyroidism: (five A) construct

- Assess
  - Assess details of screening and confirmatory tests and re-confirm that treatment is necessary.
  - Ask about clinical symptoms and signs: constipation, prolonged jaundice, coarseness of skin.
  - Listen to mother's concerns and build confidence with her by reassurance.

- Advice
  - Give the following information (please refer to annex3)
  - Thyroid gland: its position and function in the body (production of thyroxin).
  - Thyroxin hormone: can be given by mouth, it is an effective treatment.
  - The child may require lifelong treatment.
  - The disease will have good outcome if treated without interruption.

- Ask parents for any queries answer and assure accordingly

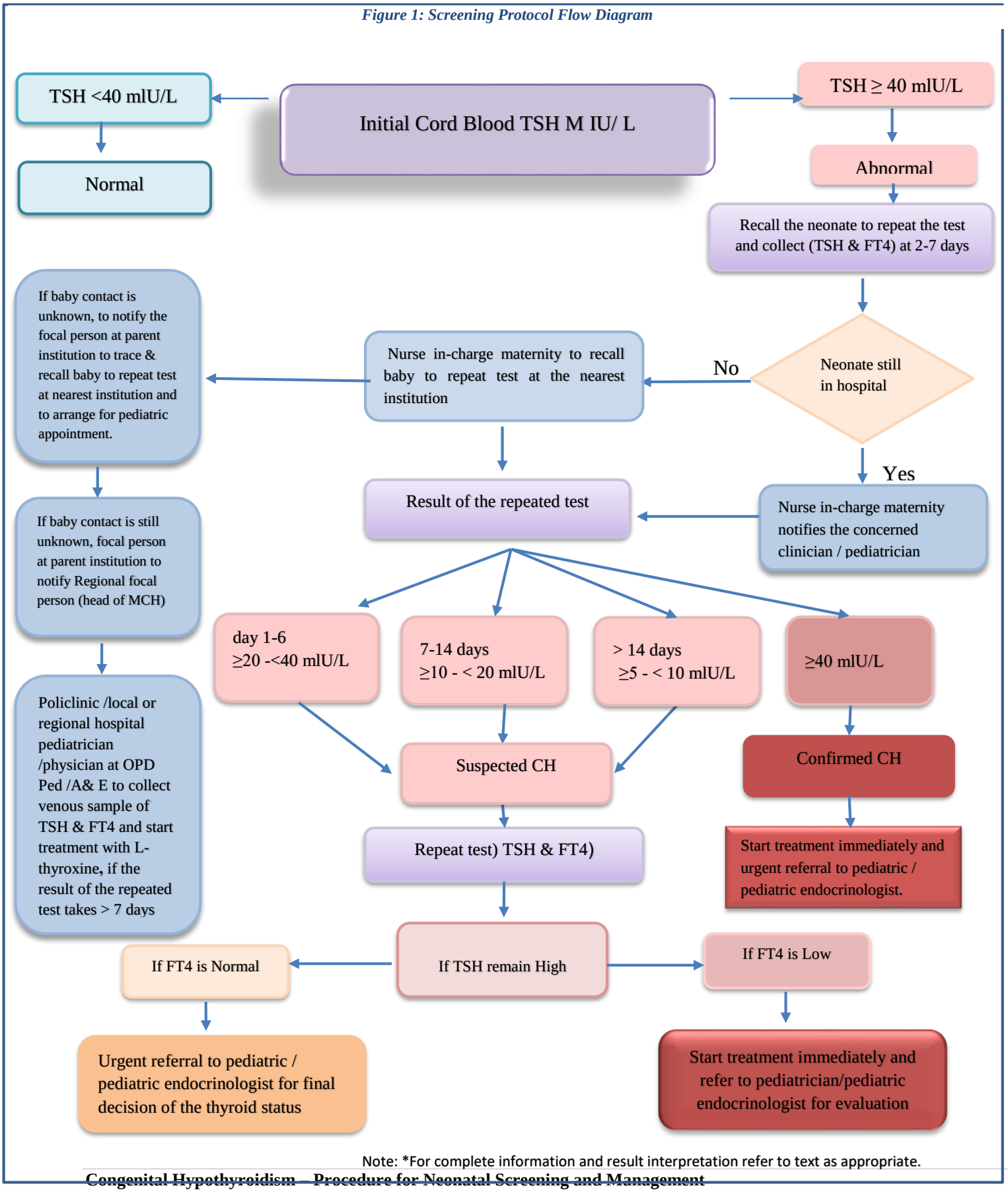
- Agree

- Agree with the parents on next steps:
- To start the treatment with thyroxin.
- To do thyroid ultra sound/scan (if not done).

