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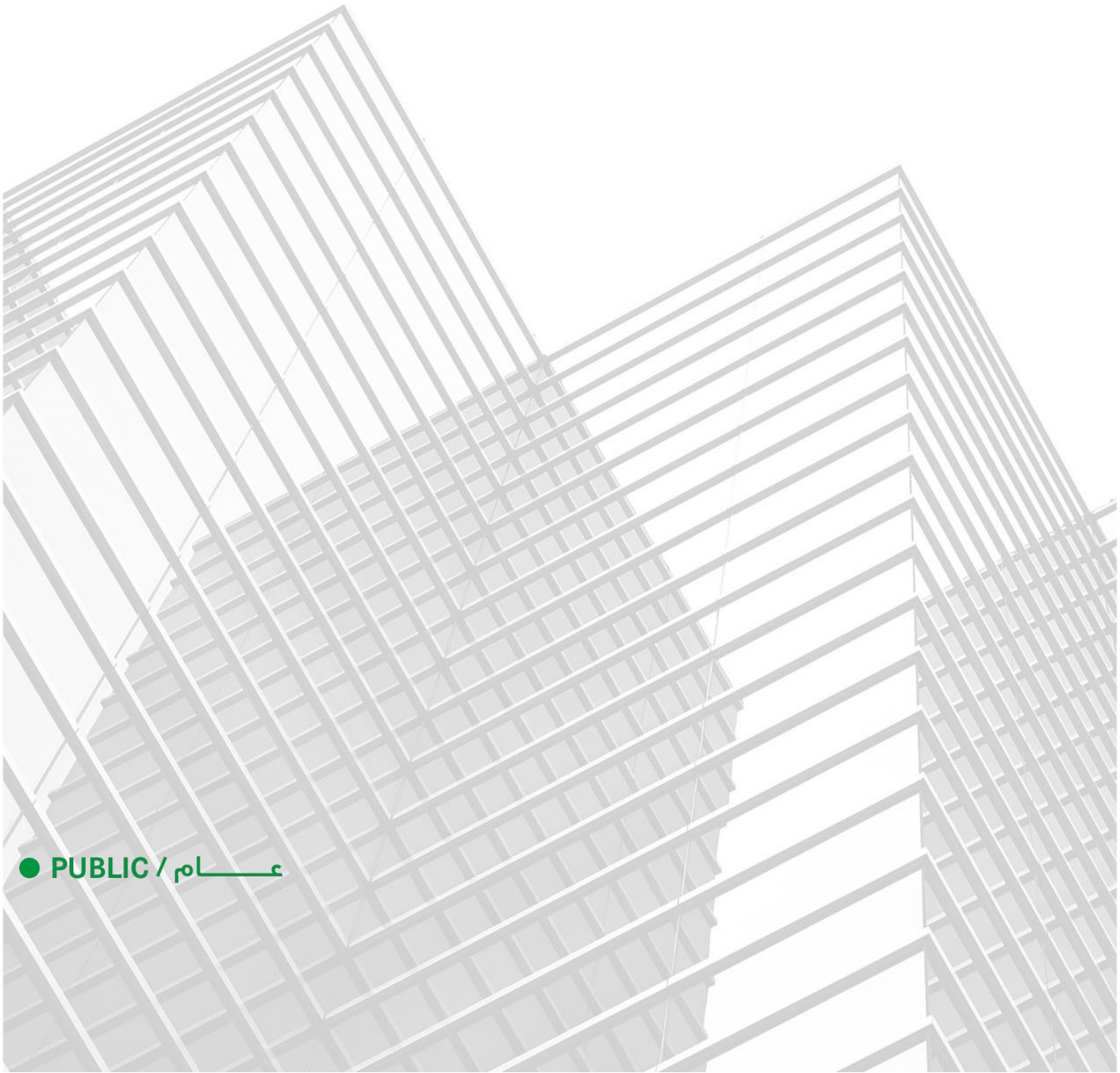
DEPARTMENT OF HEALTH

Standard for Newborn Screening Requirements



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1.Standard Purpose and Scope

1.1 Purpose: The purpose of this standard is to standardize quality of care for all newborns through continuous quality improvement principles, and to provide consistent and clear approaches and pathways to newborn screening.

1.2 This standard is intended to ensure that all infants in the Emirate of Abu Dhabi receive high-quality, evidence-based newborn screening elements at birth. The goal is early detection, management and follow up for those affected, ensuring timely referral and intervention. Without detection and treatment, infants with selected disorders are at risk of possible permanent developmental and physical damage or death.

1.3 Scope: This standard applies to all licensed maternal healthcare facilities and professionals providing newborn screening services, in the Emirate of Abu Dhabi.

1.4 This document aims to define the responsibilities and authorities of all stakeholders involved in the screening process and to highlight the importance of health awareness through proper counselling and health education of parents.

2. Definitions and Abbreviations

No.	Term / Abbreviation	Definition
2.1	AABR	Automated Auditory Brainstem Response: is a direct measurement of the neural response to sound
2.2	ADPHC	Abu Dhabi Public Health Center
2.3	CH	Congenital Hypothyroidism
2.4	CCHD	Critical Congenital Heart Disease
2.5	DoH	Department of Health
2.6	EHS	Emirates Health services
2.7	G6PD	Glucose-6-phosphate dehydrogenase (G6PD) deficiency is an inherited disorder caused by a genetic defect
2.8	HCF	Health Care Facilities
2.9	PRBC	Packed Red Blood Cell Transfusion
2.10	Presumptive Positive screening result	The screening result is positive
2.11	NB Screening	Newborn Screening
2.12	NBS Program	Newborn Screening Program
2.13	LIS	Laboratory information system
2.14	DBS	Dried Blood sample
2.15	KPI	Key Performance Index
2.16	MOHAP	Ministry of Health and Prevention
2.17	MSUD	Maple Syrup Urine Disease

3. Standard Requirements and Specifications

3.1 All DoH licensed eligible maternity and birthing service providers (facilities and professionals) are responsible for ensuring that all infants born in the facility are offered newborn screening that comprises of the below mandatory elements. The newborn screening program in the Emirate of Abu Dhabi includes:

3.1.1 Newborn comprehensive physical examination:

All newborns should have comprehensive physical examination within the first 24 hours after birth and before the discharge of the newborn which must be conducted by a neonatologist or a pediatrician as a part of the routine postnatal care. This is different from the initial examination carried out at the time of birth by one of the health professionals attending the birth. The neonatologist or the pediatrician responsible for conducting the newborn comprehensive physical examination should assess risk factors in newborns. Refer to **appendix (1) Newborn Screening physical examination.**

The core purpose of the examination is to confirm the sex of the baby and detect the ambiguous genitalia, congenital malformations, deformities, and serious anomalies that might impact the health of the child and require immediate attention. It also represents an opportunity to assess and reassure parents about minor anomalies or normal variants.

3.1.2 Hearing screening:

Must be completed by the licensed registered nurse at the healthcare facility. The hearing screening should be performed as close to discharge as possible, preferably 12 hours or more after birth, but the screening can be performed sooner, if needed, for preterm infants. Refer to **appendix (2) newborn hearing screening**. The primary purpose of newborn hearing screening test is to identify those infants at risk for permanent congenital hearing loss.

3.1.3 Critical congenital heart disease screening (CCHD):

Using pulse oximetry, the CCHD screening must be performed by the licensed registered nurse at the health care facility 24 hours after birth. The purpose of the screening is to detect the critical congenital heart defects by measuring oxygen saturation using pulse oximetry. Refer to **appendix (3) newborn critical congenital heart diseases screening.**

3.1.4 Newborn blood spot screening (heel prick test):

The primary goal of newborn blood spot screening is to identify newborns who are likely to have genetic, metabolic, congenital endocrine disorder and enzyme disorders and who require further evaluations, diagnosis and early interventions to prevent disabilities and death. All newborns should be screened within 24-48 hours based on NBS national committee recommendations. Specimen should be sent to the "Diagnostic center for Genetic and Neonatal Screening" EHS – Ministry of Health & Prevention Lab, as per cabinet law number (15) year 2020 in regards Newborn screening low, field 8). Refer to the below appendixes for further details:

- **Appendix (4): Newborn heel prick sampling procedure, handling, packing and shipping**
- **Appendix (5): Preterm < 33 weeks gestational age or Very Low Birth Weight Infants < 1500g +/- PRBC Transfusion pathway**
- **Appendix (6): Full term and Preterm Infant > 33 weeks or > 1500 g +/- PRBC Transfusion.**
- **Appendix (7): Newborn blood test screens the listed disorders in appendix (7).**

3.1.5 Appropriate referral and follow-up care to be provided for newborns within the timeframes specified:

Timely referrals and follow-up care for newborns are crucial to ensure early intervention and healthy development of the infants. Refer to **appendix (8) Critical and non-critical disorders.** It's important to start the treatment as soon as possible as it may include special formula, diet restrictions, supplements, medications, and close monitoring. Refer to:

- **appendix (9) Reporting of Newborn blood spot screening results**
- **appendix (10) Pathway of Newborn Blood Spot Screening**

3.1.6 Parents' health education:

All prospective parents must receive general information about the newborn screening during the prenatal period antenatal care and postnatally (within 24 hours of the birth or at the time of screening) through one to one or group education and as a health-related materials which includes:

3.1.6.1 The importance of NB screening, as it is crucial in detecting infants who, despite appearing healthy at birth, may have a serious and possibly fatal disorder that requires prompt treatment.

3.1.6.2 Parents should be given information about what to expect after the screening.

3.1.6.3 Discussing with parents that, while unlikely, there is a chance that their newborn will need another newborn screening.

3.1.6.4 Encourage the parents to have the repeat screen collected in a timely manner, if requested.

3.1.6.5 If parents refuse any of the newborn screening tests, this must be documented using the consent form in appendix (11) Newborn screening – Parent refusal form or attested refusal form used by HCF. This form must be signed by the parent(s) and signed copies must be kept in the newborn's hospital medical records and a copy provided to the parents.

3.1.7 Data reporting to DoH e-Notification system:

Screening results must be reported to DoH through the DoH e-notification link as follows: https://bpmweb.doh.gov.ae/UserManagement/MainPage.html. Maternal health care facilities and neonatologists should list in the discharge summary that all the newborn screening tests are done to ensure that the NBS has been collected.

3.1.8 Payment for Newborn Screening: Newborn screening performed during the inpatient delivery. It's considered part of the delivery service and shall be covered by the insurance plan under the maternity benefit. In the case that the infant was discharged from the hospital before the heel prick test, hospitals must arrange for it at no additional charge and to ensure the infant is tested before 72hrs from birth. Additionally, hospitals must report the testing using the established rules for venipuncture.

3.1.8.1 Subsequent encounters for newborn baby screenings where no abnormal findings were detected shall be considered as a preventive service.

3.1.8.2 For Thiqa holders: Newborn baby screenings are covered under the Thiqa scheme for preventative care.

3.1.8.3 For non-Thiqa holders (Basic and Enhanced policy holders), coverage should be consistent with their insurance policy and policy schedule of benefits as approved by DoH.

3.1.8.4 Subsequent encounter because of the Newborn Screening, and where abnormal findings were detected, such encounters shall be billed and reimbursed under the insurance plan but as a medical condition and not a preventive service.

