

# CASE REPORT

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## The Nature and Function of the Case Report in Psychology and Medicine

The **case report** stands as a foundational methodology within clinical medicine and psychology, providing detailed observations, analyses, and contextual information regarding individual patients or unique clinical occurrences. Unlike large-scale epidemiological studies or randomized controlled trials (RCTs), which focus on generalizability and statistical significance across populations, the case report offers profound insights into rare diseases, unusual presentations of common disorders, or novel therapeutic approaches. Its primary function is descriptive, meticulously documenting the patient's history, symptoms, diagnostic process, treatment regimen, and eventual outcome. This depth of information, often spanning psychological, biological, and social dimensions, is invaluable for generating new hypotheses, highlighting exceptions to established medical rules, and fostering clinical reasoning among practitioners.

Historically, case reports have been instrumental in the identification of new syndromes and the understanding of complex pathophysiology. For instance, early descriptions of conditions like AIDS or Creutzfeldt-Jakob disease often began as isolated case reports before robust population data became available. In the field of psychology, case reports, particularly those popularized by psychodynamic traditions, allow for the exploration of intricate intrapsychic dynamics and the trajectory of long-term therapeutic interventions that cannot be captured by short-term experimental designs. They serve as essential educational tools, enabling clinicians and researchers to recognize subtle symptom patterns and understand the variability inherent in human disease, thus reinforcing the importance of individualized patient care. While they possess limitations regarding external validity, their internal richness provides a critical counterbalance to quantitative research methodologies, documenting clinical reality as experienced by both the patient and the treating team.

The structure of a robust case report follows established academic conventions to ensure clarity and replicability, facilitating its utility for the broader scientific community. Essential components include a comprehensive introduction that situates the case within existing literature, a detailed presentation of the patient's clinical course, a thorough discussion section comparing the findings to previous research, and a conclusive summary highlighting the main learning points. The meticulous attention to detail required in constructing these reports underscores their ethical responsibility, demanding strict adherence to patient confidentiality and informed consent. Furthermore, case reports often bridge the gap between basic science and clinical application, illustrating how genetic, biochemical, or environmental factors translate into specific clinical phenotypes, thereby contributing significantly to **translational medicine**.

## Methodological Framework and Structure of a Case Report

A well-constructed case report adheres to a rigorous methodological structure designed to

maximize clarity and scientific utility. The initial section, the **Introduction**, establishes the clinical relevance of the case by providing a concise review of the relevant literature concerning the disease or presentation in question. It clearly states why this particular case is unique, challenging, or instructive--whether due to rarity, an unusual co-morbidity, or a novel treatment outcome. This sets the stage for the reader, ensuring they understand the context and the specific knowledge gap the report aims to address. The introduction must be precise, often culminating in a succinct statement of the case's objective.

Following the introduction is the core of the document, the **Case Presentation**. This segment provides a chronological and detailed narrative of the patient's journey. Key elements meticulously documented here include demographic data (age, gender, ethnicity), relevant medical and family history, the initial symptoms and presentation upon admission, and findings from the comprehensive physical examination. Crucially, all diagnostic procedures must be itemized, including laboratory results, imaging studies, and psychological assessments, presenting the data objectively and often in sequence as they were obtained. This section also outlines the differential diagnoses considered by the clinical team before the definitive diagnosis was reached, reflecting the complex decision-making process inherent in clinical practice. Transparency in reporting both confirming and contradictory data is paramount to maintain scientific integrity.

The subsequent sections detail the **Management and Intervention** strategies employed, followed by the clinical **Discussion**. The management section describes the specific therapeutic interventions--pharmacological, surgical, or psychological--including dosages, duration, and any modifications made over the course of treatment, along with the patient's response. The discussion section is arguably the most analytical component; here, the authors interpret the findings in light of the current scientific knowledge. They compare the patient's symptoms, diagnosis, and treatment response with established literature, exploring potential mechanisms underlying the unique presentation. This critical synthesis allows the authors to draw meaningful conclusions about pathophysiology, diagnostic challenges, or optimal treatment protocols, transforming raw clinical data into actionable scientific knowledge.

### **Illustrative Example: Congenital Adrenal Hyperplasia (CAH) Background**

To illustrate the utility of the case report methodology, we examine a case involving **Congenital Adrenal Hyperplasia (CAH)**, a rare group of autosomal recessive disorders that severely impact endocrine function and sexual development. CAH is primarily caused by mutations in the *CYP21A2* gene, leading to a deficiency of the enzyme 21-hydroxylase. This enzymatic defect disrupts the normal synthesis pathways for cortisol and aldosterone in the adrenal cortex. The absence or severe deficiency of **cortisol**, a crucial stress hormone, triggers a compensatory overproduction of adrenocorticotrophic hormone (ACTH) by the pituitary gland. This excessive stimulation leads to the accumulation of precursor hormones, which are shunted into the androgen

synthesis pathway, resulting in elevated levels of male sex hormones (androgens).

