

CONGENITAL HYPOTHYROIDISM

CONGENITAL HYPOTHYROIDISM

Congenital hypothyroidism is the lack of adequate amounts of functional thyroid hormone in the newborn period. Thyroid hormone is important in many metabolic functions and is essential for normal growth and development. Congenital hypothyroidism is one of the most common conditions detected by newborn screening, with an incidence rate of 1:5,000.

Clinical Features in Children With Untreated Hypothyroidism

Untreated congenital hypothyroidism results in a child with profound mental retardation, delayed growth and a complex of characteristic features (cretinism). Affected infants with untreated congenital hypothyroidism may appear relatively normal for several months of age though serious irreversible damage may be occurring. In the absence of a universal screening program, diagnosis of Congenital Hypothyroidism before age 2-3 months is rare.

Clinical symptoms or signs of untreated Congenital Hypothyroidism may include prolonged neonatal jaundice, constipation, lethargy, poor muscle tone, feeding problems, a large tongue, puffy face, large fontanel, distended abdomen, umbilical hernia, and hypothermia. These signs and symptoms are not reliable indicators of congenital hypothyroidism. Laboratory test results are the only reliable means of diagnosing congenital hypothyroidism in the newborn infant.

Causes of Congenital Hypothyroidism

The most common causes are total or partial failure of development of the thyroid gland or its development in an abnormal location (an ectopic gland). Less commonly, congenital hypothyroidism results from damage to fetal thyroid by medications (anti-thyroid drugs or excess iodine) used by the mother during pregnancy, or results from primary failure of the hypothalamic- pituitary axis with the pituitary failing to produce adequate amounts of thyroid stimulating hormone (TSH). In some cases congenital hypothyroidism results from a genetic defect in thyroid hormone synthesis.

Laboratory Screening Tests

The Kansas State Laboratory tests for TSH levels in all newborns. See Table 3 for normal values and laboratory criteria for requesting repeat samples.

TABLE 3
Normal Values and Laboratory Criteria for Requesting Repeat Samples

ANALYTE	NORMAL RESULTS	RESULTS REQUIRING PHONE FOLLOW-UP	RESULTS REQUIRING MAIL FOLLOW-UP
TSH	< 20.0µIU/mL	≥60.0µIU/ml	≥ 20.0 - < 60.0µIU/ml

All phoned results are followed by faxed or mailed confirmation. All tests are screening tests. Abnormal results need full evaluation before a diagnosis is confirmed.

Confirmatory Testing

When the infant's primary care provider is notified that the screening results are ≥60.0µIU/ml, a referral to a genetic consultant/pediatric endocrinologist should be made. In cases in which the TSH is unequivocally elevated, treatment can be started as soon as the serum is obtained, pending final confirmation. For borderline screening results, ≥ 20.0 - < 60.0µIU/ml, a repeat newborn screen or TSH level is recommended.

Treatment

Treatment of congenital hypothyroidism is relatively simple and effective in most children. Thyroxine is administered (Synthroid or Levothyroid), in pill form, is crushed, and administered once daily. Generic thyroid preparations should not be prescribed. Because some children present certain complex issues, evaluation and treatment are best handled in consultation with a genetic consultant/Pediatric Endocrinologist. T4 and TSH levels need to be monitored as the infant grows. As part of comprehensive care, children should also have periodic developmental testing. If treatment is started early and maintained, development can be expected to be normal.

Screening Practice Considerations

Detection of hypothyroidism does **NOT** depend on protein, lactose ingestion or other nutritional factors. Eighty-five to ninety percent of infants with hypothyroidism are detected on the 1st specimen even if it is collected a few hours after birth. For this reason, it is important to obtain a screening specimen on every infant prior to discharge from the hospital or birth center. **Practitioners must remain alert to clinical symptoms in older infants despite normal initial screening. If a practitioner clinically suspects hypothyroidism, he/she should arrange for testing regardless of the results of the newborn screen.**

False positive TSH results may occur in infants when the specimen is collected within the first twenty four hours after birth, when there is a physiologic rise of TSH that is transient.

Prompt confirmatory testing is required even if there is evidence to suggest that one of the situations associated with false positive screens is present (e.g., early specimen collection or prematurity). The presence of any of these does not exclude the possibility of disease.

