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Case Report

Microscopic Müllerian Duct Remnants in a 46,XY Individual with Genetically Confirmed 17α-Hydroxylase/17,20-Lyase Deficiency: an Unexpected Histopathological Finding

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Abstract
17α-hydroxylase/17,20-lyase deficiency (17OHD) is a rare form of congenital adrenal hyperplasia characterized by impaired cortisol and sex steroid synthesis with accumulation of mineralocorticoid precursors. Although hypertension, hypokalemia, and sexual infantilism represent the classical phenotype, atypical presentations are increasingly recognized. We report a 15-year-old phenotypically female adolescent presenting with primary amenorrhea, absent secondary sexual characteristics, and a *46,XY* karyotype. Despite biochemical evidence of complete 17α-hydroxylase/17,20-lyase deficiency, no hypertension was documented on repeated office and home blood pressure measurements, and serum potassium concentrations remained within the reference range during serial testing. Molecular analysis identified a homozygous pathogenic variant in *CYP17A1* (c.374G>A; p.Arg125Gln). Laparoscopy demonstrated no macroscopic Müllerian structures; however, histopathological examination of the bilateral gonadectomy specimens revealed bilateral microscopic Müllerian duct remnants within the perigonadal fibrous tissue. Serum anti-Müllerian hormone concentration was 12.58 ng/mL, within the expected range for a pubertal 46,XY individual. Targeted sequencing of *AMH* and *AMHR2*, together with additional genes relevant to gonadal development and 46,XY disorders of sex development, identified no pathogenic variants, likely pathogenic variants, or variants of uncertain significance. This case expands the clinicopathological spectrum of complete 17α-hydroxylase/17,20-lyase deficiency by demonstrating that microscopic Müllerian duct remnants may persist despite a serum AMH concentration within the expected range, with no molecular explanation identified by targeted genetic analysis.
Keywords: Congenital adrenal hyperplasia; Steroid 17-alpha-hydroxylase; Anti-Müllerian hormone; Müllerian duct; Differences of sex development

What is already known on this topic?
Complete 17α-hydroxylase/17,20-lyase deficiency is a rare form of congenital adrenal hyperplasia that typically presents with hypertension, hypokalemia, sexual infantilism, and complete regression of Müllerian structures in 46,XY individuals due to preserved anti-Müllerian hormone (AMH) secretion.

What this study adds?
Microscopic Müllerian duct remnants may persist in genetically confirmed 46,XY 17α-hydroxylase/17,20-lyase deficiency despite a normal adolescent serum AMH concentration and no reportable variants in *AMH, AMHR2*, or selected gonadal-development genes. The finding underscores the diagnostic value of meticulous histopathological evaluation when macroscopic Müllerian structures are absent.

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Introduction
Congenital adrenal hyperplasia due to combined 17α-hydroxylase/17,20-lyase deficiency (17OHD) is a rare autosomal recessive disorder caused by biallelic pathogenic variants in *CYP17A1*, accounting for approximately 1% of all congenital adrenal hyperplasia cases. Impaired 17α-hydroxylase and 17,20-lyase activities disrupt cortisol and sex steroid biosynthesis while leading to accumulation of mineralocorticoid precursors, particularly deoxycorticosterone and corticosterone (1–5). Consequently, affected individuals typically present with sexual infantilism, delayed puberty, hypertension, and hypokalemia, although considerable phenotypic variability has been reported and normotensive presentations are increasingly recognized (1,2).
In 46,XY individuals with 17OHD, Sertoli cell differentiation and anti-Müllerian hormone (AMH) secretion are generally preserved, resulting in normal regression of the Müllerian ducts despite markedly impaired androgen production (6,7). Accordingly, persistence of Müllerian structures is considered an uncommon finding in genetically confirmed 17OHD. Careful gonadal evaluation, including histopathological examination, is also important in individuals with 46,XY disorders of sex development because of its diagnostic value and its role in assessing gonadal differentiation and potential germ cell tumor risk (8–10).
We report a genetically confirmed 46,XY adolescent with complete 17α-hydroxylase/17,20-lyase deficiency in whom microscopic Müllerian duct remnants were unexpectedly identified on histopathological examination despite the absence of macroscopic Müllerian structures at laparoscopy, normal serum AMH concentrations with no molecular explanation identified by the targeted genetic analyses performed.

