



**Abu Dhabi Newborn Screening Program
Healthcare Professional's Manual
First Edition 2009**



Health Authority-Abu Dhabi
Reliable Excellence in Healthcare

Newborn Screening Program in the Emirate of Abu Dhabi 2009

Health Care Professional's Manual

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Health Authority – Abu Dhabi
Reliable Excellence in Health Care

Acknowledgment

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We appreciate your support in showing us ways of improving and strengthening this program.

The Newborn Screening Program in the Emirate of Abu Dhabi Contact Information:

Phone 800 800 or +971 2 4193 543

Email mshehhi@haad.ae

Fax +9712 4444 285

Building Address (for visitors, couriers, deliveries):

Health Authority Abu Dhabi (HAAD)

Non-Communicable Disease Section, Office Number IR-11

Public Health and Research Department

Public Health and Policy Division

The Emirate of Abu Dhabi

United Arab Emirates (UAE).

Mailing Address:

Health Authority of Abu Dhabi

Non-Communicable Disease Section

P.O. Box 5674

United Arab Emirates (UAE).

Newborn Screening Program in the Emirate of Abu Dhabi 2009

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1. Introduction

The Health Authority of Abu Dhabi developed this manual to be a reference guide for health care professionals involved in newborn screening in the Emirate of Abu Dhabi. Primary care practitioners, Obstetricians and Pediatricians who are involved in the prenatal, initial and postnatal care of newborns, nurses, laboratory personnel, should find this manual helpful in coordinating further laboratory diagnosis and follow-up of babies with an abnormal newborn screen.

The manual includes an overview of the Newborn Screening Program in the Emirate of Abu Dhabi, the newborn screening pathway. It provides a brief overview of the conditions screened in the newborn screening program in the Emirate of Abu Dhabi and information on proper screening practices– newborn screening specimen collection, testing methodologies, normal values, criteria for abnormal results, hemoglobinopathy interpretation chart, and blood spot screening results, notification, referral, treatment and resources.

The information in this manual is updated periodically in the online version, on the Health Authority website (www.haad.ae). Check there for the most current information on contact persons, testing methods, and so forth.

2. Overview of the Newborn Screening Program in the Emirate of Abu Dhabi 2009.

A standardized, an integrated and comprehensive Newborn Screening program in the Emirate of Abu Dhabi was launched by the Health Authority of Abu Dhabi in March 6th. 2010. The program is implemented in the Public and Private sectors in the Emirate of Abu Dhabi.

All newborns in the Emirate of Abu Dhabi should have access and are required to receive newborn screening as per the mandate of the Health Authority of Abu Dhabi unless their parent or guardian refuse such testing based on religious beliefs and practices; in which case, a refusal form must be signed by the parents and documented in the newborn's medical record.

The objective of this program is that every newborn in the Emirate of Abu Dhabi be screened for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth. Many of these disorders are metabolic in nature, which means they interfere with the body's ability to use nutrients to produce energy and maintain healthy tissue. Other types of disorders that may be detected through newborn screening include problems with hormones or blood disorders. These metabolic and other inherited disorders can interfere with an infant's normal physical and mental development in a variety of ways. In some instances they can even lead to death.

The newborn screening program in the Emirate of Abu Dhabi comprises of; a comprehensive newborn physical examination, Hearing Screening, Education, Screening Tests, Follow up, Diagnosis, Counseling, Treatment and Management, and Evaluation, Quality Assurance and Information System. It screens eleven (11) conditions, as listed below. These conditions were selected on the basis of assessment of public health needs, potential benefit and cost-effectiveness of early detection and management of newborns diagnosed with these condition(s) and prevention of hazardous complications and improving health outcomes.

Although additional disorders may be added as determined by the Health Authority of Abu Dhabi Public Health under the advisement of the Newborn Screening Advisory group, the newborn screening panel in the Emirate of Abu Dhabi currently includes the following disorders:

Metabolic Disorders	1. Phenylketoneuria (PKU) 2. Galactosemia (GALT) 3. Biotinidase (BIO)
Blood Disorders	4. Sickle Cell Disease (Hb S/S) 5. Sickle Cell Disease (Hb S/C) 6. Thalassemia (Hb S/A) 7. Glucose 6-phosphate dehydrogenase Deficiency (G6PD)
Endocrine Disorders	8. Congenital Hypothyroidism (CH) 9. Congenital Adrenal Hyperplasia (CAH)
Respiratory Disorders	10. Cystic Fibrosis (CF)
Genetic Disorders	11. Hearing Loss

3. Components of the Newborn Screening Program in the Emirate of Abu Dhabi 2009.

The developed Newborn screening program in the Emirate of Abu Dhabi 2009 is composed of fourteen (14) components:

1. Education on Newborn Screening Program in the Emirate of Abu Dhabi.
2. Newborn Identification System of Newborn Screening Program in the Emirate of Abu Dhabi.
3. Newborn Comprehensive Physical Examination of Newborn Screening Program in the Emirate of Abu Dhabi.
4. Early Detection of Hearing Loss of Newborn Screening Program in the Emirate of Abu Dhabi.
5. Informed Choices, Consent, Refusal of Newborn Screening Program in the Emirate of Abu Dhabi.
6. Newborn Screening Specimen Collection of Newborn Screening Program in the Emirate of Abu Dhabi.
7. Dispatch Transport of Newborn Screening Specimen Card of Newborn Screening Program in the Emirate of Abu Dhabi.
8. Newborn Specimen Screening, Reporting of Newborn Screening Program in the Emirate of Abu Dhabi.
9. Referral Follow Up of Newborn Screening Program in the Emirate of Abu Dhabi.
10. Diagnosis, Counseling of Newborn Screening Program in the Emirate of Abu Dhabi.
11. Treatment, Management of Newborn Screening Program in the Emirate of Abu Dhabi.
12. Evaluation of Newborn Screening Program in the Emirate of Abu Dhabi.
13. Quality Assurance of Newborn Screening Program in the Emirate of Abu Dhabi.
14. Information System of Newborn Screening Program in the Emirate of Abu Dhabi.

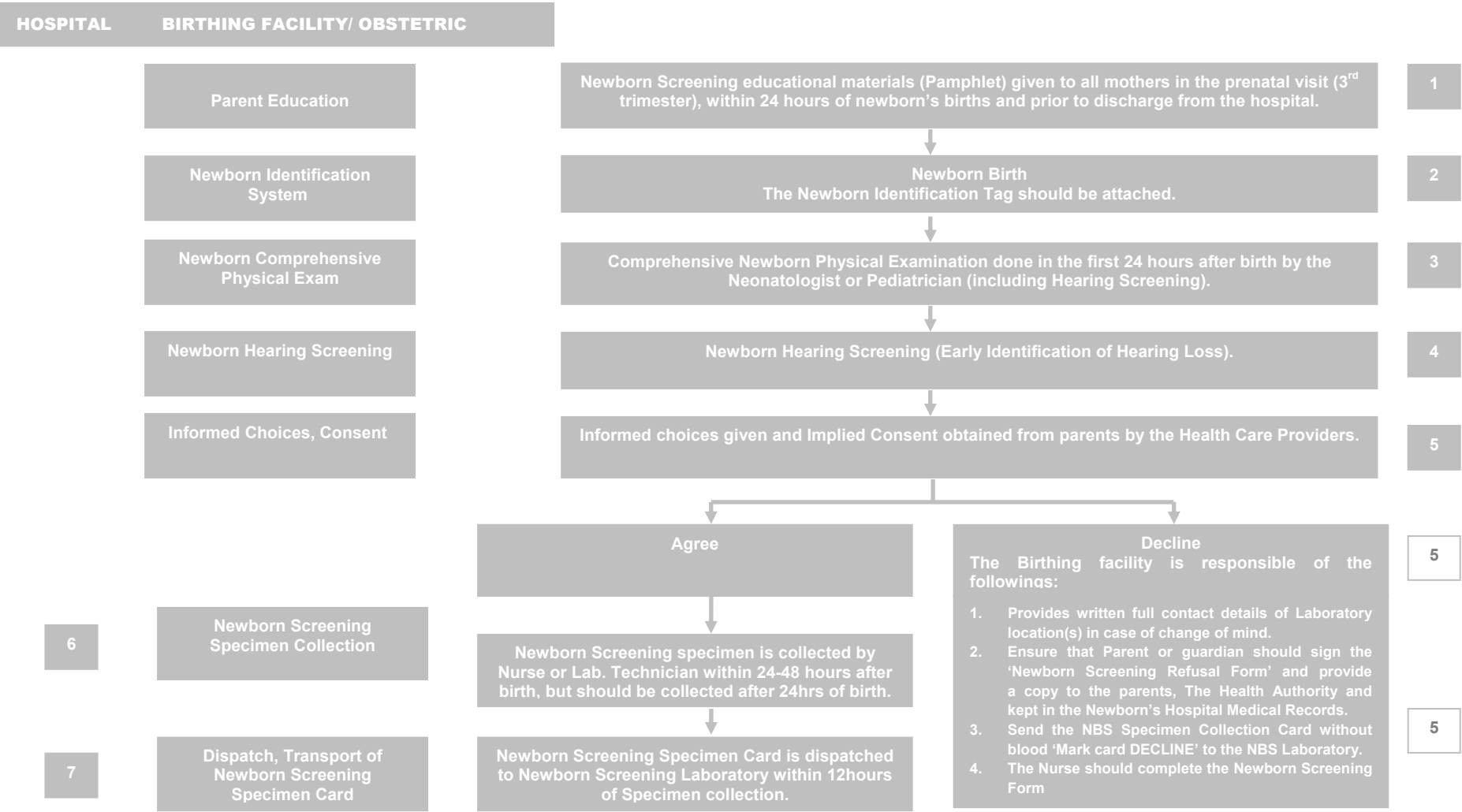
4. Procedures and Process of the Newborn Screening Program in the Emirate of Abu Dhabi 2009.

1. Parent Education	All Prospective parents should receive information on Newborn Screening during the prenatal period (in the 3rd trimester), and postnatal (within 24 hours of newborn's births). Parent knowledge should be reinforced after delivery by educational materials and discussion as needed by the newborns in hospital Parent information providers (health care professionals in the prenatal health care and labor and delivery services (Obstetricians, Registered Nurses). Neonatologists and Neonatal Registered Nurses in case of newborns in the NICU/SCBU. Family Physicians and Registered Nurses are the Parent information providers on newborn Screening in the Primary Health Care Settings.
2. Newborn Identification System	The attending staff at the Birthing facility, which is part of the Newborn Screening Center should attach the Newborn Identification Tag to one wrist and one ankle of newborn at the time of the delivery.
3. Newborn Comprehensive Physical Examination	All Newborns should have Comprehensive Physical Examination including newborn screening for Hearing Loss within 24 hrs after birth conducted by a Neonatologist or a Pediatrician.
4. Early Detection of Hearing Loss	All Newborns should have Hearing Screening as a part of Newborn Comprehensive Physical Examination (done within 24 hrs after birth). The initial Newborn hearing screening is the first hearing screening preferably done 12 hours or more after birth performed by trained Health Care Personnel.
5. Parental Informed Choices, Consent and Refusal	All Prospective parents should receive information about newborn screening. Explicit, written parental consent is not required for the newborn screening. Implied consent is the accepted practice. An exemption from the Newborn screening because of any reason is allowed. Refusal of Newborn Screening Tests Form should be Signed by documented in writing and kept in the Newborn's Hospital Medical Records and copies should be provided to parents and the Department of Public Health, Health Authority of Abu Dhabi.
6. Newborn Screening Specimen Collection	All Healthy Newborns should have Newborn Screening Blood Specimen Collection (Heel Prick Test) within 24-48 hours after birth. The Initial First Newborn Screening is the first newborn screening performed at the birthing facility by trained Health Care Personnel either a Registered Nurse or Lab Technician.

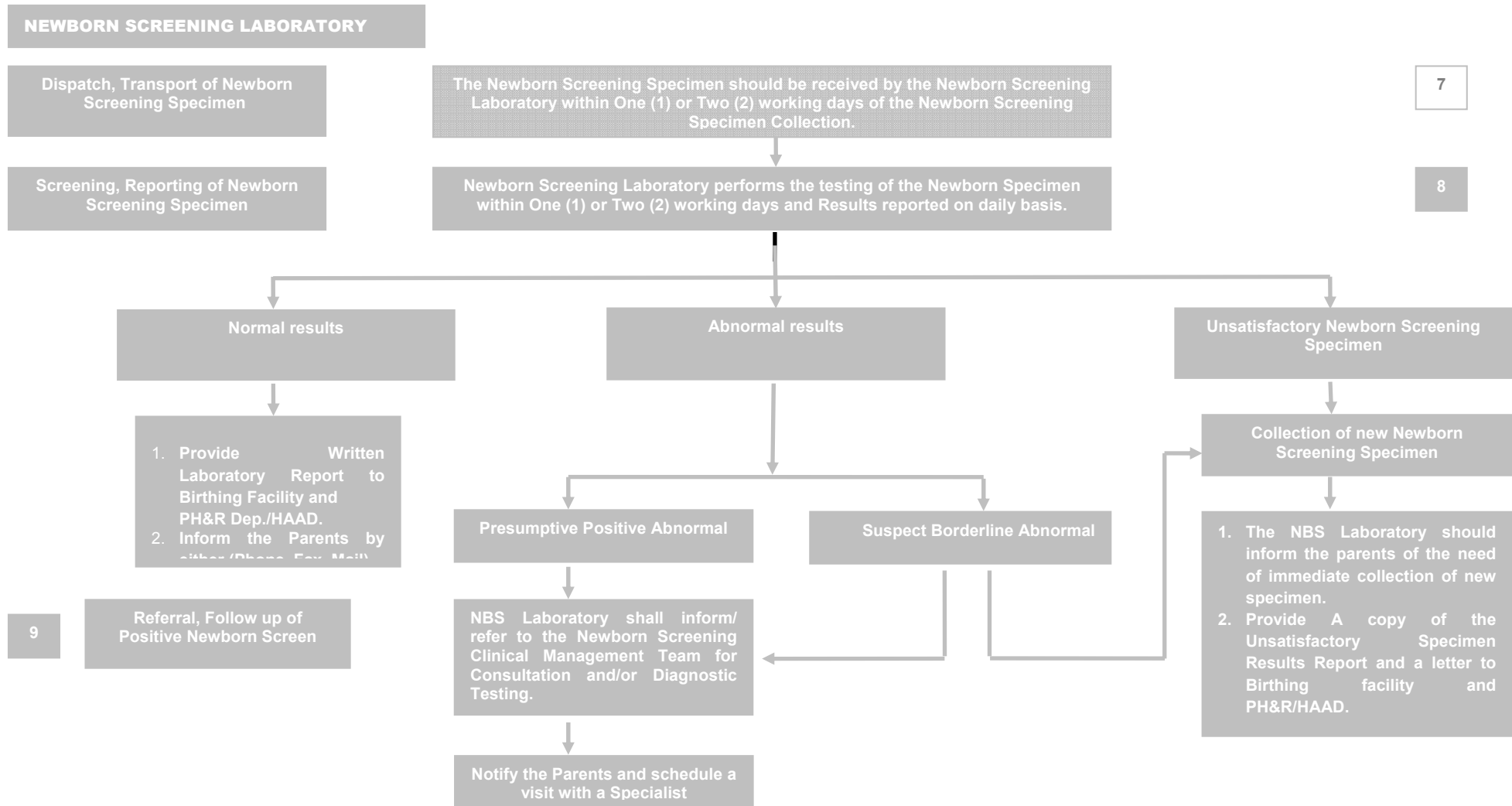
4. Procedures and Process of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