A small percentage of cases of congenital hypothyroidism do not develop until after the first weeks of life. Therefore, as with other screening tests, in the presence of clinical symptoms, evaluation for congenital hypothyroidism should be performed despite normal newborn screening results.

Medical Consultants for Children with Congenital Hypothyroidism

Medical consultants are available to provide consultation for the follow-up, evaluation, and long-term management of children with Congenital Hypothyroidism through the State of Kansas.

Date: 7/2008

Dr. James Casey
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Kenneth Dykstra
Wichita Clinic
Wichita, KS
Office: 316-689-9989

It is strongly recommended that prior to repeating the newborn screen practitioners should confer with one of the consultants. The consultant may recommend repeating the State Newborn Screen, or they may suggest labs to draw and analyze in a practitioner's local lab. The consultant may wish to see the newborn in the office and do the lab work and assessment there. Whatever the case, we want to prevent unnecessary lab draws or inappropriate testing on these infants. In summary, please contact the consultant in your vicinity prior to drawing the repeat blood work.

Overview of Follow-up Procedure Abnormal Newborn Screening for Congenital Hypothyroidism

- 1) Newborn Screening follow-up team reviews the laboratory reports that are faxed overnight from the lab or records the information provided per telephone call from the laboratory on a white phone information sheet.
- 2) If the TSH result is $< 20 \mu\text{IU/ml}$: the results are considered **normal**.
 - a) Follow-up team does not receive results.
 - b) Lab will fax or mail results to doctor listed on NBS card.
- 3) If the TSH result is $\geq 20 \mu\text{IU/ml}$ and $< 60 \mu\text{IU/ml}$, the results are considered **borderline**.
 - a) Lab will contact follow-up team via nightly fax with baby's information and test results.
 - b) Follow-up team will print CH borderline letter and mail to healthcare provider.
 - c) Follow-up team will print CH parent letter and CH parent information sheet and mail to baby's parents to inform them that their child has an abnormal result. NOTE: If infant is in the NICU, no parent letter is sent.
 - d) Lab will fax or mail results to doctor listed on NBS card.
 - e) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 1 month from date of letter. NOTE: Name changes are documented on the copy of the lab report. Surname changes are also documented in WebIZ as an alias.
 - f) When complete, paperwork is filled by infant's date of birth.
- 4) If the TSH result is $\geq 60 \mu\text{IU/ml}$, the results are considered **presumptive**.
 - a) Lab will contact follow-up team via phone with baby's information and test results. Follow-up team will document information on white phone slip.
 - b) Follow-up team will print out baby's information from DHEL database on a green sheet of paper, attach the white phone slip and write "CH" and the result on green sheet.
 - c) Follow-up team will enter data into Access database under TSH.
 - d) Follow-up team will call healthcare provider listed on report and:
 - i) Verify that they are seeing the baby.
 - ii) Inform them of results.
 - iii) Ask them to notify parents and arrange appointment with specialist.
 - iv) Confirm doctor's fax number.
 - v) Inform them that a letter will be faxed to their office with the results and instructions.
 - e) Follow-up team will print CH presumptive letter and CH physician report form and fax (or mail, if no fax) to healthcare provider.
 - f) Follow-up team will print CH presumptive letter and CH physician report form and fax (or mail, if no fax) to healthcare provider.
 - g) Follow-up team will print CH parent letter and CH parent information sheet and mail to baby's parents to inform them that their child has an abnormal result. NOTE: If infant is in the NICU, no parent letter is sent.

- h) Follow-up team will enter data into Excel spreadsheet "Presumptive Totals" located on the "H" drive.
- i) Lab will fax or mail results to doctor listed on NBS card.
- j) Lab will fax results to follow-up team. Report is attached to green sheet.
- k) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 1 month from date of letter. NOTE: Name changes are documented on the lab report. Surname changes are also documented in WebIZ as an alias.
- l) When complete, paperwork is filled by infant's date of birth.



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED TSH

CONDITION: CONGENITAL HYPOTHYROIDISM (CH)

DIFFERENTIAL DIAGNOSIS: Primary congenital hypothyroidism; secondary congenital hypothyroidism; transient CH; thyroxine binding globulin (TBG) deficiency.