- Assist
  - Assist in the management plan as follows:
  - Prescription for medication.
  - Give brochure on hypothyroidism and request them to read.

- Arrange
  - Arrange scan appointment if not done.
  - Give appointment for follow up visits.
  - To do thyroid ultrasound/scan (if not done).

Figure 1: Screening Protocol Flow Diagram



Note: *For complete information and result interpretation refer to text as appropriate.

M. LABORATORY PROTOCOLS

1. Reception of specimens and accompanying documentation

a) Specimens received for screening are checked for:

i. Quantity of blood is sufficient (>2 ml)
ii. Blood collected is in the correct container (plain container – clotted blood)
iii. There are no leakages
iv. The container is unbroken
v. The age of the specimen is acceptable (<3 days and has been stored at 4°c).
vi. That the specimen is not in any other way invalid.

b) The documents accompanying the specimen:

i. Are legible
ii. Patient identification is complete
iii. Dates, time of sampling etc. are complete.

c) Notification of problems associated with the specimen or documentation:

i. Maternity ward nurse-in-charge she be notified about any problems associated with the sample/documentation within 24 hours of receipt.
ii. The maternity ward nurse in-charge she be informed of the reason for the invalidity of the specimen so that a repeated sample from the neonate can be initiated as soon as possible.
iii. A written record is kept for each incident of non-compliance with the specimen or documentation protocol.

2. Sample Entry

After verification of sample integrity and compliant documentation, each specimen will be entered

into the laboratory computer and assigned a unique laboratory number according to standard laboratory protocols for the receipt of all laboratory specimens.

3. Analytical Procedure

I. The specimen is centrifuged and the serum separated and stored at 4°c if to be assayed for TSH within 3 days, otherwise keep frozen until ready for analysis.
II. The specimen will be analyzed in the normal way following the procedures prescribed in the Standard Operating Procedures (SOP) for TSH analysis along with other clinical specimens.
III. Any abnormal result will be repeated on the original sample for TSH.
IV. If the TSH is high the maternity ward nurse in-charge of the birthing facility from where the sample was collected and sent, should be notified immediately in order to facilitate a follow-up blood specimen collection and dispatch (see Positive Sample Follow-up Procedure).
V. All required Quality Control (QC) samples would be run in conjunction with the test samples.
VI. Only when all QC samples are in compliance with acceptable criteria will the test results be accepted and released.
VII. All test results and QC results will be maintained on the host computer (test results) and on the instrument file (QC results).
VIII. A hard copy of all test results along with unique identifying number will be retained in the laboratory.
IX. A copy of all cord blood TSH results will be sent to the maternity nurse.

4. Data accumulation and summation:

The delivery health facility carries the responsibility of keeping records of cases and dispatching them appropriately to respective health care facility.
The following section highlights the process and necessary requirements.

Documentation, accumulation, and summation data includes:

I. Number of live born
II. Number of TSH tests performed
III. Number of results tracked
IV. Numbers confirmed
V. Numbers lost with documentation of reasons
VI. Date of diagnosis (confirmation after the second blood sample).

Appropriate follow up data should be reported/sent to:

- Head of Woman and Child Health Section in the respective governorate of focal person in the health institution.
- Department of Woman and Child Health to maintain data base on CH screening (quarterly report):

Final case disposition (affected, not affected, lost to follow-up) from the secondary care should include:

- Date of evaluation to confirm screening results.
- Date of diagnosis/case disposition.
- Date of initiation of Treatment/intervention (if applicable).
- Test results on which diagnosis was based.
- Name of person who communicates the diagnosis information.
- For diagnosed cases (i.e. affected), referral and follow up information to the primary care.
- For cases with uncertain diagnosis, clinical surveillance and action plan to achieve case resolution.
- Identification of the person recording/entering the information.

ROLES AND RESPONSIBILITIES OF FOCAL POINTS IN RESPECTIVE LOCATIONS

Focal point in the labor room:

- ✓ Fill the details of all newborns, born within the last 24 hours in the enclosed form (Annex1).
- ✓ Send the form to the lab of the same health institution.
- ✓ In case if the lab is in another health institution, Please find a channel of communication after agreement with the other parties to report to the lab.
- ✓ Repeat the test if required if the baby is still in the hospital.