7. Dispatch, Transport of Newborn Screening Specimen Card	All Birthing facility in the Emirate of Abu Dhabi that is part of Newborn Screening Center should ensure that all Newborn Screening Specimen Card is Dispatched to the Newborn Screening Laboratory within 12 hours of Newborn screening specimen collection.
8. Newborn Specimen Screening, Reporting	The Newborn Screening Laboratory undertake the (Initial First) Screening of Newborn Specimen within One (1) to Two (2) working days of the Newborn Specimen receipt by Newborn Screening Laboratory and report the Newborn Screening results on a daily basis.
9. Referral and Follow Up	The Newborn Screening Laboratory should set up a referral system of Newborn's with Presumptive Positive Abnormal Screening Results to the Newborn Screening Clinical Management Team for Consultation and/or Diagnostic Testing.
10. Diagnosis and Counseling	Every newborn with Presumptive Positive Abnormal screening results receives proper diagnostic and counseling services.
11. Treatment and Management	Every newborn should receive high quality standard of Treatment and Management Services.
12. Evaluation	The Newborn Screening Management Team shall develop an Evaluation Plan that defines the selected outcome data, assigns responsibility for their monitoring, and outlines the periodicity with which evaluation are to occur. The Evaluation of the Newborn Screening Program in the Emirate of Abu Dhabi should encompass both short-term and long-term activities.
13. Quality Assurance	
14. Information System	

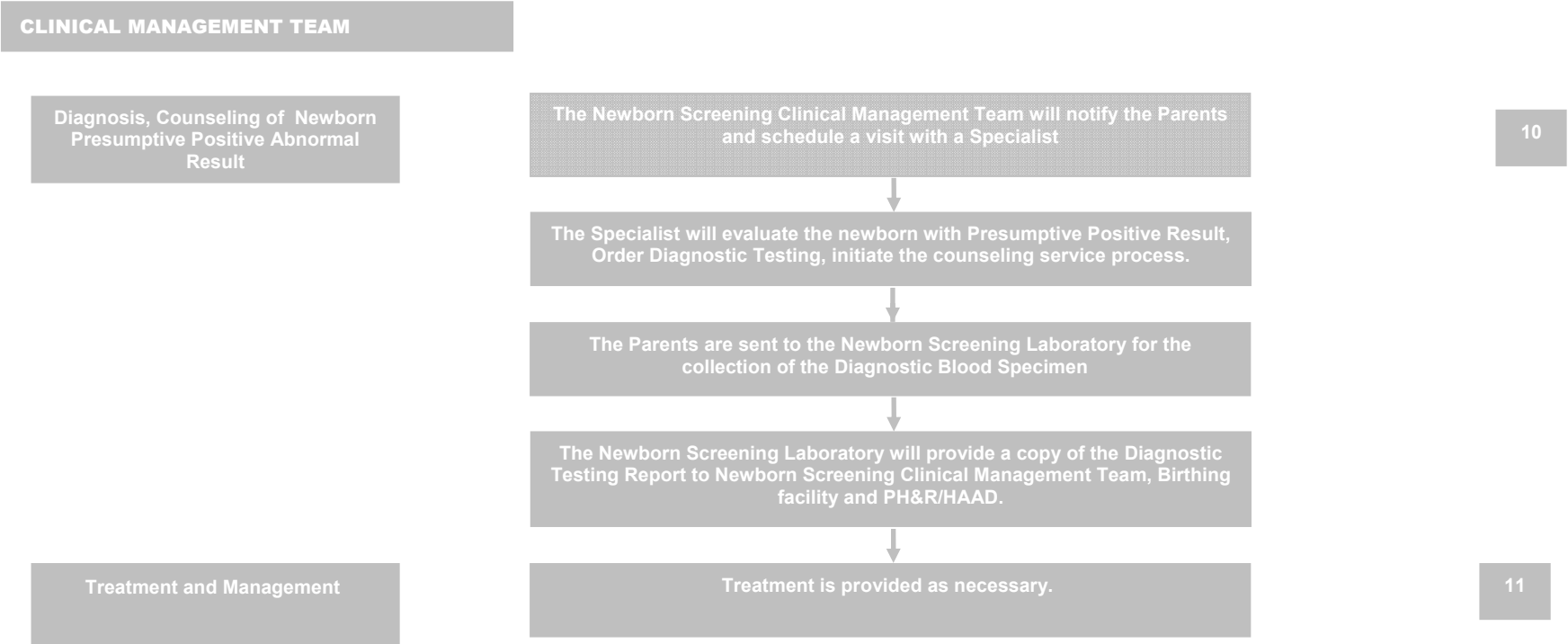
5. Pathway of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 .



5. Pathway of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).



5. Pathway of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).



6. Newborn Comprehensive Physical Examination.

All Newborns should have Comprehensive Physical Examination within the 24 hours after birth conducted by a Neonatologist or a Pediatrician as a part of the Routine Newborn Screening. This is different from the initial examination carried out at the time of birth by one of the health professionals attending the birth. The Newborn Comprehensive Physical Examination should also include newborn screening for Hearing loss. The Neonatologist or the Pediatrician responsible of conducting the Newborn Comprehensive Physical Examination should assess risk factors in newborn these are;

1. Birth before 37 wk or after 42wk gestation.
2. Birth weight <2500 or >400gm.
3. Deviations in expected size for stage of development.
4. History of fetal neonatal sibling death or serious illness.
5. Poor condition at delivery (Apgar 0-4at 1min) or resuscitation required at delivery or subsequently.
6. History of maternal infection or other illness during pregnancy ,premature raptures of membranes, serve social problems (e.g. Teenage pregnancy, drug addiction),absent or late prenatal care ,abnormal gestational weight gain, prolonged infertility, four or more previous pregnancies ,35yrsor more maternal age (especially if primiparous),or ingestion of drugs.
7. Multiple pregnancy or gestation commencing within 6mo of a previous pregnancy.
8. Delivery by cesarean section or any unusual obstetrical complications, including hydramnios, abruption placentae, placenta previa, or abnormal presentation.
9. Significant malformation or suspicion of malformation.
10. Anemia or blood group incompatibility.
11. Severe maternal emotional problems, such as hyperemesis gravidarum.
12. Serious accidents or general anesthesia drug pregnancy.

The Comprehensive physical examinations of newborn should take place in the context of assessments which include;

1. Detailed Health History
 - Review of all information obtained during the prenatal visit including; risk factors, problems arising or suspected from prenatal screening, prenatal diagnostic procedures.
 - Significant intrapartum or postpartum complications.
 - Gather extensive medical and family hist
 - ory and address parental concerns.
 - Assessment of the newborn risk factors.
2. Feeding
 - Assessment of the Newborn feeding and nutrition as recommended by the AAP statement "Breastfeeding and the Use of Human Milk"(2005) [URL: <http://aappolicy.aappublications.org/cgi/content/full/pediatrics;115/2/496>].
3. Drugs and Immunization
 - Check appropriate administration of Vitamin K, and Immunization as per the Extended Program on Immunization approved by UAE National Vaccination Committee.
4. Parental clinical Evaluation
 - Maternal physical and emotional assessment to Identify issues such as; depression, domestic violence, substance abuse, learning difficulties or mental health problems.
5. Explain problems such as jaundice that might not be observable in the newborn but could be significant a few days or weeks later.
6. Convey information about services and inform families how they can request and negotiate additional help, advice, and support as needed.
7. Review the Newborn's scores of Dubowitz/Ballard Exam for Assessment of the Gestational Age and Apgar Score.
8. Assessment of the newborn risk factors.

6. Newborn Comprehensive Physical Examination (cont.).

General Observation

- level of consciousness.
- General Appearance (Resting Posture, Tone, Spontaneous Activity, Respiratory efforts).
- Skin (color, texture, nails, presence of rashes or birthmarks).
- Inspect facies at rest.

Measure the newborn's Vital Signs;

- Body Temperature
- Blood Pressure
- Pulse
- Check Respiratory Rate

Extensive anthropomorphic measurements should be measured , these are;

- The following anthropomorphic measurements should be measured, recorded and plotted (Occipital frontal circumference, Height, Weight).
- Measurement of inner canthal and outer canthal distances with calculations of interpupillary distances.
- Measurements of ear size and ear placement on the head.
- Measurement of philtrum length.
- Measurement of internipple distance.
- Measurements of finger and palm lengths.
- Abdominal Circumference.

The Head and Neck Region

- Head
 - Appearance, shape, presence of molding.
 - Transilluminate head.
 - Fontanelles.
- Eyes
 - Determine pupil response to light.
 - Check for Optical Opacities.
 - Funduscopic examination.
- Separate protocol is recommended for Screening of Retinopathy in premature, as recommended by the AAP statement "Screening Examination of Premature Infants For Retinopathy of Prematurity"(2006).
- Nose.
- Mouth- palate, tongue, throat.
- Ears, including
 - Assessment of tympanic membranes. (See Appendix 4: Standards and Guidelines of Early Identification of the Hearing Loss).
- Neck
 - Assessment of thyroid gland size.
 - Clavicles.

6. Newborn Comprehensive Physical Examination (cont.).

The Thorax

- Inspect shape to detect any a symmetry and integrity of skin. Palpate bony structures.
- Examine both breasts for to detect any a symmetry in shape, enlargements, discharge per nipples.

The Respiratory System

- Auscult breath sounds.

Cardiovascular System

- Auscult Heart sounds, Rate, Rhythm.
- Check murmurs.
- Check femoral pulse and peripheral pulses.

The abdomen

- Assessment of organ size.
- Assessment for abnormal masses.
- Check the condition of the Umbilical Cord.

The Genitalia and Anus

- Check genitalia for form.
- Measurement of size of genital components.
- Check anus for patency.
- Check for undescended testicles in males.

The Spine

- Inspect shape to detect any a symmetry and palpate bony structures.

The extremities(Limbs, Hands, feet and Digits)

- Assess proportion and symmetry.
- Specific measurement of any joint limitations.
- Check for foot deformities.

The Hips

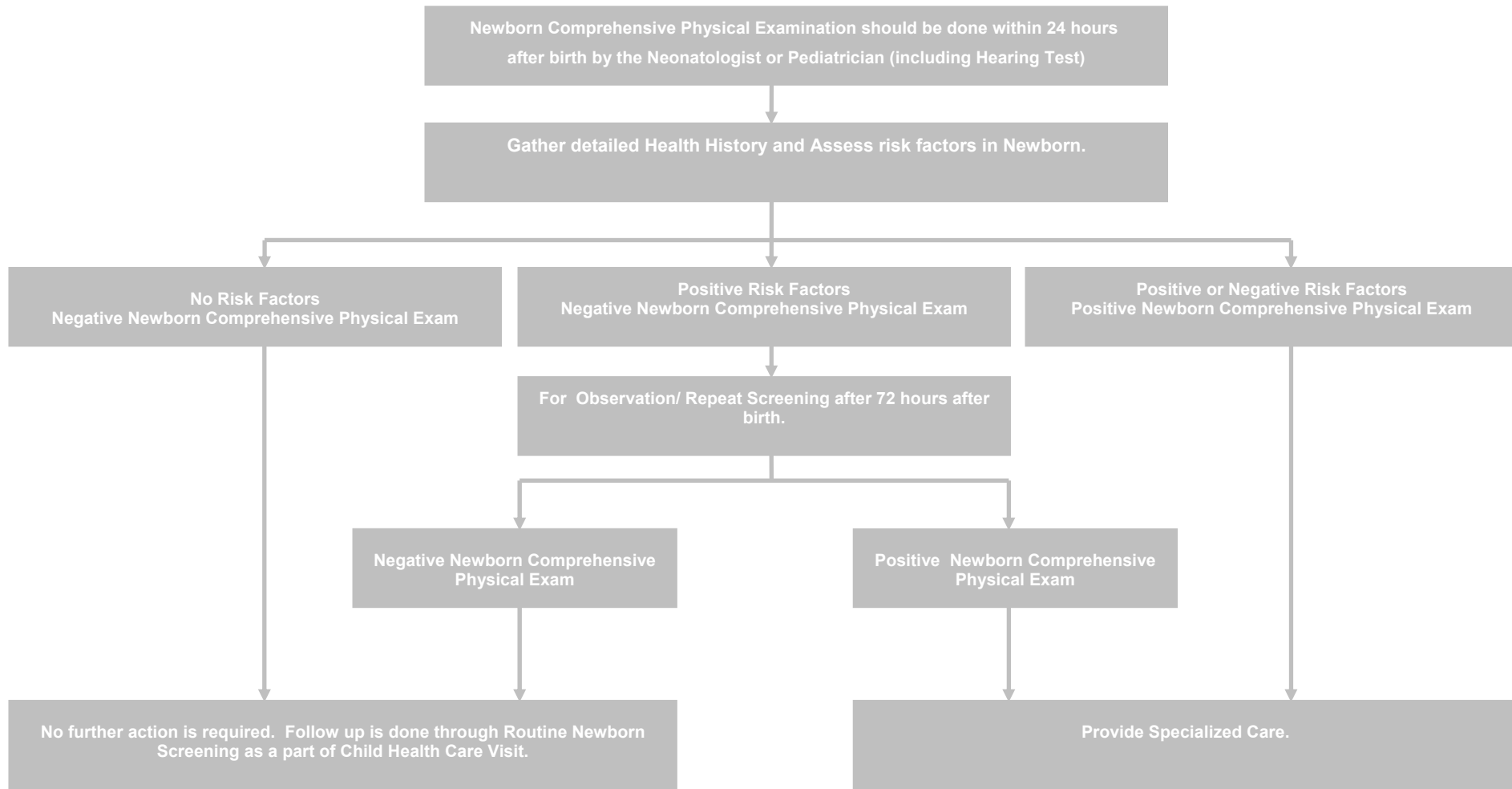
- Check a symmetry of the limbs and skin folds.
- Perform Barlow and Ortolani's maneuvers.

The skin for abnormalities, which often includes Woods light examination of fluorescent depigmented areas.

The Central Nervous

- Observe tone, Behavior, Movements and posture.
- Elicit Tendon reflexes.

Pathway of Newborn Comprehensive Physical Examination of Newborn Screening Program in the Emirate of Abu Dhabi.



7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009.

A. Metabolic Disorders

1. Phenylketonuria (PKU)

Phenylketonuria (fennel-key-ton-uria) commonly known as PKU) is an inherited disorder that increases the levels of a substance called phenylalanine in the blood. It is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. The gene that is related to phenylketonuria- is the mutations in the PAH gene cause phenylketonuria. The PAH gene provides instructions for making an enzyme called phenylalanine hydroxylase. This enzyme converts the amino acid phenylalanine ("phe" for short) in to into another amino acid, tyrosine. If gene mutations reduce the activity of phenylalanine hydroxylase, phenylalanine from the diet is not processed effectively. As a result, "phe" accumulates to a toxic level in the blood and other parts of the body. Because nerve cells in the brain are particularly sensitive to phenylalanine levels, excessive amounts of this substance prevent the brain from growing and developing normally and can cause brain damage.

Classic PKU, the most severe form of the disorder, occurs when phenylalanine hydroxylase activity is severely reduced or absent. People with untreated classic PKU have levels of phenylalanine high enough to cause severe brain damage and other serious medical problems. Mutations in the PAH gene that allow the enzyme to retain some activity result in milder versions of this condition, such as variant PKU or non-PKU hyperphenylalaninemia.

Changes in other genes may influence the severity of PKU, but little is known about these additional genetic factors.

Phenylalanine is a building block of proteins (an amino acid) that is obtained through the diet. It is found in all proteins and in some artificial sweeteners. If PKU is not treated, phenylalanine can build up to harmful levels in the body, causing intellectual disability and other serious health problems.

The signs and symptoms of PKU vary from mild to severe. The most severe form of this disorder is known as classic PKU. Infants with classic PKU appear normal until they are a few months old. Without treatment with a special low-phenylalanine diet, these children develop permanent intellectual disability. Seizures, delayed development, behavioral problems, and psychiatric disorders are also common. Untreated individuals may have a musty or mouse-like odor as a side effect of excess phenylalanine in the body. Children with classic PKU tend to have lighter skin and hair than unaffected family members and are also likely to have skin disorders such as eczema. Less severe forms of this condition, sometimes called variant PKU and non-PKU hyperphenylalaninemia, have a smaller risk of brain damage. People with very mild cases may not require treatment with a low-phenylalanine diet.