Statement of Ethics
This case report was conducted in accordance with institutional and international ethical standards. According to the institutional policy of Şişli Hamidiye Etfal Training and Research Hospital, formal ethics committee approval was not required for this single-patient case report. Written informed consent was obtained from the patient and her parents for participation and for publication of the clinical data and accompanying images included in this report. The study was conducted in accordance with the ethical principles of the Declaration of Helsinki.

Case Presentation
A phenotypically female adolescent was referred at the age of 15 years because of primary amenorrhea and absent secondary sexual characteristics. She was born at term after an uneventful pregnancy to consanguineous parents, with no family history of disorders of sex development (DSD). On physical examination, her height was 154 cm (SDS: -1.1), weight was 53 kg (SDS: –0.95), and body mass index (BMI) was 22.5 kg/m² (SDS: +0.22). She had Tanner stage I breast development and absent axillary and pubic hair. External genitalia

appeared completely female. Systemic examination was otherwise unremarkable. Blood pressure at presentation was 115/70 mmHg (systolic SDS: +0.81; diastolic SDS: +0.60). Although ambulatory blood pressure monitoring was not performed, repeated office and home blood pressure measurements obtained over one week remained within the normal range.

Initial endocrine evaluation demonstrated hypergonadotropic hypogonadism, with markedly elevated gonadotropin concentrations (FSH 74.2 IU/L and LH 21.8 IU/L) in the setting of delayed puberty, prompting further investigation of adrenal steroidogenesis. Serum concentrations of total testosterone (0.025 ng/mL), estradiol (<15 ng/L), androstenedione (<0.01 ng/mL), and dehydroepiandrosterone sulfate (DHEA-S, 1.21 µg/dL) were markedly decreased. Serum 17-hydroxyprogesterone concentration was 0.21 ng/mL. As measurement of deoxycorticosterone (DOC) was unavailable, serum progesterone was used as an indirect marker of steroidogenic blockade and was found to be elevated (6.12 ng/mL). Basal serum cortisol was decreased (1.5 µg/dL) and demonstrated a suboptimal response to ACTH stimulation (peak cortisol: 13.6 µg/dL), accompanied by an elevated plasma ACTH concentration (204 ng/L), consistent with impaired cortisol synthesis resulting in increased ACTH secretion. Plasma renin activity was suppressed (0.38 ng/mL/h), whereas aldosterone concentration was markedly decreased (<0.37 ng/dL), consistent with mineralocorticoid precursor excess. Serum potassium was within the normal range at presentation (4.25 mmol/L) and remained within the reference range on serial measurements during the initial evaluation. No other electrolyte abnormalities were detected. **Serum anti-Müllerian hormone (AMH) concentration was 12.58 ng/mL (published reference interval for pubertal 46,XY males approximately 8–35 ng/mL), consistent with ongoing Sertoli cell AMH secretion.** The complete hormonal profile is summarized in Table 1.

Pelvic ultrasonography demonstrated the absence of Müllerian derivatives. Bilateral gonads with sonographic features consistent with testes were identified within the inguinal canals. Cytogenetic analysis revealed a *46,XY* karyotype. Molecular genetic analysis identified a homozygous pathogenic variant in *CYP17A1* (c.374G>A, p.Arg125Gln), a previously reported variant associated with complete 17α-hydroxylase/17,20-lyase deficiency (11–13).

