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Fumarate Hydratase-Deficient Leiomyomas: A Retrospective Case Series with Clinical and Familial Findings Suggestive of Hereditary Leiomyomatosis and Renal Cell Carcinoma Syndrome

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ABSTRACT

OBJECTIVES: Fumarate hydratase (FH)-deficient uterine leiomyomas are a rare variant of smooth muscle tumors that may signal underlying hereditary syndromes like HLRCC, making their recognition crucial for gynecologists and pathologists.

STUDY DESIGN: This retrospective case series included ten patients who underwent surgery for uterine fibroids (benign smooth muscle tumors) between November 2015 and October 2023. Immunohistochemistry (IHC)-a lab technique using antibodies to detect specific proteins in tissue-was performed to confirm fumarate hydratase (FH) deficiency. After confirmation, patients underwent renal ultrasonography (kidney ultrasound) and had available imaging reviewed (CT and MRI, both advanced modalities). Each patient also underwent skin assessment for cutaneous leiomyomas (skin tumors) and received counseling for FH deficiency. Demographic, clinical, surgical, and familial data were collected and analyzed descriptively.

RESULTS: The mean age of patients was 40.2 years (range: 27-52), and the mean BMI was 26.9 ± 3.8 kg/m². Five patients underwent myomectomy, including one laparoscopically. The remaining five had a hysterectomy via Pfannenstiel incision. All diagnoses of FH-deficient leiomyomas were confirmed by immunohistochemistry. Two patients exhibited notable co-pathologies: one with adenomyosis and the other with a low-grade appendiceal mucinous neoplasm (LAMN). Family history findings included one patient with a sibling diagnosed with renal cell carcinoma, one patient with a sibling diagnosed with endometrial cancer, and another patient with first-degree relatives affected by uterine leiomyoma and endometrial intraepithelial neoplasia. Germline FH mutation analysis was not performed for any patient. Fertility-preserving procedures were offered to women of reproductive age, but no pregnancies were recorded during follow-up.

CONCLUSION: FH-deficient leiomyomas (fibroids lacking fumarate hydratase activity) should be considered in patients with large, multiple, or early-onset fibroids. Histopathology (examination of tissue under a microscope) and IHC (immunohistochemistry, a specialized lab technique for identifying proteins in tissue) are essential for diagnosis. Germline FH testing (testing for inherited mutations in the FH gene) was not systematically performed in our cohort. However, genetic referral (to a genetics specialist) was offered to 5 patients in the most recent year. This reflects a shift toward integrating molecular assessment into clinical practice. Our experience highlights the need to incorporate genetic counseling, routine renal imaging (regular kidney scans), and dermatologic evaluation (skin checks) into follow-up, especially for patients with features suggestive of a syndromic condition (that is, symptoms that could indicate a genetic syndrome).

Keywords: Uterine leiomyoma; Fumarate hydratase deficiency; Myomectomy; Immunohistochemistry; Hereditary leiomyomatosis and renal cell carcinoma

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Introduction

Hereditary leiomyomatosis and renal cell carcinoma (HLRCC) is a rare autosomal dominant syndrome. Autosomal

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dominant inheritance means that a single copy of the altered gene is sufficient to cause the disorder, whereas germline mutations are inherited changes present in all cells. HLRCC is caused by pathogenic variants in the fumarate hydratase (FH) gene and is defined by a triad: cutaneous leiomyomas (benign skin smooth muscle tumors), uterine leiomyomas (uterine fibroids), and type II papillary renal cell carcinoma (an aggressive kidney cancer frequently affecting young individuals) (1,2). The FH gene mutations were first identified in siblings with progressive encephalopathy and fumarase deficiency by Bourgeron et al. in 1994 (3). Subsequent research has expanded the known spectrum of FH-related disorders. As of February 2025, 474 FH gene variants have been documented in 368 individuals globally (4), encompassing a phenotypic continuum from autosomal recessive fumarase deficiency (requiring biallelic mutations) to autosomal dominant HLRCC syndrome, illustrating the range of consequences associated with FH dysfunction.

