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Lipoid Congenital Adrenal Hyperplasia Due to Steroidogenic Acute Regulatory Protein (StAR) Deficiency in a 46,XY Child With a Female Phenotype: A Case Report

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Abstract

Primary adrenal insufficiency in childhood is frequently associated with inherited disorders. Lipoid congenital adrenal hyperplasia due to steroidogenic acute regulatory protein (StAR) deficiency is a rare disorder characterized by impaired mitochondrial cholesterol transfer and deficient adrenal and gonadal steroidogenesis. We report a five-year-old child raised as a girl who had been diagnosed with primary adrenal insufficiency at three months of age and was receiving hydrocortisone and fludrocortisone. Physical examination showed female-appearing external genitalia without virilization and Tanner stage I development. Biochemical evaluation demonstrated markedly elevated adrenocorticotrophic hormone with profoundly low morning cortisol and globally reduced steroidogenesis, including low 17-hydroxyprogesterone, androstenedione, dehydroepiandrosterone sulfate, and free testosterone. Karyotyping revealed 46,XY. Anti-Müllerian hormone and inhibin B concentrations were compatible with preserved Sertoli cell secretory activity and supported the presence of testicular tissue despite severely impaired androgen production. A contrast-enhanced abdominal and pelvic magnetic resonance imaging study was subsequently re-evaluated in the picture archiving and communication system by an expert radiologist.

Targeted review identified structures compatible with bilateral undescended testes, with the right probable gonad in the inguinal canal and the left probable gonad in the abdominal cavity. Magnetic resonance imaging and transabdominal ultrasonography also demonstrated a tubular structure posterior to the bladder that was considered compatible with a rudimentary vaginal canal; no definite uterine structure was identified on targeted review. Molecular testing identified a homozygous pathogenic truncating variant in *STAR* (NM_000349.3:c.577C>T; p.Arg193*), establishing the diagnosis of lipoid congenital adrenal hyperplasia due to StAR deficiency. This case highlights the value of integrating steroid profiles, cytogenetic findings, Sertoli cell biomarkers, molecular testing, and expert imaging review in children with primary adrenal insufficiency and 46,XY differences of sex development.

Categories: Radiology, Genetics, Endocrinology/Diabetes/Metabolism

Keywords: 46 xy dsd, adrenal insufficiency, anti-müllerian hormone, lipoid congenital adrenal hyperplasia, pediatric endocrinology, star deficiency, star gene, steroidogenesis

Introduction

Primary adrenal insufficiency (PAI) comprises a heterogeneous group of disorders characterized by deficient glucocorticoid production, with or without mineralocorticoid and adrenal androgen deficiency. In children, inherited monogenic disorders account for a substantial proportion of cases, and congenital adrenal hyperplasia (CAH) is an important cause of PAI [1-3].

Lipoid congenital adrenal hyperplasia (LCAH) is a rare and severe disorder of steroidogenesis caused most commonly by biallelic pathogenic variants in the *STAR* gene. The encoded steroidogenic acute regulatory protein (StAR) facilitates cholesterol transfer from the outer to the inner mitochondrial membrane, where cholesterol becomes available for CYP11A1-mediated conversion to pregnenolone. Severe loss of StAR function therefore produces a proximal defect that can compromise glucocorticoid, mineralocorticoid, and sex-steroid synthesis [4-7].

In individuals with a 46,XY karyotype, severe impairment of fetal testicular steroidogenesis may result in marked undervirilization and female-appearing external genitalia. Sertoli cell secretory activity, however, may remain relatively preserved; anti-Müllerian hormone (AMH) and inhibin B can therefore provide useful evidence of functioning testicular tissue during childhood [8,9]. Because the clinical and biochemical phenotype can overlap with other proximal defects of steroidogenesis, particularly CYP11A1 deficiency, molecular testing is central to establishing the specific etiology.

