Child With Suspected Adrenal Insufficiency

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:
• Fatigue
• Weight loss
• Nausea, vomiting
• Abdominal pain
• Weakness
• Salt craving
• Morning headaches
• Hyperpigmentation of gums, palmar creases, scars, elbows, areolae, scrotum, sun UN-exposed areas
• Dizziness
• Dehydration
• Hypoglycemia: pallor seizures/LOC, moodiness
• Orthostatic hypotension
 	
Family History:
Autoimmune diseases
Adrenal disease
 	
Differential Diagnosis
	Blood tests:
	• Sodium
	• Potassium
	• Glucose
	• Cortisol (7-8 am)
	• ACTH
	• Renin
	
	Other tests to consider after consultation with Pediatric Endocrinologist
	• Adrenal autoantibodies
	  ○ Anti-CYP11 (21-hydroxylase Ab)
	  ○ Anti-cortex adrenal Ab
	• ACTH stimulation test
	• Very long chain fatty acids
		Urgent:
		Nearly always urgent referral and treatment needed
		
		Find a Pediatric Endocrinologist
			Previous growth data/growth charts
			
			Pertinent medical records
			
			Recent laboratory and radiologic studies
				Additional Information
				
				http://www.nadf.us
				
				http://www.niddk.nih.gov/health-information/health-topics/endocrine/adrenal-insufficiency-addisons-disease/Pages/fact-sheet.aspx
				
				Adrenal Insufficiency: A Guide for Families
				
				References

Differential Diagnosis of Adrenal Insufficiency in Childhood

- Autoimmune
  - Isolated
  - Autoimmune Polyglandular Syndrome
- Acquired
  - Hemorrhage, infection, infiltration, drugs
- Hypopituitarism/ ACTH deficiency
- Defects of steroid biosynthesis
  - Congenital adrenal hyperplasia
  - Congenital lipoid adrenal hyperplasia
- Adrenal Dysgenesis
  - Adrenal Hypoplasia Congenita
  - SF-1 deficiency
  - Pallister- Hall syndrome
- Metabolic and cholesterol disorders
  - X-linked adrenoleukodystrophy
  - Wolman disease
  - Smith-Lemli Opitz
  - Kearns-Sayre
- Familial glucocorticoid deficiency/ ACTH resistance
- Allgrove syndrome: alacrima-achalasia-adrenal insufficiency neurologic (ALADIN) disorder

Additional information

- Children with primary adrenal insufficiency can present with hyponatremia, hyperkalemia and hypoglycemia
- Children with secondary adrenal insufficiency can present with mild hyponatremia and hypoglycemia
- Need to consider adrenal insufficiency in an infant presenting with ambiguous genitalia

Treatment

- In acute adrenal insufficiency, child will need intravenous sodium
- Hydrocortisone and fludrocortisone
- Patients and caregivers must be carefully and repeatedly trained on how and when to administer stress dose steroids to prevent an adrenal crisis.

Suggested References and Additional Reading

- Hsieh, S. and P. C. White (2011). "Presentation of primary adrenal insufficiency in childhood." J Clin Endocrinol Metab 96(6): E925-928.
- Malikova, J. and C. E. Fluck (2014). "Novel insight into etiology, diagnosis and management of primary adrenal insufficiency." Horm Res Paediatr 82(3): 145-157.
- Charmandari, E., N. C. Nicolaides, et al. (2014). "Adrenal insufficiency." Lancet 383(9935): 2152-2167.

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