In women, uterine leiomyomas frequently represent the earliest and most prevalent clinical manifestation of HLRCC. Up to 80–100% of affected females develop multiple, symptomatic fibroids (2,5), which often arise at a younger age and necessitate surgical intervention, such as myomectomy (uterus-sparing fibroid removal) or hysterectomy (uterine removal). These interventions are typically required about a decade earlier than in women with sporadic leiomyomas (5).

While renal cell carcinoma (RCC) occurs less frequently in HLRCC than leiomyomas, its prognosis is poor due to its aggressive nature. Early detection of FH deficiency is crucial for initiating clinical surveillance and facilitating genetic counseling. Uterine leiomyomas with FH deficiency should prompt consideration of a syndromic process, especially in cases with early-onset, multiplicity, or large tumors. Emerging data indicate that FH-deficient leiomyomas can also develop sporadically owing to somatic mutations, complicating diagnosis (6).

Management should be tailored to patient age, symptomatology, and fertility goals. Myomectomy is generally prioritized for patients of reproductive age, although disease extent can restrict minimally invasive options (7,8). Regardless of renal findings at presentation, a diagnosis of FH deficiency warrants oncogenetic consultation. Evaluation of familial cancer risk is imperative (1,5).

With this background, we present ten patients with FH-deficient uterine leiomyomas (fibroids lacking the fumarate hydratase enzyme). We aim to describe their clinical features, surgical outcomes, and key familial findings in the context of HLRCC. This sets the stage for the methods and results below.

Material and Method

This retrospective, observational case series was conducted at the Department of Obstetrics and Gynecology at the University of Health Sciences, Istanbul Training and Research Hospital. Medical records from November 1, 2015, to November 1, 2023, were reviewed.

Due to the retrospective design, all leiomyoma (benign uterine smooth muscle tumor) specimens during the study period were examined. Immunohistochemical (IHC) staining-a laboratory process that uses antibodies to detect specific proteins in tissue-was not routinely available before 2021 due to cost constraints and limited awareness. In earlier years, IHC analysis was performed only in cases with clinical or pathological suspicion. After 2021, its use became more routine as awareness and access increased.

A total of ten female patients were diagnosed with FH-deficient uterine leiomyomas based on histopathological evaluation and IHC findings.

The study was approved by the Clinical Research Ethics Committee of the University of Health Sciences, Istanbul Training and Research Hospital (Approval No: 295, Date: 10.11.2023). All procedures followed the Declaration of Helsinki, an international set of research ethics guidelines.

Because the study was retrospective, informed consent was waived. Patient confidentiality was strictly maintained.

Inclusion criteria were as follows: Patients who underwent surgical resection of uterine fibroids with histopathologically confirmed FH-deficient leiomyomas based on immunohistochemical staining.

Clinical features, such as a personal or family history of RCC and cutaneous findings suggestive of HLRCC, were recorded as variables rather than inclusion criteria.

Exclusion criteria included: Cases with incomplete medical records, insufficient pathology material for IHC analysis, or patients who could not be reached for follow-up.

Incidental findings, including low-grade neoplasms such as low-grade appendiceal mucinous neoplasm (LAMN), were recorded but were not considered exclusion criteria.

Genetic counseling was not an inclusion criterion; however, referral was offered to selected patients in the most recent period.

Data collection: Data were obtained retrospectively by reviewing the hospital's electronic health records and pathology reports. After confirmation of FH deficiency, all patients were invited for clinical reassessment. Postoperative evaluations included renal ultrasonography and a review of any available CT or MRI imaging. Each patient also had a dermatological evaluation for cutaneous leiomyomas. Patients were informed about FH deficiency and counseled on the importance of routine follow-up.

Data collection tool: A structured data collection form was developed by the investigators based on a literature review. The form captured sociodemographic characteristics, clinical symptoms, surgical details, pathological features, family history, and follow-up outcomes.

Statistical analysis

Descriptive statistics were used to summarize the data. Continuous variables such as age, BMI, hemoglobin levels, and fibroid dimensions were presented as mean ± standard deviation (SD) and range (minimum-maximum). Categorical variables, including type of surgery and comorbidities, were reported as frequencies. No inferential statistical analysis was performed due to the small sample size.