METABOLIC DESCRIPTION: Lack of adequate thyroid hormone production.

ACTION TO BE TAKEN IMMEDIATELY:

- Contact family immediately to inform them of the newborn screening test results.
- Consult pediatric endocrinologist; referral to endocrinologist if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Diagnostic tests should include serum **free T4** and **thyroid stimulating hormone (TSH)**; consultant may also recommend **total T4 and T3 resin uptake**. Test results include **reduced free T4 and elevated TSH** in primary hypothyroidism. **TSH is reduced or inappropriately normal** in secondary (hypopituitary) hypothyroidism. **Low total T4 and elevated T3 resin uptake** are consistent with TBG deficiency.

CLINICAL EXPECTATIONS: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanel, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

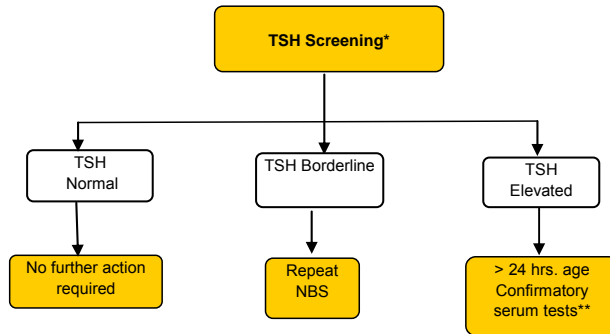
Pediatric Endocrinology
KU Medical Center
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Dr. Kenneth Dykstra
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TSH (CONGENITAL HYPOTHYROIDISM)



Abbreviations/Key

NBS = Newborn screening
TSH = Thyroid stimulating hormone

**** = Confirmatory Serum Tests**

Free T4 (or)
Total T4 and T3 resin uptake (T3RU)
TSH

***NOTE:**

Baby must be > 24 hours old when sample is taken for TSH results to be valid.

TSH Results

TSH Normal = < 20 μ IU/mL
TSH Borderline = 20-59.9 μ IU/mL
TSH Elevated = \geq 60.0 μ IU/mL

Action steps are shown in gold (shaded) boxes; results are in plain boxes.

DISCLAIMER: These algorithms and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines

EXAMPLE OF BORDERLINE CONGENITAL HYPOTHYROIDISM PHYSICIAN LETTER



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

**Abnormal TSH Lab Report
Kansas Newborn Screening Program**

Date

Doctor's Name
Address Line 1
Address Line 2

RE: Baby's Name
DOB: xx/xx/xxxx

MOTHER'S NAME: Mother's Name
MOTHER'S PHONE: xxx-xxx-xxxx

Specimen date: xx/xx/xxxx

TSH: Result μ IU/mL Expected value: < 20.0 μ IU/mL

The newborn screening result above indicates a **BORDERLINE** result because hypothyroidism cannot be ruled out. A slightly elevated TSH level between 20 – 59.9 μ IU/ml can be transient, particularly if the specimen was collected within the first 24 hours of life, or an early sign of congenital hypothyroidism.

RECOMMENDATION:

Repeat the test within 2 to 3 days of receiving this notice, and send the filter paper specimen to the State lab. (If transfused, repeat filter paper on day 4 after transfusion.) If repeat testing is performed at an outside lab, please fax results to 785-296-2950 as we are unable to access those results.

In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or birth attendant to obtain repeat specimens when needed to complete the screening process.

Consultation is available from:

Pediatric Endocrinology
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Kenneth Dykstra
Wichita Clinic
Wichita, KS
Office: 316-689-9989

Additional information is available on the Kansas Newborn Screening Website at:
http://www.kdheks.gov/newborn_screening/info_professionals.htm.

You may contact the Newborn Screening Program at (785) 291-3363 or 1-800-332-6262 if you have any questions or concerns.

EXAMPLE OF PRESUMPTIVE CONGENITAL HYPOTHYROIDISM LETTER TO PHYSICIANS



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

**Abnormal Thyroid Activity (CH) Lab Report
Kansas Newborn Screening Program**

Date

Doctor's Name
Address Line 1
Address Line 2

RE: Baby's Name
DOB: xx/xx/xxxx

MOTHER'S NAME: Mother's Name
MOTHER'S PHONE: xxx-xxx-xxxx

Specimen date: xx/xx/xxxx

TSH: Result Expected value: < 20.0µIU/mL

The newborn screening result above is highly suggestive of congenital hypothyroidism. Untreated congenital hypothyroidism leads to developmental delay or mental retardation and poor growth.

