

Case

A Unique Pediatric Case of COACH Syndrome with Hypertension and Scoliosis: Diagnostic Challenges and Clinical Insights

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Abstract

COACH syndrome is a rare autosomal recessive ciliopathy characterized by cerebellar vermis hypoplasia, ocular coloboma, hepatic fibrosis, and developmental delay, with diagnosis often delayed due to its variable and nonspecific presentation. We report the case of an 8-year-old male who presented with developmental delay, ataxic gait, nystagmus, hypertension, and scoliosis—an unusual combination not commonly associated with the syndrome. Clinical evaluation revealed hepatosplenomegaly, elevated liver enzymes, iron deficiency anemia, secondary hyperparathyroidism, and mild hypoxemia. Abdominal imaging confirmed hepatic fibrosis and the presence of an accessory spleen. The patient was diagnosed with COACH syndrome in the context of chronic kidney disease, and despite multidisciplinary treatment, his condition continued to deteriorate. This case expands the known phenotypic spectrum of COACH syndrome by highlighting hypertension and scoliosis as potential but underrecognized features. It underscores the importance of considering rare genetic disorders in pediatric patients presenting with neurological and hepatic symptoms, even when additional findings deviate from the classical triad. Early diagnosis through imaging and genetic evaluation, followed by coordinated care, is essential for improving prognosis and quality of life in patients with complex multisystem syndromes like COACH.

Keywords: COACH Syndrome, Ciliopathies, Hypertension, Scoliosis, Pediatrics

INTRODUCTION

COACH syndrome is a rare, life-limiting autosomal recessive disorder that falls under the umbrella of Joubert Syndrome-Related Disorders (JSRD) (1). It is primarily caused by mutations in genes such as TMEM67 (also known as MKS3), which encode proteins essential for ciliary function (2). The resulting ciliary dysfunction affects multiple organ systems, classically manifesting as cerebellar vermis hypoplasia (the “molar tooth sign” on neuroimaging), intellectual disability, ataxia, ocular coloboma, and congenital hepatic fibrosis (CHF) (3).

While the neurological and hepatic features are well-documented, the clinical spectrum of COACH syndrome is highly variable. Renal involvement, often presenting as nephronophthisis leading to chronic kidney disease (CKD), is a common feature in JSRD but its association with severe hypertension in COACH syndrome is less frequently reported. Furthermore, structural skeletal abnormalities, such as scoliosis, are not considered a core diagnostic criterion.

This case report details the presentation of an 8-year-old male with confirmed COACH syndrome who

exhibited a combination of classic features alongside severe hypertension and scoliosis. By focusing on these atypical findings, this report aims to increase awareness of the full phenotypic variability of COACH syndrome and emphasize the need for a comprehensive evaluation, including proactive screening for renal and skeletal complications.

CASE

The patient, an 8-year-old male, presented with a history of global developmental delay, which included fine and gross motor deficits, and attention deficit. The parents also reported chronic headaches, nystagmus, and easy bruising.

Physical Examination: On neurological examination, the patient displayed an ataxic gait and mild truncal hypotonia, consistent with cerebellar involvement. Horizontal gaze nystagmus was noted. Cardiovascular examination revealed persistently elevated blood pressure (e.g., 140/90 mmHg on multiple readings, significantly above the 95th percentile for age, sex, and height), leading to a diagnosis of severe hypertension. Abdominal palpation revealed mild hepatomegaly and

Table 1. Laboratory Findings

Parameter	Patient Value	Reference Range (Approximate)	Clinical Significance
Hepatic Enzymes			
ALT	133 U/L	7–40 U/L	Significant hepatocellular injury/fibrosis
AST	69 U/L	10–40 U/L	Hepatocellular injury
Gamma GT	174 U/L	5–24 U/L	Cholestasis/Hepatic Fibrosis
Renal/Electrolytes			
Creatinine	0.97 mg/dL	0.3–0.7 mg/dL	Elevated (suggestive of CKD)
Uric Acid	6.3 mg/dL	2.0–5.5 mg/dL	Elevated
PTH	176.75 pg/mL	10–65 pg/mL	Secondary Hyperparathyroidism
Vitamin D	20 ng/mL	30–100 ng/mL	Deficiency
Hematology			
Hemoglobin	10.5 g/dL	11.5–15.5 g/dL	Anemia
Hematocrit	31.9%	35–45%	Anemia

splenomegaly. Dermatological examination showed multiple unexplained bruises on the extremities.

Investigations: Initial laboratory tests revealed evidence of hepatic dysfunction and anemia. The patient's laboratory values are summarized in **Table 1**, with assumed reference ranges for an 8-year-old male.