Based on the clinical, biochemical, and genetic findings, a diagnosis of complete 17α-hydroxylase/17,20-lyase deficiency was established, and hydrocortisone replacement therapy was initiated. Psychiatric evaluation confirmed that the patient had a stable female gender identity. Following multidisciplinary evaluation and detailed counseling regarding the diagnosis, gonadal malignancy risk, fertility potential, and treatment options, bilateral laparoscopic gonadectomy was performed after written informed consent had been obtained. Intraoperatively, a blind-ending vaginal pouch was observed, and no macroscopic Müllerian structures were identified. Gross examination demonstrated bilateral testis-like gonads. Histopathological examination confirmed immature testicular tissue composed of seminiferous tubules in both gonads. Bilateral Müllerian-type epithelium with tubal differentiation was identified within the perigonadal fibrous tissue of the gonadectomy specimens. To further characterize these unexpected epithelial structures, immunohistochemical staining for WT1 and PAX8 was performed. WT1 immunoreactivity highlighted Sertoli cells within the immature seminiferous tubules, whereas bilateral nuclear PAX8 immunoreactivity was observed in the Müllerian-type epithelial structures. Müllerian differentiation was established on the basis of the combined histomorphological and immunohistochemical findings rather than PAX8 expression alone (Figure 1A–E). Immunohistochemical staining for OCT3/4 was negative, and no morphologic evidence of gonadoblastoma, germ cell neoplasia in situ, or other germ cell neoplasia was identified in the examined sections. Additional immunohistochemical evaluation for AMH could not be performed because the archived tissue block was considered unsuitable for further staining following pathological review. Given the unexpected identification of microscopic Müllerian remnants despite a normal serum AMH concentration, additional molecular analysis was undertaken. Targeted next-generation sequencing included *AMH* and *AMHR2*, together with additional genes relevant to gonadal development and 46,XY disorders of sex development, including *C14orf39, DHX37, SPIDR, MND1* and *XRCC2*. All coding exons and exon-intron boundaries were analyzed, and no pathogenic variants, likely pathogenic variants, or variants of uncertain significance were identified in the analyzed genes.

Estrogen replacement therapy was initiated for pubertal induction, while hydrocortisone replacement was continued. The patient remains under regular multidisciplinary follow-up.

Discussion

This report describes a genetically confirmed case of complete 17α-hydroxylase/17,20-lyase deficiency (17OHD) in a 46,XY adolescent presenting with female external genitalia, no documented hypertension on repeated office and home blood pressure measurements, serially normal serum potassium concentrations, and bilateral microscopic Müllerian duct remnants identified within the perigonadal fibrous tissue despite normal serum anti-Müllerian hormone (AMH) concentrations, the absence of macroscopic Müllerian structures, and no pathogenic variants, likely pathogenic variants, or variants of uncertain significance in the analyzed genes relevant to AMH signaling, gonadal development, and 46,XY disorders of sex development. To our knowledge, this represents one of the few reported cases in which microscopic Müllerian remnants have been documented histopathologically in genetically confirmed 17OHD despite a serum AMH concentration suggesting preserved Sertoli cell function. Although hypertension and hypokalemia have traditionally been regarded as cardinal manifestations of 17OHD, recent evidence indicates that these features are not universal. The recent meta-analysis by Willemsen et al. and the large Turkish cohort reported by Siklar et al. demonstrated that a subset of patients remain normotensive and normokalemic despite complete enzymatic deficiency, emphasizing the broad phenotypic spectrum of this disorder (1,2). Our patient exhibited the characteristic endocrine profile of complete 17OHD, including markedly elevated ACTH and gonadotropins together with profoundly reduced cortisol and sex steroid concentrations. Repeated office and home blood pressure measurements obtained over one week remained within the normal range, although ambulatory blood pressure monitoring was not performed. Serum potassium concentrations also remained within the reference range on serial measurements during the initial evaluation. These findings further support that the absence of documented hypertension on repeated measurements should not exclude the diagnosis of 17OHD [1,2].

The homozygous *CYP17A1* c.374G>A (p.Arg125Gln) pathogenic variant identified in our patient has previously been shown to abolish both 17α-hydroxylase and 17,20-lyase activities and has been associated with complete 17OHD (11–13). Despite the severe biochemical phenotype consistently observed with this variant, published cases have demonstrated variability in clinical manifestations, particularly regarding blood pressure and age at diagnosis. Our findings further illustrate that considerable phenotypic heterogeneity may exist even among individuals carrying the same pathogenic variant.

