Electronic Medical Record (EMR) Summary

Patient ID: PID64651196

Name: Priya

Age: 3, Sex: Female

Visit ID: VISIT10708227

Date: 2025-05-17 14:32

Clinical Reasoning Summary

Definition & Key Concerns

This case likely represents congenital adrenal hyperplasia (CAH), a group of autosomal recessive disorders characterized by impaired cortisol production. The most common form is 21-hydroxylase deficiency. The baby's ambiguous genitalia, vomiting, lethargy, and signs of dehydration are suggestive of an adrenal crisis, which is a life-threatening condition that requires immediate treatment.

Differential Diagnosis

1. Congenital Adrenal Hyperplasia (CAH): The most likely diagnosis given the ambiguous genitalia and signs of adrenal crisis.
2. Adrenal Hypoplasia: This condition can present similarly to CAH, but ambiguous genitalia is less common.
3. Salt-wasting nephropathy: This can cause similar symptoms but is less likely due to the presence of ambiguous genitalia.

Can?t-Miss Diagnosis

The critical high-risk condition that must be ruled out is Congenital Adrenal Hyperplasia, as it can lead to life-threatening adrenal crisis if not promptly treated.

Suggested Investigations

Key electrolytes to check include sodium and potassium, as CAH can lead to hyponatremia and hyperkalemia. Other tests include serum 17-hydroxyprogesterone, androstenedione, and cortisol levels. A karyotype analysis can be done to confirm the sex of the baby.

Management Plan

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Initial management involves fluid resuscitation with 0.9% saline and correction of electrolyte abnormalities. Hydrocortisone should be started immediately (dose: 25 mg/m2/day divided into three doses). Fludrocortisone may also be needed for mineralocorticoid replacement. The baby should be referred to a pediatric endocrinologist for long-term management.

Reference Insight

According to UpToDate, immediate treatment of adrenal crisis includes fluid resuscitation, correction of hypoglycemia, and administration of stress doses of hydrocortisone. Long-term management of CAH involves glucocorticoid and mineralocorticoid replacement, and monitoring for complications.

Rare Disease Alerts

Congenital Adrenal Hyperplasia (matched 3 symptoms)

Prescription

None provided