

Diagnostic Verification Survey of Endocrinologists Treating Children Identified by Newborn Screening as Having Congenital Hypothyroidism

Patient Information:

Child's Birth Date	Child's Name		Parent's Name	
	Last	First	Last	First
DD-Mon-YY	LastName	FirstName	LastName	FirstName

- 1.) Are you or your group's practice providing care for the patient listed in the above table [check one]?
 - a. Yes (if yes, proceed to question 2)
 - b. No; **If no, do you know the patient's current health care provider?**
 Name: _____
 Address: _____

 Phone: _____
 Fax: _____

- 2.) What state performed the Newborn Screening for this patient?
 - a. Illinois
 - b. Indiana
 - c. Kentucky
 - d. Michigan
 - e. Ohio
 - f. Wisconsin

- 3.) What additional laboratory studies (besides newborn screen) were used to confirm diagnosis of Congenital Hypothyroidism (CH)? [check all that apply]
 - a. TSH
 - b. Free T4
 - c. Total T4
 - d. Free T3
 - e. Total T3
 - f. Thyrotropin Binding Inhibitory Immunoglobulin (TBII)
 - g. Anti-Thyroid Peroxidase Antibodies
 - h. Maternal Thyroid Function and/or Antibody Testing
 - i. Other

- 4.) What imaging studies were used at CH diagnosis? [check all that apply]
 - a. Thyroid Ultrasound
 - b. Thyroid Technetium Scan
 - c. Thyroid Uptake Scan (I^{123})
 - d. Brain MRI/CT
 - e. None

- 5.) Was the etiology of the patient's hypothyroidism established?
 - a. No (proceed to question #7)
 - b. Yes, at onset or within 6 months of treatment
 - c. Yes, after 6 months but before 3 years of age
 - d. Yes, after 3 years of age

6.) What was the etiology of the hypothyroidism?

- a. Agenesis or Dysgenesis (thyroid gland developmental defect)
- b. Dyshormonogenesis (defects in thyroid hormone synthesis)
- c. Mutations in thyroid development genes or TSH receptor
- d. Defects in hypothalamus or pituitary (central or secondary/tertiary hypothyroidism)
- e. Maternal Iodine deficiency
- f. Other (please specify): _____

7.) Is the patient currently being treated for CH [check one]?

- a. Yes (proceed to question #13)
- b. No

8.) When was the treatment stopped? [check one]

- a. Less than 6 months of age
- b. 6 months – 1 year of age
- c. 1 – 2 years of age
- d. 2 – 3 years of age
- e. Greater than 3 years of age

9.) What is the reason that the treatment was discontinued? [check all that apply]

- a. FT_4 and TSH concentrations remain in reference range off treatment
- b. Parental non-compliance
- c. Lost to follow-up (cannot locate infant)
- d. Other (please specify): _____

10.) If the patient's CH diagnosis was re-evaluated, what testing was used? [check all that apply]

- a. Thyroid challenge was conducted by decreasing/discontinuing the dosage of levothyroxine with follow-up Free T4/T4 and TSH levels
- b. Thyroid ultrasound
- c. Thyroid technetium scan
- d. Thyroid uptake scan (I^{123})
- e. The patient's CH diagnosis has NOT been re-evaluated (proceed to question #13)
- f. Other (specify): _____

11.) What was the patient's CH re-evaluation result? [check one]

- a. Diagnosis Confirmed
- b. Diagnosis Not confirmed

12.) How did you make your decision to re-evaluate the patient's CH diagnosis? [check one]

- a. Re-evaluation is part of my practice protocol
- b. Thyroid function testing during therapy indicated possibility of transient hypothyroidism
- c. Other

13.) If patient's CH diagnosis has not been re-evaluated, do you plan to do so? [check one]

- a. No, diagnosis confirmed to my satisfaction at initial presentation
- b. No, patient's subsequent TSH levels since diagnosis have required dosage changes
- c. No, parental refusal
- d. No, patient lost to follow-up
- e. Yes, within the next 6 months
- f. Yes, within the next 6-12 months
- g. Yes, within the next 1-2 years

14.) If the patient's CH diagnosis will be re-evaluated, what testing will be used? [check all that apply]

- a. Thyroid challenge was conducted by decreasing/discontinuing the dosage of levothyroxine with follow-up Free T4/T4 and TSH levels
- b. Thyroid ultrasound
- c. Thyroid technetium scan
- d. Thyroid uptake scan (I^{123})
- e. The patient's CH diagnosis has NOT been re-evaluated
- f. Other (specify): _____

15.) What is your protocol for thyroid lab testing following treatment cessation? [check all that apply]

- a. 2-4 weeks after discontinuation
- b. 4-8 weeks after discontinuation
- c. 2-3 months after discontinuation
- d. 3-6 months after discontinuation
- e. 6-12 months after discontinuation
- f. 1-2 years after discontinuation
- g. 2-3 years after discontinuation

16.) Who provides genetic counseling to family after diagnosis of CH is made? [check all that apply]

- a. Face-to-face counseling is provided my practice
- b. Printed literature is provided to family
- c. Internet references are provided to family
- d. Patient is referred to Genetics specialist for counseling
- e. No counseling is provided
- f.

17.) What form of education is given to family after diagnosis of CH is made? [check all that apply]

- a. Face-to-face education is provided by my practice
- b. Printed literature is provided to family
- c. Internet references are provided to family
- d. No education is provided

18.) Systematic Notification for CH Challenge: Please indicate the level of benefit to CH patients and your practice if the state newborn screening program systematically contacted you/practice when CH patients reach their 3rd birthday? **[check one]**

- a. Extremely helpful
- b. Somewhat helpful
- c. Not helpful

Thank you very much for you assistance and contribution to our efforts to improve the Newborn Screening Program!!