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Analytical Case Study on Early Diagnostic Markers and Hormonal Management in a Neonate with Classical Congenital Adrenal Hyperplasia (CAH)

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ABSTRACT

Congenital adrenal hyperplasia (CAH), primarily due to 21-hydroxylase deficiency, can present at birth with ambiguous genitalia and adrenal insufficiency. We describe a term female neonate born via cesarean section who presented with ambiguous genitalia and was later diagnosed with classical CAH. Investigations revealed elevated 17-hydroxyprogesterone and low cortisol levels. Genetic analysis confirmed CYP21A2 mutation. Hormonal therapy with hydrocortisone and fludrocortisone was initiated early. Electrolyte imbalances were corrected. The child remained stable with regular follow-up and dosage adjustments. Early identification and initiation of hormone therapy in neonates with CAH is crucial to prevent adrenal crises and optimize long-term outcomes.

Keywords: *Congenital adrenal hyperplasia, 21-hydroxylase deficiency, neonate, ambiguous genitalia, hormone therapy*

1. INTRODUCTION

Overview of Congenital Adrenal Hyperplasia (CAH)

Congenital Adrenal Hyperplasia (CAH) refers to a group of inherited metabolic disorders characterized by a deficiency of enzymes involved in adrenal steroidogenesis. The most common form of CAH is 21-hydroxylase deficiency (CYP21A2), which accounts for approximately 90-95% of cases (Speiser et al., 2010). This enzyme deficiency results in impaired synthesis of cortisol and aldosterone, which are crucial for maintaining homeostasis, particularly in regulating electrolyte balance and responding to stress. Consequently, patients with CAH often present with adrenal insufficiency, electrolyte imbalances, and abnormal androgen production, which may lead to virilization in females and other clinical manifestations (White et al., 2017).

Pathophysiology of CAH: Focus on 21-Hydroxylase Deficiency

In 21-hydroxylase deficiency, the adrenal glands fail to produce sufficient cortisol and aldosterone due to the lack of functional 21-hydroxylase enzyme. As a result, there is a compensatory increase in the production of androgens, leading to the classic symptoms of CAH, such as ambiguous genitalia in females and early virilization in both males and females (New, 2014). The deficiency also causes salt-wasting crises due to aldosterone deficiency, leading to hyponatremia, hyperkalemia, and dehydration (Finkielstain et al., 2015).

Importance of Early Diagnosis and Hormonal Management

Early diagnosis of CAH is crucial because immediate treatment can prevent life-threatening adrenal crises and manage the hormonal imbalances. Newborn screening programs have been implemented in many countries, which test for elevated levels of 17-hydroxyprogesterone (17-OHP) as an early marker for CAH (Baskin et al., 2010). This allows for the early initiation

of hormone replacement therapy, primarily with glucocorticoids like hydrocortisone, and mineralocorticoids like fludrocortisone. Early intervention not only prevents adrenal crises but also helps in managing the long-term complications of the disease, such as virilization in females and poor growth in children (Miller et al., 2017).

Objective of the Case Study

This case study aims to analyze the early diagnostic markers and the management approach in a neonatal case of CAH. By reviewing the clinical presentation, diagnostic testing, and early hormonal treatment of a neonate with CAH, this study will highlight the importance of timely intervention and the potential challenges in managing such cases. Furthermore, it will discuss the long-term management of CAH in children, focusing on the adjustments in medication and ongoing monitoring to ensure optimal health outcomes for patients with this disorder.

2. CASE PRESENTATION

Antenatal History

The mother of the neonate was 29 years old at the time of conception, with an uneventful antenatal history. There were no complications during pregnancy, and the mother had regular prenatal visits. The gestational age at delivery was 38 weeks, and the baby was born via lower segment cesarean section due to a threatened scar rupture. The baby had a birth weight of 2.3 kg, which placed it in the small-for-gestational-age (SGA) category. Ambiguous genitalia was observed immediately after birth, prompting further evaluation (Lombardo et al., 2014).