Babies born to mothers with PKU and uncontrolled phenylalanine levels (women who no longer follow a low-phenylalanine diet) have a significant risk of intellectual disability because they are exposed to very high levels of phenylalanine before birth. These infants may also have a low birth weight and grow more slowly than other children. Other characteristic medical problems include heart defects or other heart problems, an abnormally small head size (microcephaly), and behavioral problems. Women with PKU and uncontrolled phenylalanine levels also have an increased risk of pregnancy loss.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

Screening Test of Phenylketoneuria (PKU)

PKU can be diagnosed by newborn screening in virtually 100% of cases based upon detection of the presence of hyperphenylalaninemia using the Guthrie microbial or other assays on a blood spot obtained from a heel prick. PKU is diagnosed in individuals with plasma phenylalanine (Phe) concentrations higher than 1000 $\mu\text{mol/L}$ in the untreated state; non-PKU HPA is diagnosed in individuals with plasma Phe concentrations consistently above normal (i.e., $>120 \mu\text{mol/L}$), but lower than 1000 $\mu\text{mol/L}$ when on a normal diet.

Treatment of Phenylketoneuria (PKU)

At the present time, a diet low in "phe" is the only treatment for PKU. If the diet is started early enough and closely followed, the child's development will be normal in almost all cases. A low-phenylalanine diet must be started within the first days of life for all newborns whose blood phenylalanine levels are above 10 mg/dL (600 $\mu\text{mol/L}$). Some "phe" is essential for growth. Too much "phe" is harmful. The diet must be carefully planned to allow enough "phe" for the child to grow normally, yet not enough to produce the harmful effects of excessive "phe". This balance between too much and too little "phe" is different for each child. A child's needs depend on the severity of the enzyme deficiency and the child's age, growth rate, and current state of health. The right amount of "phe" for the child is determined through blood tests that measure the amount of "phe" in the child's blood. The dietary control must keep the phenylalanine levels between 2 and 5 mg/dL (120 and 300 $\mu\text{mol/L}$) until 10 y of age. Thereafter, a progressive and controlled relaxation of the diet is allowed, keeping levels below 15 mg/dL until the end of adolescence and below 20 mg/dL (1200 $\mu\text{mol/L}$) in adulthood. A lifelong follow-up is recommended for PKU women to prevent for maternal PKU.

The diet prescription is adjusted accordingly by your physician and nutritionist. Several special formula-milk substitute products are available. These special formulas make it possible to plan a diet that is low in "phe" but adequate in protein, calories, and essential vitamins and minerals. In addition to special formulas, the child with PKU can have foods that are low in protein and "phe" in measured amounts. This includes most fruits and vegetables, some cereals and candies and special breads, cookies and pastas. All high-protein foods such as milk, meat, eggs and cheese, which contain large amounts of "phe", must be completely eliminated from the diet. This diet record, with the exact kinds and amounts of food eaten, will need to be kept just before each monitoring blood test. The nutritionist will use this record to decide what changes, if any, need to be made in the diet prescription.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

2. Galactosemia (GALT)

Galactosemia (ga-lac-to-se-me-a) is a rare hereditary condition caused by the body's inability to breakdown galactose (a sugar found in milk and milk products). Breast milk and most infant formula contain a sugar called lactose. When lactose is ingested the body breaks it down (digestion) into sugars called glucose and galactose. Before the body can use galactose it must be broke down further with the help of an enzyme. This enzyme (galactose-1-phosphate uridyl transferase) is a chemical that changes galactose into a form the body can use for energy. Ninety five percent of people with galactosemia are missing this enzyme and without it galactose builds up in the body. The high levels of galactose poison the body causing serious damage like a swollen and inflamed liver, kidney failure, stunted physical and mental growth, and cataracts in the eyes. Placing the child on a special diet within the first few days of life may prevent this damage.

Galactosemia is inherited when both parents pass a galactosemia gene to their child. The father and mother are carriers of the disorder; carriers of galactosemia will not get sick. But when two carriers have a child together there is a 1 in 4 (25%) that the child will have galactosemia, a 2 in 4 chance (50%) that the child will be a carrier of the disease, and a 1 in 4 chance (25%) that the child will not be a carrier nor have the disease. These are the chances with each birth. Researchers have identified several types of galactosemia. These conditions are each caused by mutations in a particular gene, and affect different enzymes involved in breaking down galactose. Classic galactosemia, also known as type I, is the most common and most severe form of the condition. Galactosemia type II (also called galactokinase deficiency) and type III (also called galactose epimerase deficiency) cause different patterns of signs and symptoms.

If infants with classic galactosemia are not treated promptly with a low-galactose diet, life-threatening complications appear within a few days after birth. Affected infants typically develop feeding difficulties, a lack of energy (lethargy), a failure to gain weight and grow as expected (failure to thrive), yellowing of the skin (jaundice), liver damage, and bleeding. Other serious complications of this condition can include overwhelming bacterial infections (sepsis) and shock. Affected children are also at increased risk of delayed development, clouding of the lens of the eye (cataract), speech difficulties, and intellectual disability. Females with classic galactosemia may experience reproductive problems caused by ovarian failure.

Galactosemia type II causes fewer medical problems than the classic type. Affected infants develop cataracts, but otherwise experience few long-term complications. The signs and symptoms of galactosemia type III vary from mild to severe and can include cataracts, delayed growth and development, intellectual disability, liver disease, and kidney problems. (Genetic Home Reference; A service of the US National Library of Medicine ® available online: <http://ghr.nlm.nih.gov/condition=galactosemia>)

Screening Test of Galactosemia (GALT)

Levels of galactose and galactose-1-phosphate in the blood are determined by enzyme assay. The enzyme galactose-1-phosphate uridyl transferase is measured on the same dried blood spot if the metabolite levels are elevated. Rarely, other forms of galactosaemia, mainly galactokinase deficiency, are diagnosed by this test.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

Treatment of Galactosemia (GALT)

The treatment for galactosemia is to restrict galactose and lactose from the diet for life. Since galactose is a part of lactose, all milk and all foods that have milk in them must not be eaten. This is not just cow's milk, but any animal's milk including goat's milk and human breast milk. This includes dairy products like butter, cheese, and yogurt. Other foods that have small amounts of milk products in other forms such as whey, casein, and curds must also be eliminated.

Children with galactosemia should be followed by pediatric metabolic specialists and nutritionists. These health professionals provide the child with appropriate medical care and educate the family concerning special diet requirements. Families are taught to read labels carefully when shopping for food for the child with galactosemia. Many prepared foods have hidden ingredients containing galactose. Families learn to question physicians and pharmacists about prescribed medicines for their child since many medicines contain fillers that include galactose.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

3. Biotinidase (BIO)

Biotinidase deficiency is an inherited condition that affects the way a person's body uses the vitamin, biotin. Biotin is an important vitamin that helps enzymes called carboxylases make certain fats and carbohydrates and break down proteins. Biotin is essential for proper growth and development. A person with biotinidase deficiency cannot use the bound biotin found in food. This means that the biotin is not available for use. Low levels of biotin may cause seizures, developmental delay, hearing loss and other serious and sometime life threatening illness.

Biotinidase is a genetic condition caused by changes in the BTB (Biotinidase) gene. The BTB gene is responsible for making the enzyme called biotinidase. Biotinidase frees the bound biotin in protein. This free biotin can then help carboxylases make fats, carbohydrates, and break down protein. When there is an alteration in the BTB gene, biotinidase levels go down and the free biotin is too low. Biotinidase deficiency is inherited in an autosomal recessive pattern, which means two copies of the BTB gene must be changed for a person to be affected with biotinidase. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "carriers", with one copy of the changed BTB gene and one copy of the normal BTB gene. When both parents are carriers, there is a one-in-four (or 25 percent) chance that both will pass a changed BTB gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25 percent) chance that they will each pass on a normal BTB gene, and the child will be free of the condition. There is a two-in-four (or 50 percent) chance that a child will inherit a changed BTB gene from one parent and a normal BTB gene from the other, making it a carrier like its parents. These chances are the same in each pregnancy for these parents.

Infants with biotinidase deficiency appear normal at birth, but develop serious symptoms after the first few weeks or months of life, but the age of onset varies. Children with profound biotinidase deficiency, the more severe form of the condition, symptoms include low muscle tone, seizures, developmental delay, loss or absence of hair, hearing loss and optic nerve atrophy. These symptoms can become serious enough to lead to coma and death. If left untreated, the disorder can lead to hearing loss, eye abnormalities and loss of vision, problems with movement and balance (ataxia), skin rashes, hair loss (alopecia), and a fungal infection called candidiasis. Immediate treatment and lifelong management with biotin supplements can prevent many of these complications. Partial biotinidase deficiency is a milder form of this condition. Affected children experience hypotonia, skin rashes, and hair loss, but these problems may appear only during illness, infection, or other times of stress.

Screening Test of Biotinidase (BIO)

Individuals with profound Biotinidase deficiency have less than 10% of mean normal serum Biotinidase enzyme activity. Individuals with partial Biotinidase deficiency have 10%-30% of mean normal serum Biotinidase enzyme activity. Both profound and partial Biotinidase deficiency is usually identified by newborn screening using colorimetric assay.

Treatment of Biotinidase (BIO)

Biotinidase deficiency is treated with free biotin, or biotin that is not bound to protein or other molecules. In patients diagnosed through screening, treatment will clear the skin rash and alopecia and improve the neurological status. It is necessary that treatment be managed by the doctor to be sure that the biotin is in the free form and in sufficient amounts. However, hearing problems may occur in spite of treatment. Treatment should begin as soon as possible following a diagnosis and will continue throughout an individual's life. Children and adults with biotinidase deficiency require follow-up care at a medical center or clinic that specialize in this condition. In addition, regular blood tests are used to monitor your child's health.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

B. Blood Disorders

1. **Sickle Cell Disease (Hb S/S)**
2. **Sickle Cell Disease (Hb S/C)**
3. **Thalassemia (Hb S/A)**
4. **Glucose 6 Phosphate Deficiency (G6PD)**

Sickling Haemoglobinopathies are inherited disorders that result in production of an abnormal form of hemoglobin S, C or E, or a decreased synthesis of a beta globin chain. Sickle cell disease is a term for hemoglobin disorders characterized by the predominate production of hemoglobin S, which can distort red blood cells into a sickle, or crescent, shape. Any sign of illness in an infant with sickling disease is a potential medical emergency.

Mutations in the HBB gene cause sickle cell disease. Hemoglobin consists of four protein subunits, typically, two subunits called alpha hemoglobin and two subunits called beta hemoglobin. The HBB gene provides instructions for making beta hemoglobin. Various versions of beta hemoglobin result from different mutations in the HBB gene. One particular HBB mutation produces an abnormal version of beta hemoglobin known as hemoglobin S (HbS). Other mutations in the HBB gene lead to additional abnormal versions of beta hemoglobin such as hemoglobin C (HbC) and hemoglobin E (HbE). HBB mutations can also result in an unusually low level of beta-hemoglobin; this abnormality is called beta thalassemia.

In people with sickle cell disease, at least one of the beta hemoglobin subunits in hemoglobin is replaced with hemoglobin S. In sickle cell anemia, which is a common form of sickle cell disease, hemoglobin S replaces both beta hemoglobin subunits in hemoglobin. In other types of sickle cell disease, just one beta hemoglobin subunit in hemoglobin is replaced with hemoglobin S. The other beta hemoglobin subunit is replaced with a different abnormal variant, such as hemoglobin C. For example, people with sickle-hemoglobin C (HbSC) disease have hemoglobin molecules with hemoglobin S and hemoglobin C instead of beta hemoglobin. If mutations that produce hemoglobin S and beta thalassemia occur together, individuals have hemoglobin S-beta thalassemia (HbSBetaThal) disease.

Abnormal versions of beta hemoglobin can distort red blood cells into a sickle shape. The sickle-shaped red blood cells die prematurely, which can lead to anemia. Sometimes the inflexible, sickle-shaped cells get stuck in small blood vessels and can cause serious medical complications.

Acute and chronic tissue injury can occur when sickled cells cause vascular occlusion. Sickling diseases can cause severe pain anywhere in the body, but most often in the hands, arms, chest, legs and feet. Complications may include, but are not limited to, the following:

- **Sepsis:** The first sign of infection may be a fever of 101° F or greater. These children require immediate medical attention. Children with sickle cell diseases are very susceptible to pneumococcal infections.
- **Acute chest syndrome:** A serious condition caused by infection and/or trapped sickled red blood cells in the lungs. Symptoms may include dyspnea, coughing and chest pain.
- **Hand-and-foot syndrome:** This painful swelling of the hands and feet is due to severe vascular occlusion.
- **Splenic sequestration crisis:** Early signs include pallor, enlarged spleen and pain in the abdomen due to accumulation of sickled cells within the spleen. This complication can result in circulatory collapse and shock, with sudden death, if not recognized and treated immediately.
- **Aplastic crisis:** The bone marrow temporarily stops producing red blood cells resulting in severe anemia. The child may appear pale, tired, and less active than usual.
- **Stroke:** Cerebral vascular occlusion due to sickled cells can affect even very young children. Any loss of consciousness or weakness in an extremity should be evaluated promptly.
- **Painful episodes :** The pain of sickling disorders is acute and can be quite severe; even very young children may require prescription medications for pain relief.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

Screening Test of Blood Disorders

Newborn screening for sickle cell disease is performed by high performance liquid chromatography (HPLC) testing to determine the presence of abnormal hemoglobins (Hgb) in whole blood. Unaffected infants will have mostly fetal hemoglobin (Hgb F) and some adult hemoglobin (Hgb A). HPLC has been shown effective in detecting Haemoglobinopathies characterized by synthesis of an abnormal hemoglobin molecule immediately after birth. A baby testing positive for a form of sickle cell disease will have Hgb F with Hgb S and possibly, another abnormal hemoglobin such as Hgb C, Hgb E or beta thalassemia. All abnormal newborn screening test results indicating a sickle cell disorder require appropriate confirmatory blood tests, sometimes including testing of parents and siblings for actual diagnosis. Referral to a pediatric hematologist for evaluation and diagnostic testing is recommended within the first month of life and should not be delayed until the infant is older. If newborn screening results indicate less serious hemoglobin disorders or traits, referral to a pediatric hematologist for parental education and counseling is recommended. Even small transfusions may cause false negative screening test results and any results indicating that the baby was transfused require repeat testing 90 days after the last transfusion.

There are several recommended testing methods for diagnosis of sickling disorders and other hemoglobinopathies: Hemoglobin electrophoresis including both cellulose acetate and citrate agars (one is not sufficient), isoelectric focusing and high performance liquid chromatography are considered proven, reliable and accurate methods for defining an infant's hemoglobin phenotype. All siblings of infants diagnosed with a sickle cell disease should be tested; genetic counseling services should be offered to parents.

Treatment of Blood Disorders

The National Institutes of Health clinical guidelines for management of sickle cell diseases state, "Penicillin prophylaxis should begin by 2 months of age for infants with suspected sickle cell anemia, whether or not the definitive diagnosis has been established." Antibiotic therapy should continue until at least 5 years of age. Normal dosage for an infant is 125 mg of penicillin twice a day until 3 years of age, when dosage is increased to 250 mg twice a day. An alternative antibiotic is available for children who are allergic to penicillin therapy. Prescription pain medication also may be indicated during sickling crises. Health care monitoring and maintenance with appropriate immunizations is imperative to the health of the baby, and pneumococcal conjugate vaccine immunizations also are recommended, beginning at 2 months of age.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

4. Glucose 6-phosphate dehydrogenase Deficiency (G6PD)

Glucose-6-phosphate dehydrogenase deficiency is a genetic disorder that occurs most often in males. This condition mainly affects red blood cells, which carry oxygen from the lungs to tissues throughout the body. In affected individuals, a defect in an enzyme called glucose-6-phosphate dehydrogenase causes red blood cells to break down prematurely. This destruction of red blood cells is called hemolysis.