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We report a pediatric case of LCAH due to a homozygous pathogenic *STAR* variant in a 46,XY child with female-appearing external genitalia. The case illustrates the diagnostic integration of adrenal and gonadal steroid profiles, Sertoli cell biomarkers, cytogenetics, molecular testing, and targeted radiologic reassessment for gonadal localization and internal genital anatomy.

Case Presentation

In December 2024, at five years of age, a child residing in a rural area of Boyacá, Colombia, and socially raised as a girl was referred for pediatric endocrinology follow-up. She was born at 38 weeks of gestation by vaginal delivery as the product of the fourth pregnancy, with appropriate birth weight and length. Her parents were non-consanguineous. She had been diagnosed with PAI at three months of age, and no adrenal crises had been documented since diagnosis. At the time of evaluation, she was receiving chronic replacement therapy with hydrocortisone 7.5 mg/day divided into three doses and fludrocortisone 0.1 mg/day.

On physical examination, her weight was 23 kg, her height was 117 cm, and her body mass index was 16.8 kg/m², with anthropometric parameters appropriate for age. There was no hyperpigmentation, vomiting, dehydration, or evidence of hemodynamic compromise. The external genitalia were female-appearing, without signs of virilization. Pubertal development corresponded to Tanner stage I for both breast and genital development.

Biochemical assessment showed marked adrenocorticotrophic hormone (ACTH) elevation with a profoundly low morning cortisol concentration and globally reduced adrenal and gonadal steroidogenesis. Cytogenetic analysis using GTG banding demonstrated a 46,XY karyotype in 27 metaphases. During follow-up, free testosterone remained suppressed, whereas AMH and inhibin B concentrations were compatible with preserved Sertoli cell secretory activity and supported the presence of testicular tissue. Plasma renin activity remained within the laboratory reference interval while the patient was receiving fludrocortisone, consistent with an ongoing mineralocorticoid replacement effect in the overall clinical context. The laboratory findings, units, and available assay-specific reference intervals are summarized in Table 1.

Date	Laboratory test	Result	Units	Reference interval
Initial evaluation (December 2024)	Adrenocorticotrophic hormone	2000	pg/mL	7.2-63.3
	Morning cortisol	0.06	µg/dL	0.6-24.0 (1-5 years)
	Androstenedione	<0.24	ng/mL	Female, 2-6 years: <0.34
	17-Hydroxyprogesterone	<0.03	ng/mL	Female, 3-14 years: 0.07-1.70
	Free testosterone	<0.01	pg/mL	ND-1.3
	Dehydroepiandrosterone sulfate	3.40	µg/dL	24.4-209.7
	Sodium	140.1	mmol/L	136-146
	Potassium	3.41	mmol/L	3.6-5.1
Follow-up (April 2025)	Adrenocorticotrophic hormone	2000	pg/mL	7.2-63.3
	Dehydroepiandrosterone sulfate	0.20	µg/dL	2.8-85.2
	17-Hydroxyprogesterone	0.07	ng/mL	0.08-2.58
	Plasma renin activity	0.12	ng/mL/h	0.06-4.69
	Aldosterone	<3.7	ng/dL	Upright: 3.7-43.2; supine: 3.7-31.0
	Free testosterone	<0.24	pg/mL	Girls, 1-10 years: 0.24-1.43; boys, 1-10 years: 0.24-0.78
	Follicle-stimulating hormone	5.60	mIU/mL	Girls, 1-10 years: 0.46-5.98
	Luteinizing hormone	0.54	mIU/mL	Girls, 1-10 years: 0.03-3.9
	Anti-Müllerian hormone	17.32	ng/mL	Female, 0-29 years: 0.69-6.23; male, 0-29 years: 0.32-7.36
	Inhibin B	129.28	pg/mL	Girls: <83; boys: <352

TABLE 1: Endocrine and biochemical findings at the initial evaluation and follow-up

Reference intervals are those reported by the performing laboratories or documented in the clinical record unless otherwise indicated. Sex-specific reference intervals are reproduced where relevant because the patient has a 46,XY karyotype and was registered as female by the laboratories; these intervals should not be used in isolation to infer gonadal sex. Reference intervals may vary according to assay methodology and laboratory platform.