Results

During the study period, surgical data included 661 myomectomy cases and approximately 2,100 hysterectomies performed for fibroid-related indications. Among these, 10 patients were diagnosed with FH-deficient uterine leiomyomas, corresponding to approximately 0.4% of our cohort; however, this should not be interpreted as a true incidence due to non-uniform application of FH immunohistochemistry during the study period. The mean age of affected patients was 40.2±8.0 years (range: 27-52), and the mean body mass index (BMI) was 26.9±3.8 kg/m² (20.3-35.6) (Table I). The most common presenting symptoms were abnormal uterine bleeding (n=4) and chronic pelvic pain (n=3). None of the patients presented with cutaneous leiomyomas, and no RCC was detected during the evaluation period.

Anemia was observed in half of the patients at admission, with mean hemoglobin levels of 11.5±2.4 g/dL (7.2-14.4). Preoperative hemoglobin improved to a mean of 12.1±1.7 g/dL, while discharge values averaged 9.9±1.1 g/dL. Preoperative ultrasound revealed fibroids measuring between 43 mm and 85 mm (65.4±15.0 mm), and the largest fibroid on pathological examination reached 95 mm (69.7±19.1 mm). Renal ultrasonography was performed in all patients and was unremarkable.

Comorbidities were present in four patients: diabetes mellitus (DM) in Case 1; hypertension (HT) and DM in Case 4; HT and transient ischemic attack in Case 7; and HT with valvular heart disease in Case 10. No major chronic illnesses were reported in the remaining six patients. These comorbidities were taken into account during surgical planning and postoperative follow-up.

Macroscopically, the excised tumors appeared as firm, whitish, elastic masses with homogeneous cut surfaces, consistent with leiomyoma nonmorphology. An illustrative example is provided in Case 3, who underwent myomectomy for a 95-mm lesion (Figure 1). Pathological examination confirmed FH-deficient leiomyomas in all ten cases.

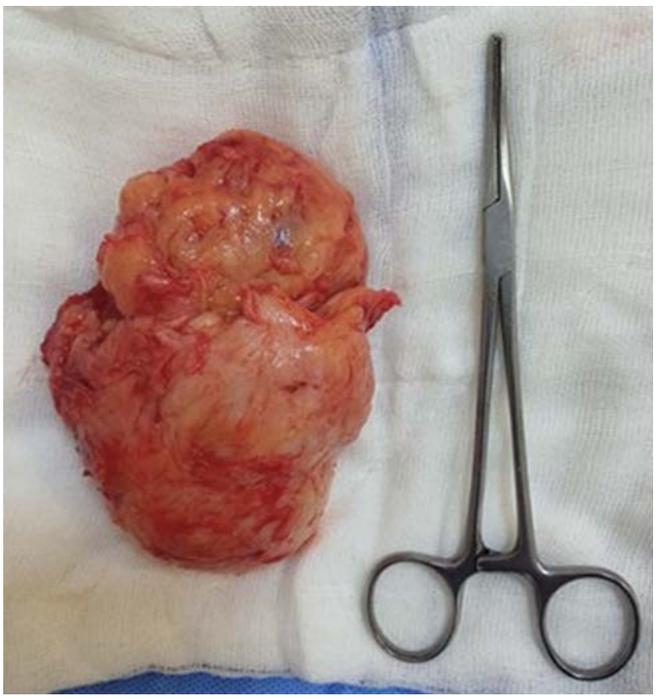

Figure 1: *Macroscopic appearance of the excised uterine leiomyoma from Case 3. The specimen measured 95 × 65 × 65 mm, with a firm, whitish, elastic texture and a homogeneous gray-white cut surface, consistent with fumarate hydratase-deficient leiomyoma.*

Histopathological features such as bizarre nuclei, mild atypia, and low mitotic activity were described in several pathology reports; however, detailed morphologic features characteristic of FH deficiency were not consistently reported across all cases.