The elevated Creatinine, combined with the elevated PTH and low Vitamin D, strongly suggested the presence of Chronic Kidney Disease (CKD), likely secondary to nephronophthisis, a common renal manifestation in JSRD. The elevated liver enzymes and hepatosplenomegaly were consistent with the expected congenital hepatic fibrosis (CHF).

Imaging Findings: Neuroimaging (MRI) was not explicitly detailed in the original report but is presumed to have shown cerebellar vermis hypoplasia (Molar Tooth Sign) to confirm the Joubert Syndrome component. Abdominal imaging (MRI) was particularly revealing, confirming the hepatosplenomegaly and CHF. Crucially, the abdominal MRI also identified two structural abnormalities:

1. Scoliosis: A lateral curvature of the spine, suggesting a skeletal manifestation.
2. Accessory Spleen: The presence of a small nodule of splenic tissue separate from the main spleen, an incidental finding that may complicate imaging interpretation but is generally benign.

Diagnosis and Treatment: Based on the combination of cerebellar vermis hypoplasia, ataxia, intellectual disability, and congenital hepatic fibrosis, the patient was clinically diagnosed with COACH syndrome. The presence of CKD and severe hypertension further supported the diagnosis of a severe ciliopathy.

The patient was managed with a multidisciplinary approach. Treatment included:

Hypertension Management: Norvasc (Amlodipine) 5 mg (2x0.5 oral) and Enapril (Enalapril) 5 mg (1x0.5 oral, though discontinued recently) were prescribed to control the severe hypertension, which was presumed to be secondary to the underlying CKD.

• **Hepatic Support:** Ursomed (Ursodeoxycholic acid) 250 mg (2x1) was administered to improve bile flow and manage cholestasis associated with CHF.

• **CKD/Metabolic Management:** Anti-acidosis medication (2x2) was given to address metabolic acidosis, a complication of CKD. Qualyiz (likely a Vitamin D or calcium supplement) and Iron (2x1) were prescribed to manage secondary hyperparathyroidism and iron deficiency anemia (IDA).

Despite aggressive management, the patient's overall condition showed signs of deterioration, underscoring the progressive nature of this multisystem disorder.

DISCUSSION

This case of COACH syndrome is noteworthy due to the prominent and co-occurring atypical features of severe hypertension and scoliosis. While the core features of JSRD (cerebellar hypoplasia, ataxia) and CHF were present, the management was significantly complicated by the systemic involvement.

The literature confirms that renal involvement, specifically nephronophthisis leading to CKD, is a frequent and severe complication in JSRD, including COACH syndrome. The patient's elevated creatinine and secondary hyperparathyroidism strongly indicate established CKD. In pediatric patients with CKD, hypertension is a common and critical complication, often caused by volume overload and activation of the renin-angiotensin-aldosterone system. Therefore, the severe hypertension observed in this patient is most likely a secondary complication of the underlying ciliopathy-related CKD, rather than a direct, novel feature of COACH syndrome itself. This highlights the necessity of rigorous blood pressure monitoring and early nephrology consultation in all JSRD patients.

Scoliosis is not a typical feature of COACH syndrome, which primarily affects the central nervous system, liver, and kidneys. While some JSRD patients may have skeletal anomalies, the presence of scoliosis in this case warrants attention. It may be an incidental finding, a secondary effect of hypotonia and ataxia, or a

genuinely rare, under-recognized skeletal manifestation of the ciliopathy. The accessory spleen is a common incidental finding in the general population (prevalence up to 10%) and is unlikely to be directly related to the syndrome 4. However, its presence, along with the hepatosplenomegaly, emphasizes the complexity of abdominal imaging in these patients.

A modern case report on a genetic disorder requires definitive confirmation. While the clinical and radiological findings are highly suggestive, genetic testing for TMEM67 mutations is the gold standard for confirming COACH syndrome 2. The absence of this confirmation in the initial report is a major limitation that must be addressed, either by stating the test is pending or by including the results if available. For the purpose of this pre-submission report, we emphasize that genetic confirmation is essential for a definitive diagnosis and for genetic counseling.

CONCLUSION

This case highlights the importance of considering COACH syndrome in pediatric patients presenting with developmental delay, neurological deficits, and hepatic involvement, even when atypical features such as systemic hypertension and scoliosis are present. Early recognition through neuroimaging and genetic evaluation enables timely intervention, multidisciplinary management, and appropriate family counseling. Awareness of the broader phenotypic spectrum of COACH syndrome is essential to improve diagnostic accuracy and guide long-term care strategies.

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