A pelvic ultrasound had initially described a small tubular structure interpreted as a retroverted uterus measuring 19×8×5 mm, with a linear endometrial echo; the ovaries were not visualized. Because this interpretation was discordant with the 46,XY karyotype and the Sertoli cell biomarkers, a contrast-enhanced abdominal and pelvic magnetic resonance imaging (MRI) study was subsequently performed in February 2026, when the patient was six years and one month old. The MRI report described adrenal glands with usual morphology. The ultrasound and MRI studies were subsequently re-evaluated in the picture archiving and communication system by an expert radiologist.

Targeted review of the pelvic MRI identified an oval structure in the right inguinal region, hypointense on T1-, T2-, and short tau inversion recovery-weighted sequences and surrounded by fluid signal, considered compatible with a probable undescended right testis in the inguinal canal. A second oval structure with similar signal characteristics was identified in the left iliac fossa, in contact with sigmoid colon loops, and was considered compatible with a probable intra-abdominal undescended left testis (Figure 1).

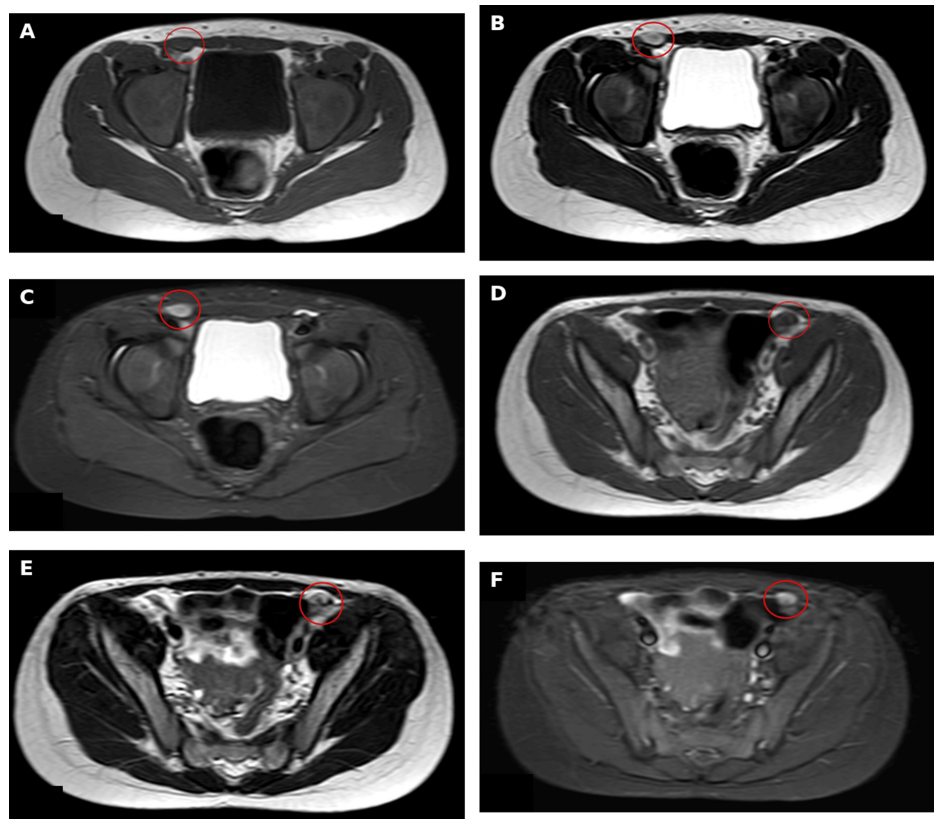


FIGURE 1: Magnetic resonance imaging findings suggestive of bilateral undescended testes

Axial pelvic magnetic resonance imaging demonstrates an oval structure in the right inguinal canal (red circle), compatible with a probable undescended right testis, on T1-weighted (A), T2-weighted (B), and short tau inversion recovery (C) sequences. A second oval structure is seen in the left iliac fossa (red circle), compatible with a probable intra-abdominal undescended left testis, on T1-weighted (D), T2-weighted (E), and short tau inversion recovery (F) sequences.