The final newborn screening lab report will be sent when all testing is completed.

RECOMMENDATION:

Immediate consultation with one of the consultants listed below is essential for diagnostic testing and genetic counseling. Please call to arrange this **urgent** appointment.

In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.

Consultation is available from:

Pediatric Endocrinology
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Kenneth Dykstra
Wichita Clinic
Wichita, KS
Office: 316-689-9989

****PLEASE COMPLETE AND RETURN THE ENCLOSED PHYSICIAN REPORTING FORM WHEN FOLLOW UP IS COMPLETE****

Additional information is available on the Kansas Newborn Screening Website at:
http://www.kdheks.gov/newborn_screening/info_professionals.htm.

You may contact the Newborn Screening Program at (785) 291-3363 or 1-800-332-6262 if you have any questions or concerns.

EXAMPLE OF CONGENITAL HYPOTHYROIDISM (CH) PHYSICIAN'S REPORT FORM



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

**CONGENITAL HYPOTHYROIDISM (CH) NEWBORN SCREENING
PHYSICIAN REPORTING FORM**

****Return this form When Follow-Up is Complete ****

Date

Doctor's Name
Address Line 1
Address Line 2

If not a current patient of this practice, record name and contact information for Primary Care Physician and return form.

RE: Baby's Name
DOB: xx/xx/xxxx

Baby's name if different than listed

DIAGNOSIS EXCLUDED: Date Excluded: _____

Baby does **NOT** have Congenital Hypothyroidism

DIAGNOSIS CONFIRMED: Date Diagnosis Confirmed: _____

Baby has Congenital Hypothyroidism

Lab Results: (please fill in and attach copy of specialist's report)

TSH: _____ Free T4: _____

Additional lab results: _____

Date treatment began: _____

Baby has Other Hypothyroidism

Please specify (secondary, transient, other): _____

Date treatment began, if indicated: _____

FORM CONTINUES ON BACK

Kansas Newborn Screening Program



Congenital Hypothyroidism Information for Health Professionals

Congenital hypothyroidism (CH) is inadequate thyroid hormone production in newborn infants. This can occur because of an anatomic defect in the gland, an inborn error of thyroid metabolism, or iodine deficiency. There are several types of primary CH, the most common form resulting from abnormal fetal development of the thyroid gland. The thyroid gland may be absent, ectopic, or malformed. Transient hypothyroidism may occur in some infants as a result of maternal exposure to excess iodine, antithyroid medications (propylthiouracil or PTU), or exposure of the infant to maternal antithyroid antibodies. The use of iodine-based skin disinfectants on neonates, especially premature neonates, can inhibit thyroxine production resulting in transient hypothyroidism. Untreated maternal hypothyroidism also can result in low fetal levels of thyroxine.

✓ **Clinical Symptoms**

Most newborns do not have symptoms. Subtle clinical signs, which usually present after the newborn period, of CH include the following symptoms: feeding problems, lethargy, prolonged postnatal jaundice, delayed stooling and constipation, enlarged protruding tongue, hoarse cry, protruding abdomen with an umbilical hernia, cold mottled skin, sluggish reflexes, patent posterior fontanelle with widely spread cranial sutures or delayed skeletal maturation for gestational age. If untreated, thyroxine deficiency causes irreversible mental and physical retardation

✓ **Incidence**

CH occurs in 1 out of every 5,000 births. The incidence of CH is higher in Hispanic individuals and lower in black individuals. There is a 2:1 incidence in females compared with males, and there is an increased risk in infants with Down syndrome. Incidence is believed to be still greater among Native American and Asian populations.

✓ **Genetics of congenital hypothyroidism**

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.

✓ **How do people inherit congenital hypothyroidism?**

Most cases of congenital hypothyroidism are sporadic and 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. 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.