4 Key stakeholder roles and responsibilities

4.1 Healthcare facilities participating in DoH's newborn screening program should:

4.1.1 Provide the mandated set of screening tests in accordance with DoH policies and standards, laws and regulations of the Emirate of Abu Dhabi.

4.1.2 Adhere with policies and standards on patient education and consent.

4.1.3 Conduct the screening and collect specimens for laboratory screening tests from newborns, where agreed by parents and supply laboratories with samples. mandated timeframes.

4.1.4 Ensure appropriate patient discharge, referral, and follow-up, where required, consistent with DoH standards.

4.1.5 Apply policies and standards on managing patient medical records including:

- 4.1.5.1 Developing effective recording systems
- 4.1.5.2 Maintaining patient records
- 4.1.5.3 Maintaining confidentiality
- 4.1.5.4 Privacy and security of patient information
- 4.1.5.5 Educating patients on services provided
- 4.1.5.6 Satisfying the requirements of patient informed consent and patient rights and responsibilities charter

4.1.6 Comply with DoH data standards and procedures

4.1.7 Comply with DoH requests to inspect and audit records and cooperate

4.1.8 Comply with DoH authorized auditors as required by DoH

4.1.9 Comply with requirements for information technology (IT) and data management including sharing of screening/diagnosis, pathology results, and electronic patient records management systems.

4.1.10 **Reporting and referral requirements:**

4.1.10.1 Healthcare facilities should nominate a group of employees on behalf of the facility to be responsible for reviewing the medical records relevant to the screening and submitting the screening results to DoH E- Notification system electronically.

4.1.10.2 Healthcare providers should inform parents of abnormal screening test results and the need for secondary or confirmatory testing and follow up, including referral to a specialist healthcare provider.

4.1.10.3 Healthcare providers are responsible for calling back parents and their newborn to collect a new specimen and re-test, when required within the mandated timeframes.

4.1.10.4 Parents do not need to be notified of normal screening results, but all results (whether normal or abnormal) should be within the infant's electronic medical record or equivalent.

4.1.10.5 Newborn blood spot sampling (heal prick) reporting and referral: If the first newborn screening sample was "unsatisfactory". This means that the sample could not be tested properly. When infant 's sample cannot be properly tested, a repeat sample is needed as soon as possible to perform newborn screening.

4.1.10.6 If the infant is transferred to another facility for continuing care, the newborn screening status should be endorsed (i.e., where applicable the due date/timing of next blood screen should be endorsed).

4.1.11 **Follow up and management in regards positive cases:**

4.1.11.1 A positive screen does not mean that the infant is affected with a disease; diagnostic testing must be done to confirm or rule out a disease. A negative screen does not rule out a disease; any infant symptomatic of a disease should have the appropriate diagnostic evaluation.

4.1.11.2 Parents should be notified that when the test is done that any abnormalities will be communicated to them. Unusual results may mean that infants may need to under a repeat screen and/or blood test. Parents will be notified of additional referrals that

may become indicated.

4.1.11.3 The positive newborn screening result should be referred to the pediatric medical specialist. The newborn screening coordinator is responsible for assisting the infant's family and facilitating the referral procedure. The positive newborn screening cases are assessed by the designated consultant to confirm the diagnosis and initiate the management as soon as the diagnosis is confirmed.

4.1.11.4 For blood spot screening: Abnormal screening results are designated by the screening laboratory staff as "positive screen" or "presumptive result".

4.1.11.5 Borderline result: Indicates that the screening was slightly abnormal and that the infant needs a repeat newborn screening sample. If the repeat is still borderline, it will be reported as "persistent borderline" and is treated in the same manner as a positive screen. Refer to appendix (8) Reporting of newborn blood spot screening results.

4.1.11.6 Documentation that a sample was collected should be registered in the system Report can be saved electronically in the laboratory information system.

4.1.12 **Procedure in case of parental refusal: If parents refuse to have the test, the following steps should be followed:**

4.1.12.1 Confirm the parents/guardian have received appropriate education regarding the importance of the newborn screenings.

4.1.12.2 Members of the medical treatment team must meet with the parents and clearly outline the benefits & risks of the NBS and the risks of not completing the test. This is to ensure that the parents have a good understanding of the possible consequences.

4.1.12.3 Medical staff should clearly document in the progress notes the parents' refusal of the test and the nature of the discussion. The refusal for informed consent form should be completed and countersigned by the parents. Appendix (11): newborn screening refusal form

4.1.12.4 The signed informed consent form needs to be kept in the newborn's medical record and a copy provided to the parents. Document the refusal for the test in child's health record book.

4.1.12.5 Parents/guardians can change their mind at any stage, but it should be explained to them that some disorders will not be detected if newborn screening is not performed at the correct time.

4.2 Healthcare Facility's Administration (Technical Director) should:

4.2.1 Implement the newborn screening standard at the health care facility

4.2.2 Designate a coordinator and an alternate staff to be responsible for the newborn screening program in the health care facility.

4.2.3 Provide support in terms of human resources, material and equipment that facilitate implementation of newborn screening program at the health care facility's level. Ensure that all babies born in the facility undergoes all newborn screening elements.

4.3 Healthcare Professionals/ Physicians should:

4.3.1 Counsel, educate parents/guardians and ensure that they receive accurate information about NBS during antenatally screening and soon after delivery.

4.3.2 Perform Newborn screening elements for all infants born in their facility within the first (24-48 hours) of age which includes comprehensive physical examination, hearing screening test, Congenial heart disease, and newborn blood spot sampling (Heel Prick).

4.3.3 Counsel parents of infants with positive results and ensure prompt referral to subspecialty.

4.3.4 In case of refusal, provide counselling and ensure that parents/guardian understand the consequences of not performing the test.

4.3.5 Document the refusal, ensure that parent/guardian sign the newborn refusal form appendix (4) and place a copy of the form in the newborn's medical file (electronic or hard copy).

4.3.6 Counsel parents of infants with presumptive positive screening results and ensure prompt referral to the Referral treating center.

4.3.7 Document the positive results and referral in the Newborn's medical file.

4.4 Coordinator of the newborn screening program should:

4.4.1 Facilitate and prepare education/training session for all staff involved in the newborn
4.4.2 screening process in the health care facility.
4.4.3 Provide orientation for all staff about the NBS program.
4.4.4 Report and submit data for newborn screening and the results through the DoH e-notification link as follow: https://bpmweb.doh.gov.ae/UserManagement/MainPage.html
4.4.5 Communicate with parents/guardians of infants with positive results and coordinate for referrals and booking appointments.
4.4.6 If the newborn is not screened before discharge: The newborn screening coordinator should inform the parents about the necessity of the sample collection as soon as possible (not more than 48 hours).

4.5 Nurses/Midwives/Lab technicians should:

4.5.1 Provide appropriate patient education and information regarding the screening test to parents/guardians.

4.5.2 Assure completion of the required/ mandatory information on the blood spot collection device.

4.5.3 Follow the procedure of obtaining blood spot sampling provided in the guideline.

Recall of infants with unsatisfactory or repeat sample. Refer to appendix (12) Specimens acceptance & rejection criteria.

5. Parents/Guardians should:

5.1 Read the newborn screening education material and understand its importance.
5.2 Participate in newborn screening program.
5.3 Cooperate with medical health provider and attend in proper time for screening test if discharge before 24 hours.
5.4 Go to the healthcare facility immediately in case a repeat sample is required.
5.5 Update the contact numbers in the medical records and provide correct information.
5.6 In case of refusal to participate in NBS program, sign a refusal form.
5.7 In case of positive results, it is the parent's responsibilities to attend the appointments given by program coordinator.

6.Monitoring and Evaluation

Healthcare providers are required to submit and report their performance. They shall be evaluated in accordance with the statistical data provided, as well as the accuracy of reporting, KPIs which includes:

KPI	Definition	Targeted Level
Uptake rate of newborn screening annually	Number of newborns participated in the Newborn screening program from the total number of newborns delivered in the healthcare facility	92 %
Total number of positive screening test findings in the following: CCHD, hearing, Physical examination & heal prick test.	Number of newborns with abnormal screening results in any of the screening test components.	NA

Auditing will be performed by DoH or third party.

6. Enforcement and Sanctions

6.1 Healthcare providers and professionals performing the Newborn Screenings must comply with the terms and requirements of this standard and other related regulatory requirements.
6.2 DoH may impose sanctions in relation to any breach of requirements under this standard in accordance with disciplinary regulation of the healthcare sector.

7. Relevant Reference Documents

No.	Reference Name	Relation Explanation / Coding / Publication Links
1.	DoH Health care Providers Manual	https://www.doh.gov.ae/en/resources/policies
2.	DoH Healthcare Professionals Manual:	https://www.doh.gov.ae/en/resources/policies
3.	DoH Healthcare Regulators Manual	https://www.doh.gov.ae/en/resources/policies
4.	DoH Healthcare Insurers Manual	https://www.doh.gov.ae/en/resources/policies
5.	DoH Standards	https://www.doh.gov.ae/en/resources/standards
6.	Laws & Legislations	https://www.doh.gov.ae/en/about/law-and-legislations
7.	DoH Circulars	https://www.doh.gov.ae/en/resources/Circulars
8.	Newborn Screening Ontario.	https://www.newbornscreening.on.ca
9.	American College of Medical Genetics and Genomics & Advanced Solutions International	https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx

Appendix No.	Appendix Name
Appendix 1	Newborn Screening physical examination
Appendix 2	Newborn hearing screening Charter 1: Hearing Screening for Healthy Newborns Charter 2**:** Hearing Screening for High-Risk Newborns
Appendix 3	Newborn critical congenital heart diseases screening Charter 3: Clinical pathway for newborn Critical Congenital Heart Diseases Screening
Appendix 4	Newborn heel prick sampling procedure, handling, packing and shipping
Appendix 5	Preterm < 33 weeks gestational age or Very Low Birth Weight Infants < 1500g +/- PRBC Transfusion pathway
Appendix 6	Full term and preterm Infant > 33 weeks or > 1500 g +/- PRBC Transfusion
Appendix 7	Newborn blood test screens the listed disorders.
Appendix 8	Critical and non-critical disorders
Appendix 9	Reporting of Newborn blood spot screening results
Appendix 10	Pathway of Newborn Blood Spot Screening
Appendix 11	Newborn screening – Parent refusal form
Appendix 12	Blood specimen’s acceptance and rejection criteria

Appendix (1): Newborn screening physical examination

A complete physical exam is an important part of newborn care. Each body system is carefully checked for signs of health and normal function.