In addition, MRI demonstrated a linear structure between the urinary bladder and rectum without a clear connection to other pelvic organs. On targeted review, this was considered compatible with a rudimentary vaginal canal. The corresponding transabdominal ultrasonographic image showed a tubular hypoechoic structure posterior to the bladder, concordant with the MRI finding. Thus, the structure initially interpreted as a small uterus on ultrasonography was considered on expert reassessment to be more consistent with a rudimentary vaginal canal; the reviewed studies did not provide definitive evidence of a uterus (Figure 2).

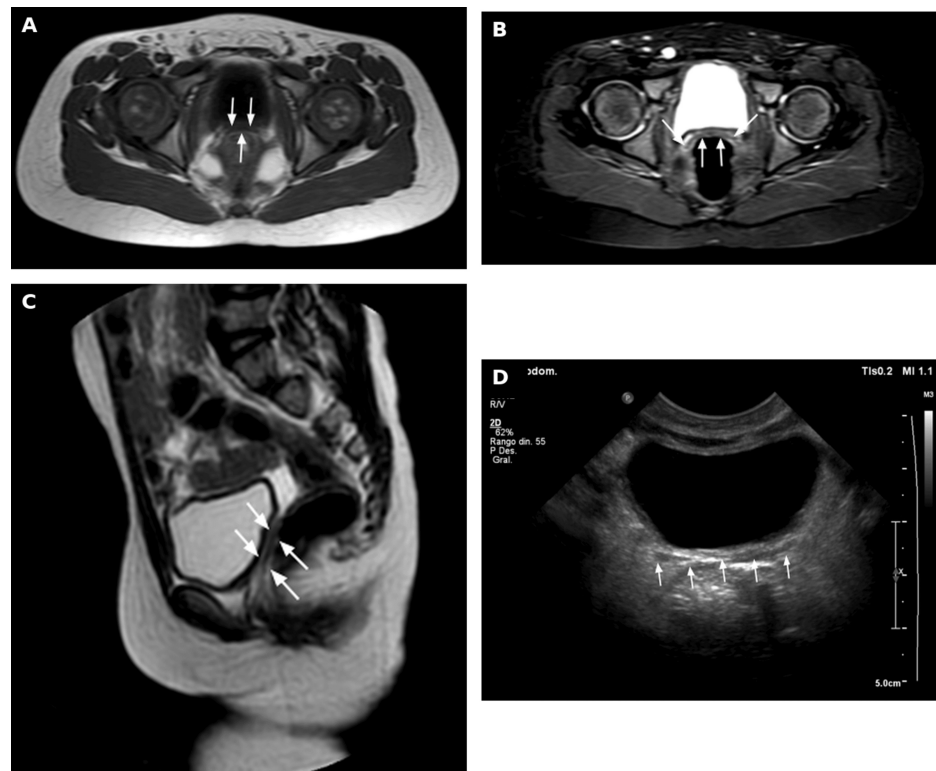


FIGURE 2: Magnetic resonance imaging and ultrasonographic correlation of a probable rudimentary vaginal canal

Pelvic magnetic resonance imaging demonstrates a linear structure between the urinary bladder and rectum (arrows) on axial T1-weighted (A), axial short tau inversion recovery (B), and sagittal T2-weighted (C) sequences. The structure was considered compatible with a rudimentary vaginal canal. Transabdominal ultrasonography (D) demonstrates a corresponding tubular hypoechoic structure posterior to the urinary bladder (arrows).