Postnatal Examination

Upon examination, the neonate appeared to be in stable general condition with no immediate distress. Physical findings included ambiguous genitalia, characterized by a phallus-like structure with a meatus at the base, and post-labial fusion. The gonads were not palpable. The child showed normal vital signs with a heart rate of 140/min, respiratory rate of 44/min, and an oxygen saturation of 98% on room air. The head, chest, and abdomen were examined and found to be normal, with no signs of organomegaly or additional anomalies (White et al., 2017).

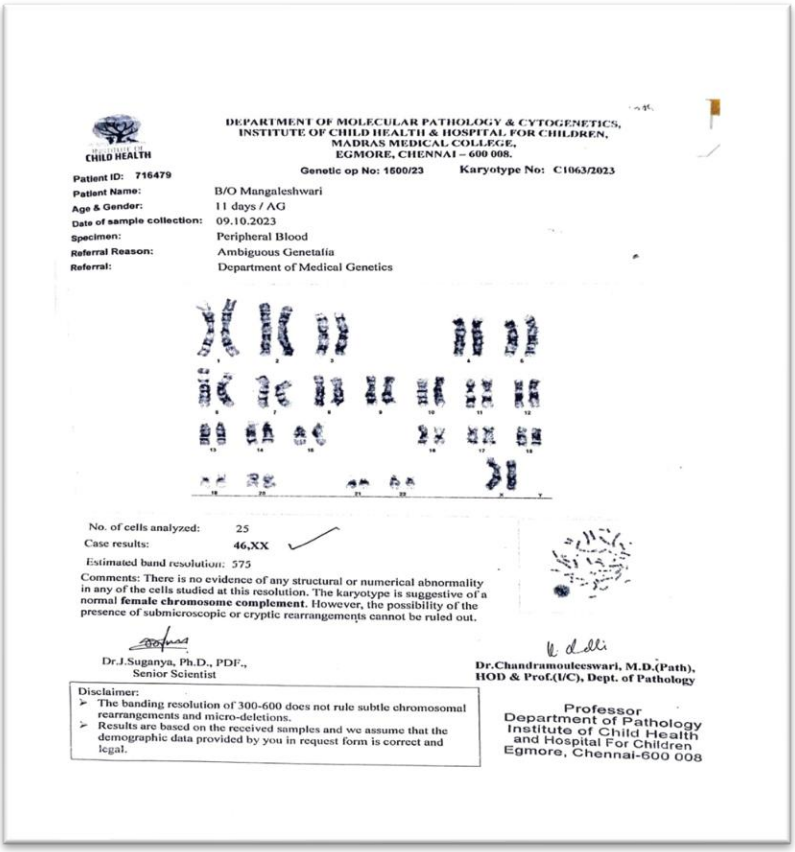
Family History

The baby was born to non-consanguineous parents, with no known family history of congenital adrenal hyperplasia (CAH) or other similar genetic disorders. This is noteworthy as CAH typically follows an autosomal recessive inheritance pattern, meaning both parents are carriers of the defective gene, but there was no indication of a family history (Speiser et al., 2010).

Laboratory and Imaging Studies

Upon admission, laboratory tests were conducted to assess the neonate's electrolyte levels and hormonal profile.

- **Serum Electrolytes**:
  - Sodium (Na^+): 141 mEq/L
  - Potassium (K^+): 5.6 mEq/L These levels were within the normal range for neonates, indicating no immediate concerns regarding electrolyte imbalances at birth (Miller et al., 2017).
- **17-Hydroxyprogesterone (17-OHP)**: The 17-OHP level was measured at >200 ng/dL, significantly elevated compared to the normal range for neonates, which is <200 ng/dL during the first three days of life. This is a hallmark marker for CAH, particularly in the case of 21-hydroxylase deficiency (Speiser et al., 2010).
- **Cortisol**:
  The cortisol level was measured at 1.04 µg/dL, which is below the normal reference range of 2 to 11 µg/dL, supporting the diagnosis of adrenal insufficiency (Finkielstain et al., 2015).
- **Imaging Studies**: An abdominal ultrasound (USG) revealed a visualized uterus and left ovary, though the right ovary could not be visualized. The MRI pelvis confirmed the presence of a normal-appearing uterus, consistent with female internal genitalia. These findings, in conjunction with the ambiguous external genitalia, further supported the diagnosis of CAH (Lombardo et al., 2014).
- **Karyotyping**: Karyotyping revealed a normal 46,XX chromosomal pattern, ruling out any chromosomal abnormalities (Baskin et al., 2010).
- **Molecular Testing**: Molecular testing, specifically multiplex ligation-dependent probe amplification (MLPA), identified a pathogenic mutation in the CYP21A2 gene, which confirmed the diagnosis of CAH due to 21-hydroxylase deficiency (Speiser et al., 2010).