The health care provider also looks for any signs of illness or birth defects. Physical exam of a newborn often includes assessment of the following:

Physical examination	Comments
Vital signs	1. Temperature. Able to maintain stable body temperature of 97.0°F to 98.6°F (36.1°C to 37°C) in normal room environment. 2. Heartbeat. Normally 120 to 160 beats per minute. It may be much slower when an infant sleeps. 3. Breathing pattern and respiratory rate of Normally 40 to 60 breaths per minute. 4. Blood pressure. Normally an upper number (systolic) is between 60 and 80, and a lower number (diastolic) between 30 and 45. 5. Oxygen saturation. Normally 95% to 100% on room air.
General appearance	1. Physical activity, muscle tone, posture, and level of consciousness or whether an infant is awake and alert. 2. Skin (color, texture, nails, presence of rashes, hemangiomas, or birthmarks).
Head and neck	1. Appearance, shape, and shaping of the head from passage through the birth canal (molding)The open soft spots between the bones of the baby's skull (fontanels). 2. Bones across the upper chest (clavicles)
Extensive anthropomorphic measurements	1. Occipital frontal circumference. 2. Weight. 3. Height
Face. Eyes, ears, nose, cheeks	1. Pupil response to light. 2. Corneal Opacities. 3. Red Reflex. 4. Ears (low set ears, pre-auricular pits, accessory auricles).
Mouth	Roof of the mouth (palate), tongue, throat, central cyanosis, cleft lip
Respiratory System	Breath sounds, Breathing pattern.
Cardiovascular System	1. Heart sounds, Rate, Rhythm. 2. Murmurs. 3. Femoral pulse and peripheral pulses.
Abdomen	1. Appearance 2. Palpable organs 3. Umbilicus – granulomas, bleeding 4. Hernia orifices
Genitals and anus.	1. Ambiguous genitalia 2. Hypospadias 3. imperforate anus 4. Cryptorchidism
Spine	Shape to detect any a symmetry and palpate bony deformities; dimples, spinal clefts, unusual tufts of hair, hemangiomas. Mongolian spot
Extremities	1. Proportion and symmetry. 2. Specific measurement of any joint limitations. 3. Foot deformities.
Hips	1. Asymmetry of the limbs and skin folds. 2. Barlow and Ortolani's maneuvers.
Central Nervous	1. Tone, Behavior, Movements and Posture. 2. Tendon reflexes, grasp, sucking and rooting reflex

Appendix (2): Newborn hearing screening for Healthy newborns

1. Introduction

The primary goal of newborn hearing screening is to identify newborns who are likely to have hearing loss and who require further evaluation. Finding newborns with health issues that may result in late onset hearing loss and creating a strategy for ongoing assessment of their hearing status are secondary goals.

2. General Considerations

- A follow-up system for infants who do not pass inpatient hearing screening, who are at risk for late-onset and progressive hearing loss, or who are missed by the inpatient screening program—to include repeat screening or referral for appropriate evaluation and early intervention.
- Maintain a quality assurance process to evaluate the effectiveness of newborn hearing screening.

3. Hearing Screening procedure

Healthy Newborns (With no risk factors)

- **Timing:** should be performed within 48 hours after birth, and preferably after 12 hours of birth.
- **Method used**: one stage screening protocol with Automated Auditory Brainstem Response (AABR).
- **Procedure setting:**
    - Ensure that the equipment is working properly and that a quiet screening environment is available.
    - The infant should be asleep or resting quietly for the test and positioned to reduce muscle artifact.
    - The screener visually inspects the outer part of the ear canal to ensure that the canal is clear of debris and then places the transducer.
    - Both ears are screened during each session.
    - Each screening test should not exceed two attempts of trying per ear.
- **Special consideration on time Test:**
    - Consider performing after 34 weeks gestational age.
    - Babies should be in a stable condition without any medical equipment attached (i.e. with no life support, no nasal cannula and in an open cot).
    - If the corrected age at the time of discharge for the preterm baby is more than 3 months, the baby should be referred directly to the audiologist for diagnostic hearing assessment.
    - All babies admitted in the NICU for five or more days will need to have an additional evaluation by 6 months corrected age even if they pass the initial hearing test.
    - Babies admitted for less than 5 days will have hearing screening performed at point of discharge from the neonatal unit or from the postnatal ward.
    - For babies who develop a co-morbidity (such as meningitis, etc.) prior to discharge, the hearing screening test should be repeated, and an urgent referral given to specialized otolaryngology clinic with audiology unit.
    - Test duration takes about 20 minutes, the time maybe longer if a baby is restless or the environment is noisy

- **Pass or Refer Criteria for AABR: (refer to chart 1)**
    - **Pass**: Reliable evoked responses present at 35 dBnHL on **both** ears.

      Parents advised to monitor the speech and language developmental milestones and attend the well child visits.

    - **Refer**: No recordable responses at 35 dBnHL in **one** or both ears.

      Rescreen with AABR at **3 weeks of age** at the same place of the first screening (Maternity ward):
        - **Pass:** Advise Parents to monitor the speech and language developmental milestones and attend the well child visits.
        - **Refer:** Refer for diagnostic audiological evaluation prior at **3 months of age**

4. High Risk Newborns

- High risk Newborns include the followings:

Prenatal risk factors	- Family history of hereditary childhood sensorineural hearing loss. - In-utero infection, such as cytomegalovirus, rubella, syphilis, herpes, and toxoplasmosis. - Craniofacial anomalies include those with morphological abnormalities of the pinna and ear canal. - Birth weight is less than 1500 g. - Hyperbilirubinemia at a serum level requiring exchange transfusion. - Ototoxic medications, including but not limited to the aminoglycoside, are used in multiple courses or in combination with loop diuretics. - Bacterial meningitis. - APGAR score of 0-4 at 1 minute or 0-6 at 5 minutes. - Mechanical ventilation. - Stigmata or other findings associated with a syndrome known to include sensorineural and/or conductive hearing loss. - Any Neonatal Intensive Care Unit (NICU) stays for longer than 5 days.
Perinatal or Postnatal risk factors	- Culture positive infection associated with sensorineural hearing loss including confirmed bacterial and viral meningitis or encephalitis. - Events associated with hearing loss: ▪ Significant head trauma especially basal skull, temporal bone fractures. ▪ Chemotherapy.

- **Pass or Refer Criteria for AABR: refer to chart 2**
    - **Pass**: Reliable evoked responses present at 35 dBnHL on **both** ears.

      -Parents advised to monitor the speech and language developmental Milestones.

      -Referral for Diagnostic Audiology Evaluation **within 6 months of age.**

    - **Refer**: No recordable responses at 35 dBnHL in **one** or both ears.

      -Refer to an audiologist for diagnostic audiology evaluation **within 1 month of age.**

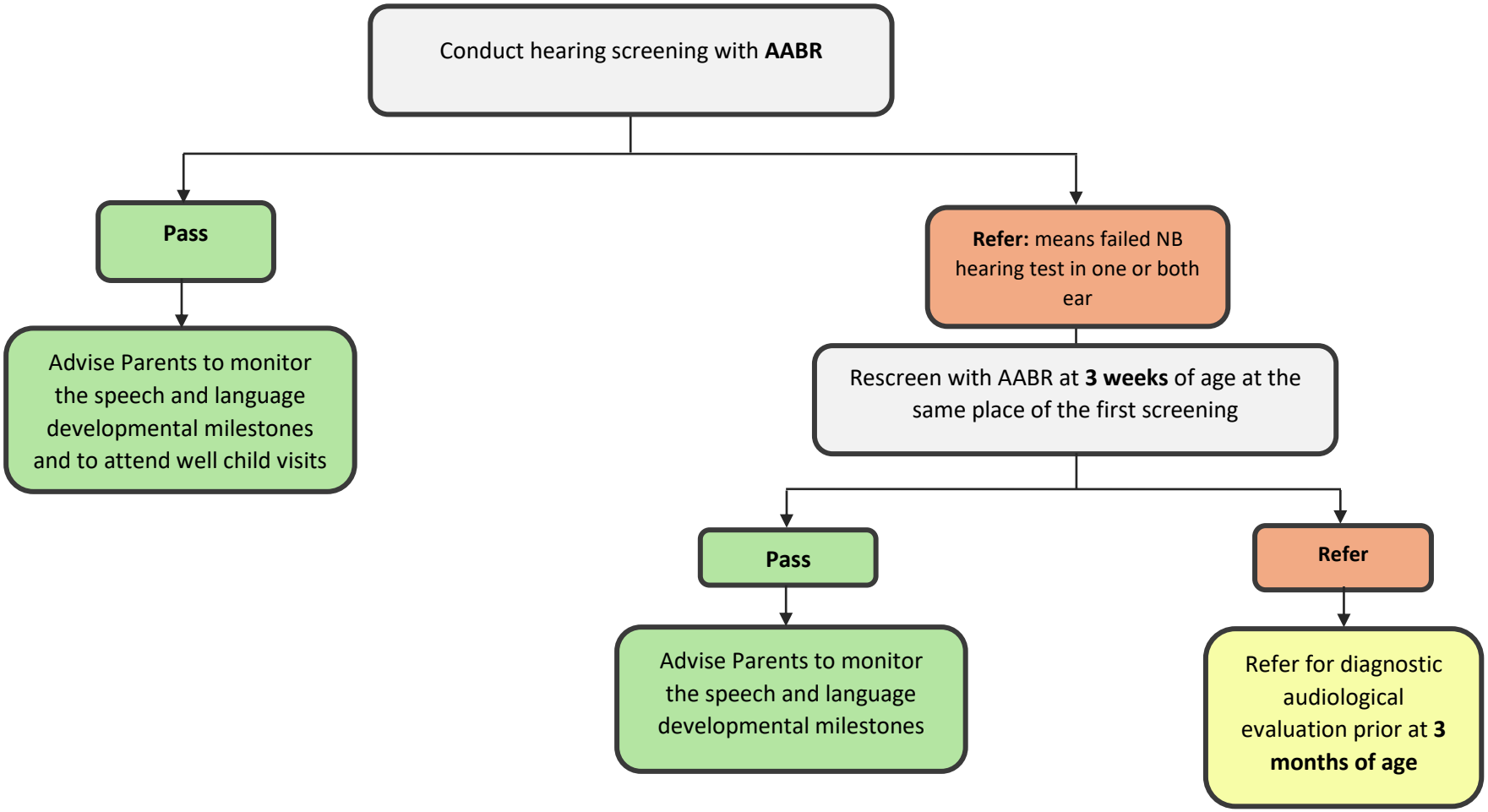
5. For re-admissions in the first month of life

All Newborns who have initially passed a hearing screening are rescreened if readmitted to the hospital in the first month of life with conditions associated with potential hearing loss (e.g. Hyperbilirubinemia requiring exchange transfusion or culture- positive sepsis), is recommended to have a repeat hearing screening either before discharge or through a referral to the ENT clinic, following the same protocol of high-risk infants with one stage AABR screening.