Molecular testing using a CAH gene panel identified a homozygous pathogenic truncating variant in *STAR* (NM_000349.3:c.577C>T; p.Arg193*). Parental segregation analysis was not available at the time of this report. Parental testing would be useful to document the segregation of the variant and to support autosomal recessive genetic counseling. The molecular finding, together with the early-onset PAI, globally reduced steroidogenesis, 46,XY karyotype, female-appearing external genitalia, preserved Sertoli cell biomarkers, and radiologic findings compatible with bilateral undescended testes, established the diagnosis of LCAH due to StAR deficiency. The patient remained clinically stable on hydrocortisone and fludrocortisone replacement, with continued multidisciplinary follow-up in pediatric endocrinology and medical genetics.

Discussion

LCAH is one of the most severe inherited disorders of steroidogenesis. StAR acts upstream of adrenal and gonadal steroid synthesis by facilitating cholesterol delivery to the inner mitochondrial membrane, where CYP11A1 catalyzes conversion of cholesterol to pregnenolone [4-7]. Severe disruption of this step produces a biochemical pattern that differs from more common forms of CAH, particularly 21-hydroxylase deficiency, in which steroid precursors proximal to the enzymatic block accumulate and androgen excess is typical [3].

The biochemical phenotype in this patient strongly suggested a proximal defect in steroidogenesis. Markedly elevated ACTH with profoundly low morning cortisol was consistent with PAI, while low 17-hydroxyprogesterone, androstenedione, dehydroepiandrosterone sulfate, and free testosterone indicated globally impaired steroid production rather than diversion toward androgen excess. This profile narrowed the differential diagnosis toward defects at the beginning of the steroidogenic pathway, principally StAR deficiency and CYP11A1 deficiency, making molecular testing decisive for etiologic confirmation.

The pathophysiology of classic StAR deficiency has traditionally been described by the two-hit model. The first hit is the acute reduction in StAR-dependent cholesterol transfer and steroidogenesis; limited StAR-independent steroid production may initially persist. The second hit reflects progressive accumulation of cholesterol and cholesterol esters in steroidogenic cells, with cellular injury and further loss of steroidogenic capacity [7]. This framework helps explain the evolution of adrenal and gonadal dysfunction and some of the

phenotypic variability observed among affected patients.

The homozygous *STAR* variant identified in our patient, NM_000349.3:c.577C>T (p.Arg193*), introduces a premature termination codon and is expected to result in severe loss of function. The p.Arg193* variant has previously been reported in LCAH [10]. The patient's early-onset adrenal insufficiency, female-appearing external genitalia despite a 46,XY karyotype, and profound impairment of androgen production are concordant with a classic phenotype. A recent systematic review of *StAR* deficiency demonstrated substantial genotype-phenotype heterogeneity and showed that nonsense and frameshift variants are predominantly associated with the most severe 46,XY phenotypes, whereas variants retaining partial activity can produce later-onset or nonclassic disease [11,12].

The 46,XY difference of sex development in this case is physiologically consistent with severe fetal Leydig cell steroidogenic failure. External genital virilization depends on adequate fetal androgen synthesis; severe *STAR* dysfunction can therefore result in female-appearing external genitalia. In contrast, Sertoli cell secretory activity is not dependent on the same steroidogenic pathway. AMH and inhibin B are therefore valuable markers of functioning testicular tissue in prepubertal children with 46,XY differences of sex development [8,9]. In our patient, suppressed testosterone together with measurable AMH and inhibin B supported a dissociation between severely impaired Leydig cell steroidogenesis and preserved Sertoli cell secretory activity.

The targeted radiologic reassessment added important anatomic evidence to this endocrine interpretation. MRI demonstrated structures compatible with bilateral undescended testes, with the right probable gonad in the inguinal canal and the left probable gonad in the abdominal cavity. These findings are concordant with the 46,XY karyotype and the Sertoli cell biomarkers and substantially strengthen the inference that testicular tissue is present. Because the gonads have not been surgically or histologically confirmed, however, the imaging findings should appropriately remain described as probable rather than definitive.