The most distinctive finding in our patient was the histopathological identification of microscopic Müllerian duct remnants. In 46,XY individuals with 17OHD, Sertoli cell differentiation and AMH secretion are generally preserved, resulting in normal Müllerian duct regression (6,7). Persistent Müllerian structures are therefore considered exceptional. Panesar et al. previously reported withdrawal vaginal bleeding following estrogen replacement in a phenotypically female individual with 46,XY 17OHD, suggesting the presence of hormonally responsive Müllerian tissue (14). However, neither serum AMH concentrations nor molecular evaluation of the AMH signaling pathway was available in that report. In contrast, our patient had normal serum AMH concentrations and underwent targeted sequencing of AMH and AMHR2, together with additional genes relevant to gonadal development and 46,XY disorders of sex development, without identification of pathogenic variants, likely pathogenic variants, or variants of uncertain significance. The normal serum AMH concentration measured during adolescence documents current Sertoli cell secretion but cannot retrospectively establish adequate AMH bioactivity during the critical fetal window of Müllerian duct regression. The absence of macroscopic Müllerian derivatives indicates that regression was largely effective. We therefore interpret the focal microscopic Müllerian-type epithelium with tubal differentiation as a possible consequence of incomplete local regression or tissue remodeling rather than as evidence of generalized AMH deficiency or AMHR2 resistance. No molecular explanation for this finding was identified by the genetic analyses performed. This mechanistic interpretation remains speculative. Because PAX8 expression is not specific for Müllerian epithelium and may also be observed in Wolffian-derived male genital tract epithelium, Müllerian differentiation was established on the basis of the combined histomorphological and immunohistochemical findings rather than PAX8 expression alone. Importantly, although no macroscopic Müllerian structures were identified during laparoscopy, histopathological examination demonstrated bilateral Müllerian-type epithelium with tubal differentiation within the perigonadal fibrous tissue, highlighting the value of meticulous histopathological assessment in individuals with disorders of sex development (15,16).

Recent evidence has further emphasized the importance of meticulous gonadal histopathological evaluation in patients with 46,XY disorders of sex development. Batatinha et al. reported estrogen-producing testicular tumors in patients with 17OHD and highlighted the value of

detailed pathological assessment for identifying clinically relevant gonadal abnormalities and guiding long-term management (8–10). Although no neoplastic changes were identified in our patient, the unexpected detection of microscopic Müllerian duct remnants reinforces the diagnostic value of comprehensive histopathological examination, even when preoperative imaging and intraoperative findings appear unremarkable. Beyond tumor surveillance, detailed histopathological evaluation may also reveal unexpected developmental abnormalities that are not evident clinically or radiologically, as demonstrated in the present case. This issue may be particularly relevant to 46,XY individuals with 17α-hydroxylase/17,20-lyase deficiency who are raised male and retain their gonads, because gonadal tumor risk remains incompletely defined. Detailed histopathological assessment, supplemented where appropriate by germ cell markers such as OCT3/4, may contribute to individualized risk assessment.

This report has several limitations. Functional assessment of AMH signaling was not performed. Although additional immunohistochemical evaluation for AMH expression within Sertoli cells was requested, it could not be performed because the archived tissue block was considered unsuitable for further staining after pathological review. Immunohistochemical evaluation with additional sex cord and germ cell markers (including TSPY, DMRT1, SOX9, SF1, 3β-HSD, FOXL2, and WNT4) was also not performed. However, OCT3/4 immunostaining was negative, and no morphologic evidence of gonadoblastoma, germ cell neoplasia in situ, or other germ cell neoplasia was identified. In addition, ambulatory blood pressure monitoring was not performed; therefore, masked hypertension could not be definitively excluded, although repeated office and home blood pressure measurements obtained over one week remained within the normal range. Finally, this report describes a single patient, limiting the generalizability of the findings.

Conclusion

In conclusion, this case expands the clinicopathological spectrum of complete 17α-hydroxylase/17,20-lyase deficiency by demonstrating an atypical presentation with repeatedly normal blood pressure and serum potassium despite biochemical evidence of mineralocorticoid precursor excess. The unexpected identification of bilateral microscopic Müllerian duct remnants despite normal serum AMH concentrations and the absence of a molecular explanation highlights the clinicopathological heterogeneity of this disorder and underscores the value of comprehensive histopathological and molecular evaluation in individuals with 46,XY disorders of sex development.

Conflict of Interest Statement

The authors declare no conflicts of interest.

Author Contributions

LGBO and AU conceived the study, managed the patient, interpreted the clinical findings, and drafted the manuscript. EI participated in the clinical management of the patient, contributed to data collection and interpretation, and critically revised the manuscript. SC performed the molecular genetic analyses, interpreted the genetic findings, and critically revised the manuscript. CT performed the histopathological evaluation and immunohistochemical analyses, interpreted the pathological findings, and critically revised the manuscript. BAB contributed to the interpretation of the clinical and laboratory findings and manuscript revision. ADC contributed to the study design, interpretation of the findings, and critical revision of the manuscript. All authors reviewed and approved the final version of the manuscript and agree to be accountable for all aspects of the work.