The clinical manifestations of CAH are varied, ranging from the life-threatening **salt-wasting crisis**, which typically presents in neonates due to severe aldosterone deficiency, to the milder, non-classic forms discovered later in life. In genetically female infants (46, XX), the prenatal exposure to excessive androgens leads to varying degrees of virilization of the external genitalia, often presenting as ambiguous genitalia (e.g., clitoromegaly, labial fusion). This requires prompt and accurate diagnosis, as the psychological and social implications of atypical sex development are profound, necessitating careful medical, surgical, and psychological intervention. Furthermore, the risk of a salt-wasting crisis--characterized by dehydration, hyponatremia, and hyperkalemia--makes CAH a pediatric emergency requiring immediate hormonal replacement therapy.

The estimated prevalence of CAH due to 21-hydroxylase deficiency is approximately 1 in 10,000 to 1 in 18,000 live births in the general population, classifying it as a rare disease that often requires specialized management protocols. The complexity of its presentation--involving both metabolic imbalance and genital ambiguity--highlights the need for multidisciplinary team involvement, including endocrinologists, genetic counselors, urologists, and pediatric psychologists. Understanding the genetic and biochemical basis of CAH is crucial for effective long-term management, which aims to normalize hormone levels, prevent adrenal crises, and address the psychosocial aspects associated with the condition, thereby ensuring the best possible quality of life for the affected individual.

### Detailed Case Presentation

This specific case report details the presentation and management of a rare, severe manifestation of CAH in an infant. The patient, a female infant, was referred to the Neonatal Intensive Care Unit (NICU) at 2 weeks of age due to suspicion of an endocrine disorder following initial clinical observation. The infant was the product of a full-term gestation, born to a healthy, non-consanguineous couple, with an unremarkable prenatal history and a birth weight of 3.2 kg, suggesting normal fetal growth trajectory. However, upon detailed postnatal examination, several physical findings raised immediate clinical concern regarding sexual differentiation and hormonal imbalance, necessitating specialized care and investigation.

The physical examination was highly suggestive of excessive androgen exposure *in utero*. The infant exhibited significant virilization of the external genitalia, specifically noted as pronounced **clitoromegaly** and partial **labial fusion**, resulting in genitalia that appeared ambiguous according to the Prader scale. While the infant did not initially present with overt signs of a salt-wasting crisis (such as profound lethargy or severe dehydration), the genital ambiguity prompted immediate laboratory screening. Initial diagnostic investigations revealed critically elevated levels of androgens (specifically 17-hydroxyprogesterone) coupled with markedly decreased circulating

levels of **cortisol**. This biochemical profile strongly confirmed the clinical suspicion of 21-hydroxylase deficiency, the most common form of CAH.

To definitively confirm the diagnosis and classify the genetic etiology, specialized testing was performed. **Genetic analysis** revealed a homozygous mutation in the *CYP21A2* gene, confirming the diagnosis of classic CAH. This genetic finding supported the severity of the enzyme deficiency suggested by the profound hormonal imbalances observed. The prompt diagnosis at 2 weeks of age allowed the clinical team to initiate life-saving and corrective treatment before the onset of a potentially fatal adrenal crisis, emphasizing the critical role of newborn screening and vigilant clinical assessment in recognizing rare congenital disorders that have immediate life-threatening potential if left untreated.

### Clinical Management and Therapeutic Interventions

Given the confirmed diagnosis of classic CAH and the risk of acute adrenal insufficiency, immediate therapeutic intervention was mandatory. The management strategy focused on replacing the deficient hormones--cortisol (glucocorticoid) and aldosterone (mineralocorticoid)--and suppressing the excessive production of androgens by the hyperactive pituitary-adrenal axis. The infant was immediately initiated on **dexamethasone**, a potent synthetic glucocorticoid, to replace the deficient cortisol and suppress the release of ACTH, thereby aiming to reduce the pathological androgen production. Concurrently, **fludrocortisone** was administered to supplement the mineralocorticoid activity, essential for maintaining electrolyte and fluid balance, preventing the severe hypotension and hyponatremia characteristic of salt-wasting CAH.

In addition to pharmacological interventions, supportive care was crucial. The infant was placed on a specialized **low-salt diet** regimen to assist in mitigating potential electrolyte abnormalities, particularly hyperkalemia, which is a common consequence of mineralocorticoid deficiency. The patient's metabolic status was rigorously monitored through daily clinical assessments and regular laboratory tests, focusing specifically on serum sodium, potassium, and plasma renin activity (PRA) to fine-tune the fludrocortisone dosage. Monitoring also included periodic ultrasounds of the pelvic region and regular measurements of androgen levels (e.g., 17-hydroxyprogesterone) to assess the efficacy of the glucocorticoid suppression therapy.