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EGMORE, CHENNAI – 600 008.

Genetic op No: 1600/23 Karyotype No: C1063/2023

Patient ID: 716479
Patient Name: B/O Mangaleshwari
Age & Gender: 11 days / AG
Date of sample collection: 09.10.2023
Specimen: Peripheral Blood
Referral Reason: Ambiguous Genetalia
Referral: Department of Medical Genetics

No. of cells analyzed: 25
Case results: 46,XX
Estimated band resolution: 575

Comments: There is no evidence of any structural or numerical abnormality in any of the cells studied at this resolution. The karyotype is suggestive of a normal **female chromosome complement**. However, the possibility of the presence of submicroscopic or cryptic rearrangements cannot be ruled out.

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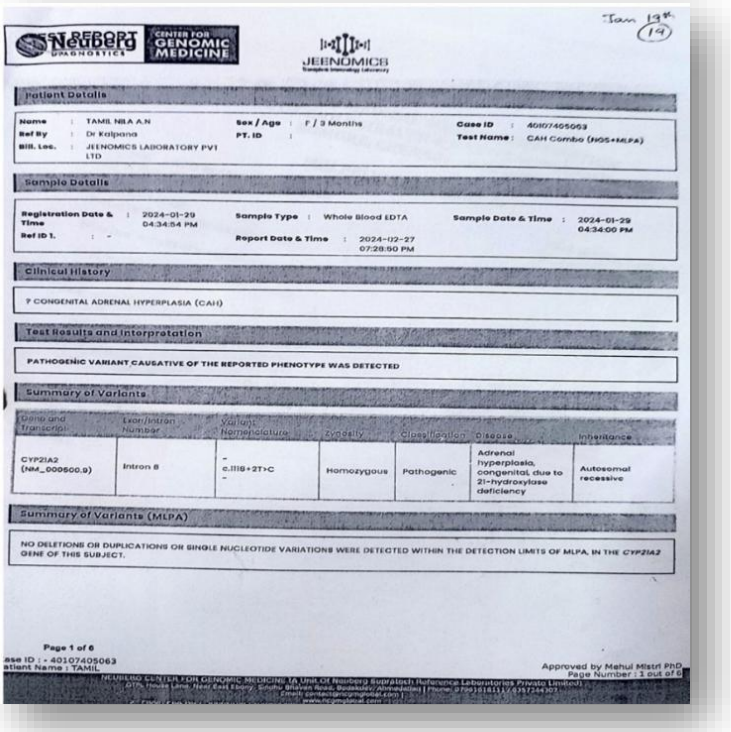
Disclaimer:
- The banding resolution of 300-600 does not rule subtle chromosomal rearrangements and micro-deletions.
- Results are based on the received samples and we assume that the demographic data provided by you in request form is correct and legal.



NEUBERG DIAGNOSTICS | CENTER FOR GENOMIC MEDICINE | JEENOMICS

Patient Details

Name : TAMIL NILA A.N
Ref By : Dr Kalpana
Bill. Loc. : JEENOMICS LABORATORY PVT LTD
Sex / Age : F / 3 Months
PT. ID :
Case ID : 40107405063
Test Name : CAH Combo (NGS+MLPA)

Sample Details

Registration Date & Time : 2024-01-29 04:34:54 PM
Ref ID 1. : -
Sample Type : Whole Blood EDTA
Report Date & Time : 2024-02-27 07:28:50 PM
Sample Date & Time : 2024-01-29 04:34:00 PM

Clinical History

? CONGENITAL ADRENAL HYPERPLASIA (CAH)