Focal point in the Lab.:

- ✓ The form (Annex 1) that relates to the TSH of this cohort of newborns must be filled in within 48 hours and sent to the Head of Woman and Child Health in the respective governorate or focal point in the healthcare institution.
- ✓ Newborns with cord TSH values ≥40 mIU/Lor invalid samples must be reported to the focal point in the labor room and in case the baby is discharged to report it to the Head of Woman and Child Health in the respective governorate.
- ✓ Complete the data regarding TSH values and send it to the Head of Woman and Child Health in the respective governorate.

Head of Woman and Child Health in the respective governorate:

- ✓ Locate the parent institution of newborns with cord TSH value ≥ 40 mIU/L or with the invalid samples that must be repeated.
- ✓ Send the form to the focal point in the respective parent institution to take action.
- ✓ To send the CH quarterly report to the Department of Woman and Child Health (Annex 6).

Focal point at the parent institution:

- ✓ Receive the form from the Head of Woman and Child Health and call the families of the babies who need to have a repeat TSH blood sample because of invalid samples. This sample will be a venous sample and details should be entered in the usual electronic lab request form.
- ✓ Arrange an immediate referral of the baby who has cord TSH ≥40 mIU/Lto the Pediatrician for the initiation of treatment and for repeating TSH sample.

DOCUMENT HISTORY AND VERSION CONTROL

Document History and Version Control			
Version	**Description of Amendment**	**Author**	**Review Date**
01	Initial Release	Team for Updating Congenital Hypothyroidism Guidelines, Child Health Team, Department of Woman & Child Health	June 2010
02	Updating of Guidelines	Child Health Team, Department of Woman & Child Health	May/2021
03	Updating of Guidelines	Child Health Team, Department of Woman & Child Health	May 2023

Written by	**Reviewed by**	**Approved by**
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ANNEXURES

ANNEX 1: NEONATAL SCREENING FOR CONGENITAL HYPOTHYROIDISM AND FOLLOW –UP FORM

LIST OF LIVE BIRTHS IN THE LAST 24 HOURS (DATA)

HOSPITAL……. ….. GOVERNORATE……. FOCAL PERSON…….

NAME……..…….DESIGNATION…….

	I.P.NO	DATE OF BIRTH	MOTHER'S NAME	ANC NO	PARENT INST	CONTACT NO	TSH RESULT		REMARKS
							VALUE	PENDING	IF NEED TO REPEAT THE SAMPLE
1									
2									
3									
4									
5									

In the delivery units	*All the information should be completed by the labor room staff, then send to the lab. on daily 24 hours' basis. (source of information is the maternity register)
In the hospital laboratory	*The last 2 shaded columns are for lab use, who will fill the needed data and then send it to the regional focal point/head of Woman and Child Health at governorate in the next 48 hours.
In the region	the regional focal persons will fax it to all parents institutions focal persons
In the parent Institutions	*the EPI focal persons will note down the value of TSH on the child health record during the first visit of the child in the child health clinic. *EPI focal persons will recall the babies who need a repeat sample & venous sample will be collected in the parent institution/delivery hospital. write down the lab, request form and fill the special neonatal TSH request form label as neonatal TSH–missed & send to lab.
In the hospital laboratory	The laboratory focal person should continue updating the list of newborn tested for TSH-missed, send the new list to the regional focal persons after obtaining the latest results

ANNEX 2: INTERPRETATION OF THYROID FUNCTION TEST IN DIFFERENT THYROID RELATED DISORDERS

Disorder	**TSH**	**FT4**
Primary hypothyroidism	H	L
Primary hyperthyroidism	L	High N to H
Central/hypothalamic/pituitary hypothyroidism	L, N, H*	L
Euthyroid sick syndrome	L, N, H*	L to low-N
TSH adenoma or pituitary resistance	N to H	H
Compensated hypothyroidism	H	N
Thyroxine-binding globulin (TBG) deficiency	N	N (Low Total T4); H TBG

H: High; L: low; N: normal.

*Result can be normal, low, or slightly raised.

ANNEX 3: INFORMATION FOR COUNSELING PARENTS OF A CHILD WITH HYPOTHYROIDISM

What is congenital hypothyroidism?

Congenital hypothyroidism is a disorder that affects infants from birth (congenital), resulting from the severe deficiency of thyroid function (hypothyroidism), usually due to failure of the thyroid gland to develop correctly. Sometimes the thyroid gland is absent or ectopic (in an abnormal location). As a result, the thyroid gland does not produce enough thyroxine/T4 after birth. This may result in abnormal growth and development, as well as slower mental functions.

