Child With Abnormal Newborn Screen for Congenital Hypothyroidism

Suggestive history and physical findings	Initial laboratory and/or radiologic work-up can include:	When to refer	Items useful for consultation	Additional information

Symptoms/signs:
 	
Often, no signs or symptoms, but may see:
• Wide fontanelle
• Lethargy
• Poor suck/poor feeding
• Delayed meconium
• Umbilical hernia
• Macroglossia
• Persistent jaundice
• Hypothermia
• Bradycardia
 	
Differential Diagnosis
	Blood tests:
	• Repeat thyroid function ASAP with a serum sample:
	  ○ TSH
	  ○ Free T4
		Urgent:
		• All cases should be referred to a pediatric endocrinologist immediately in order to initiate treatment
		• Best course of action is to call your local pediatric endocrinologist; often therapy with levothyroxine will be initiated before the first in-person appointment
		• Prompt treatment is critical for developmental outcome
		
		Find a Pediatric Endocrinologist
			Birth history
			
			Maternal medical history/medications taken during pregnancy
				Additional Information
				
				References

Differential diagnosis of congenital hypothyroidism

- Failure of normal thyroid gland development (thyroid dysgenesis)
- Failure of thyroid gland hormonal production (thyroid dyshormonogenesis)
- TSH Deficiency (Central hypothyroidism)
- Iodine deficiency (most common cause worldwide)
- Exposure to maternal medications: amiodarone, methimazole, carbimazole, propylthiouracil, lithium
- Transfer of maternal antibodies that block activation of the TSH receptor
- History of maternal Graves disease may cause neonatal hypo- or hyperthyroidism

Additional Information:

- If newborn screen is obtained before 24h of age, you may see a false positive elevation in TSH due to the physiologically normal TSH surge
- Children with central hypothyroidism (TSH deficiency) may be missed in states where newborn screening measures TSH levels. Consider obtaining serum TSH and Free T4 in infants who exhibit signs or symptoms of hypothyroidism despite a normal TSH on newborn screen.
    - In central hypothyroidism, the infant will have a low Free T4 level with a low or inappropriately normal TSH.
    - Central hypothyroidism should also be suspected in children with mid-line defects and/or other pituitary hormone deficiencies.
- Congenital Hypothyroidism is treated with levothyroxine tablets. Tablets should be crushed and mixed with a very small amount of breastmilk, non-soy based formula or water and administered via spoon or syringe. It should not be mixed into a bottle
- Levothyroxine should not be administered at the same time as multivitamins containing calcium or iron or with soy containing products.
- New commercially available compounded levothyroxine solutions can be considered.
- Consult a pediatric endocrinologist prior to obtaining thyroid imaging or scanning

Suggested References and Additional Reading:

- Bauer AJ, Wassner AJ. Thyroid hormone therapy in congenital hypothyroidism and pediatric hypothyroidism. *Endocrine*. 2019 Jul; 66:51-62.

- Leger J et al. European Society for Paediatric Endocrinology consensus guidelines on screening, diagnosis, and management of congenital hypothyroidism. *JCEM* 2014; 99(2): 363-84

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