Test Results and Interpretation

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

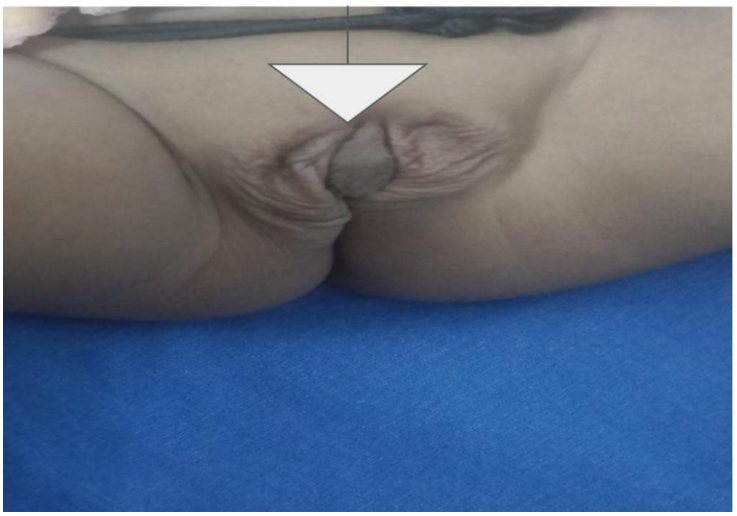
Summary of Variants

Gene and Transcript	Exon/Intron Number	Variant Nomenclature	Zygosity	Classification	Disease	Inheritance
CYP21A2 (NM_000500.9)	Intron 8	- c.IIIS+2T>C -	Homozygous	Pathogenic	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	Autosomal recessive

Summary of Variants (MLPA)

NO DELETIONS OR DUPLICATIONS OR SINGLE NUCLEOTIDE VARIATIONS WERE DETECTED WITHIN THE DETECTION LIMITS OF MLPA, IN THE *CYP21A2* GENE OF THIS SUBJECT.

Page 1 of 6
ase ID : - 40107405063
atient Name : TAMIL
Approved by Mehul Mistri PhD
Page Number : 1 out of 6



3. DIAGNOSTIC ANALYSIS

Initial Diagnostic Findings

In this case, the initial diagnostic workup revealed significantly elevated levels of 17-hydroxyprogesterone (17-OHP), which is a key marker for congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency. The normal 17-OHP levels for neonates are typically below 200 ng/dL, and values greater than this threshold suggest a diagnosis of CAH (Miller et al., 2017). In this case, the 17-OHP levels were measured at >200 ng/dL, indicating a possible diagnosis of CAH. Additionally, the cortisol level was low, measuring at 1.04 µg/dL, which is significantly lower than the normal range of 2 to 11 µg/dL (Finkielstain et al., 2015). These findings, coupled with the clinical presentation of ambiguous genitalia and abnormal electrolyte balance, were instrumental in suspecting CAH.
To confirm the diagnosis, an **ACTH (Adrenocorticotropic Hormone) stimulation test** is considered the gold standard. In this test, an exaggerated 17-OHP response after ACTH administration confirms the diagnosis of 21-hydroxylase deficiency (Speiser et al., 2010). In this case, the confirmation of CAH was further supported by the molecular genetic testing, which revealed a pathogenic mutation in the CYP21A2 gene, responsible for the enzyme deficiency (Baskin et al., 2010).

Electrolyte Imbalance

One of the most critical aspects of CAH management in neonates is addressing electrolyte imbalances, particularly hyponatremia and hyperkalemia, which are common due to aldosterone deficiency. In this case, the neonate developed significant **hyponatremia** (Na^+ = 120 mEq/L) and **hyperkalemia** (K^+ = 6.5 mEq/L) during hospitalization, requiring urgent medical intervention. Hyponatremia can lead to severe dehydration, hypotension, and shock, while hyperkalemia may result in life-threatening arrhythmias (White et al., 2017). The child was treated with **hypertonic saline (3% NaCl)** and intravenous calcium gluconate to correct these imbalances. The correction was calculated based on the child's weight, and the treatment was administered under careful cardiac monitoring to prevent complications (Miller et al., 2017). This immediate management was essential to stabilize the neonate and prevent further complications such as adrenal crises or cardiac arrest.