Addressing the anatomical consequences of prenatal virilization was another critical component of the long-term management plan. At four months of age, following stabilization of the hormonal profile, the infant underwent corrective surgical procedures known as **feminizing genitoplasty** and clitoroplasty. These surgeries are highly specialized and aim to reconstruct the external genitalia to align with the assigned female gender, optimizing anatomical function and minimizing future psychosocial challenges associated with atypical development. Following the initial stabilization phase and subsequent surgical intervention, the infant's glucocorticoid regimen was transitioned

from dexamethasone to **hydrocortisone**, a shorter-acting and generally preferred replacement therapy for infants, allowing for a more physiological dosing schedule. By six months of age, subsequent laboratory monitoring confirmed that the patient's androgen levels had normalized, indicating successful hormonal control and suppression of the adrenal hyperactivity.

## Analysis and Discussion of Findings

This case report provides a compelling illustration of the severe, classic presentation of CAH and underscores the efficacy of timely, integrated medical and surgical management. The infant presented with significant virilization despite being diagnosed within the first two weeks of life, suggesting a profound 21-hydroxylase deficiency confirmed by the homozygous *CYP21A2* mutation. The rapid initiation of combined glucocorticoid and mineralocorticoid replacement therapy was crucial not only for managing the immediate risks associated with cortisol deficiency but also for preventing the onset of acute salt-wasting, which can be fatal if diagnosis is delayed beyond the neonatal period. The choice of initial high-dose glucocorticoid (dexamethasone) followed by transition to hydrocortisone reflects standard pediatric endocrine protocols aimed at achieving swift hormonal control while minimizing the long-term growth suppression effects associated with prolonged high-dose steroid use.

The successful normalization of androgen levels by six months of age validates the effectiveness of the hormonal suppression regimen. However, the discussion must also center on the complex ethical and psychological considerations surrounding the surgical management of ambiguous genitalia. The decision to perform **feminizing genitoplasty** at four months of age aligns with some clinical recommendations arguing for early intervention to facilitate gender assignment and parental bonding, although this approach remains a subject of ongoing debate within the pediatric and psychological communities regarding timing and necessity. The surgical team's objective was to achieve a functional and aesthetically acceptable outcome, which is critical for the patient's future psychosexual development and social integration. Continuous psychological support and monitoring for both the patient and the family are paramount throughout childhood and adolescence to address potential body image issues or challenges related to chronic disease management.

Ultimately, this case reinforces the diagnostic pathway for CAH: clinical suspicion based on genital ambiguity (in females) or unexplained salt-wasting (in both sexes), followed by biochemical confirmation (elevated 17-OHP and low cortisol), and definitive genetic testing. The positive outcome observed--normalized androgen levels and successful surgical correction--highlights that even severe forms of CAH can be managed effectively when diagnosed early and treated aggressively by a specialized, multidisciplinary team. This outcome also contributes to the body of evidence supporting early hormonal intervention to mitigate the damaging effects of excessive androgen exposure on developing tissues and to ensure proper growth and pubertal development.

later in life.

## Conclusion and Implications for Clinical Practice

This detailed case report of classic Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency serves as a powerful reminder of the importance of recognizing rare, life-threatening endocrine disorders in the neonatal period. The case demonstrated that prompt, aggressive medical intervention--specifically the immediate replacement of deficient cortisol and mineralocorticoid using dexamethasone/fludrocortisone, subsequently transitioned to hydrocortisone--is essential for stabilizing the patient and preventing catastrophic adrenal crises. The careful monitoring of electrolytes and androgen precursors played a central role in guiding treatment adjustments, ensuring therapeutic goals were met while minimizing adverse effects associated with high-dose steroid use.

Furthermore, the successful execution of **feminizing genitoplasty** and clitoroplasty underscores the necessity of integrating specialized surgical correction into the comprehensive management plan for female infants severely virilized by CAH. The good outcome observed in this patient--normalized hormone levels and anatomical correction by six months--validates the multidisciplinary approach involving pediatric endocrinology, genetics, surgery, and mental health professionals. Future longitudinal studies tracking the patient's psychological development and quality of life will be necessary to fully evaluate the long-term success of this integrated management strategy.

In conclusion, this case report significantly contributes to the clinical understanding of CAH by illustrating a successful diagnostic and management paradigm for a severe phenotype. It reinforces key clinical practices: the need for heightened clinical suspicion in infants with ambiguous genitalia, the immediate necessity of hormonal replacement therapy, and the requirement for long-term follow-up to manage chronic steroid use and potential psychosocial complexities. Case reports, such as this one, remain an indispensable tool for educating clinicians, ensuring that rare yet critical conditions like CAH receive the prompt and appropriate care necessary to secure a positive prognosis.

## References

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