✓ **Treatment**

Immediate diagnosis and treatment of congenital hypothyroidism in the neonatal period is critical to normal brain development and physical growth. Treatment is usually effective if started within the first few weeks of life. Delayed treatment may result in decreased intellectual capacity. Recommended treatment is lifetime daily administration of levo-thyroxine. Dosage will need to be gradually increased as the infant grows. Avoid generic thyroid preparations, at least in the first three years of life.

✓ **Screening Methodology**

Primary newborn screening for CH utilizes fluorometric assay to determine the thyroid stimulating hormone level. False positive and false negative results are possible with this screening.

✓ **What to do After Receiving Presumptive Positive CH Screening Results**

- Seriously elevated TSH requires prompt follow-up and, when notified of these results, the clinician should immediately check on the clinical status of the baby.
- Refer the infant to a pediatric endocrinologist.
- Collection of serum TSH and free T4 level also is recommended.
- Suspect abnormal results (moderately elevated TSH) indicate the need for repeat filter paper screening.
- Call KS Newborn Screening Program at 785-291-3363 with questions about results
- Report Clinical Findings to Newborn Screening Program at 785-291-3363
- Same birth siblings (twins, triplets) of infants diagnosed with CH should be re-screened; additional testing of these siblings also may be indicated.

✓ **Confirmation of Diagnosis**

The diagnosis is confirmed with a repeat measurement of TSH and free thyroxine using a blood sample. Further tests can include thyroid scans and x-rays. Diagnostic testing is arranged by a specialist at your regional treatment center.

✓ **Communication of Results to Parents**

If a baby has a presumptive positive congenital hypothyroidism newborn screening result, additional testing needs to be performed to confirm a diagnosis. In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.

If a baby is diagnosed with CH, the following points should be conveyed to parents:

- ***Parents should understand that treatment for primary congenital hypothyroidism will be lifelong.***
- ***Parents should understand that treatment is not curative and that all morbidity cannot necessarily be prevented. Long-term management, monitoring and compliance with treatment recommendations are essential to the child's well-being. A multidisciplinary approach is recommended and should include the following specialties: pediatrics and endocrinology. Infants and children with congenital hypothyroidism should have regular follow-up appointments with a pediatric endocrinologist. Periodic hearing evaluations also are recommended for children with CH, as hearing disorders are sometimes associated with congenital hypothyroidism.***
- ***Genetic counseling services may be indicated. A list of counselors and geneticists, whose services are available in Kansas should be given to the parents if they have not already seen a geneticist.***

For consultation, contact:

Pediatric Endocrinology
KU Medical Center
Kansas City
Office: 913-588-6326

Dr. Majed Dasouki
KU Medical Center
Kansas City
Office: 913-588-6326

Dr. Kenneth Dykstra
Wichita Clinic
Wichita, KS
Office: 316-689-9989

07/29/09

EXAMPLE OF PARENT LETTER FOR CONGENITAL HYPOTHYROIDISM



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

Date

Mother's Name
Address Line 1
Address Line 2

RE: Baby's Name DOB: xx/xx/xxxx

Dear Parent:

Best wishes on the birth of your baby! Shortly after your baby was born, a small blood sample was taken for a test called the Newborn Screen. This test helps parents find out if their baby has certain health problems. A baby can look healthy, but may have a harmful illness that can be found by doing this blood test.

The result of your baby's blood test shows that more testing needs to be done for congenital hypothyroidism. ***This does not necessarily mean your child is ill.***

THIS IS WHAT YOU NEED TO DO NOW:

1. Call your baby's doctor. Say that you have received a letter stating that your baby's Newborn Screen test was not normal. Set up a time for your baby to have a second test done as soon as you can.
2. We have < Doctor's Name > listed as your baby's doctor, and we have notified him/her of your baby's test result. If this is NOT your baby's doctor, please call the Newborn Screening Program at 785-296-0109 so we can contact the right doctor.

DO NOT DELAY. YOUR BABY'S HEALTH DEPENDS ON YOU.

If your baby does not have a doctor, or if you have questions about this letter, please call Kansas Newborn Screening at 785-296-0109.

Sincerely,

A handwritten signature in cursive script that reads "Jamey Kendall".

Jamey Kendall BSN, RN
Kansas Newborn Screening
Follow-up Coordinator
Jamey Kendall BSN, RN
Newborn Screening Program Coordinator

A handwritten signature in cursive script that reads "Linda A. Williams".