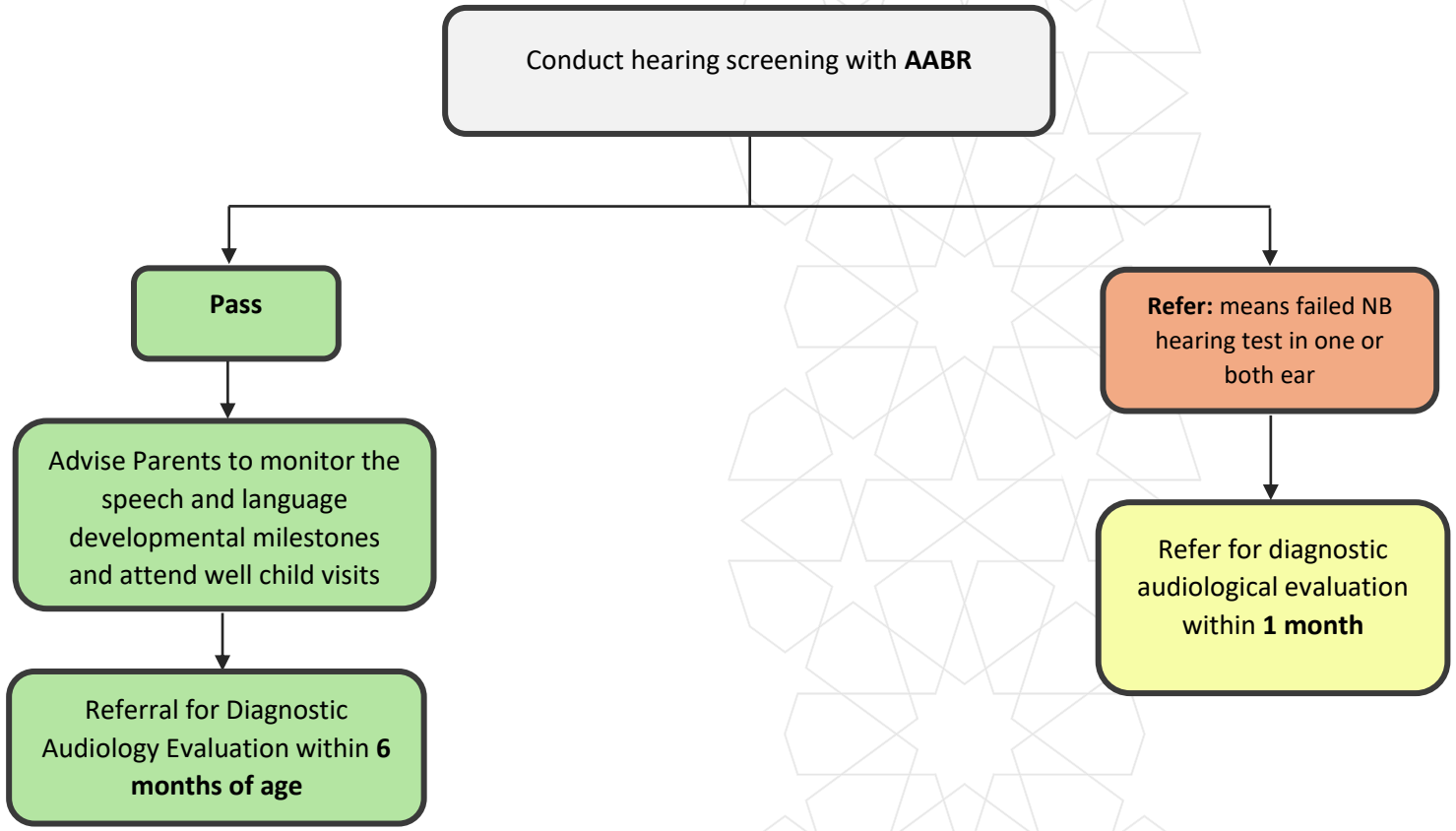
6. Standard Universal Precautions

- Decontamination, cleaning, disinfection, and sterilization of multiple-use equipment before reuse must be carried out according to the infection control policies and procedures and according to the manufacturer's instructions.

Charter (1): Hearing Screening for Healthy Newborns



Charter (2): Hearing Screening for High-Risk Newborns



[1] *Newborn Screening Manual. A GUIDE FOR NEWBORN CARE PROVIDERS. Ontario's newborn screening system. Edition 2.1, February 2018*

Appendix (3): Newborn critical congenital heart diseases screening

The primary target of CCHD screening is to detect the "Seven critical cyanotic heart diseases". The screening may also pick up other heart malformations and non-cardiac causes of cyanosis.

1. **Seven critical cyanotic heart diseases**
   - Hypoplastic left heart syndrome
   - Pulmonary atresia with intact septum
   - Total anomalous pulmonary venous return
   - Transposition of the great arteries
   - Truncus arteriosus
   - Tetralogy of Fallot
   - Tricuspid atresia

2. **Secondary targets**
   - Aortic Arch Atresia or Hypoplasia
   - Coarctation of the aorta
   - Double outlet right ventricle
   - Ebstein's anomaly
   - Interrupted aortic arch
   - Other single ventricle defect (other than hypoplastic left heart syndrome and tricuspid atresia).

3. **Additional non-cardiac causes of cyanosis potentially identified by CCHD screening**
   - Sepsis
   - Persistent Pulmonary Hypertension of the Infant (PPHN)
   - Respiratory Illnesses

4. **Inclusion Criteria**
   - All healthy newborns
   - Newborns in the Neonatal Intensive care Unit (NICU)

5. **Exclusions Criteria**
   - Newborns who had already echocardiography done,
   - For babies requiring oxygen more than 24 -36 hours of age and already cause of hypoxemia identified.
   - Preterm infant less than 35 weeks (required admission to NICU/SCBU).

6. **Screening Procedure**

   Method used: Pulse oximetry is a simple and non-invasive screening tool that measures oxygen levels in the infant's blood

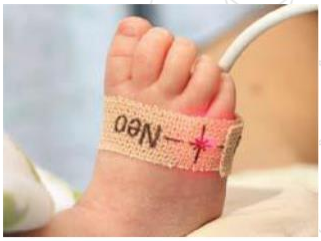
   Timing:
   - Screening should begin after 24 hours of age to 36 hours of age.
   - If discharge prior to 24 Hours of age is planned, screening shall be completed as close to discharge as possible.
   - Waiting until 24 hours of life will decrease the false positive results
   - To attract attention

7. **Preparation:**
   - Educate parent(s) about Newborn screening program including pulse oximeter screening for CCHD detection
   - Ensure that the environment is quiet.
   - It is preferred that the infant is awake, alert but quiet during the test.
   - Do not test an actively crying or cold stressed infant.
   - Bright lights and bilirubin lights should be turned off prior to screening.

8. **Procedure for screening with pulse oximetry**
    - Explain the procedure for the Parents/guardians.
    - Obtain the pulse oximeter(s) and disposable/reusable sensor(s).
    - Use high-quality equipment and machine settings that can limit the number of false-positive test results.
    - Turn pulse oximeter(s) on and allow time for self-test to run.
    - Select the application site:
        - make sure that the skin is clean and dry before applying the sensor.
        - Place the photodetector portion of the sensor on the fleshy portion of the infant's hand or foot.
        - Place the light emitter portion of the sensor on the top of the hand or foot directly opposite the photodetector.
        - **Right hand** (pre-ductal) around the right palm with the light under the 4th or 5th finger AND
        - **Either foot** (post-ductal) and place outer aspect of foot with the light under the 4th or 5th toe, either foot is acceptable.





- Calm and swaddle infant as necessary, pulse oximetry takes just a few minutes to perform when the infant is quiet, alert, and warm.
- Check waveform / signal quality as appropriate to the equipment.
- Keep pulse oximeter probe on hand/foot until pulse indicates accuracy.
- Test is considered complete once 10 seconds of reliable pulse oximetry has been achieved.

9. **Interpretation of the results**

- **First Measurement**
    - **PASS=** SpO2 ≥ 95% and ≤3 % difference between right hand and foot.
      No further action is required, notify parent(s) of the result and document the result in CCHD record and patient medical record.
    - **FAIL** = SpO2 < 90% on hand or foot
      Monitor infant for signs and symptoms of cardiac or respiratory distress and refer for immediate assessment

- **RETEST** = SpO2 is 90-94% or > 3 % difference between right hand and foot, repeat screen in one hour.

- **Second Measurement**: Keep pulse oximeter on and monitor infant, repeat screen in one-hour x1 (2 total screens):
  - **PASS** = SpO2 is ≥ 95% in both the hand and foot and ≤ 3% difference
    No further action is required, notify parent(s) of the result and document the result in CCHD record and patient medical record.
  - **FAIL** = SpO2 < 95% on right hand or foot and/or > 3 % difference between hand and foot.
    - Repeat the screening in one hour after the second screening (third screening)
    - Monitor infant for signs and symptoms of cardiac or respiratory distress.

10. Assessment of Newborns with Positive Screens:

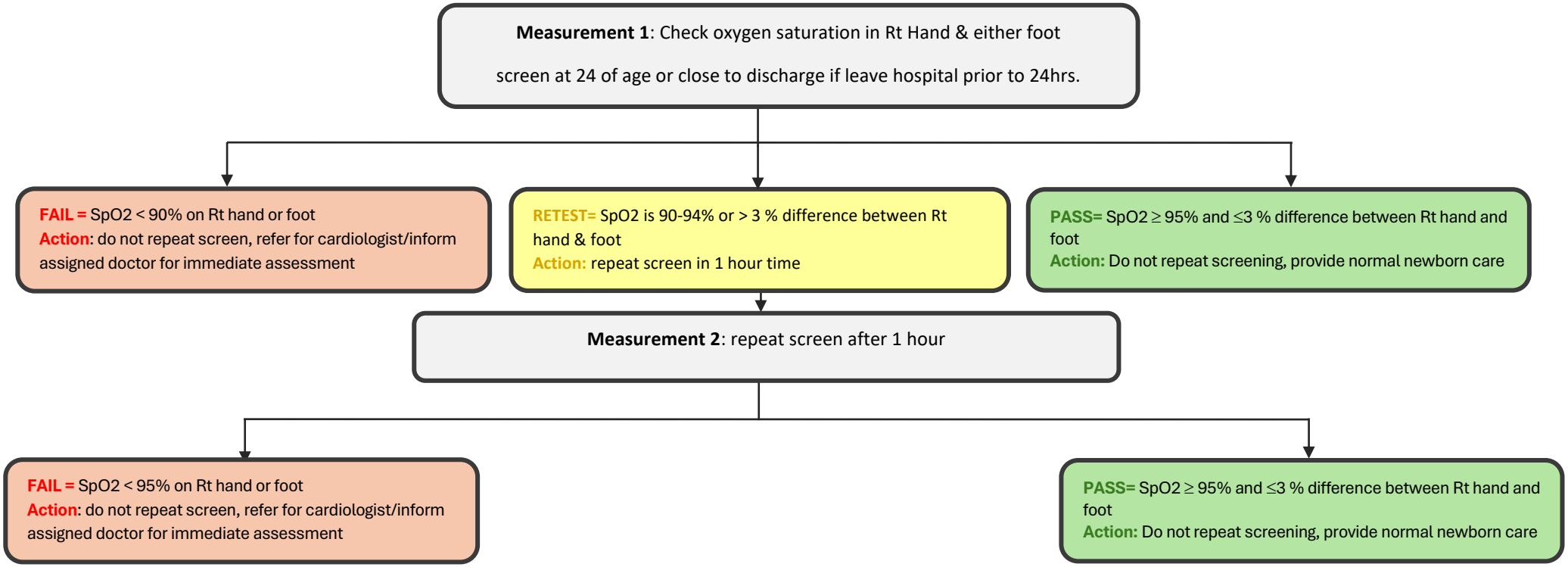
- A neonate with hypoxemia shall not be discharged from the hospital without excluding potentially life-threatening conditions. Newborns with positive screening results shall undergo evaluation to identify the cause of hypoxemia.
- **Causes:** Neonatal hypoxemia can be due to cardiac, pulmonary, metabolic, neurological, infectious, and hematologic disorders.
- **Evaluation:** Infants with persistent hypoxemia shall have an immediate complete History and clinical examination including the following:

  - Continuous pulse oximeter monitoring, preductal (right hand) and post ductal (either foot) SPO2 check. It is important to have a reading check for 4 limbs BP and ensure that the baby is calm while checking.
  - Chest XR, ECG, blood gas, Lactate.
  - Echocardiography.
  - Sepsis works up including CBC, CRP, blood culture.
  - Blood glucose, electrolytes and minerals.
  - Evaluation and respiratory support including oxygen therapy, and if needed to start on Prostaglandin infusion, shall not be delayed while awaiting an echocardiogram.