The most common medical problem associated with glucose-6-phosphate dehydrogenase deficiency is hemolytic anemia, which occurs when red blood cells are destroyed faster than the body can replace them. This type of anemia leads to paleness, yellowing of the skin and whites of the eyes (jaundice), dark urine, fatigue, shortness of breath, and a rapid heart rate. In people with glucose-6-dehydrogenase deficiency, hemolytic anemia is most often triggered by bacterial or viral infections or by certain drugs (such as some antibiotics and medications used to treat malaria). Hemolytic anemia can also occur after eating fava beans or inhaling pollen from fava plants (a reaction called favism).

Glucose-6-dehydrogenase deficiency is also a significant cause of mild to severe jaundice in newborns. Many people with this disorder, however, never experience any signs or symptoms.

Mutations in the G6PD gene cause glucose-6-phosphate dehydrogenase deficiency. The G6PD gene provides instructions for making an enzyme called glucose-6-phosphate dehydrogenase. Glucose-6-Phosphate Dehydrogenase (G6PD) functions throughout the body, but its deficiency is seen predominantly in its effects on the red blood cells.

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A striking characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

This enzyme is involved in the normal processing of carbohydrates. It also protects red blood cells from the effects of potentially harmful molecules called reactive oxygen species. Reactive oxygen species are byproducts of normal cellular functions. Chemical reactions involving glucose-6-phosphate dehydrogenase produce compounds that prevent reactive oxygen species from building up to toxic levels within red blood cells. If mutations in the G6PD gene reduce the amount of glucose-6-phosphate dehydrogenase or alter its structure, this enzyme can no longer play its protective role. As a result, reactive oxygen species can accumulate and damage red blood cells. Factors such as infections, certain drugs, or ingesting fava beans can increase the levels of reactive oxygen species, causing red blood cells to be destroyed faster than the body can replace them. A reduction in the amount of red blood cells causes the signs and symptoms of hemolytic anemia. Researchers believe that carriers of a G6PD mutation may be partially protected against malaria, an infectious disease carried by a certain type of mosquito. A reduction in the amount of functional glucose-6-dehydrogenase appears to make it more difficult for this parasite to invade red blood cells. Glucose-6-phosphate dehydrogenase deficiency occurs most frequently in areas of the world where malaria is common.

Glucose-6-Phosphate Dehydrogenase (G6PD) functions throughout the body, but its deficiency is seen predominantly in its effects on the red blood cells. G6PD anchors the production of NADPH and glutathione to protect the body from oxidative insults. Erythrocytes are especially sensitive to oxidative damage. G6PD deficiency can result in neonatal jaundice and in life threatening reactions to several medications, foods and infections. G6PD deficiency affects 400 million people around the world and is the most common genetic enzyme deficiency in man. Population and epidemiology information point to G6PD deficiency as providing some resistance to malaria.

Babies with G6PD deficiency appear normal at birth. They may experience neonatal jaundice and haemolysis that can be so serious as to cause neurologic damage or even death. Barring such severe complications in the newborn period, infants with G6PD deficiency generally experience normal growth and development. Exposure to certain antimalarial drugs and sulfonamides, infection stress (such as upper respiratory or GI infections), environmental agents (e.g. moth balls), and eating certain foods (e.g. fava beans), each of which impact the patient's ability to handle oxidative reactions, can cause acute hemolytic anemia.

7. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

Screening Test of Glucose 6-phosphate dehydrogenase Deficiency (G6PD)

Newborn screening for G6PD deficiency can be done by enzyme analysis or primary DNA screening. Confirmatory testing using a quantitative assay should be performed for diagnosis of G6PD deficiency.

Treatment of Glucose 6-phosphate dehydrogenase Deficiency (G6PD)

Infants with G6PD deficiency may be at increased risk for pathological newborn jaundice and may warrant close monitoring for associated complications during the newborn period. Otherwise, treatment of G6PD deficiency is avoidance. For the infant, this means avoidance of several medications routinely prescribed for infections and illness. Strict attention to the ingredients of prepared foods and restaurant meals is required as fava beans are a frequent addition to prepared foodstuffs. Patients should not be exposed to moth balls containing naphthalene. The adverse affects of infection on patients with G6PD Deficiency can be acute and life threatening. Over exertion from exercise and work leading to dehydration and hypoglycemia can precipitate clinical symptoms. As mentioned above, patients mindful of these limitations can lead a normal life of exercise. Because the diagnosis and therapy of this disorder is complex, the pediatrician is advised to manage the patient in close collaboration with a consulting pediatric hematology specialist. It is recommended that parents travel with a letter of treatment guidelines from the patient's physician.

6. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

C. Endocrine Disorders

1. Congenital Hypothyroidism (CH)

Congenital hypothyroidism is a condition that affects infants from birth (congenital) and results from a partial or complete loss of thyroid function (hypothyroidism). The thyroid gland is a butterfly-shaped tissue in the lower neck. It makes iodine-containing hormones that play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism).

Congenital hypothyroidism occurs when the thyroid gland fails to develop or function properly. In 80 to 85 percent of cases, the thyroid gland is absent, abnormally located, or severely reduced in size (hypoplastic). In the remaining cases, a normal-sized or enlarged thyroid gland is present, but production of thyroid hormones is decreased or absent.

Congenital hypothyroidism can lead to brain damage and developmental delay if not diagnosed early and treated throughout life. Females are affected twice as often as males. While percentages of specific etiologies vary from country to country. If untreated, congenital hypothyroidism can lead to intellectual disability and abnormal growth. In the United States and many other countries, all newborns are tested for congenital hypothyroidism. If treatment begins in the first month after birth, infants usually develop normally.

Mutations in the DUOX2, PAX8, SLC5A5, TG, TPO, TSHB, and TSHR genes cause congenital hypothyroidism. Gene mutations cause the loss of thyroid function in one of two ways. Mutations in the PAX8 gene and some mutations in the TSHR gene prevent or disrupt the normal development of the thyroid gland before birth. Mutations in the DUOX2, SLC5A5, TG, TPO, and TSHB genes prevent or reduce the production of thyroid hormones, even though the thyroid gland is present. Mutations in other genes that have not been well characterized may also cause congenital hypothyroidism. Most cases of congenital hypothyroidism are sporadic, which means they occur in people with no history of the disorder in their family. An estimated 15 to 20 percent of cases are inherited. Many inherited cases are autosomal recessive, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition. Some inherited cases (those with a mutation in the PAX8 gene or certain TSHR mutations) have an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Infants with CH are usually born at term or after term. Some may exhibit decreased activity, poor feeding, jaundice, hypotonia, or hoarse cry. Some physical features may include cardiac septal defects, macroglossia, large fontanelles, umbilical hernia, mottled/dry skin, pallor, myxedema, goiter, or coarse facial features. Profound mental retardation is the most serious effect of untreated CH. Affected infants whose treatment is delayed can also develop neurologic problems such as spasticity and gait abnormalities, dysarthria or mutism and autistic behavior.

Screening Test of Congenital Hypothyroidism (CH)

Diagnosis of primary hypothyroidism is confirmed by demonstrating decreased levels of serum thyroid hormone (total or free T4) and elevated levels of TSH.

Treatment of Congenital Hypothyroidism (CH)

Recommended treatment is lifetime daily administration of levo-thyroxine. Only the tablet form of levo-thyroxine should be prescribed. The U.S. Food and Drug Administration has not approved liquid suspensions. Suspensions prepared by pharmacists may lead to unreliable dosage. The tablets should be crushed daily, mixed with a few milliliters of water, formula or breast milk and fed to the infant. Levo-thyroxine should not be mixed with soy formula or with formula containing iron, as these products interfere with absorption of the medication. Dosage will need to be gradually increased as the infant grows.

6. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

2. Congenital Adrenal Hyperplasia (CAH)

21-hydroxylase deficiency (also known as congenital adrenal hyperplasia) is an inherited disorder that affects the adrenal glands. These glands are located on top of the kidneys and produce a variety of hormones that regulate many essential functions in the body. Two of these hormones, cortisol and aldosterone, are produced from cholesterol through the activity of an enzyme called 21-hydroxylase. Cortisol has numerous functions such as maintaining blood sugar levels, protecting the body from stress, and suppressing inflammation. Aldosterone, sometimes called the salt-retaining hormone, acts on the kidneys to regulate the levels of salt and water in the body, which affects blood pressure. People with 21-hydroxylase deficiency have a shortage of the 21-hydroxylase enzyme, which impairs the conversion of cholesterol to cortisol and aldosterone. When the precursors of cortisol and aldosterone build up in the adrenal glands, they are converted to male sex hormones called androgens. Androgens are normally responsible for the appearance of secondary sex characteristics in males (virilization). Elevated levels of androgens can affect the growth and development of both males and females. There are three types of 21-hydroxylase deficiency. Two types are classic forms, known as the simple virilizing and salt-loss types. Simple virilizing 21-hydroxylase deficiency causes a buildup of potent androgens that leads to the masculinization (development of male characteristics) of external genitalia in females at birth. The development of the internal reproductive organs (uterus and ovaries) in these patients is normal. Salt-loss 21-hydroxylase deficiency results from an almost complete loss of enzyme activity. In these cases, so little aldosterone is produced that the kidneys do not reabsorb sodium. In the third type of 21-hydroxylase deficiency, known as the nonclassic form, levels of functional 21-hydroxylase enzyme are moderate. Both males and females with the nonclassic type can display signs and symptoms of androgen excess after birth. (Genetic Home Reference; A service of the US National Library of Medicine ® available online: <http://ghr.nlm.nih.gov/condition=21hydroxylasedeficiency>)

Screening Test of Congenital Adrenal Hyperplasia (CAH)

Newborn screening test is for the 21-OH form of CAH. Screening is based on an immunoassay for the precursor steroid, 17-alpha-hydroxyprogesterone (17-OHP), the metabolic product just prior to the cortisol synthesis step. The levels of 17-OHP are generally elevated in both salt-losing and non-salt-losing forms of 21-OH CAH.

Treatment of Congenital Adrenal Hyperplasia (CAH)

Treatment for classic 21-OHD CAH includes glucocorticoid replacement therapy, which needs to be increased during periods of stress. Individuals with the salt-wasting form of 21-OHD CAH require treatment with 9α-fludrohydrocortisone and often sodium chloride. Bilateral adrenalectomy may be indicated for individuals with severe 21-OHD CAH who are homozygous for two null mutations and who have a history of poor control with hormonal replacement therapy. Females who are virilized at birth may require feminizing genitoplasty. Precocious puberty is treated with analogs of luteinizing hormone-releasing hormone (LHRH). Surveillance includes monitoring of glucocorticoid/mineralocorticoid replacement therapy every three to four months while children are actively growing and less often thereafter, and monitoring for testicular adrenal rest tumors in males every three to five years after onset of puberty. (Gene Reviews. Available online: <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=cah>

6. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

D. Respiratory Disorders

1. Cystic Fibrosis (CF)

Cystic fibrosis (CF) is an inherited disorder that results in abnormal, thick secretions in the digestive and respiratory systems. The clinical symptoms of cystic fibrosis vary between affected individuals. Some affected infants have meconium ileus, an intestinal obstruction caused by thick secretions at birth, but most infants appear healthy at birth. Poor growth and abnormal bowel movements appear in most children within the first year of life due to abnormal secretion by the pancreas gland, which results in malabsorption of fat and other nutrients. In the lungs, thick secretions lead to frequent cough, wheezing and susceptibility to respiratory tract infections. Respiratory symptoms sometimes occur during the first few weeks of life, but may not occur for years.

Screening Test of Cystic Fibrosis (CF)

Initial screening of newborn bloodspots measures IRT. This pancreatic exocrine product is significantly elevated in over 90% of affected newborns. An elevated IRT should prompt additional genetic evaluation or sweat testing to confirm the diagnosis. If the patient being screened had meconium ileus or other bowel obstruction, IRT screening is not reliable and additional screening or diagnostic tests should be considered as indicated.

Treatment of Cystic Fibrosis (CF)

Early diagnosis by newborn screening has allowed earlier combined anti-inflammatory and antibiotic therapies to combat upper respiratory infections and nutritional supplementation to avoid nutritional deficits. Dramatic progress has been made in improving quality of life for these newborns.

Because the diagnosis and therapy of cystic fibrosis is complex, the pediatrician is advised to manage the patient in close collaboration with a consulting pediatric pulmonologist. It is recommended that parents travel with a letter of treatment guidelines from the patient's physician.

6. The screened conditions in the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

E. Genetic Disorders

1. Hearing Loss

Although most babies can hear normally, 2 to 3 of every 1,000 babies are born with some degree of hearing loss. Without newborn hearing screening, it can be difficult to detect hearing loss in the important first months and years of your baby's life. About half of the children with hearing loss have no risk factors for it.

Newborn hearing screening can detect possible hearing loss in the first days of a baby's life. If a possible hearing loss is found, further tests will be done to confirm the results. If a hearing loss is confirmed, treatment and early intervention can start promptly. Early intervention helps babies with hearing loss and their families learn important communication skills.

That is why the American Academy of Pediatrics recommends that all babies receive newborn hearing screening before they go home from the hospital. Babies learn from the time they are born. One of the ways they learn is through hearing. If they have problems with hearing and do not receive the right treatment and early intervention services, babies will have trouble with language development. For some babies early intervention services may include the use of sign language and/or hearing aids. Studies show that children with hearing loss who receive appropriate early intervention services by age 6 months usually develop good language and learning skills. (American Academy of Pediatrics: http://www.aap.org/publiced/br_hearingscreen.htm)

Screening tests of Hearing Loss

There are 2 screening tests that may be used.

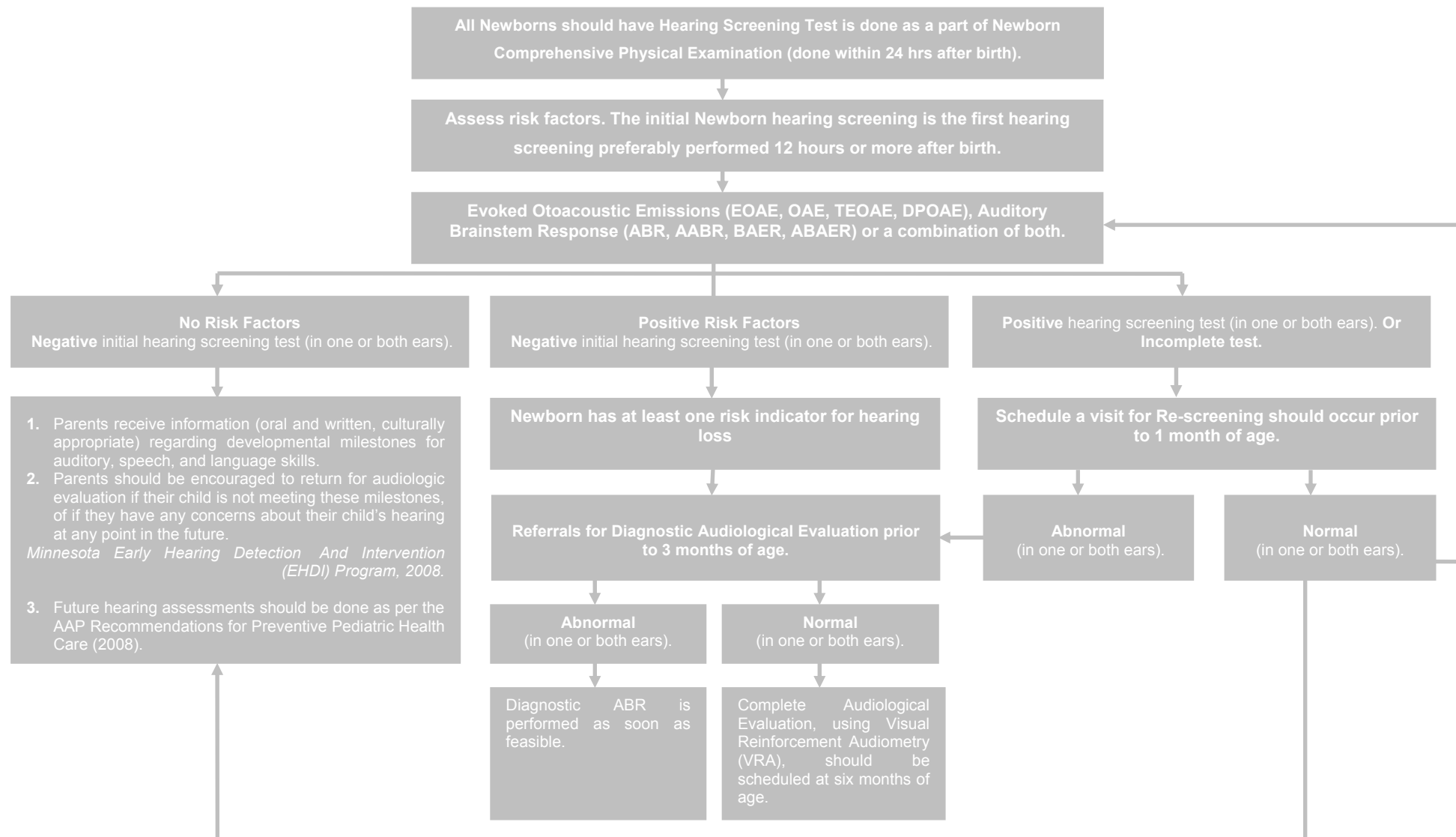
- Auditory brainstem response (ABR): This test measures how the brain responds to sound. Clicks or tones are played through soft earphones into the baby's ears. Three electrodes placed on the baby's head measure the brain's response.
- Otoacoustic emissions (OAE): This test measures sound waves produced in the inner ear. A tiny probe is placed just inside the baby's ear canal. It measures the response (echo) when clicks or tones are played into the baby's ears.