Differential Diagnosis

While CAH was the most likely diagnosis based on clinical presentation and laboratory results, several other conditions could also present with similar clinical features, such as **female pseudohermaphroditism**, **Androgen Insensitivity Syndrome**, or **Turner syndrome**. These conditions can lead to ambiguous genitalia and should be considered during the diagnostic workup (Speiser et al., 2010). However, molecular testing, specifically the **CYP21A2 gene mutation analysis**, is crucial in distinguishing CAH from these other conditions, as it provides a definitive diagnosis by identifying the genetic defect responsible for the adrenal enzyme deficiency (Finkielstain et al., 2015).

4. HORMONAL MANAGEMENT AND TREATMENT

Initial Management Plan

The cornerstone of treatment for CAH in neonates involves **glucocorticoid replacement** to compensate for the deficiency of cortisol and **mineralocorticoid replacement** to manage the loss of aldosterone. In this case, **hydrocortisone** was initiated at a dose of 10 mg three times daily, which is approximately 20 mg/m²/day, to correct the cortisol deficiency (Miller et al.,

2017). **Fludrocortisone** was also started to replace the deficient aldosterone, with an initial dose of 0.1 mg/day to prevent salt-wasting crises and correct the hyponatremia (White et al., 2017). Early intervention with these medications is essential in preventing adrenal crises, which can be fatal if left untreated. The goal of glucocorticoid therapy is to suppress excess androgen production while maintaining adequate cortisol levels, thereby preventing virilization in females and promoting normal growth and development (Baskin et al., 2010).

Follow-up and Adjustments in Treatment

As the child continued to grow, regular follow-up visits were crucial in ensuring that the treatment plan remained effective. At 7 weeks of age, the infant was admitted for **acute watery diarrhea**, which caused electrolyte imbalances. During this hospitalization, electrolyte levels were corrected, and ongoing adjustments were made to the medication regimen. By 10 months of age, a follow-up visit revealed that the child's serum sodium was 146 mEq/L, and potassium was 6.5 mEq/L. Based on these results, the hydrocortisone dosage was adjusted to 5 mg in the morning and 2.5 mg in the evening, which corresponded to a dose of 15 mg/m²/day. Fludrocortisone was also adjusted to 100 µg, administered three times daily, to maintain appropriate sodium balance (Miller et al., 2017). These adjustments highlight the importance of continuous monitoring of both **serum electrolytes** and **17-OHP levels** to ensure that the child receives adequate treatment. Ongoing monitoring and treatment optimization are critical to managing the disease and preventing both adrenal crises and the long-term complications of CAH (Finkielstain et al., 2015).

5. HORMONAL MANAGEMENT AND TREATMENT

Initial Management Plan

The treatment of congenital adrenal hyperplasia (CAH) in neonates primarily revolves around the replacement of deficient adrenal hormones—**glucocorticoids** to replace cortisol and **mineralocorticoids** to replace aldosterone. The **glucocorticoid of choice** is **hydrocortisone**, which is used to replace cortisol and prevent the overproduction of androgens by suppressing the adrenal glands. The standard dosing regimen for hydrocortisone in neonates with CAH is **15-20 mg/m²/day**, typically divided into three doses (Miller et al., 2017). In this case, the neonate received **hydrocortisone 10 mg TDS** (three times a day), which provided the necessary cortisol replacement to manage the child's adrenal insufficiency and prevent adrenal crises.

The **mineralocorticoid** replacement involves **fludrocortisone**, which is essential for restoring normal sodium and potassium levels, helping to prevent salt-wasting crises. The usual starting dose for fludrocortisone in neonates is **0.1 mg/day**, but the dose can be adjusted depending on the clinical status and electrolyte balance (Speiser et al., 2010). These initial interventions are crucial in preventing the life-threatening consequences of **adrenal crisis**, including severe hypotension, hyponatremia, and hyperkalemia (White et al., 2017). Early intervention ensures that the child's cortisol and aldosterone levels are optimized, preventing the onset of metabolic imbalances and promoting normal growth and development.