[2] *Ewer, A. K., & Martin, G. R. (2016). Newborn pulse oximetry screening: Which algorithm is best? Paediatrics, 138(5).* https://doi.org/10.1542/peds.2016-1206

-Martin, G. R., Ewer, A. K., Caviglia, A., Hom, L. A., Saarinen, A., Sontag, M., Burns, K. M., Kemper, A. R., & Oster, M. E. (2020). Updated Strategies for pulse oximetry screening for Critical congenital heart. Disease. Pediatrics, 146(1). https://doi.org/10.1542/peds.2019-1650

Charter (3): Clinical pathway for newborn Critical Congenital Heart Diseases Screening



[3]

[3] - *Jegatheesan, P., Song, D., Angell, C., Devarajan, K., & Govindaswami, B. (2013). Oxygen saturation nomogram in newborns screened for critical congenital heart disease. Paediatrics, 131(6), e1803–e1810. https://doi.org/10.1542/peds.2012-3320*

-UpToDate. (n.d.). UpToDate. https://sso.uptodate.com/contents/evaluation-of-suspected-critical-congenital-heart-disease-chd-in-the-newborn

- Clinical screening and diagnosis for critical congenital heart defects. (2024, May 15). Congenital Heart Defects (CHDs). https://www.cdc.gov/heart-defects/hcp/screening/index.html

Appendix (4): Newborn heel prick sampling procedure, handling, packing and shipping

1. Timing of collection:

1.1 Full term or Preterm infants ≥ 33 weeks gestational age and ≥1500g:

a. A newborn screening sample should be collected for all newborns between 24 and 48 hours of baby's age (Not less than 24 hours) prior to discharge from hospital.
b. If the newborn is discharged before 24 hours the first sample should be taken, and the parents should be instructed to attend the birth hospital for a repeat screen test within 24-48 hours of age.

1.2 Premature < 33weeks of gestational age at birth, or birth weight < 1500:

a. The first sample should be collected between 24 to 48 hours of age.
b. The second sample should be collected at 3 weeks of age or prior to discharge whichever comes first.

1.3 Required Blood transfusion (Packed Red Blood Cells -PRBC):

a. In the newborn screening program, a blood transfusion is defined as recipient of packed red blood cells (PRBC). Indicate "NO" for transfusion status on the newborn screening filter Card if a newborn has only received fresh frozen plasma (FFP) and/or platelets.

b. For Full term or Preterm infants = 33 weeks gestational age or =1500g:

- **First sample:** prior to blood transfusion if possible & regardless of newborn's age and gestational age.
- **Second sample**: 48-72 hours post-transfusion only if the first sample was taken before 24 hours of age.
- If the first sample was not taken before blood transfusion, a sample should be taken at 48-72 hours post transfusion and another sample at 120 days after the last blood transfusion.

c. For preterm< 33 weeks or Birth weight < 1500g:

- **First sample**: prior to blood transfusion if possible & regardless of newborn's age and gestational age
- **Second sample**: 48-72 hours post-transfusion only if the first sample was taken before 24 hours of age.
- **Third sample**: 3 weeks of age or prior to discharge whichever comes first.
- If the blood sample was not collected prior to the transfusion, three blood samples should be collected:
  - 1st sample at 48-72 hours post transfusion.
  - 2nd sample at 3 weeks of age or prior to discharge whichever comes first.
  - 3rd sample at 120 days after the last blood transfusion

1.4 Total Parenteral Nutrition (TPN):

- First sample: Prior to treatment if possible & regardless of newborn's age and gestational age
- Second sample: 24-48 hours of age only if the first sample was taken before 24 hours of age.
- Third sample: 3 days after cessation of TPN.
- Mark "TPN" and write the last Total Parental Nutrition date in the screening form if given.

2 Handling transporting, storage and retention of the specimen.

1. Preparation for Specimen Collection:

- Perform hand hygiene.
- Parts of the filter paper circles should not be touched before, during or after collection to avoid specimen contamination.
- The information details (especially two contact numbers) on the blood, as per the consent document contact information
- Spot cards should be completed at the time of sampling.
- The identity of the neonate should be checked with the laboratory test request form. See appendix (10) for guidance on the quality of the sample.

2. ***Filter Paper Handling and Transport Procedure***:

- The specimen card should be dried in a horizontal position.
- The Blood spots should be dried thoroughly at room temperature for at least 4 hours.
- The specimen card should be kept away from direct heat or sunlight and stored in ventilated areas not in closed areas such as drawers /refrigerator.
- The patient information section should be verified and filled out before sending the specimen.
- The list of the specimens that were sent should be documented. Dried specimens should be inserted into an envelope (no plastic bags) & transported at 15 to 22 degrees Celsius within 24 hours of collection. For weekend & holiday specimens should be stored at room temperature and sent at the earliest possible.

3. **Laboratory Report Policy**: whenever it becomes evident that erroneous laboratory test results have been reported, an amended report must be completed with all corrections and the reason for the error documented. Both the amended and erroneous laboratory reports are considered legal documents and must be retained together as per facility accreditation standard & Health authority regulations.

3.1 Notify the clinician as soon as possible of the error and the amended results. If patient care has been affected by the erroneous results, the Laboratory Director must also be notified. Clerical or transcription error: Enter the correct laboratory result into LIS. In the comments area for the corrected result, indicate time and date, your initials, and which clinician was notified of the correction. Results will be retransmitted to PNC as "corrected."

3.2 Technical or testing error: re-run test if possible or call the patient back for the new specimen. Enter the corrected results into LIS with the reason for the error. On the amended report, indicate the time and date that the clinician was notified of the correction. Results will be retransmitted to PNC as "corrected.

3.3 If laboratory testing was done on a specimen from the wrong patient, notify the clinician to contact the correct patient to submit a new specimen as soon as possible. Document in the lab report that a specimen error was made indicating time and date with your initials that the clinician was notified. The result will be retransmitted to PNC as "corrected". When testing of the new specimen is completed, attach an amended report to the erroneous report (labeled as such).

4. Specimen Storage and Retention Period

4.1 The newborn screening dried blood spot card should be stored in the refrigerator (2-8C) or at ambient temperature (18-25) until testing is completed. After testing is finished, the cards are stored in a walk-in refrigerator until disposal. After testing, the card is stored for one year pending requests for follow-up on additional testing to confirm screening test results, for use in internal quality assurance, and instrumentation and/or methodology validation studies. At the end of one year, these specimen cards containing dried blood spots will be destroyed according to "Procedure for Laboratory Safety Practices".

4.2 Specimens that are presumptive positive for any of the diseases included in the newborn screening panel are stored at laboratory infinitely at -20 ºC.

4.3 Retained DBS and associated demographic information can be used for the following purposes:

4.3.1 Re-analysis to confirm the original test results.

4.3.2 Internal method development and method validation studies, including the setting of appropriate cut-offs or normal ranges.

4.3.3 Quality assurance audits.

4.3.4 Aiding another laboratory in developing or validating a newborn screening method (which requires a statement from the laboratory requesting the specimens that specify how the specimens will be used, and written approval from the Laboratory Director).

4.4 Disposal DBS will be autoclaved and then handled as medical waste, which involves off-site incineration as per "Procedure for Laboratory Safety Practices".

4.5 Release of Specimens.

Access to newborn screening specimens is restricted to Laboratory staff involved with specimen receipt, testing, data entry, and laboratory management only.

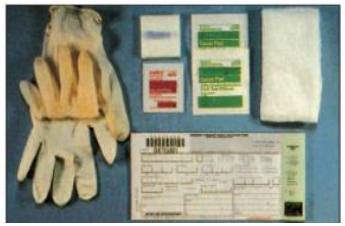
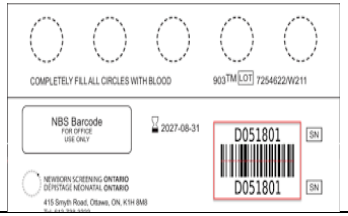
4.*6 DBS may be transferred to other entities as delineated below:*

4.6.1 An entity that has a contract with the Laboratory to perform additional (i.e., Second Tier) testing in response to an out-of-range screening result.

4.6.2 A health care provider at the request of the patient, legal guardian, or legal representative after completing and signing a written request form.

4.6.3 A researcher with written, informed consent from the patient, legal guardian or representative, if the research project has been reviewed and approved by MOHAP or Local Authority.

Newborn heel prick sampling procedure, handling, packing and shipping

<table>
<tr>
<td> 
 
</td>
<td>Sampling:

➢ Check the identification of the <u>newborn.</u> All information details should be completed on the filter card at the time of sampling including two contact numbers. All fields on the form must be completed.

➢ Before collecting the sample, the following materials must be gathered:
• Sterile lancet with tip less than 2.4 mm long
• Sterile 70% alcohol pads
• Sterile gauze pads
• Warm, moist towel or compress
• Fully completed, in-date filter paper blood collection form.
• Sterile gloves</td>
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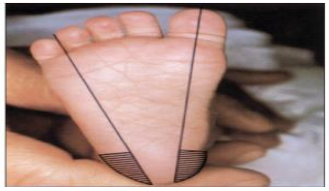
<tr>
<td>  </td>
<td>➢ The dried blood spot (DBS) circles shouldn’t be contaminated with spillage or by touching before or after blood collection to avoid specimen contamination.</td>
</tr>
<tr>
<td>  </td>
<td>➢ Hatched area indicates safe areas for puncture site.</td>
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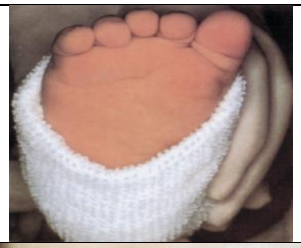
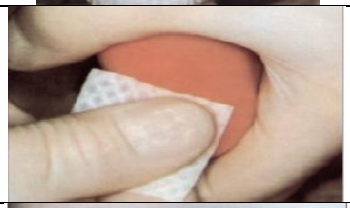
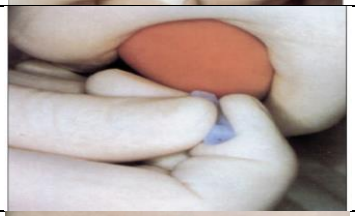
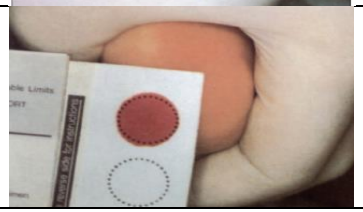
	➢ Utilize appropriate measures to enhance comfort for the baby during blood spot collection (e.g., breastfeeding, skin-to-skin contact)
	➢ Warm puncture site with a soft cloth, moistened with warm water (not hot) for 3 to 5 minutes.
	➢ Clean the puncture site with a sterile alcohol Prep and allow to air dry or wipe dry with sterile gauze pad.
	➢ Puncture skin and wipe away the first drop of blood with a sterile gauze pad. Allow a second large blood drop to form. To obtain sufficient blood the infant's heel should be punctured with a sterile lancet to a depth of 2.0 mm.
	➢ Lightly touch the filter paper to the large drop of blood. ➢ Allow the blood to soak through and completely fill the circle with a single application of large blood drop. (To enhance blood flow, very gentle intermittent pressure may be applied to the area surrounding the puncture site). ➢ Apply blood to one side of filter paper only.