The reassessment also clarified an apparent discrepancy in the internal genital anatomy. Pelvic ultrasonography had initially suggested a small uterine structure, but expert review of MRI and ultrasonographic images considered the tubular structure posterior to the bladder to be more consistent with a rudimentary vaginal canal. The reviewed studies did not provide definitive evidence of a uterus. This distinction is biologically relevant because preserved Sertoli cell secretory activity and AMH production in a 46,XY child would generally favor Müllerian regression during fetal development [8,9]. Importantly, identification of a vaginal structure is not equivalent to demonstration of persistent Müllerian derivatives, and imaging in young children with differences of sex development may be limited by small structure size and incomplete tissue characterization.

Normal-appearing adrenal morphology on MRI does not contradict the diagnosis. In a recent systematic review of genetically confirmed *StAR* deficiency, adrenal hyperplasia was reported in only a proportion of patients, underscoring that adrenal imaging is neither required nor sufficiently sensitive to establish or exclude the disorder [11]. In this case, the biochemical phenotype and the biallelic pathogenic *STAR* variant provide substantially stronger diagnostic evidence than adrenal morphology. Beyond conventional morphologic imaging, shear-wave elastography has been investigated as a quantitative technique for assessing adrenal tissue stiffness. Gürün and Akdulum reported preliminary reference values for adrenal stiffness in healthy newborns [13]; however, the potential diagnostic utility of this technique in congenital adrenal disorders, including lipid congenital adrenal hyperplasia, remains to be established.

Identification of probable bilateral undescended testes also has implications for longitudinal care. Gonadal location and function should be considered in future surveillance and in individualized discussions regarding pubertal development, hormone replacement, fertility potential, and gonadal management.

Contemporary care for 46,XY differences of sex development emphasizes multidisciplinary evaluation and shared decision-making with the patient and family as developmentally appropriate [14].

Treatment of PAI caused by *StAR* deficiency requires lifelong glucocorticoid replacement and, when mineralocorticoid deficiency is present, mineralocorticoid replacement. Education regarding stress dosing, recognition of adrenal crisis, and access to emergency glucocorticoid treatment are core components of care [1,2]. Persistently elevated ACTH should not be used in isolation as a target for escalating glucocorticoid doses because complete biochemical normalization may require excessive exposure. Longitudinal assessment should integrate clinical status, growth, blood pressure, electrolytes, renin, and evidence of glucocorticoid or mineralocorticoid under- or overtreatment [1,2].

The diagnosis in this patient is supported by the convergence of multiple complementary findings. The combination of a steroid profile showing severe impairment of adrenal and gonadal steroidogenesis, a 46,XY karyotype, preserved Sertoli cell biomarkers, targeted expert radiologic identification of probable bilateral undescended testes, and a homozygous pathogenic *STAR* variant provides a consistent clinicobiochemical, anatomical, and molecular framework for classic LCAH due to *StAR* deficiency.

Conclusions

LCAH due to StAR deficiency should be considered in children with early-onset PAI when the biochemical profile shows markedly elevated ACTH with globally reduced cortisol, steroid precursors, and androgens. In 46,XY individuals, severe impairment of fetal Leydig cell steroidogenesis can result in female-appearing external genitalia despite relatively preserved Sertoli cell secretory activity. In this patient, the combination of PAI, a 46,XY karyotype, severely impaired androgen production, preserved Sertoli cell biomarkers, imaging findings compatible with bilateral undescended testes, and a homozygous pathogenic *STAR* variant established the diagnosis.

Targeted radiologic reassessment was particularly valuable in reconciling the endocrine findings with the internal anatomy and in distinguishing a probable rudimentary vaginal canal from a previously suspected uterine structure. Molecular confirmation and multidisciplinary longitudinal follow-up remain essential for genetic counseling, adrenal replacement therapy, evaluation of pubertal development, and future individualized gonadal management.

Additional Information

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All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

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Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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