Both tests are quick (about 5 to 10 minutes), painless, and may be done while your baby is sleeping or lying still. Either or both tests may be used.

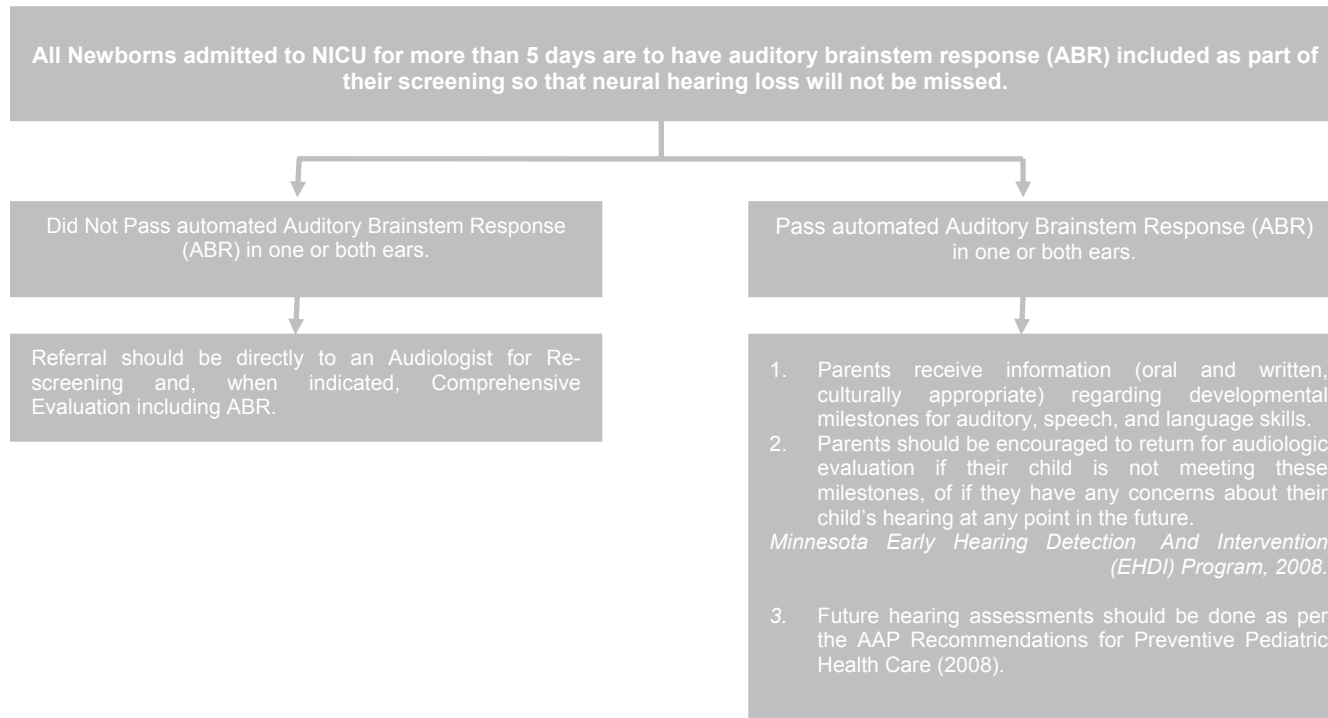
Treatment of Hearing Loss

Every baby with hearing loss should be seen by a hearing specialist (audiologist) experienced in testing babies and a pediatric ENT doctor (otolaryngologist). If the hearing loss is permanent, hearing aids and speech and language services may be recommended for the baby. Early intervention programs should be offered to children with hearing loss, beginning at the time the child's hearing loss is identified. The outlook is good for children with hearing loss who begin an early intervention program before the age of 6 months. Research shows these children usually develop language skills on par with those of their peers.

Pathway of the Initial Hearing Screening (Early Detection of Hearing Loss) for Healthy Newborns



Pathway of the Initial Hearing Screening (Early Detection of Hearing Loss) for NICU Newborns



8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009.

The Health Authority of Abu Dhabi has adapted the below Clinical and Laboratory Standards Institute (CLSI) approved Standards and guidelines for the Newborn Screening Program in the Emirate of Abu Dhabi 2009.

- The Clinical and Laboratory Standards Institute. Blood Collection on Filter Paper for Newborn Screening Programs; (CLSI). Approved Standards-Fifth Edition (LA4-A5).
- The Clinical and Laboratory Standards Institute. Protection of Laboratory Workers from Occupationally Acquired Infections; (CLSI) Approved Guidelines-Third Edition (M29-A3).
- The Clinical and Laboratory Standards Institute. Newborn Screening Guidelines for Premature and /or Sick Newborns; Proposed Guideline. Clinical and Laboratory Standards Institute (CLSI). (I/LA31-P)
- The Clinical and Laboratory Standards Institute. Collection, Transport, Preparation, and Storage of Specimens for Molecular Methods; Approved Guidelines (MM13-A).
- The Clinical and Laboratory Standards Institute (CLSI). Newborn Screening Follow Up Approved Guidelines (I/LA27-A).
- The Clinical and Laboratory Standards Institute (CLSI). Procedures for the Collection of Diagnostic Blood Specimen by Venipuncture; Approved Standards-6th Edition (H-A6).
- The Clinical and Laboratory Standards Institute (CLSI). Procedures and Devices for the Collection of Diagnostic Capillary Blood Specimens; Approved Standards-6th Edition (H04-A6).

To obtain a copy of the above documents, please contact:

The Newborn Screening Program in the Emirate of Abu Dhabi.

Phone 800 800 Extension 543

Email mshehhi@haad.ae

Fax +9712 4444 285

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

1. Newborn Screening Specimen Card



The specimen collection cards for dried blood spots have two distinct parts: a filter paper section to collect the blood drops and a demographic section to record data about the newborn. **The Clinical and Laboratory Standards Institute. Blood Collection on Filter Paper for Newborn Screening Programs; (CLSI). Approved Standards-Fifth Edition (LA4-A5)** includes the Protocol for testing the absorption characteristics of filter paper and filter paper specifications.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

2. Newborn Screening Specimen Collection

A. Timing

1. In the hospital newborn nursery, infant blood specimens are collected on Newborn Screening Specimen Collection Forms. Instructions should be printed on the back of the form to assist in collecting a completed specimen. Initial Forms allow for submission of blood spots and hearing screening information on the first infant specimen. Repeat Forms are available and are designed only for submission of blood spots on any specimen collected after the initial specimen.
2. A specimen will be collected from all newborns, before being discharged from the hospital or birthing facility.
3. A specimen collected between 24-48 hours of life is considered optimum for healthy newborn screening. Specimens should not be collected earlier than 24 hours of age, with the exception of special circumstances.

Special circumstances include:

- **Early discharge**

If the infant is to be discharged at less than 24 hours of age, collect specimen prior to discharge. Inform parents that infant must be rescreened within the second day of life.*

- **Transfers**

If the infant requires transfer to another facility, if at all possible, specimen should be collected prior to transfer, regardless of infant's age. If a specimen cannot be collected prior to transfer, the transferring facility is responsible for informing the admitting facility of the need for specimen collection.

- **Transfusions**

If the infant requires transfusion, specimen should always be collected prior to transfusion, regardless of infant's age. If this is not possible and the infant was transfused prior to specimen collection, indicate the last transfusion date prior to the specimen collection on the filter paper collection form.

Specimen collection immediately after transfusion **will affect** all newborn screening results. If a post-transfusion specimen is necessary, collection should be delayed for 48 hours post-transfusion.

If the infant's initial specimen was collected post-transfusion, a second specimen should be collected 48 hours post-transfusion, and a third specimen is required 90 days after the final transfusion.

- Transfusions **will invalidate** screening for classical galactosemia.
- Transfusions **will invalidate** screening for biotinidase deficiency.
- Transfusions **will invalidate** screening for hemoglobinopathies.

- **Premature and sick infants**

- If the infant's condition is medically unstable, the specimen should be collected at 24-48 hours of age.
- All infants admitted to a neonatal intensive care unit (NICU) or special care nursery should have a routine second specimen collected at 14 days of age or prior to discharge from the unit, whichever comes first.
- The "NICU" check box on the specimen card should be marked on specimens from all infants admitted to a NICU or special care nursery. Retest box should be marked for all repeat specimens.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

▪ Infants receiving special feedings

- Infants requiring soy formula, hyperalimentation or total parenteral nutrition (TPN), and those not yet receiving milk (galactose) feedings require documentation of feeding type or status on the specimen card.
- The feeding type box should be clearly marked for “breast,” “soy,” “other,” “TPN” or “NPO” (nothing by mouth). This information is important to IDPH laboratory staff to assure reliable testing.
- Soy formula or lack of milk feeding will affect screening for galactosemia.
- Hyperalimentation and TPN may affect tandem mass spectrometry screening for some amino acid, fatty acid oxidation and organic acid disorders. If screening results suggest TPN effects, another specimen is requested when the infant has been off TPN for 48 hours, or on day 14 of life if the baby was admitted to NICU or special care nursery.
- If indicated by the feeding type information provided, IDPH laboratory staff will perform additional testing, the galactose-1-phosphate uridyl transferase (GALT) enzyme assay to screen for classical galactosemia on specimens from infants receiving soy formula, and those infants who have not yet received an oral feeding (NPO), when the specimen card is marked accordingly.

▪ Infants receiving antibiotics

- When infants are receiving antibiotics at the time of specimen collection, the “antibiotic” box of the specimen collection card should be marked, as the presence of antibiotics and some other medication metabolites (valproic and benzoic acids) may be detected by tandem mass spectrometry. In these cases, a repeat specimen will be requested.

4. The INITIAL newborn screening specimen form is used for the FIRST specimen collected from an infant; for all subsequent specimens collected from the infant, the REPEAT newborn screening specimen form is used.
5. INITIAL specimens from sick or premature infants should be collected before a blood transfusion or between 24-48 hours of life. All sick or premature infants should have a REPEAT specimen collected between 7-14 days of life.
6. All specimens should be sent to the Screening Laboratory within 24 hours of collection.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

B. Method of Newborn Screening Specimen Collection

1. A Heel-stick is the method recommended by the Center for Disease Control and The National Committee for Clinical Laboratory Standards (NCCLS).
2. If heel stick is not possible, use of a syringe to collect blood from an umbilical catheter is recommended by CLSI.
 - If heel stick is not a viable option, see CLSI recommended procedures for specimen collection, Approved Standard LA4-A5.
 - Some screening results may vary slightly between heel stick specimens and venous or capillary specimens.
3. Collection of specimens in capillary tubes is not recommended by IDPH or CLSI.
 - Heparinized capillary tubes should only be used when heel stick or collection by syringe is not possible.
 - Extreme caution in applying blood samples from capillary tubes is required to avoid damage to the filter paper and allow uniform application of the sample to avoid "layering."
 - Layering can affect the validity and reliability of screening results. If heel stick is not an option, see the CLSI recommended procedures for specimen collection.

Ideally, specimen collection should take place between 24-48 hours of age. Unless medically contraindicated, specimens are required on all newborns prior to transfusion, after transfer, or early discharge, even if the specimen has to be drawn prior to 24 hours of age. Under those circumstances, a repeat specimen will be necessary using the following timelines:

1. Initial sample collected prior to 24 hours of age: repeat sampling within 72 hours
2. Initial sample collected prior to 24 hours of age with a subsequent transfusion of blood products: repeat sampling within 72 hours from last transfusion
3. Initial sample collected after transfusion has taken place and there is no pre-transfusion specimen: repeat sampling at 90 days from last transfusion

Specimens should always be collected by following instructions found on the back of the filter paper specimen cards. When collecting a specimen, follow these general guidelines:

1. Do not squeeze tissue to obtain blood
2. Do not use devices that contain EDTA (ethylene-di-amine-tetra-acetic acid)
3. Do not apply specimen to both sides of specimen card
4. Do not expose card to heat, moisture, or direct sunlight
5. Do not hold specimens to form batches
6. Make sure all information on specimen card is filled out completely and accurately. Correctly report the newborn's birthing hospital.
7. Mail initial filter paper within 12 hours of collection to the correct testing laboratory.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

B. Method of Newborn Screening Specimen Collection (cont.)

The heel-stick is the usual practice in obtaining blood spot. Venipuncture may be attempted for complicated heel-stick.

The Newborn Screening Program uses standards developed by the Clinical and Laboratory Standards Institute for blood collection on filter paper specifically for newborn screening programs: NCCLS. Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard—Fifth Edition.

The primary goal of this standard is to ensure the quality of blood spots collected from newborns. Poor quality specimens place an unnecessary burden on the screening facility, because unnecessary trauma to the infant and anxiety to the infant's parents, potentially delay the detection and treatment of the affected infant, and may contribute to a missed or late diagnosed case.

When the Newborn Screening Program receives an unacceptable specimen, program staff requests a repeat sample from the birthing center. Blood collected from the heel is the standard for newborn screening. The medial and lateral parts of the underfoot are preferred (*see diagram on back of newborn screening card*). Blood should never be collected from: the arch of the foot, fingers, earlobe, a swollen or previously punctured site, or IV lines containing other substances (TPN, blood, drugs, etc.).

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

C. Blood Sample Collection and handling procedure



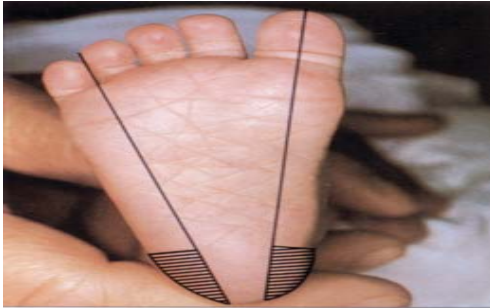
Procedure 1 Necessary equipment: sterile lancet with tip approximately 2.0mm, sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, Powder-free gloves are best worn while collecting. gloves. Lotion, Vaseline, and other substances which can interfere with analysis should be kept off the newborn's skin.