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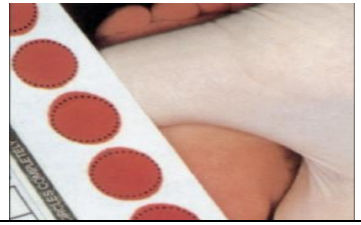
<td>  </td>
<td>

Sequentially fill the remaining circles with blood drops in the same manner as the previous step. If blood flow is diminished, repeat the previous steps with sterile equipment.
Infection control measures should be applied during skin puncture and should be consistent with the health facility standard procedures.

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Specimen Handling and Integrity:

Following the application to the blood spot collection card:

Allow the DBS cards to dry in a horizontal position for a minimum of 4 hours in a ventilated and not in a closed areas such as drawers and refrigerators
Avoid touching or smearing the blood spots.
Avoid exposing the specimen to direct sunlight, heat, alcohol, intense fluorescent lights, fumes from preservatives such as formaldehyde, or fumes from various disinfectants such as Clorox® as they can interfere with the analysis or render the specimen unacceptable.
Send the dry specimen to the NBS laboratory as soon as possible after the blood spots have dried.
Samples should be sent within 24 hours of collection. Delays could have serious consequences for affected infants and may render the sample unsatisfactory.

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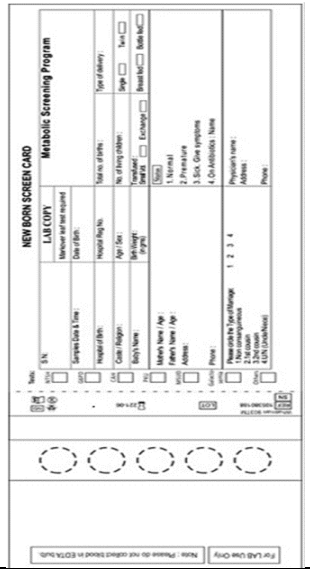


Packaging and Shipping:

- The patient information section should be verified and filled out before sending the DBS card (attached Card)

- ***Use the basic triple-packaging system to ship the filter paper cards:***

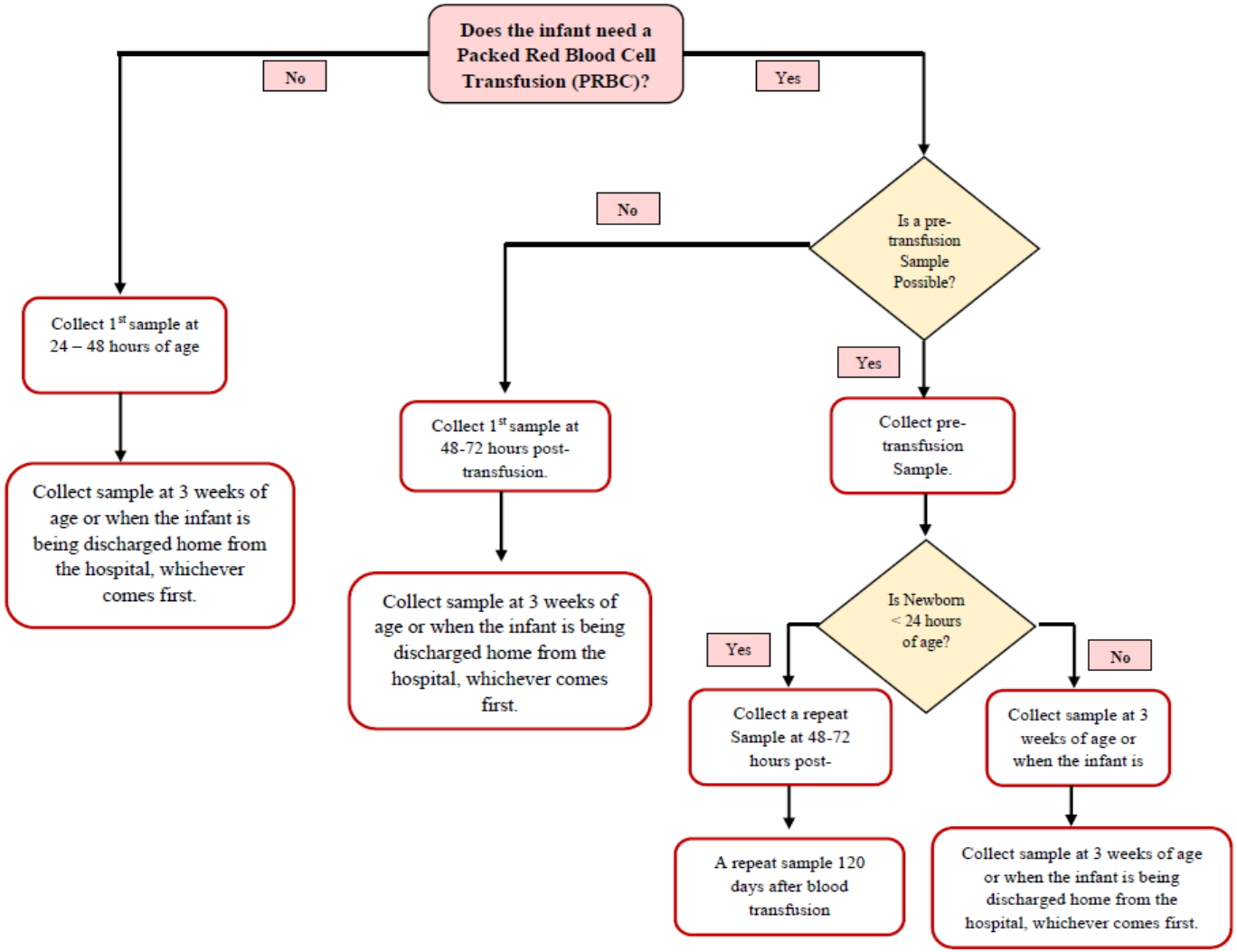
  1. The primary container is the filter paper matrix that contains the absorbed and dried blood.
  2. A secondary container must enclose the filter paper. The secondary container should have a fold-over flap or an inner envelope to secure the contents.
  3. To place the secondary envelope in a bigger sturdy envelope. This will provide safety from occupational exposure and maintain optimal specimen integrity.

- DBS cards should be inserted into an envelope (not plastic bags) & transported at 15 to 22ºC within 24 hours after collection.

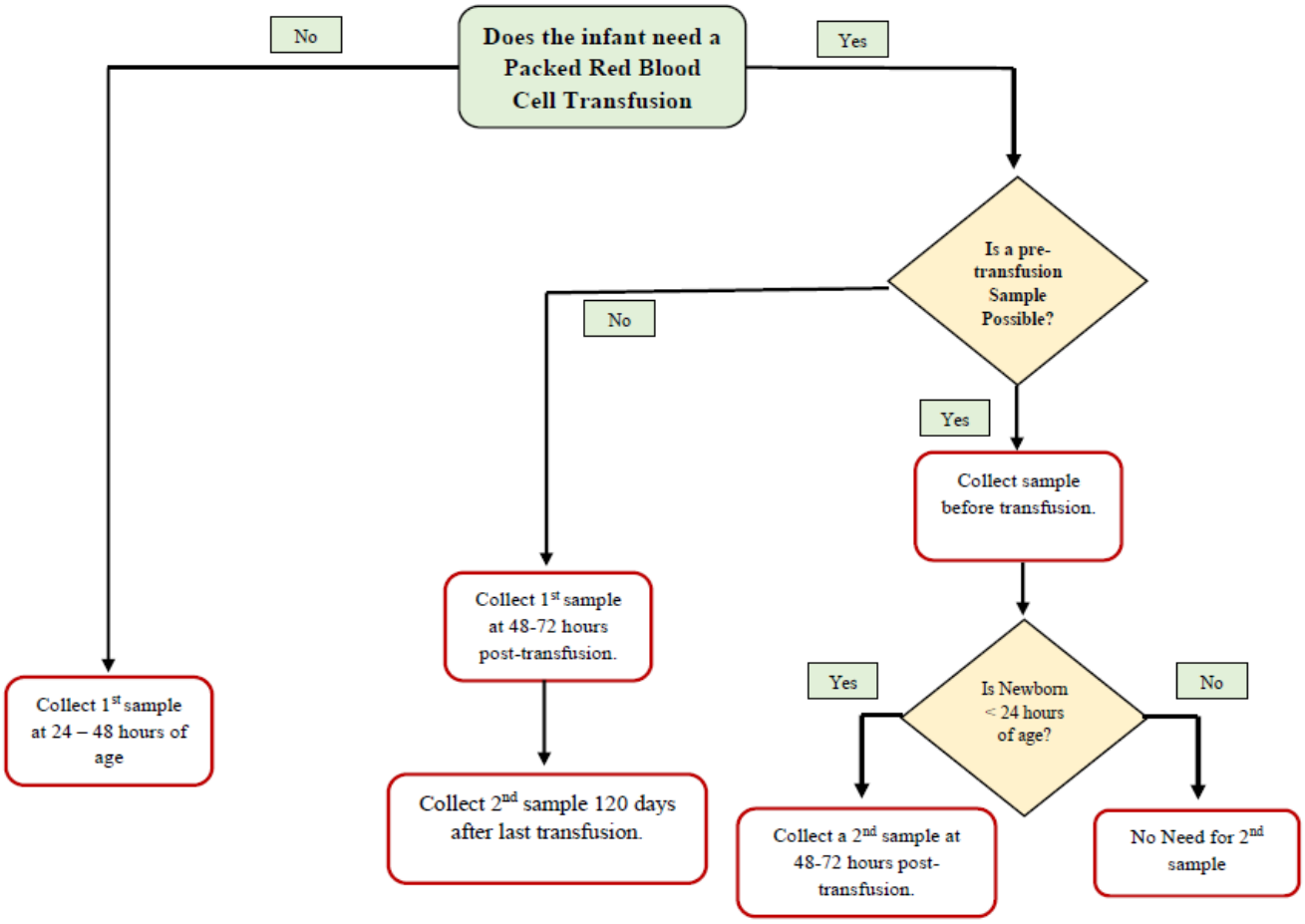
- In Special Conditions Specimens should be stored & placed in a sealable plastic bag in a refrigerator (2- 8 °C) and shipped as soon as possible on the next day.

- The list of the newborn screening details along with the DBS cards should be prepared and sent (hard & soft copies) from the health care facility to the NBS lab.