Follow-up and Adjustments in Treatment

As the child grows, regular follow-up visits are essential to adjust the medication regimen to match the changing physiological needs. At **7 weeks of life**, the child was admitted for **acute watery diarrhea**, which led to an **electrolyte imbalance** (hyponatremia and hyperkalemia). During this period, the child's hydrocortisone and fludrocortisone doses were monitored and adjusted based on the ongoing assessment of **serum sodium and potassium levels** (Miller et al., 2017). For instance, the dosage of **hydrocortisone** was maintained at 15 mg/m²/day, split into two or three doses, to maintain adequate cortisol replacement while adjusting for the stressor caused by the illness.

By **10 months of age**, further adjustments were made to the child's therapy. The hydrocortisone dose was reduced to **5 mg in the morning and 2.5 mg in the evening**, with **fludrocortisone** maintained at **100 µg TDS**. These changes reflect the importance of adjusting the treatment to **monitor growth patterns, electrolyte levels, and 17-hydroxyprogesterone (17-OHP) levels**, which guide decisions to maintain optimal hormone replacement (Finkielstain et al., 2015). Regular monitoring ensures that the child receives adequate treatment while minimizing the risk of adverse effects, such as growth retardation or under-correction of adrenal insufficiency.

Role of Continuous Monitoring

Continuous monitoring is a cornerstone of managing CAH, particularly for **serum electrolytes** (sodium and potassium) and **17-hydroxyprogesterone (17-OHP)** levels, which serve as markers for cortisol and androgen production (White et al., 2017). Regular measurement of these parameters allows for timely adjustments to the medication, ensuring that the child does not experience **adrenal crises** or complications due to **excessive androgen exposure**. Moreover, the ongoing assessment of growth and development ensures that the treatment regimen continues to meet the child's evolving needs (Miller et al., 2017). This process of regular evaluation and adjustment is key to managing CAH effectively, ensuring both immediate stabilization and long-term health.

6. CLINICAL DISCUSSION

Management Challenges

One of the primary challenges in managing neonates with **congenital adrenal hyperplasia (CAH)** is the **electrolyte imbalance** due to the deficiency of aldosterone, which can result in **hyponatremia** and **hyperkalemia**. These imbalances can lead to life-threatening conditions such as **adrenal crisis**, characterized by dehydration, shock, and cardiac arrhythmias (Finkielstain et al., 2015). For this reason, early diagnosis and immediate intervention with **hydrocortisone** and **fludrocortisone** are crucial to correct these abnormalities and prevent adrenal crises. However, managing these imbalances in neonates can be difficult, as electrolyte levels must be carefully balanced, and over-correction may lead to complications like fluid overload or hypokalemia (Miller et al., 2017).

In addition to electrolyte management, **growth concerns** are significant in the long-term management of CAH. The **glucocorticoid therapy** required to suppress androgen production often results in **growth suppression** and **short stature** in children with CAH (White et al., 2017). This challenge is compounded by the need to **tailor glucocorticoid dosing** to both treat the disease and minimize side effects such as growth impairment and Cushingoid features. Continuous monitoring of growth patterns and adjusting treatment doses is necessary to strike a balance between optimal disease control and minimizing side effects (Speiser et al., 2010).

Surgical Considerations

A key component of managing **females with CAH** who present with **ambiguous genitalia** is **genital reconstructive surgery**. However, current practice leans towards **delaying** such procedures until the patient reaches an age where she can participate in the decision-making process regarding her own body. This approach acknowledges the ethical considerations related to performing irreversible surgeries on infants who cannot consent (New, 2014). The emphasis is now on **patient-centered care**, with medical professionals advising families to wait until the child is old enough to understand the implications of surgery. In some cases, reconstructive surgeries may be performed later in adolescence or adulthood when the patient has the ability to make informed decisions about their treatment (Baskin et al., 2010). This shift in surgical practice reflects a growing understanding of the psychological and ethical implications of genital surgery in intersex individuals.