A photograph of a completed blood collection form. The form has a barcode at the top left with the number 06752821. It contains various fields for handwritten information, including dates, times, and names. The form is titled "ABU DHABI NEWBORN SCREENING PROGRAM" and "SUBMITTER COPY".

Procedure 2 Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come into contact with spillage or by touching before or after blood collection. Keep "SUBMITTER COPY" if applicable.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

D. Blood Sample Collection and handling procedure (cont.)



Procedure 3 Hatched area indicates safe areas for puncture sites.



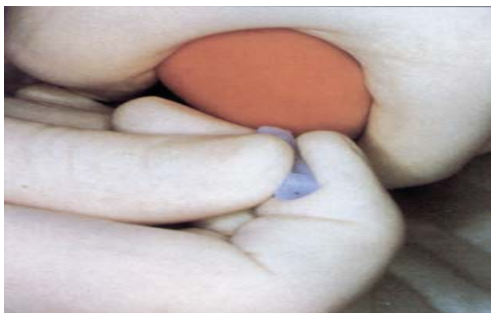
Procedure 4 Warm site with soft cloth, moistened with warm water up to 41 ° C, for three to five minutes. and position the leg lower than the heart to increase venous pressure before collecting the sample.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

D. Blood Sample Collection and handling procedure (cont.)



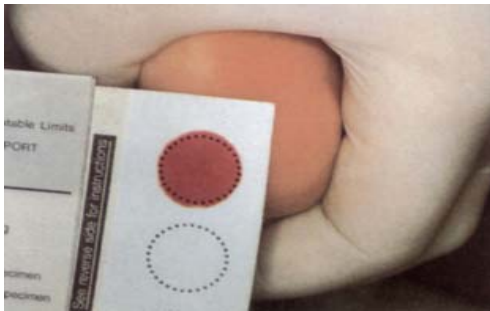
Procedure 5 Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad or allow to thoroughly air dry.



Procedure 6 Puncture heel using a sterile lancet or a heel incision device to make an incision 1 mm deep and 2.5 mm long. When collecting from small, premature infants, it is safer to make the incision more shallow. Wipe away first blood drop with sterile gauze pad. Allow another large blood drop to form.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

C. Blood Sample Collection and handling procedure (cont.)



Procedure 7 Lightly touch filter paper to LARGE blood drop. Allow blood to soak through and completely fill circle with 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.



Procedure 8 Fill remaining circles in the same manner as step 7, with successive blood drops. If blood flow is diminished, repeat step 5 through 7. Care of skin puncture site should be consistent with your institution's procedures. Do not apply multiple layers of blood drops to the same circle. The circles are measured and contain a set volume of blood (think of them as small flat test tubes). Layering can interfere with the accuracy of the test by providing a non-standard amount of blood or non-uniform analyte concentration. Excessive milking or squeezing of the puncture site can result in an unsatisfactory specimen because of hemolysis breaking down the blood cells to be analyzed or mixing tissue fluids into the specimen which can dilute it.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

D. Blood Sample Collection and handling procedure (cont.)



Procedure 9 Dry blood spot on a dry, clean, flat, non-absorbent surface for a minimum of four hours, which is essential to maintain the integrity of the blood spots.



Procedure 10 mail completed form to testing laboratory within 12 hours of collection.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

3. Unsatisfactory Newborn Screening Specimen



VALID SPECIMEN

Allow sufficient quantity of blood to soak through to completely fill the preprinted circle on the filter paper. Fill all required circles with blood. Do not layer successive drops of blood or apply blood more than once in the same collection circle. Avoid touching or smearing spots.



INCOMPLETE SATURATION

Uneven saturation; blood did not soak through the filter paper. Possible causes:

Removing filter paper before blood has completely filled circle or before blood has soaked through to opposite side; improper capillary tube application; allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.

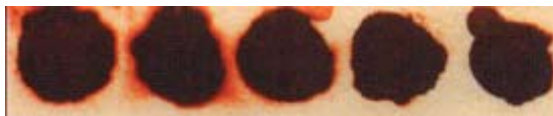


LAYERED CLOTTED OR SUPERSATURATED

Possible causes: Touching the same circle on filter paper to blood drop several times; filling circle on both sides of filter paper; application of excess blood; clotted swirl marks from improper capillary application. Use of unheparinized capillary tube.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

3. Unsatisfactory Newborn Screening Specimen (cont.)



DILUTED, DISCOLORED OR CONTAMINATED

Possible causes: squeezing or milking of area surrounding the puncture site; allowing filter paper to come in contact with gloved or ungloved hands, or substances such as alcohol, formula, antiseptic solutions, water, hand lotion, powder, etc., either before or after blood specimen collection; exposing blood spots to direct heat; allowing blood spots to come in contact with tabletop, etc. while drying the sample.



QUANTITY NOT SUFFICIENT

Quantity of blood on filter not sufficient for testing. Possible causes: Removing filter paper before blood has completely filled circle; not allowing an ample sized blood drop to form before applying to filter; inadequate heel stick procedure.



SPECIMEN ABRADED

Filter scratched, torn or abraded. Possible causes: Improper use of capillary tubes. To avoid damaging the filter paper fibers, do not allow the capillary tube to touch the filter paper. Actions such as “coloring in” the circle, repeated dabbing around the circle, or any technique that may scratch, compress, or indent the paper should not be used.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

3. Unsatisfactory Newborn Screening Specimen (cont.)

BLOOD ON OVERLAY COVER

Overlay cover came in contact with wet blood specimen. Sample is unsatisfactory for testing because blood soaked from back of filter onto the gold colored backing of the form. The filter circles are designed to hold a specific quantity of blood. If the wet filter is allowed to come in contact with the paper backing of form, blood can be drawn out of filter making the quantitative tests performed by the Newborn Screening Laboratory invalid. Allow blood spots to thoroughly air dry for at least 2 hours in a horizontal position, away from direct heat and sunlight. Do not allow the blood to touch any surface during drying, including other parts of the form.

OLD SPECIMEN

Specimen greater than 15 days old when received at NBS Laboratory. The collection card should be transported or mailed to the Newborn Screening Laboratory within 24 hours after specimen collection. Avoid the practice of holding onto specimens to wait for more to accumulate before mailing, also referred to as “batching” the specimens. Although batching may seem more efficient, it’s not worth it in the long run because a delay in screening and treatment can cause irreparable damage to a child with metabolic disease.

OLD FORM

Sample received on out-of-date form. The current newborn screening specimen collection form has a printed form number located next to the form bar code that begins with “B05”. If a form number begins with any other letters or digits and does not have a printed bar code, then it is an outdated form and should not be used.



SERUM RINGS

Serum separated into clear rings around blood spot. Possible causes: Card dried vertically (on side) instead of flat; squeezing excessively around puncture site; allowing filter paper to come in contact with alcohol, hand lotion, etc.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

3. Unsatisfactory Newborn Screening Specimen (cont.)

REFUSAL

Parents refused to have Newborn Screening. Parents who object to testing on religious grounds shall state those objections in writing. The written objection shall be filed with the attending physician, certified nurse midwife, public health facility, ambulatory surgical center or hospital. It is important that the signed refusal of testing be kept with the baby's medical records. Upon receipt, the attending physician, certified nurse midwife, public health facility, ambulatory surgical center or hospital shall send a copy of the written objection to the Department of Health and Senior Services to the attention of Genetics and Healthy Childhood, 930 Wildwood Dr., Jefferson City, MO 65109.



NO BLOOD

Filter submitted without blood.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

4. Testing Methodologies, Normal Values, Criteria for Abnormal Results

Newborn Conditions included in the Newborn Screening Program in the Emirate of Abu Dhabi 2009.	ICD 9 and 10 Codes	Compound Assessed	Testing Method
1. Phenylketoneuria (PKU)	270.1, E70.0	Phenylalanine	Any of these Methods: 1. Guthrie Bacterial Inhibition Assay (BIA) 2. Automated fluorometric assay 3. Tandem mass spectrometry
2. Congenital Hypothyroidism (CH)	243, E03	Human thyroid stimulating hormone (hTSH)	Quantitative time-resolved fluoroimmunoassay
3. Congenital Adrenal Hyperplasia (CAH)	255.2, E25.0	<i>First-Tier Screen:</i> 17 α -hydroxyprogesterone (17-OHP) by quantitative time-resolved fluoroimmunoassay Any specimens that have an abnormal first-tier screen will automatically be repeated with the second-tier screen (a repeat specimen is not necessary for second-tier screening).	Any of these methods: 1. Quantitative time-resolved fluoroimmunoassay 2. Tandem Mass Spectrometry
4. Sickle Cell Disease (Hb S/S)	282.6, D57.0	Normal and Variant hemoglobin	Isoelectric Focusing
5. Sickle Cell Disease (Hb S/C)	282.6, D57.2		
6. Thalassemia (Hb S/A)	282.49, D56.0		
7. Galactosemia (GALT)	271.1, E74.2	Galactose-1-phosphate uridyltransferase deficiency	Fluorometric Assay

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

4. Testing Methodologies, Normal Values, Criteria for Abnormal Results (cont.)

Newborn Conditions included in the Newborn Screening Program in the Emirate of Abu Dhabi 2009.	ICD 9 and 10 Codes	Compound Assessed	Testing Method
8. Biotinidase (BIO)	266.1, E53.9	Biotinidase enzyme	Qualitative Colorimetric Assay
9. Cystic Fibrosis (CF)	277.00, E84.9	<i>First-Tier Screen:</i> Immunoreactive trypsinogen (IRT) by Quantitative time-resolved Fluoroimmunoassay Any specimens that have an abnormal first- tier screen will automatically proceed to the second-tier screen (a repeat specimen is not necessary for second-tier screening).	Quantitative time-resolved Fluoroimmunoassay
10. Hearing Loss	289, H90.5		Both of these methods: 1. Auditory brainstem response (ABR) 2. Otoacoustic emissions (OAE)
11. Glucose 6-phosphate dehydrogenase Deficiency (G6PD)	282.2, D55.0	Glucose-6-Phosphate Dehydrogenase Enzyme	Enzyme analysis or primary DNA screening. Confirmatory testing using a quantitative assay should be performed for diagnosis of G6PD deficiency.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

5. Haemoglobinopathies Interpretation Chart

Description of Result	Observation	Interpretation
Transfused?	A>F	Results suggest that the infant was transfused (A>F on hemoglobinopathy screening). A pre-transfusion specimen was collected on [date], and Hemoglobinopathy results were normal. No further testing is required for hemoglobinopathies.
Transfused?	A>F	Results suggest that the infant was transfused (A>F on hemoglobinopathy screening), and Hemoglobinopathy results cannot be interpreted. Please send repeat newborn screening specimen at 90 days post transfusion.
Low A	F low A	Decreased amounts of adult hemoglobin are present. Recommend performing hemoglobin electrophoresis at nine months of age to confirm result.
F only	Hemoglobin F only (undetectable A)	Abnormal result possibly caused by prematurity, diabetic mother, or beta thalassemia in the baby. Check mother's blood glucose. If normal, obtain CBC and hemoglobin electrophoresis on the infant at 4-6 months of age.
ABNORMAL	FS	This newborn screen indicates that the child may have Sickle Cell Disease (hemoglobin S no A). Physician should immediately contact a hematology specialist and obtain hemoglobin electrophoresis as soon as possible. Prophylactic antibiotics should be initiated before two months of age. Depending on test results, genetic counseling may benefit the family.
ABNORMAL	FS & Barts (High)	This newborn screen indicates that the child may have Sickle Cell Disease (hemoglobin S no A). The results also indicate the baby may have alpha thalassemia. Physician should immediately contact a hematology specialist and obtain hemoglobin electrophoresis as soon as possible. Prophylactic antibiotics should be initiated before two months of age. Depending on test results, genetic counseling may benefit the family.
ABNORMAL	FS & Barts (Low)	This newborn screen indicates that the child may have Sickle Cell Disease (hemoglobin S no A). The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Physician should immediately contact a hematology specialist and obtain hemoglobin electrophoresis as soon as possible. Prophylactic antibiotics should be initiated before two months of age. Depending on test results, genetic counseling may benefit the family.
ABNORMAL	FC	This newborn screen indicates that the child may have Hb C disease. Obtain hemoglobin electrophoresis at 6 months of age. Consultation with a hematology specialist is recommended.
ABNORMAL	FC & Barts (High)	This newborn screen indicates that the child may have Hb C disease. The results also indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 6 months of age. Consultation with a hematology specialist is recommended.
ABNORMAL	FC & Barts (Low)	This newborn screen indicates that the child may have Hb C disease. The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Obtain hemoglobin electrophoresis at 6 months of age. Consultation with a hematology specialist is recommended.
ABNORMAL	FSC	This newborn screen indicates that the child may have Sickle Cell Disease. Physician should immediately contact a hematology specialist and obtain hemoglobin electrophoresis as soon as possible. Prophylactic antibiotics should be initiated before two months of age. Depending on test results, genetic counseling may benefit the family.
ABNORMAL	FSC & Barts (High)	This newborn screen indicates that the child may have Sickle Cell Disease. The results also indicate the baby may have alpha thalassemia. Physician should immediately contact a hematology specialist and obtain hemoglobin electrophoresis as soon as possible. Prophylactic antibiotics should be initiated before two months of age. Depending on test results, genetic counseling may benefit the family.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

5. Haemoglobinopathies Interpretation Chart (cont.)

Description of Result	Observation	Interpretation
ABNORMAL	FSC & Barts (Low)	This newborn screen indicates that the child may have Sickle Cell Disease. The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Physician should immediately contact a hematology specialist and obtain hemoglobin electrophoresis as soon as possible. Prophylactic antibiotics should be initiated before two months of age. Depending on test results, genetic counseling may benefit the family.
ABNORMAL	FSE	This newborn screen indicates that that the child has hemoglobin type either S/E (usually harmless) or type S/O (potential for a sickling disorder). Obtain hemoglobin electrophoresis at 6 months of age. Depending on results, consultation with a hematology specialist is recommended.
ABNORMAL	FSE & Barts (High)	This newborn screen indicates that that the child has hemoglobin type either S/E (usually harmless) or type S/O (potential for a sickling disorder). The results also indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 6 months of age. Depending on results, consultation with a hematology specialist is recommended.
ABNORMAL	FSE & Barts (Low)	This newborn screen indicates that that the child has hemoglobin type S/E (Usually harmless) or type S/O (potential for a sickling disorder). The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Obtain hemoglobin electrophoresis at 6 months of age. Depending on results, consultation with a hematology specialist is recommended.
ABNORMAL	FE	This newborn screen indicates that that the child has hemoglobin type either E/E (usually harmless) or type E/beta-thal (potential for severe anemia). Obtain hemoglobin electrophoresis at 4- 6 months of age. Depending on results, consultation with a hematology specialist is recommended.
ABNORMAL	FE & Barts (High)	This newborn screen indicates that that the child has hemoglobin type either E/E (usually harmless) or type E/beta-thal (potential for severe anemia). The results also indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 4-6 months of age. Depending on results, consultation with a hematology specialist is recommended.
ABNORMAL	FE & Barts (Low)	This newborn screen indicates that that the child has hemoglobin type either E/E (usually harmless) or type E/beta-thal (potential for severe anemia). The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Obtain hemoglobin electrophoresis at 6 months of age. Depending on results, consultation with a hematology specialist is recommended.
ABNORMAL	F Unknown	The baby carries a hemoglobin type that cannot be identified by this assay. Consultation with a hematologist is recommended. Hemoglobin electrophoresis should be performed at 6 months of age and testing of other family members may be necessary.
ABNORMAL	F Unknown & Barts (High)	The baby carries a hemoglobin type that cannot be identified by this assay. The results also indicate the baby may have alpha thalassemia. Consultation with a hematologist is recommended. Hemoglobin electrophoresis should be performed at 6 months of age and testing of other family members may be necessary.
ABNORMAL	F Unknown & Barts (Low)	The baby carries a hemoglobin type that cannot be identified by this assay. The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Consultation with a hematologist is recommended. Hemoglobin electrophoresis should be performed at 6 months of age and testing of other family members may be necessary.
TRAIT	FA Barts (Low)	Baby is likely a carrier of alpha thalassemia trait (not affected). No further testing is required for hemoglobinopathies.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