Appendix (5): Preterm < 33 weeks gestational age or Very Low Birth Weight Infants < 1500g +/- PRBC Transfusion



Appendix (6): Full term and Preterm infant >33 weeks or > 1500g+/- PRBC Transfusion



Appendix (7): Newborn blood test screens the listed disorders.

Endocrine Disorders

- Congenital hypothyroidism (CH)
- Congenital adrenal hyperplasia (CAH)

1. **Hematological disorders**

- Sickle cell anemia (Hb SS)
- Sickle-cell disease (Hb S/C)
- Hb S/Beta-Thalassemia (Hb S/Th)
- Thalassemia major
- Variant hemoglobinopathies (including HbE)
- Glucose-6-phosphate dehydrogenase (G6PD deficiency)

2. **Amino Acids, Organic Acids and Fatty Acids Disorders**

a. Amino acid disorders

- Classic Phenylketonuria (PKU)
- Tyrosinemia I, II, III (TYR I, II, III)
- Argininosuccinic aciduria (ASA)
- Citrullinemia (CIT Type I &II)
- Maple syrup urine disease (MSUD)
- Defects of biopterin cofactor
- Classic Homocystinuria
- Hypermethioninaemia
- Argininemia

b. Organic Acidemia Disorders

- Glutaric acidemia type I (GA I)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemia (MMA) (MUT cblA and cblB forms
- Propionic acidemia (PA)
- Hydroxy methyl glutaric aciduria (Hydroxymethylglutaryl lyase deficiency) (HMG)
- Beta-ketothiolase deficiency (BKT)

- Holocarboxylase synthase deficiency

c. <u>Fatty Acid Oxidation Disorders</u>

- Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Trifunctional protein deficiency (TFP)
- Carnitine uptake defect (CUD)/Carnitine Transport Defect
- Glutaric acidemia type II (Multiple Acyl-CoA Dehydrogenase Deficiency) (MAD; GA-II)
- Carnitine palmityl transferase deficiency type 1
- Carnitine palmityl transferase deficiency type 2
- Carnitine/acylcarnitine Translocase Deficiency (Translocase)

d. <u>Other Enzyme Disorders</u>

- Biotinidase deficiency (BIOT)
- Galactosemia (Classic GALT deficiency)

3. Respiratory

- Cystic Fibrosis (CF)
- Very-long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

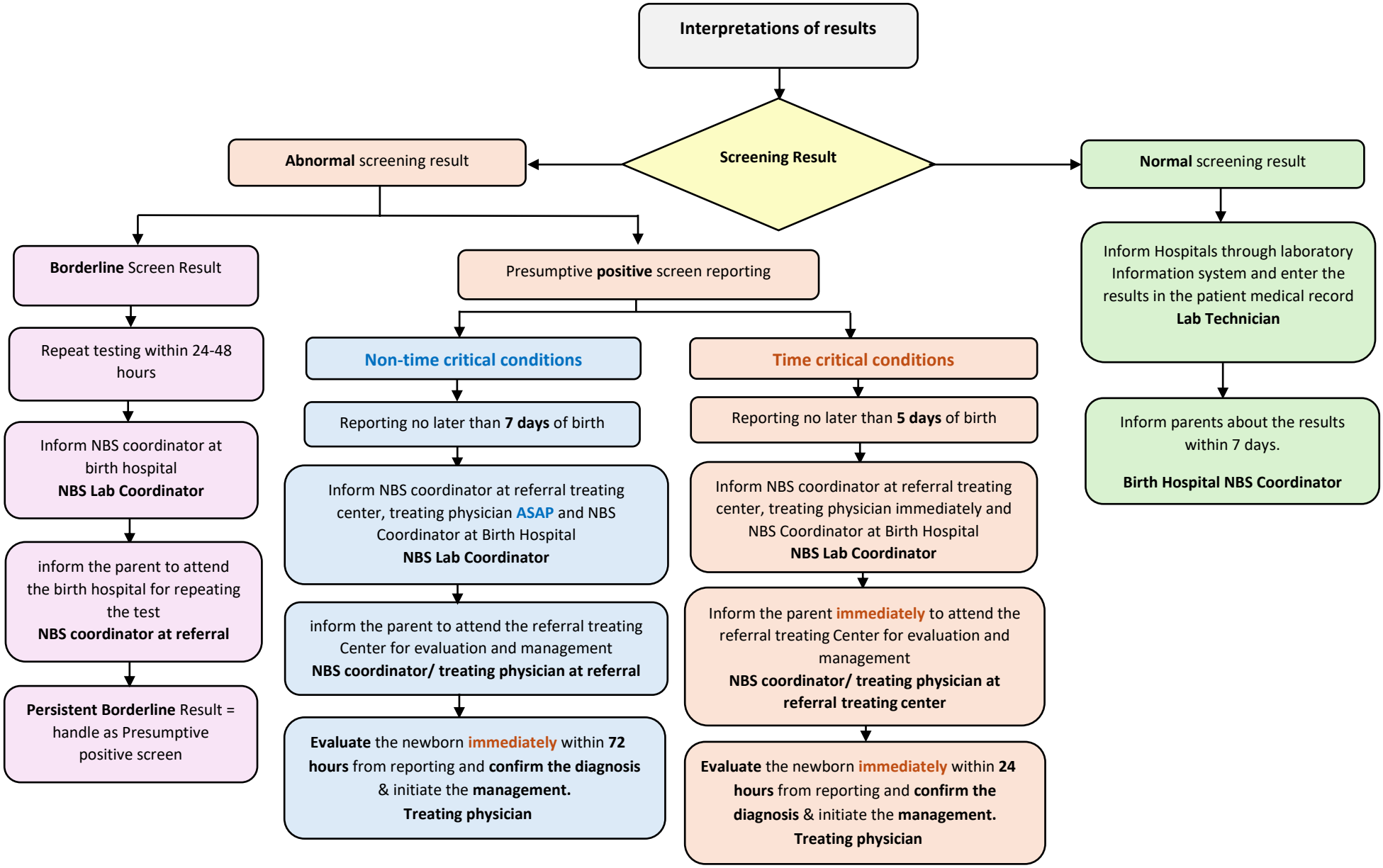
Appendix (8): Critical and non-critical disorders

These are conditions for which acute symptoms or potentially irreversible damage could develop in the first week of life, and for which early recognition and treatment can reduce the risk of illness and death.

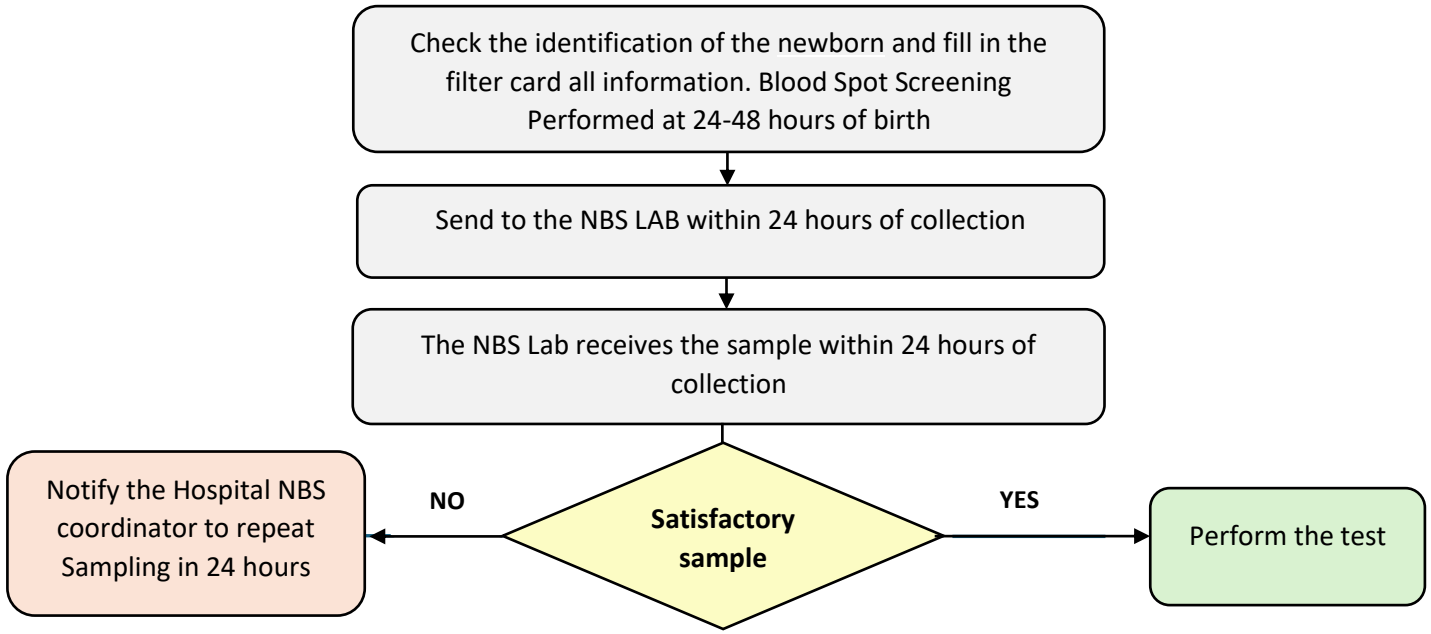
No.		**Disorders**
1	**Critical**	Argininosuccinic aciduria (ASA)
2		Citrullinemia (CIT Type I &II)
3		Maple syrup urine disease (MSUD)
4		Methylmalonic acidemia (Methylmalonyl-CoA mutase deficiency) (MUT)
5		Methylmalonic acidemia, cblA and cblB forms (MMA, Cbl A, B)
6		Propionic acidemia (PROP)
7		Congenital adrenal hyperplasia (CAH)
8	**Non-Critical**	Classic Phenylketonuria (PKU)
9		Benign hyperphenylalaninemia
10		Defects of biopterin cofactor biosynthesis
11		Defects of biopterin cofactor regeneration
12		Argininemia
13		Tyrosinemia I, II, III (TYR I, II, III)
14		Homocystinuria
15		Hypermethioninaemia
16		Glutaric acidemia type I (GA I)
17		Hydroxy methyl glutaric aciduria (Hydroxymethylglutaryl lyase deficiency) (HMG)
18		Isovaleric acidemia (IVA)
19		3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
20		Beta-ketothiolase deficiency (BKT)
21		Holocarboxylase synthase deficiency
22		Isobutyryl-CoA dehydrogenase deficiency (IBDH)
23		Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
24		Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
25		Very-long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

26		Trifunctional protein deficiency (TFP)
27		Carnitine uptake defect (CUD)/Carnitine Transport Defect
28		Glutaric acidemia type II (Multiple Acyl-CoA Dehydrogenase Deficiency) (MAD; GA-II)
29		Carnitine palmityl transferase deficiency type 1
30		Carnitine palmityl transferase deficiency type 2
31		Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
32		Carnitine/acylcarnitine Translocase Deficiency (Translocase)
33		Congenital hypothyroidism (CH)
34		Biotinidase deficiency (BIOT)
35		Galactosemia
36		Sickle cell anaemia (Hb SS)
37		Sickle-cell disease (Hb S/C)
38		Hb S/Beta-Thalassemia (Hb S/Th)
39		B-Thalassemia major
40		Variant hemoglobinopathies (including Hb E)

Appendix (9): Reporting of Newborn Blood Spot Screening Results



Appendix (10): Pathway of Newborn Blood Spot Screening



[4]

[4] *CLINICAL PRACTICE GUIDELINE. Newborn Bloodspot Screening. King Edward Memorial Hospital Obstetrics and Midwifery Clinical Care, 2017*

Appendix (11): Newborn screening – Parent refusal form

Name of Infant _____

Hospital of Birth _____

Date of Birth _____

City / Emirate _____

Medical Record Number _____

- I have been informed that newborn screening is mandated for all babies born in the United Arab Emirates
- I have received information about the Newborn Screening Program and understand there are many rare, inherited disorders for which newborns are screened for.
- I understand that the screening is done for the early detection of treatable disorders. I understand that symptoms sometimes do not appear for several weeks or months.
- I do not consent to do for my baby:

  - ☐ Newborn blood spot screening.
  - ☐ Newborn Hearing Screening.
  - ☐ Newborn Critical Congenital Heart Screening.