The thyroid is a gland that is located in the neck and is part of the endocrine system. This gland is responsible for secreting a hormone called thyroxine (T4) which plays a vital role in normal growth and development in children. This gland, is controlled by the pituitary gland. It works very much like a thermostat. The brain senses the amount of T4 and then signals the thyroid with another hormone, thyroid-stimulating hormone (TSH), or thyrotropin to produce more or less T4. When the thyroid gland produces enough T4, no extra stimulation is needed and the TSH level remains at a normal level. When there is not enough T4, the TSH rises. These characteristics of the T4 and TSH hormones allow for the screening of newborns to assess if the infant has normal or abnormal thyroid functions.

Why a child develops congenital hypothyroidism?

In most hypothyroid babies, there is no specific reason why the thyroid gland did not develop normally, although some of these children have an inherited form of this disorder. The parents should not feel the blame, as CH is NOT caused by any life style pattern or behavior of the family.

What are the symptoms of congenital hypothyroidism?

Often these babies appear perfectly normal at birth that is why screening is so vital. However, some may have one or more of the symptoms such as puffy face, swollen tongue, hoarse cry, low muscle tone, cold extremities, persistent constipation, lack of energy, excessive sleep, not growing etc.

What tests are done for confirming congenital hypothyroidism?

The thyroid functions or TFT including TSH and FT4 are confirmatory tests. A thyroid scan may be done to determine the location, or absence of the thyroid gland. Sometimes the scan may be done when the baby is three years old if it cannot be done before starting treatment.

How does one treat congenital hypothyroidism?

Treatment for CH is replacement of the missing thyroid hormone in tablet form. It is extremely important that these tablets are taken daily for life because, thyroxine (T4) is essential for all body functions. In general, the average starting dose for Levothyroxine (synthetic T4) or L-thyroxine in a newborn is between 25 and 50 mcg per day or 10 to 15 μg/kg of body weight. This value increase is dependent upon the individual needs of the child. The tablet can be crushed, and then administered in a small amount of breast milk while the child is still an infant.

Please be aware that L-thyroxine should not be mixed with Soy formula or with iron supplements as these products interfere with absorption. Blood tests will be done on a regular basis to ensure that the hormone levels are in a normal range. Thyroid hormone is necessary for normal brain and intellectual development and such development can be delayed when there is a lack of L-thyroxine.

What type of medical attention should the child receive?

Frequent visits to the doctor will be necessary with blood drawn to check if the laboratory values show normal thyroid levels. Once normal levels are reached, the blood tests will become less frequent. Generally, children are seen every 2 - 3 months, for the first three years, once normal levels have been established.

The goal is to maintain the concentration of Free T4 in the mid to upper half of the normal range (11-24 Pmol/L) for the first years of life. The TSH level should be maintained within the normal reference range for infants. The treatment for hypothyroidism is safe, simple, and effective. Successful treatment, however, depends on life-long daily medication with close follow up of hormone levels.

Making this procedure of taking medication on a routine basis needs to become a part of the lifestyle of the child in order to assure optimal growth and development.

Will other children have the disorder?

There is a small chance that the next child may have the same problem and will need to be screened after birth.

What is the outcome for a baby with hypothyroidism?

There is no cure but the serious effects of the disorder can be lessened and often prevented if medical treatment is started early and continued for life. There are a small proportion of children who have temporary (transient) CH for a period of time after birth. It is impossible to distinguish these transient hypothyroid babies from those with true CH and so these infants will be treated as well. The child will need to be reviewed and retested after 3 years of treatment to decide if the child will need lifelong treatment. In any case treatment should NOT be discontinued before 3 years. With early replacement of adequate thyroid hormone and proper follow up and care, the outcome would be favorable

ANNEX 4: CONTACT DETAILS OF THE TERTIARY CENTERS

Name of the Hospital/Center	Contact number
Royal Hospital Laboratory	24599736 / 24599717
Nuclear medicine, Royal Hospital	24627094 / 24627069 24627088 / 24627066 / 24627072
National Diabetes & Endocrine Center	24211272 / 24211230 / 24211297
Sultan Qaboos University Hospital	24144328/26 24144263

ANNEX 5: REPORT ON NEONATAL SCREENING FOR CONGENITAL HYPOTHYROIDISM (QUARTERLY REPORT)

	Q1	Q2	Q3	Q4	Total
Total Live birth					
Total Screened					
Initial TSH ≥40					
Positive cases on repeated TSH					