Linda A. Williams, MT(ASCP)
Kansas Newborn Screening
Follow-up Coordinator



Congenital Hypothyroidism Information for Parents

➤ Overview

Congenital hypothyroidism (CH) is a condition that affects infants from birth (congenital) and results from a partial or complete loss of thyroid function (hypothyroidism).

➤ What is congenital hypothyroidism?

Congenital hypothyroidism occurs when the thyroid gland fails to develop or function properly. The thyroid gland is a butterfly-shaped gland in the lower neck. It makes hormones that are important for regulating growth, brain development, and metabolism (chemical reactions in the body).

In 80 to 85 percent of all babies with congenital hypothyroidism, the thyroid gland is absent, abnormally located, or too small. In the remaining 15-20% of cases, the thyroid gland looks normal, but it does not produce any or enough hormones. If untreated, congenital hypothyroidism can lead to developmental delay or mental retardation and poor growth. If treatment begins within the first month after birth, infants usually develop normally.

➤ Why is newborn screening done for congenital hypothyroidism?

Newborn screening is done for congenital hypothyroidism so that babies with this condition can be diagnosed and treated quickly. Immediate diagnosis and treatment of congenital hypothyroidism is important for normal brain development and physical growth. Without prompt diagnosis and treatment, infants with congenital hypothyroidism will develop varying degrees of mental retardation and abnormal growth.

➤ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has congenital hypothyroidism?

No, not necessarily. Newborn screening tests the baby's level of thyroid-stimulating hormone (TSH) but additional tests will need to be done to determine if the baby has congenital hypothyroidism or not.

➤ How common is congenital hypothyroidism?

Congenital hypothyroidism affects 1 in 5,000 infants and is more common in girls. It is more common in Hispanics than in Caucasians and even less common in African Americans.

➤ Is congenital hypothyroidism inherited?

Most cases of congenital hypothyroidism are sporadic, which means that they occur in people without a history of the disorder in their family. A few genes responsible for sporadic congenital hypothyroidism have been identified.

An estimated 15-20% of cases are inherited. Many of these cases are autosomal recessive. This means that both parents of the affected child are carriers of the condition, but they do not have the disease. Each pregnancy between carrier parents has a 25% chance of producing a child affected with congenital hypothyroidism, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier. Some inherited cases have an

autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Multiple genes are associated with congenital hypothyroidism and there may be other associated genes that have yet to be well characterized. Such defective genes prevent the thyroid gland from producing normal amounts of thyroid hormones.

➤ **What are the signs and symptoms of congenital hypothyroidism?**

Most infants with congenital hypothyroidism appear normal at birth. Some babies have subtle symptoms, but many of these symptoms are also found in babies who do not have congenital hypothyroidism. Symptoms may include feeding problems, prolonged jaundice, constipation, an enlarged or protruding tongue, hoarse cry, protruding abdomen with umbilical hernia, and sluggish reflexes. Without treatment, individuals with congenital hypothyroidism have poor growth and developmental delay or mental retardation.

➤ **How is congenital hypothyroidism diagnosed?**

False positive and false negative results are possible with this screening. If TSH is elevated or an infant is considered “high risk,” additional confirmatory blood tests will be ordered to test for thyroid function. Diagnostic testing includes looking for reduced free T4 and elevated TSH. Additional thyroid hormone tests may be ordered.

➤ **Is there a cure for congenital hypothyroidism?**

No, there is no cure for congenital hypothyroidism. However, the outcome is usually excellent in infants who are treated early and continue with lifelong treatment.

➤ **How is congenital hypothyroidism treated?**

- Lifelong treatment with oral thyroxine is required to ensure normal growth and development. Individuals with CH need to be followed closely to ensure adequate thyroid hormone levels and to monitor development.
- Periodic hearing evaluations are recommended as hearing disorders are sometimes associated with CH.
- Infants with CH have an increased incidence of heart defects, which may require treatment.

➤ **Where can I get additional information?**

1. The Thyroid Foundation of America at www.tsh.org
2. Magic Foundation at www.magicfoundation.org
3. GeneTests at <http://www.genetests.org/>

7.17.08