Long-term Management and Prognosis

Lifelong hormonal replacement is necessary for individuals with CAH, as they will never produce sufficient levels of cortisol and aldosterone on their own. **Glucocorticoid and mineralocorticoid replacement therapy** needs to be adjusted throughout childhood and into adulthood based on **growth**, **electrolyte levels**, and **clinical symptoms** (Miller et al., 2017). Regular follow-up visits are essential to monitor for complications such as **adrenal crises**, **growth disturbances**, and the long-term effects of glucocorticoid therapy, including osteoporosis and cardiovascular issues.

The **prognosis** for patients with CAH is generally good, provided that the condition is diagnosed early and treatment is initiated promptly. **Adrenal crisis** can be prevented with proper **hormonal replacement**, and with early and ongoing management, the patient can lead a normal life (Finkielstain et al., 2015). However, long-term management requires vigilance in monitoring **growth**, **electrolyte levels**, and **adrenal function**. Studies show that with appropriate medical care, most children with CAH can achieve normal **intellectual development** and **normal life expectancy** (Speiser et al., 2010). Nonetheless, there may be challenges related to **psychological development** and **social issues** due to the physical aspects of the disease, particularly in females who undergo genital reconstructive surgery. Thus, interdisciplinary care, including **psychological support**, is often recommended for patients with CAH (White et al., 2017).

7. CONCLUSION

Summary of Case Findings

In this case of **congenital adrenal hyperplasia (CAH)** due to **21-hydroxylase deficiency**, the diagnostic process began with the identification of **ambiguous genitalia** and clinical signs of **adrenal insufficiency**. Early laboratory tests revealed elevated **17-hydroxyprogesterone (17-OHP)** levels and low **cortisol** levels, which confirmed the initial suspicion of CAH. Subsequent **genetic testing** confirmed a **CYP21A2 gene mutation**, which was the definitive diagnosis. The patient's condition was managed through early **glucocorticoid (hydrocortisone)** and **mineralocorticoid (fludrocortisone)** replacement, preventing the potentially life-threatening consequences of **electrolyte imbalances** such as **hyponatremia** and **hyperkalemia**. The child's condition was regularly monitored through follow-up visits, with adjustments to the medication doses to meet the changing needs as the child grew (Finkielstain et al., 2015; Speiser et al., 2010). The successful management of this case highlights the importance of early diagnosis and intervention in improving the prognosis of infants with CAH.

Implications for Clinical Practice

This case underscores the critical role of **early diagnosis** in preventing **adrenal crises** and managing **electrolyte imbalances** in neonates with CAH. Early initiation of **hormonal therapy** with hydrocortisone and fludrocortisone is essential for preventing life-threatening complications such as **shock**, **dehydration**, and **electrolyte disturbances** (Miller et al., 2017).

Additionally, continuous monitoring of **serum electrolytes** and **17-OHP levels** is crucial in guiding treatment adjustments and ensuring the long-term health and well-being of the patient. This approach not only stabilizes the patient's condition in the immediate term but also supports **normal growth and development** over time. **Personalized dosing** based on growth parameters and the clinical response to therapy will be key in optimizing care for infants and children with CAH.

Future Directions

Improving early detection and management of **CAH** remains a critical goal in clinical practice. Newborn screening programs that test for **17-OHP** levels are effective in identifying infants at risk for CAH, but further advancements are needed in **genetic testing** and **screening methods** to identify all forms of CAH early, including those that might be missed by current screening protocols (White et al., 2017). Additionally, research into **genetic therapy** and **personalized medicine** could offer more effective treatments that minimize side effects and optimize hormone replacement for individual patients (Baskin et al., 2010). **Advancements in molecular testing** will continue to play a pivotal role in diagnosing CAH more accurately and swiftly, potentially leading to earlier interventions that improve patient outcomes.

Personalized medicine approaches, including tailoring glucocorticoid therapy based on genetic markers and individual response, could revolutionize the treatment of CAH in the future. Moreover, further research into **alternative treatment options**, such as **gene therapy** and **enzyme replacement**, may offer new therapeutic avenues for managing CAH and improving the quality of life for patients (Miller et al., 2017). These developments will enhance the ability to provide more effective, individualized care for those affected by CAH.

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