- I understand that when newborn screening conditions are not detected and treated in the newborn period, there can be permanent damage such as mental retardation, developmental delays, growth failure, and even death.

- I have discussed my decision with a health care provider.

- I understand the benefits of newborn screening. The potential dangers of not being screened have been explained to me. My decision to refuse the testing was made freely and without force or encouragement by my doctor or midwife, my baby's doctor, the hospital staff, or state officials.
- I accept all responsibility, legal and otherwise, for this decision.

Parents / guardian Name _____

witnessed by (Healthcare provider name) _____

Relationship to child _____

Healthcare provider signature _____

Parent / guardian signature _____

Date

فحص حديثي الولادة - استمارة رفض ولي الأمر

اسم الطفل _____

اسم مستشفى الولادة _____

تاريخ الميلاد _____

المدينة / الإمارة _____

رقم السجل الطبي _____

- لقد تلقيت معلومات حول أهمية إجراء فحوصات حديثي الولادة وأدركت أن هناك العديد من الاضطرابات الوراثية النادرة التي يتم فحص حديثي الولادة من أجلها.
- لا أوافق على القيام بما يلي لطفلي:
- فحص الدم لحديثي الولادة.

- فحص السمع لحديثي الولادة
- فحص القلب عن العيوب الخلقية الحرجة لحديثي الولادة.
- أفهم أن اختيار عدم إجراء الفحص لطفلي قد يؤدي إلى تأخير في تشخيص وعلاج الحالات الطبية الخطيرة التي قد تؤثر على النمو والتطور الطبيعي للطفل وفي حالات نادرة قد تؤدي إلى الوفاة.
- لقد ناقشت قراري هذا مع مقدم الرعاية الصحية.

اسم الوالدين / الوصي

_____

مشهودًا عليه (اسم مقدم الرعاية الصحية)

_____

العلاقة للطفل

_____

توقيع مقدم الرعاية الصحية

_____

توقيع ولي الأمر / الوصي

_____

التاريخ

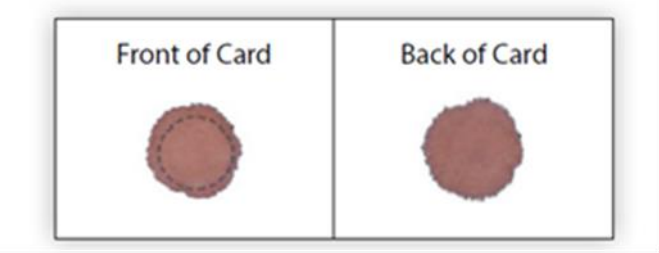
_____

Appendix (12): Blood specimen's acceptance and rejection criteria

Each specimen received at NBS Laboratory shall be reviewed for specimen quality and quantity.

a. Acceptance criteria:

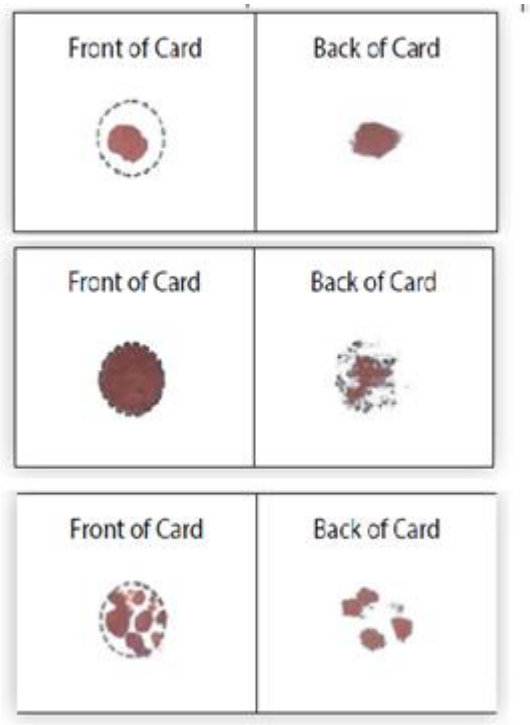
- Patient information details should be completed
- The DBS card should be inserted into an envelope (not plastic bags)
- The samples should be satisfactory for examination as shown in the figure:
    - No areas of white should be visible on the front or back of the circle.
    - The blood must fully soak through to the back of the filter paper.
    - It is estimated that 75 uL – 100 uL of blood is required to fill one circle on the filter paper.



b. Rejection criteria:

- The newborn's data information is not completed on the examination card.
- The amount of blood is unsatisfactory for examination in the following conditions:
    - **Quantity of blood insufficient & circles are not sufficiently filled:**

- The specimen appears sufficient from the back but is insufficient when viewed from the front.
- The specimen appears sufficient from the front but is insufficient when viewed from the back.
- The specimen appears insufficient from the front and the back

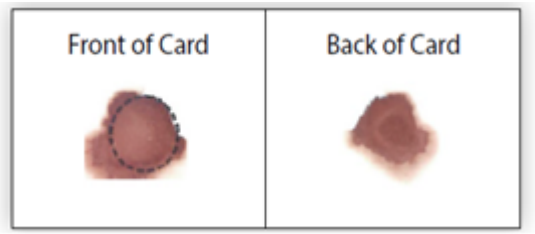


- **Blood spots are wet and/or discolored /or supersaturated /or diluted**

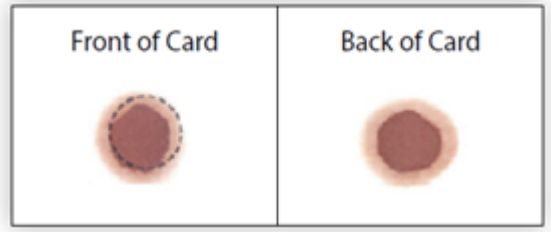
- Do not allow water, feeding formulas, antiseptic solutions, gloves powder, hand lotion, or other materials to touch with the specimen card before or after use.

Ensure that the infant's heel is dry and free of alcohol prior to performing the heel stick.

Ensure that the specimen is dry before shipping.

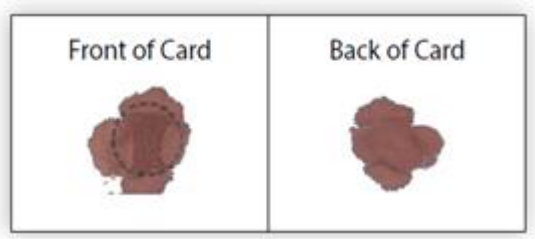


- Blood spots exhibit serum ring excessive milking or squeezing the puncture may cause haemolysis of the specimen or result in a mixture of tissue fluids.



- Blood spots appear clotted or layered

Applying successive drops of blood to already partially dried spots cause “layering” & inaccurate blood volume collection, which results in non-uniform analyte concentrations.



c. Procedure of repeat sample collection

Repeat samples are requested for all unsatisfactory specimens & whenever required for borderline results.

The laboratory Coordinator will contact the Hospital Coordinator to notify the nurse to repeat the specimen collection.

Parents should be informed of the reason for the repeat. Immediate second specimen collection is required for certain parameter.

Note: A one-week interval between samples is recommended for borderline thyroid-stimulating hormone (TS results, other parameters may require two weeks intervals.

d. Methodologies for specimen Testing

a) Screening:

- TSH, 17α- OH Progesterone, G6PD, Biotinidase, Galactosemia and IRT to be performed by immunofluorescence method.
- Sickle cell disease & Other Hemoglobinopathies to be performed by High Performance Liquid Chromatography (HPLC) /ISOELECRIC FOCUS/CAPILLARY ELECTROPHORESIS.

- Amino Acid, Organic Acid & Fatty Acid Oxidation Disorders to be performed by Tandem Mass Spectrometry (MS/MS).
- The Equipment, Reagents & Method used must be FDA approved and/or CE Mar

b) Confirmatory testing:

- Amino Acid analyser or Liquid Chromatography Mass Spectroscopy: for Amino Acid and Acylcarnitine disorders.
- Gas or Liquid Chromatography Mass Spectroscopy: for Organic Acid disorders.
- High performance Liquid Chromatography: for Haemoglobin disorders.
- Immunoassays analyser: for enzyme and hormonal disorders.
- Genetic analyser: PCR based, or sequencing based**.**

Reporting of results:

All results should be submitted by the NBS laboratory to the concerned health facility through the laboratory information system and in the patient's electronic medical records. Normal screening results: parents should be informed about the results by the Newborn screening Coordinator in the Health Facility within 7 days.

- **Abnormal screening results are defined as "Presumptive positive screen" or "borderline result":**
- **Presumptive Positive screen**: Indicates that the newborn may have a disorder and should be referred to medical specialists for confirming the diagnosis and early intervention.
- **Presumptive Positive results of time-critical conditions** (should be reported through email and phone calls by Lab newborn screening coordinator immediately to**:**

(a) The treating physician in Referral Treating Center to arrange the appointment for the newborn.

(b) The NBS coordinator at referral treating centre.

(d) The newborn screening coordinator in the birth hospital to be informed about the results.

- The Referral treating Centre NBS coordinator, or the treating physician shall inform the parents to attend the referral treating center for further evaluation and management. The reporting of result should be no later than 5 days of birth to prevent delay in the diagnosis and treatment. The treating physician should evaluate the newborn immediately within 24 hours from time of reporting and confirm the diagnosis to initiate the management.
- **Presumptive Positive results for all non-time critical conditions** appendix (12) should be reported through email and phone calls by Lab newborn screening coordinator to the Treating physician and NBS coordinator in the referral treating center as soon as possible, in addition to NBS coordinator in the birth hospital. The reporting should be no later than 7 days of birth. The Referral treating center NBS coordinator or the treating physician shall inform the parents to attend the Referral treating center for further evaluation and management. The treating physician should evaluate the newborn immediately within 72 hours from time of reporting to confirm the diagnosis and initiate the management.
- **Borderline result**: Indicates that the screening was slightly abnormal, and the newborn needs a repeat newborn screening sample testing within 24-48 hours. The NBS lab coordinator shall inform the NBS coordinator at birth hospital to inform parents to repeat the test at the same birth hospital. If the repeat is still borderline, it will be reported as “Persistent Borderline” and is treated in the same manner as a Presumptive Positive screen.
- **False negative newborn screening** identified by specialists in the clinic should be reported back to the laboratory, National Newborn screening coordinator and committee.