5. Haemoglobinopathies Interpretation Chart (cont.)

Description of Result	Observation	Interpretation
TRAIT	FA Unknown	The baby is likely a carrier of hemoglobin type that cannot be identified by this assay. Hemoglobin electrophoresis should be performed after 9 months of age and testing of other family members may be necessary.
TRAIT	FA Unknown & Barts (Low)	The baby is likely a carrier of hemoglobin type that cannot be identified by this assay. The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Hemoglobin electrophoresis should be performed after 9 months of age and testing of other family members may be necessary.
TRAIT	FAS	Baby is likely a carrier (not affected) of Sickle Cell Trait. Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT	FAS & Barts (Low)	Baby is likely a carrier (not affected) of Sickle Cell Trait. The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT	FAC	Baby is likely a carrier (not affected) of Hb C trait, common in African American newborns. Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT	FAC & Barts (Low)	Baby is likely a carrier (not affected) of Hb C trait, common in African American newborns. The results also indicate the baby is a likely a carrier of alpha thalassemia trait (not affected). Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT	FAE	Baby is likely a carrier (not affected) of hemoglobin E. Obtain hemoglobin electrophoresis and CBC at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT	FAE & Barts (Low)	Baby is likely a carrier (not affected) for both hemoglobin E and alpha thalassemia. This is not expected to cause disease and is common in Southeast Asian newborns. Obtain hemoglobin electrophoresis and CBC at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT	FAG	Baby is likely a carrier (not affected) of hemoglobin G. Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT/ THALASSEMIA	FAS & Barts (High)	Baby is likely a carrier (not affected) of Sickle Cell Trait. The results also indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT/ THALASSEMIA	FAC & Barts (High)	Baby is likely a carrier (not affected) of Hb C trait. The results also indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT/ THALASSEMIA	FAE & Barts (High)	Baby is likely a carrier for hemoglobin E and may also have alpha thalassemia. This is not uncommon in Southeast Asian newborns. Obtain hemoglobin electrophoresis and CBC at 9 months of age. Depending on results, genetic counseling may benefit the family.
TRAIT/ THALASSEMIA	FA Unknown & Barts (High)	Baby carries a hemoglobin variant that cannot be identified by this assay (also called unknown trait). The results also indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 9 months of age, and similar testing of family members may be necessary. Depending on results, genetic counseling may benefit the family.
TRAIT/ THALASSEMIA	FA Barts (High)	The results indicate the baby may have alpha thalassemia. Obtain hemoglobin electrophoresis at 9 months of age. Depending on results, genetic counseling may benefit the family.

8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

6. Follow-up on Positive Newborn Screening Results

All specialized Healthcare Providers authorized to provide tertiary care to the newborn's with Screen positive results should establish a Clinical Management Team which is composed of 2 Registered Nurses and a counselor. The Clinical Management Team must be prepared to explain to the family the meaning of a positive screening result, the possibility of a false-positive test result, and the steps that must be taken next.

Newborn Specimen Screening, Reporting and Follow up are all the primary responsibility of the Newborn Screening laboratory. In case of both Normal NBS result and the need for immediate recollection of Newborn Screening Specimen, the lab will inform the parents of the result or the need to repeat specimen collection. When Newborn Screening Laboratory identifies An Abnormal result (suspected borderline or presumptive positive Abnormal), it will immediately initiate the referral process and notify (by phone and fax) the Clinical Management Team, which in turn will notify the Parents and schedule a visit with a Specialist. depending on the condition for evaluation and order necessary diagnostic tests.

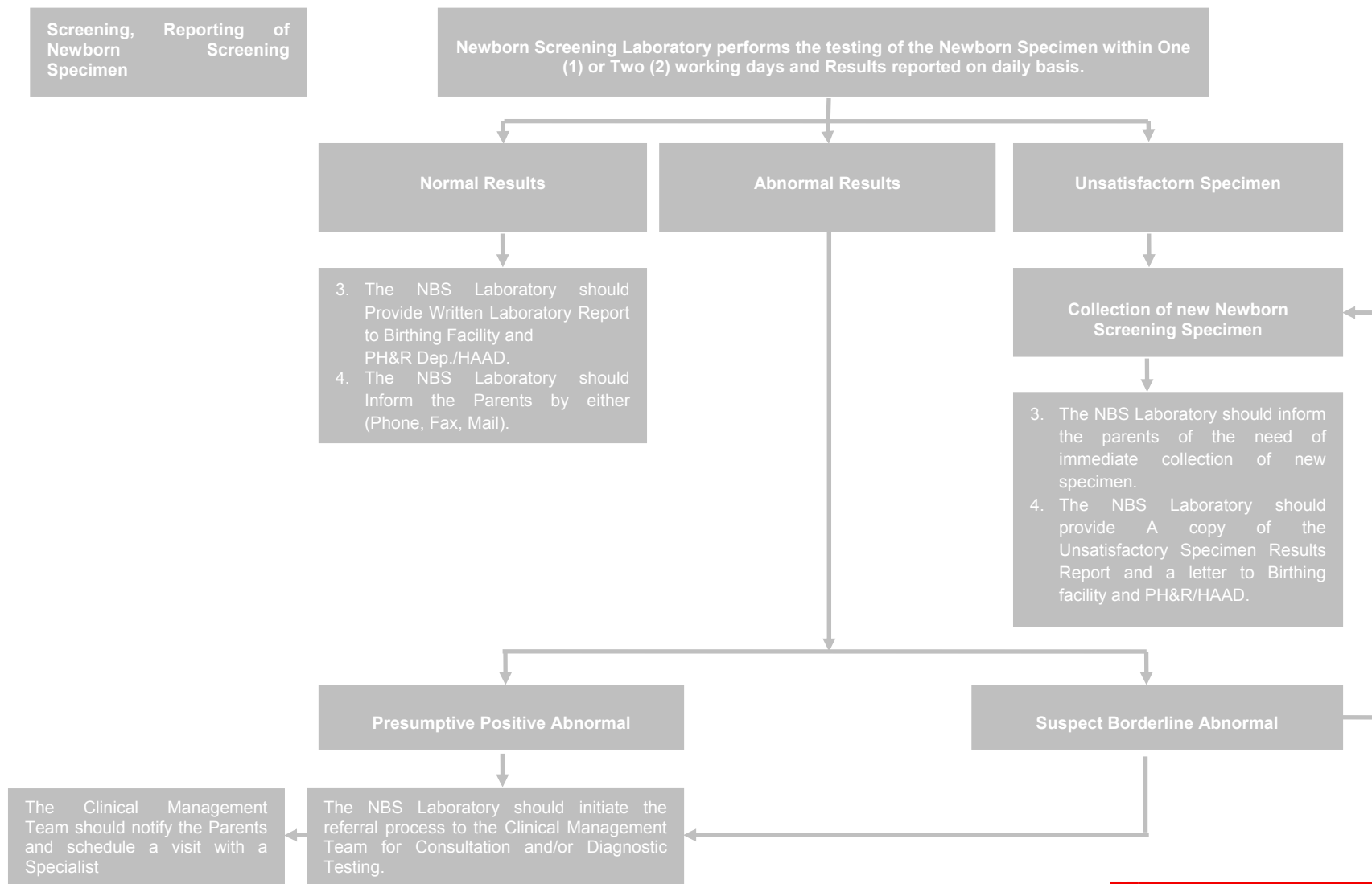
Specialists determine follow-up recommendations based on the suspected condition. Assess the health of the baby and to inform the parents of the need for additional testing and evaluation. Provide support to the parents throughout the process of diagnostic testing and evaluation. Convey information about the outcome of diagnostic testing and evaluation to NBS program public health officials once studies are complete. The Newborn Screening Program may request additional information depending on the specific disorder.

For a visual representation of the process for reporting and following-up positive results, see the Newborn Screening Follow up Chart, page (49).

7. Follow-up on Requests for Newborn Screening Specimens

If NBS Program/ Lab officials deems a specimen unsatisfactory for any reason (collected prior to 24 hours of age, layered blood, insufficient quantity of blood to complete testing, spilled substances on the card, etc.) or if the newborn has a borderline result for any assay, NBS Program /Lab officials may contact the parents directly to request a repeat specimen.

8. Newborn Screening Follow Up Chart



8. Laboratory Services of the Newborn Screening Program in the Emirate of Abu Dhabi 2009 (cont.).

9. Documentation of Newborn Screening Results

You should document newborn screening results for every child in your practice at the first well-child visit. Do not assume that no news is always good news. If you cannot locate the infant's newborn screening results, contact the birth hospital. If the hospital does not have the results on record, contact the Newborn Screening Program officials at the Health Authority of Abu Dhabi.

10. Facilitation of Parental Requests

Parents have the right to make certain requests during the newborn screening process. The health care provider should help facilitate the following requests if approached by the parent(s) or legal guardians:

- **Parents may choose not to have their child screened:** It is mandatory that all newborns have comprehensive physical examination and hearing screening. Parents and legal guardians have the option to refuse the Newborn Screening Test (Heel Prick Test) only.
- **Parents may choose to have their child's results and sample destroyed:** At any time, parents and legal guardians may ask that the results of newborn screening and the blood sample be destroyed. Should Newborn Screening Program Officials at HAAD receive such a request, the destruction of the results and blood sample will occur 24 months after the initial newborn screening takes place. Asking that the newborn screening results be destroyed means that the results will no longer be available from Newborn Screening Program officials under any circumstances; this situation may potentially lead to duplicative testing in the future.

9. Answer to Commonly Asked Questions about Newborn Screening.

Who informs parents about newborn screening program?

Although the birth hospital is ultimately responsible for informing parents about newborn screening and their options to opt out of screening, prenatal care providers can inform expectant parents about newborn screening before their baby is born.

In fact, learning about newborn screening during the prenatal period is ideal for expectant parents. Many parents are unaware that newborn screening happens in the midst of all the activities following labor and delivery. Prenatal care providers are in a unique position to inform parents about this safe and easy way to ensure a healthy start for their babies. To facilitate talking with parents, Health Authority Abu Dhabi/ Public Health & Policy recommends using the newborn screening parent pamphlet as a tool.

Who is responsible for collecting the newborn screening specimen?

Newborn screening is mandated by the Health Authority of Abu Dhabi; the statute establishes that the birth hospital is responsible for collecting the newborn screening specimen. Disorders included on the newborn screening panel may not be readily identified at birth. Symptoms may be minimal or too generalized to lead to early diagnosis, and delayed treatment can cause irreversible damage or death. Since all of the genetic disorders included on the panel are inherited in an autosomal recessive way (with the exception of congenital hypothyroidism which is usually sporadic), affected infants usually do not have affected parents or a known family history of the condition.

What if a baby is born at a Private hospital?

The newborn screening statute applies to all births in the Emirate of Abu Dhabi. The birth attendant is responsible for collecting a blood sample from a baby born in a private hospital.

How should the specimen be collected?

The heel-stick is the gold standard method for collecting newborn screening specimens because it is a relatively easy way to obtain samples, and it does not require sophisticated staff training. In addition, normal values and cutoffs for the newborn screening assays are determined from samples collected in this manner. (Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard—Fourth Edition. NCCLS document LA4-A4 [ISBN 1-56238-503-8]. NCCLS, 940 West Valley Road, Suite 1400, Wayne, Pennsylvania 19087-1898 USA, 2003.)

When should the specimen be collected?

Specimens must be collected between 24 to 48 hours of age. All normal values and references ranges for newborn screening tests are based on specimens drawn from infants during this age window. Specimen's special circumstances, specimens may be drawn when an infant is less than 24 hours of age. For example, specimens are required on all babies prior to transfusion or early hospital discharge, even if this means collecting the specimen before 24 hours of age. If a positive result is identified, the infant's health care provider will be alerted immediately. For babies less than 24 hours of age at the time of collection, results for disorders other than hemoglobinopathies, galactosemia, biotinidase deficiency, and cystic fibrosis cannot be classified as negative because normal values are not established for this age window. Therefore, if the values are normal for a specimen drawn at less than 24 hours of age, the results will be reported as "Unsatisfactory," and a repeat specimen will be required to ensure accurate screening results. Hemoglobinopathy, galactosemia, and biotinidase deficiency results are not impacted by the baby's age at the time of collection; therefore, results for these disorders will be reported as usual. If an infant is "missed," and a newborn screening specimen is not collected from an infant during the 24 to 48 hours timeframe, a specimen should still be sent immediately even if the baby is older than 48 hours of age. Ideally, all babies should be screened in the first week of life, but screening a baby late is better than not screening at all.

Can parents refuse newborn screening for their infant?

Parents and legal guardians have the option to refuse screening of their child or to choose to have the specimen and the results destroyed. Parents must indicate their refusal in writing, and a copy should be submitted to the Health Authority (HAAD) Newborn Screening Program Manager. The original form(s) should be retained in the child's medical chart with a copy provided to the parent(s) or legal guardians. The refusal form serves as documentation that the parents were informed about the possible adverse outcomes of not performing newborn screening and that they accept legal responsibility for the consequences of their decision.

9. Answer to Commonly Asked Questions about Newborn Screening (cont.)

What if an infant has a family history of a disorder detected by newborn screening?

In addition to newborn screening, infants who have a family history of a disorder detected by newborn screening should have definitive diagnostic testing for that particular disorder. This additional diagnostic testing after birth is necessary even if prenatal testing was performed. If a pregnant couple has an older affected child, please contact a medical specialist prior to the delivery to plan for immediate evaluation of the baby. Please contact the Health Authority (HAAD) Newborn Screening Program Manager if a family has a history of a disorder on the newborn screening panel.

How do I obtain a copy of newborn screening results?

A paper copy of the newborn screening results will be mailed back to the infant's birth hospital, and the birth hospital is responsible for forwarding the results to the infant's health care provider when needed.

Do newborn screens need to be repeated?

Occasionally, the baby's health care provider will be asked to collect a repeat newborn screen after the baby has already been discharged from the hospital. Blood collection can be accomplished through the birthing center laboratory.

What if a baby in my care has a positive newborn screening result?

For positive results, HAAD immediately notifies the infant's health care provider (the physician listed on the newborn screening card as responsible for infant follow-up after discharge) by phone and fax. At the same time, HAAD notifies an appropriate specialist. With the specialist's guidance, the health care provider should promptly contact the family and arrange for evaluation and diagnostic tests.

Is there newborn screening information for parents?

A parent brochure is available from the Health Authority Abu Dhabi Newborn Screening Program, and you can obtain free copies by contacting the program directly.

What about false positive results?

Any screening methodology is subject to some false positive results. False positive results may be caused by the stress of birth; the specimen being collected prior to 24 hours after birth; poor specimen collection techniques; or the way the specimen was dried, stored, or transported.

What is the Newborn Screening Program's specimen storage policy?

Newborn blood spot specimens are stored securely for an indefinite period of time. MDH may use specimens in the laboratory for quality control purposes or new test development. Researchers outside MDH may use specimens only if the specimens cannot be traced to individuals and the research has been approved by an institutional review board.

