ECRONICON OPEN ACCESS

EC PAEDIATRICS
Case Report

A Rare Dual Diagnosis: Coexistence of Congenital Adrenal Hyperplasia and Thalassemia Major in a Male Infant

Samia Saeed[1]* and Hania Khalid[2]

[1]Postgraduate Resident, Pediatric Medicine at The Children's Hospital and UCHS, Lahore, Pakistan

[2]3rd Year MBBS Student at UCMD, Lahore, Pakistan

***Corresponding Author:** Samia Saeed, Postgraduate Resident, Pediatric Medicine at The Children's Hospital and UCHS, Lahore, Pakistan.

Received: May 14, 2026; **Published:** June 18, 2026

Abstract

Congenital adrenal hyperplasia (CAH) and thalassemia major are inherited autosomal recessive disorders, each with significant clinical implications when presenting in infancy. The concurrent occurrence of both conditions in a single patient is exceedingly uncommon and poses diagnostic and therapeutic challenges. We describe a male infant initially diagnosed with salt-wasting CAH at three months of age following an adrenal crisis. At one year, he was subsequently evaluated for persistent anemia and diagnosed with thalassemia major. Genetic analysis revealed a homozygous mutation in the CYP21A2 gene. This report emphasizes the importance of early recognition, genetic assessment, and coordinated multidisciplinary care, particularly in children born to consanguineous parents [1,3].

Keywords: *Congenital Adrenal Hyperplasia (CAH); Thalassemia Major; CYP21A2 Gene*

Introduction

CAH, particularly due to 21-hydroxylase deficiency, is the most common form of this disorder and can result in life-threatening salt wasting crises in early infancy. Thalassemia major is a severe hemoglobinopathy requiring regular transfusions and is characterized by pallor, anemia, and growth delay. Both diseases follow an autosomal recessive inheritance and are more prevalent in populations with high rate of consanguinity. However, the coexistence of CAH and thalassemia major in a single individual is rarely reported in literature. This case report aims to highlight such a presentation, underscoring the value of early screening and interdisciplinary management in high-risk populations [1-4].

Case Presentation

A male infant, born at term via spontaneous vaginal delivery with a low birth weight of 1 kg, presented at 25 days of life with vomiting, loose stools, poor weight gain, and generalised hyperpigmentation.

The child was born to consanguineous parents. Initial laboratory findings were suggestive of adrenal insufficiency. At 3 months, a diagnosis of salt-wasting CAH was established based on clinical features and 17-hydroxyprogesteron levels.

The patient was started on:

- Hydrocortisone (Cortecort): 10 mg/m/day.
- Fludrocortisone (Florinef): 0.1 mg once daily.

He required one hospitalization at the time of diagnosis, after which he showed notable clinical improvement.

At one year of age, the child developed progressive pallor. Hemoglobin electrophoresis revealed:

- Hemoglobin F: 80.5%.
- Hemoglobin A: 14.6%.
- Hemoglobin A2: 4.9%.

These findings were consistent with beta thalassemia major.

Family and genetic background

The patient's family history was notable for consanguinity. An older sibling, an 11-year-old sister, was already diagnosed with thalassemia major. Genetic testing confirmed a homozygous pathogenic mutation in the CYP21A2 gene, consistent with CAH. Multiplex ligation-dependent probe amplification (MLPA) revealed no large deletions or duplications.

Physical examination:

- General: Pale appearance.
- Abdomen: Spleen palpable 2.5 cm below the costal margin.
- Genitalia: Stretched penile length of 1 cm with a single urethral opening.
- Gonads: Bilateral testes present in the scrotal sac with a volume of 2 mL each.

Laboratory findings:

- A homozygous pathogenic mutation in the CYP21A2 gene, consistent with autosomal recessive CAH type 1.
- Serum Cortisol: 24 ug/dL (23-11-2019).
- 17-Hydroxyprogesterone (17-OHP): 320 ng/dL (23-11-2019).
- 0.46 ng/dL (22-02-2021), indicating adequate hormonal control.

Final diagnosis:

- Salt-wasting Congenital Adrenal Hyperplasia (CAH) - genetically confirmed - thalassemia major - confirmed via hemoglobin electrophoresis.

Discussion

This case illustrates a rare co-existence of two distinct autosomal recessive disorders-CAH and thalassemia major-in a single infant. Early recognition of adrenal insufficiency enabled timely initiation of life saving hormonal therapy. The later development of anemia led to the diagnosis of thalassemia major, emphasizing the importance of continued clinical vigilance.

The presence of consanguinity and an affected sibling highlights the critical role of genetic counseling and family screening. Molecular testing provided essential confirmation of CAH, offering critical insights for risk stratification in future pregnancies.

This case underscores the significance of a multidisciplinary approach involving endocrinology, hematology, and genetic counseling for optimal management of patients with overlapping inherited conditions.

Conclusion

In populations with a high prevalence of consanguinity, the potential for co-inheritance of multiple genetic disorders should always be considered. Early diagnosis, molecular confirmation, and collaborative management are crucial in addressing the complexities of such presentations and improved long term outcomes [5].

Bibliography

1. Navarro-Zambrana AN and Sheets LR. "Ethnic and national differences in congenital adrenal hyperplasia incidence: a systematic review and meta-analysis". *Hormone Research in Paediatrics* 96.3 (2023): 249-258.

2. Li Z., *et al*. "Analysis of the screening results for congenital adrenal hyperplasia involving 7.85 million newborns in China: A systematic review and meta-analysis". *Frontiers in Endocrinology (Lausanne)* 12 (2021): 624507.

3. Waheed U., *et al*. "A systematic review and meta-analysis on the epidemiology of hepatitis B and hepatitis C virus among beta-thalassemia major patients in Pakistan". *Journal of Laboratory Physicians* 13.3 (2021): 270-276.

4. Arab-Zozani M., *et al*. "A systematic review and meta-analysis of stature growth complications in beta-thalassemia major patients". *Annals of Global Health* 87.1 (2021): 48.

5. Alqahtani MM., *et al*. "Prevalence of thalassemia in Saudi Arabia: a systematic review and meta-analysis". *International Journal of Medicine in Developing Countries* 8.10 (2024): 2903-2912.

Volume 15 Issue 7 July 2026