

Congenital Hypothyroidism Diagnostic Verification at Three Years of Age

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Introduction

- The standard of care for CH is to verify the diagnosis around age three by evaluating thyroid function following cessation of synthroid medication.
 - If TSH levels rise following synthroid cessation, then CH diagnosis is confirmed and treatment is resumed.
 - If TSH levels do not rise following synthroid cessation, the diagnosis is not confirmed and treatment is not resumed.
- However, the CH diagnosis verification is not universally documented meaning treatment potentially continues indefinitely for many false positives
- Thus, we do not understand the rate of false positive CH diagnoses and associated unnecessary treatment

Research Question & Aims

- **Research question:** what is the false positive rate of CH diagnosis among children with borderline thyroid function identified by Michigan's Newborn Screening program (NBS)
- **Global AIM:** To elucidate the rate of false positive CH diagnosis and associated unnecessary treatment and utilize this information to inform diagnostic and treatment standards
- **Procedural AIMS:**
 - **AIM 1:** define borderline thyroid function (inclusion criteria for follow-up)
 - **AIM 2:** identify all children having borderline thyroid function in Michigan identified as having CH by NBS
 - **AIM 3:** contact endocrinologists to conduct trial of thyroid function and record results
 - **AIM 4:** utilize results to refine CH diagnostic and treatment standards

Methods

- **Study design:** cross sectional follow-up study
- **Study population:** children with borderline thyroid function identified by the Michigan Newborn Screening program as having CH aged 3-5 years
- **Study data:** NBS lab & Endocrine Follow-Up Program Clinical data maintained by MDCH NBS Follow-up Program

Summary

- The current standard of care for CH is to verify the diagnosis at age 3; however, this rarely occurs.
- We are proposing a follow-up study to assess the rate of false positive diagnosis and associated unnecessary treatment.
- We are using NBS data to define borderline thyroid function
- The MDCH NBS Follow-up Program contracted Endocrine follow-up program will identify patients and contact endocrinologists to facilitate this study
- Results will be used to inform diagnosis and treatment standards.