10. Resources for Health Care Providers

Newborn Screening Resources

- National newborn screening and genetics resource center (NNSGRC) provides information and resources in the area of newborn screening and genetics to benefit health professionals, the public health community, consumers and government officials. <http://genes-r-us.uthscsa.edu>
- National Center for Hearing Assessment and Management. <http://www.infanthearing.org>
- The database is hosted by the **National Newborn Screening and Genetics Resource Center (NNSGRC)** and is designed to provide secure, state and territorial newborn screening information. <http://www2.uthscsa.edu/nnsis/>
- Website with information on a wide variety of syndromes and disorders. <http://www.magicfoundation.org>
- Website with information on organic acidemias. <http://www.oaaneews.org>
- Website with information on sickle cell disease. <http://www.ascaa.org>
- Website with information on galactosemia. <http://www.galactosemia.org>
- Save Babies A non-profit organization designed to inform parents about newborn genetic diseases diagnosed through newborn screening. <http://www.savebabies.org/>
- Cares Foundation Research, education, and support for CAH. <http://www.caresfoundation.org>
- Online Mendelian Inheritance in Man (OMIM). <http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>
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11. Parent Resources (Parent Brochure)



To download The Newborn Screening Program in the Emirate of Abu Dhabi 2009, please visit the Health Authority of Abu Dhabi website: <http://www.haad.ae>

Newborn Screening Program in the Emirate of Abu Dhabi 2009

Appendix 1 Parent Education Fact Sheet

Phenylketonuria (PKU)

What is phenylketonuria?

Phenylketonuria, called PKU for short, causes the body to have problems breaking down certain proteins. People with PKU cannot break down part of the protein called phenylalanine. The body usually breaks down extra phenylalanine that the body doesn't need. When phenylalanine cannot be broken down, it builds up and becomes toxic to the organs and the brain. Phenylalanine gets broken down by an enzyme (a chemical that does a job in the body). A person with PKU is missing an enzyme, or the enzyme is not working as well as it should. A special diet low in phenylalanine can prevent most symptoms of PKU.

What causes PKU?

PKU is inherited when both parents pass an abnormal PKU gene to their child. This means both parents are carriers of PKU. Carriers do not experience any health problems related to PKU. When two carriers have children together, there is a one in four (25%) chance for each child to have PKU.

How is PKU detected?

Newborn screening is done on tiny samples of blood taken from the infant's heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has PKU.

What problems can PKU cause?

Untreated PKU can cause health concerns, behavior problems, and mental retardation. People with PKU who do not eat phenylalanine have normal growth and development.

What is the treatment for PKU?

PKU can be treated. People with PKU should not eat phenylalanine and should drink a special formula. The diet is monitored to make sure that very small amounts of phenylalanine are eaten. The treatment is life-long.

Galactosemia

What is galactosemia?

A person who has galactosemia cannot break down galactose (a sugar found in milk and milk products). The body breaks down galactose into energy it can use. People with galactosemia are unable to break down galactose, and it builds up in the body. When too much galactose builds up, it becomes toxic to organs and the brain. A special diet can prevent most problems.

What causes galactosemia?

Galactosemia is inherited when both parents pass an abnormal galactosemia gene to their child. This means both parents are carriers of galactosemia. Carriers do not experience any health problems related to galactosemia. When two carriers have children together there is a one in four (25%) chance for each child to have galactosemia.

How is galactosemia detected?

Newborn screening test is done on tiny samples of blood taken from the infant's heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has galactosemia.

What problems can Galactosemia cause?

Galactosemia is different for each child. People treated for galactosemia may still have problems with speech, language, hearing, finemotor coordination, eyes, stunted growth, tremors, reproduction, and learning disabilities.

What is the treatment for galactosemia?

Galactosemia can be treated. People with galactosemia cannot eat galactose and lactose. All milk and foods that have milk in them must be avoided. This includes cow's milk, goat's milk, human breast milk, and dairy products like butter, cheese, and yogurt. Other foods that have small amounts of milk products in other forms such as whey, casein, and curds should not be eaten. The treatment is life-long.

Biotinidase (BIO)

What is biotinidase deficiency?

Biotinidase deficiency is a condition that causes the body to have trouble using biotin, an important vitamin the body needs. Biotin is found in many foods. However, a person with biotinidase deficiency needs more biotin than the amount eaten in the normal diet.

Biotinidase is an enzyme (a chemical that causes reactions in the body to take place) that helps to free or separate biotin from foods. Once biotin is free, it can be used by the body. In a person with biotinidase deficiency, the biotinidase enzyme is not working well, and the body does not have enough free biotin to use.

What causes biotinidase deficiency?

Biotinidase deficiency is inherited when both parents pass an abnormal biotinidase gene to their child. This means both parents are carriers of biotinidase deficiency. Carriers do not experience any health problems related to biotinidase deficiency. When two carriers of biotinidase deficiency have children together, there is a one in four (25%) chance for each child to have biotinidase deficiency.

How is biotinidase deficiency detected?

Newborn screening is done on tiny samples of blood taken from the infant's heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has biotinidase deficiency.

What problems can Biotinidase deficiency cause?

Biotinidase deficiency is different for each child. Untreated biotinidase deficiency can cause seizures, low muscle tone (floppiness), hearing loss, eye problems, hair loss, skin rashes, and possibly coma or death. It is very important to follow the doctor's instructions about caring for a child with Biotinidase deficiency.

What is the treatment for Biotinidase deficiency?

Biotinidase can be treated. A child needs to take extra biotin every day (more than what is normally eaten in the diet) in the form of a pill. The treatment is life-long.

Congenital Hypothyroidism (CH)

What is congenital hypothyroidism?

Congenital hypothyroidism, called CH for short, is affects the body's ability to make thyroid hormone. In children with CH, the thyroid gland (a small gland found in the front of the neck under the voice box) does not make enough thyroid hormone. Thyroid hormone acts like a chemical messenger to help control the body's metabolism (how the body makes and uses energy) and calcium balance.

What causes CH?

CH usually happens when the thyroid gland does not develop properly. CH is not usually inherited (passed through families).

How is CH detected?

Newborn screening is done on tiny samples of blood taken from the infant's heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has CH.

What problems can CH cause?

Untreated CH causes feeding problems, sleepiness, constipation, jaundice (skin looks yellow), mental retardation, and developmental delay. There is a chance that children with CH will have hearing problems. It is very important to follow the doctor's instructions about caring for a child with CH.

What is the treatment for CH?

CH can be treated. People with CH need daily medication. The medication replaces the missing thyroid hormone. The treatment is life-long.

Congenital Adrenal Hyperplasia (CAH)

What is congenital adrenal hyperplasia?

Congenital adrenal hyperplasia, or CAH for short, affects the way the body makes chemicals called hormones. These hormones are like little messengers that tell the body what to do. In children with CAH, the adrenal glands (2 walnut-sized glands that sit on top of the kidneys) do not make enough cortisol (also known as the “stress” hormone). Without enough cortisol, the body makes too much male hormone. This can cause growth and development problems. Some children also have a more serious problem called “saltwasting” crises. This happens when the body has trouble keeping the right balance of salt.

What causes CAH?

CAH is inherited when both parents pass an abnormal CAH gene to their child. This means both parents are carriers of CAH. Carriers do not experience any health problems related to CAH. When two carriers of CAH have children together, there is a 1 in 4 (25%) chance for each baby to have CAH.

How is CAH detected?

Newborn screening is done on tiny samples of blood taken from the infant’s heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has CAH.

What problems can CAH cause?

CAH is different for each child. Some female infants have an abnormality of their genitals caused by too much male hormone. Male infants often look normal at birth. If CAH is not treated, it can cause “salt-wasting” and electrolyte problems (keeping the right balance of salts in the body), dehydration (not having enough fluid in the body), heart problems, and even death. It is very important to follow the doctor’s instructions about caring for a child with CAH.

What is the treatment for CAH?

CAH can be treated. People with CAH must take daily medication. The medicine replaces the missing cortisol and sometimes other hormones that the body is also lacking. The treatment is life-long. Some girls with genital problems may need surgery.

Sickle Cell Disease and other Hemoglobinoathies

What is sickle cell disease?

Sickle cell disease affects red blood cells. Red blood cells help carry oxygen throughout the body. When a person has sickle cell disease, the red blood cells can become sickle shaped (instead of donut shaped). The sickled red blood cells can block the vessels through which blood flows. This blockage prevents blood and oxygen from reaching certain parts of the body. This blockage can cause a lot of pain and damage organs. The change from round to sickled blood cells (also called a “sickle cell crisis”) can be caused by a person becoming dehydrated (not having enough fluid in the body), getting sick or being exposed to extreme cold without protective clothing such as hats and gloves in the winter. Sometimes there is no clear cause for a sickle cell “crisis.” The red cells of a person with sickle cell disease do not live as long as the regular, round red blood cells. This leaves fewer red blood cells in the blood. Having fewer red blood cells in the blood is called “anemia.”

What causes sickle cell disease?

Sickle cell disease is inherited when both parents pass an abnormal hemoglobin gene to their child. This means both parents are carriers of certain type of hemoglobin (usually hemoglobin S) and have sickle cell trait. People with sickle cell trait do not have health problems related to sickle cell disease. When two carriers have children together, there is a one in four (25%) chance for each child to have sickle cell disease.

How is sickle cell disease detected?

Newborn screening is done on tiny sample of blood taken from the infant’s heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has sickle cell disease. This testing also helps figure out the type of sickle cell disease the child has.

What problems can sickle cell disease cause?

People with sickle cell disease can have anemia (not enough red blood cells), jaundice (skin becomes yellow), and pain episodes or “sickle cell crises.” Children with sickle cell disease may get more infections because their spleen is damaged by sickle cells (one of the spleen’s main jobs is to protect against infection). Common childhood infections can be life threatening for children with sickle cell disease if not treated right away.

What is the treatment for sickle cell disease?

Sickle cell disease can be treated. A child should take antibiotics to prevent infection and medicines to help control pain. It is also important to prevent dehydration and illness. Children with sickle cell disease should have all immunizations.

Glucose 6 Phosphate Dehydrogenase Deficiency (G6PD)

What is Glucose 6 Phosphate Dehydrogenase Deficiency (G6PD)?

Glucose-6-phosphate dehydrogenase (G6PD) is an important protein (enzyme) that helps blood cells use sugars to produce energy. Normal amounts of G6PD protect red blood cells from certain drugs or chemicals.

G6PD deficiency is an abnormally low level of the G6PD protein in the blood cells. Some people refer to this condition as “favism,” because eating fava beans can trigger symptoms in a person with G6PD deficiency. In people with this deficiency, exposure to certain drugs, foods, and chemicals can result in damage or destruction of red blood cells. Destruction of red blood cells can cause serious illness.

What causes G6PD?

This condition is usually inherited, primarily affects males, and is more common in people of African, Asian, or Mediterranean descent.

How is G6PD detected?

Newborn screening is done on tiny sample of blood taken from the infant’s heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has G6PD.

What problems can G6PD cause?

Most forms of G6PD deficiency are mild and do not cause symptoms. Symptoms may occur in a G6PD deficient person who has been ill, inhaled mothball vapors, eaten fava beans, or taken certain medications such as sulfa antibiotics or drugs used to treat malaria. Persons with a more severe type of G6PD deficiency, which is common in persons of Mediterranean descent, may develop symptoms without exposure.

Symptoms result from anemia caused by the destruction of red blood cells as a result of exposure. An affected person may feel very tired, short of breath, or suffer a number of other symptoms such as a yellowing of the skin called jaundice. If you have G6PD deficiency and experience these symptoms, consult a health care provider as soon as possible.

How can G6PD be managed?

Persons known to be G6PD deficient are prohibited from taking certain drugs or eating fava beans. If a severe episode of hemolytic anemia occurs, treatment may include blood transfusion or nasal oxygen.

Cystic Fibrosis (CF)

What is cystic fibrosis?

Cystic fibrosis, or CF, affects breathing and digestion (breaking down food). CF causes the body to make thick, sticky mucus that clogs the airways of the lungs, and it can prevent the pancreas from doing its job to help digest food. In people with CF, the sweat glands also make very salty sweat.

What causes CF?

CF is inherited when both parents carry an abnormal CF gene and pass it to their child. A person who has one abnormal CF gene is called a “carrier.” Carriers do not usually have any health problems caused by CF. When two CF carriers have children together, each baby has a one in four (25%) chance of having CF. Most kids with CF do not have a family history of the disease.

What problems can CF cause?

CF is different for each child. Symptoms may include coughing or wheezing, lung infections, too much mucus, trouble gaining weight and growing, and greasy stools. It is very important to follow the doctor’s instructions. Children with CF need to see the doctor more often than kids without CF.

What is the treatment for CF?

Although CF cannot be cured, many things can be done to treat the symptoms. People with CF must eat a healthy, high-calorie diet and take vitamins to help them grow. Some people must also take medications to get more nutrients from the food they eat. To breathe better, many people with CF need help clearing mucus from their lungs each day. Some medications can also prevent infections and help with breathing.

Hearing Loss

Infant hearing loss

Most babies who are born with a hearing loss have parents who have normal hearing and have had no experience with hearing loss. Identifying hearing loss early is important. About 33 percent of children with a hearing loss have a second disability. Parents who have an infant who is deaf may need help to find resources and connect their family to services.

Detecting hearing loss in infants

Hearing loss in babies may be found through infant hearing tests done in the hospital shortly after birth. If a baby does not pass the hearing test, another test should be done. If the baby does not pass the second time, the baby should see a hearing specialist, called an Audiologist, who can help determine if the baby really has a hearing loss, the type of loss, how much of a loss, and what the family and infant might need.

Causes of infant hearing loss

There are many things that cause hearing loss in a baby. Doctors may suggest special tests and evaluations to help figure out why a newborn is deaf:

- Hearing loss can be inherited (passed through a family). About 50 percent of the infants with a hearing loss inherited it from their families. Hearing loss can be genetic even if there are no other family members who were born deaf
- Hearing loss can be caused by certain infections, medications, or problems with birth and delivery.
- In some cases, the cause of the hearing loss may not be known.

Problems for infants and children

- A child with a hearing loss may miss important learning experiences.
- A child who is deaf may have difficulty with speech and language.
- A child may have social, educational, or emotional problems if help for the hearing loss is delayed.

Special needs

Fortunately, there is help for infants and children with hearing loss. Every child is special. By working together, parents, doctors, and special educators find the best solutions for infants with special needs and their families. When hearing loss is treated early, children who are deaf can usually do as well as children with normal hearing:

- Hearing aids may increase the sounds a baby is able to hear.
- Audiologists may fit infants for hearing aids
- Early communication is important.
- Families will need information and help to make important decisions like how to best talk and play with their deaf babies.

Early intervention and education

For children with hearing loss, early intervention and education are important to development and should begin by six months of age.

Support Resources

To join the Newborn Screening Medical support group, Please contact:

The Newborn Screening Program in the Emirate of Abu Dhabi.

Phone +971 2 4193 543

Email mshehhi@haad.ae

Fax +9712 4444 285

Medical Specialists Information

Dr. Azzam Alzoebie, MD, FAAP

Division Head, Pediatric Hematology & Oncology

Shaikh Khalifa Medical Center, Abu Dhabi, United Arab Emirates.

Phone: + 971 50 810 0315

zoeazz741@skmc.gov.ae

Dr. Fatma Al-Jasmi, MBBS, FRCPC, FCCMG

Metabolic Geneticist, Tawam Hospital, Al Ain.

Assistant Professor of Pediatric, Department of Pediatric,

Faculty of Medicine and Health Science, UAE University, Al Ain, UAE.

Phone; +97150 55 99 425

aljasmi@uaeu.ac.ae

Dr. Jozef Hertecant

Consultant Pediatrician, Tawam Hospital, Al Ain.

Clinical Assistant Professor of Pediatric, Faculty of Medicine and

Health Science, UAE University, Al Ain, UAE.

Referral physician for the National PKU Screening Program

Phone: +971 3 767 5120

jhertecant@tawamhospital.ae

Dr. NizarKherallah, M.D., F.C.C.P.

Consultant, Pediatric Pulmonologist.

Phone: + 9 7 1 2 6 1 0 2 0 0 0

nkheralla@skmc.gov.ae

Dr. Yousef M Abdulrazzaq

Professor of Pediatrics and Neonatology

Department of Pediatrics, Tawam Hospital, Al Ain.

Faculty of Medicine and Health Sciences, UAE University, Al Ain.

Phone: +971 3 703 9428

yousef@uaeu.ac.ae

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