Data Availability Statement

All data generated or analyzed during this case report are included in this published article. No additional data are available.

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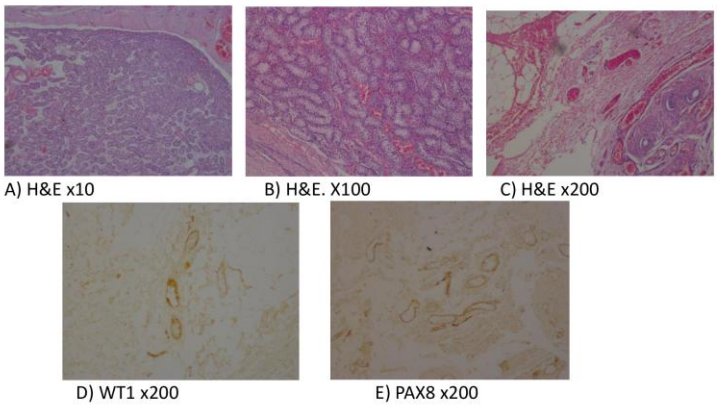

Figure 1. (A) H&E staining, ×10: Immature testicular tissue embedded in fibrotic adipose stroma. (B) Bilateral Müllerian-type epithelium with tubal differentiation within the perigonadal fibrous tissue. (C) Higher-power view of Müllerian-type epithelium with tubal differentiation within the perigonadal fibrous tissue. (D) WT1 immunostaining, ×200: WT1 immunoreactivity highlighting Sertoli cells within immature seminiferous tubules. (E) PAX8 immunostaining, ×200: Nuclear PAX8 immunoreactivity in the microscopic epithelial structures, supporting their Müllerian origin together with the histomorphological findings.

Table 1: Baseline Hormonal and Electrolyte Profile

Parameter	Case Value	Reference Range
Luteinizing Hormone (LH)	21.8 IU/L	1.5–12.4 IU/L*
Follicle-Stimulating Hormone (FSH)	74.2 IU/L	1.7–8.6 IU/L*
Total Testosterone	0.025 ng/mL	0.1–0.75 ng/mL*
Androstenedione	<0.01 ng/mL	0.75–3.1 ng/mL*
Cortisol (baseline)	1.5 µg/dL	6.7–22.6 µg/dL*
Adrenocorticotropic Hormone (ACTH)	204 ng/L	0–60 ng/L*
Progesterone	6.12 ng/mL	0.3–1.5 ng/mL*
Dehydroepiandrosterone Sulfate (DHEA-S)	1.21 µg/dL	35–430 µg/dL*
17-Hydroxyprogesterone (17-OHP)	0.21 ng/mL	0.2–1.3 ng/mL*
Estradiol (E2)	<15 ng/L	30–400 ng/L*
Plasma Renin Activity	0.38 ng/mL/h	1.5–6.5 ng/mL/h*
Aldosterone	<0.37 ng/dL	3.8–31.3 ng/dL*

Serum Sodium	142 mmol/L	135–145 mmol/L*
Serum Potassium	4.25 mmol/L	3.5–5.0 mmol/L*
Anti-Müllerian Hormone	12.58 ng/mL	Female adolescents: ~1–8 ng/mL; pubertal males: ~8–35 ng/mL**

* Reference intervals for gonadotropins and sex steroids correspond to the institutional laboratory reference ranges for pubertal females and the respective assays used at the time of testing. Because luteinizing hormone, follicle-stimulating hormone, estradiol, and progesterone concentrations vary according to Tanner stage and menstrual-cycle phase, these intervals are provided for general clinical interpretation. In this patient with a 46,XY disorder of sex development and absent spontaneous pubertal development, results were interpreted primarily in conjunction with the clinical, biochemical, and genetic findings rather than against a single sex-specific reference population.

** The AMH reference interval was interpreted according to age-appropriate 46,XY/pubertal male values because AMH reflects Sertoli cell secretion. Published pediatric AMH reference intervals vary according to age, sex, pubertal stage, and assay. The patient's serum AMH concentration of 12.58 ng/mL was within the expected range for pubertal 46,XY individuals.