Table I: *Demographic and clinical characteristics of patients with FH-deficient leiomyomas*

Variable	X±SD	Min	Max
Age (years)	40.2±8	27	52
Gravida	2.1±1.7	0	6
BMI(Body Mass Index)(kg/m²)	26.9±3.8	20.3	35.6
Baseline Hemoglobin (Hb)(g/dL)	11.5±2.4	7.2	14.4
Preoperative Hb(g/dL)	12.1±1.7	9.4	14.4
Discharge Hb(g/dL)	9.9±1.1	8.2	11.3
Largest fibroid diameter on US(mm)	65.4±15.0	43	85
Largest fibroid diameter on Pathology(mm)	69.7±19.1	45	95
Postoperative hospital stay(days)	2.7±1.3	2	6

Preoperative MRI was available for Case 9, demonstrating a large intramural leiomyoma. Coronal T2-weighted images showed a bulky pelvic mass compressing the bladder, with the uterus not clearly delineated. On sagittal views, the lesion was seen arising from the anterior uterine wall, consistent with a FIGO type 4-6 intramural fibroid, without distortion of the endometrial cavity. Axial sections further outlined the tumor margins and confirmed its intramural localization with mass effect on the uterine contour (Figure 2). Among the associated findings, Case 4 was notable for coexisting adenomyosis, while Case 7 revealed a LAMN (Table II). Due to the LAMN diagnosis, the patient was also referred for follow-up by the General Surgery department. In Case 3, a family history revealed uterine fibroids in two sisters and endometrial intraepithelial neoplasia in another. Additionally, in Case 10, a sibling had a history of endometrial cancer. In Case 4, a sibling had a history of renal cell carcinoma.

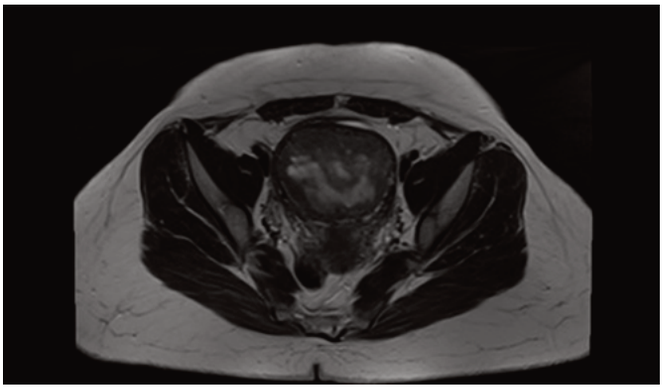

Figure 2: *Preoperative pelvic MRI of Case 9. (A) Coronal T2-weighted image demonstrates a large pelvic mass compressing the bladder, with the uterus not clearly visualized. (B) Sagittal T2-weighted image shows the lesion arising from the anterior uterine wall, consistent with a FIGO type 4-6 intramural leiomyoma, without distortion of the endometrial cavity. (C) An axial T2-weighted image delineates the tumor margins and confirms its intramural localization, with mass effect on the uterine contour.*

During follow-up, a pancreatic lesion was detected in Case 3 at postoperative month 7 and was later identified as a pseudocyst. The patient remains under clinical surveillance. No definitive association with FH deficiency has been established.

Surgical management included myomectomy (n=5) and hysterectomy (n=5). Among these, one myomectomy was performed laparoscopically (Case 5), while the remaining nine surgeries were conducted via Pfannenstiel incision. These details, along with the patients' demographic and clinical characteristics, are summarized in Table II.

These findings highlight the potential importance of histopathological recognition of FH-deficient leiomyomas in patients presenting with symptomatic or multiple fibroids. Although no RCC was detected in this series, the familial history findings underscore the importance of risk-based surveillance and genetic referral.

Follow-up data were available for all patients. Three patients were followed for approximately 5 years but were sub-

Table II: *Surgical and histopathological findings of patients with FH-deficient leiomyomas*

Case	Age	Symptom Increased fibroid size	Smoking	Medical History	Family History	Co-pathologies	Surgery
1	27	Increased fibroid size	Yes	DM	None	None	Myomectomy (abdominal)
2	45	Chronic pelvic pain	No	None	None	None	TAH+BSO
3	39	Abnormal uterine bleeding	Yes	None	Two sisters with myoma, one with endometrial intraepithelial neoplasia	None	Myomectomy (abdominal)
4	47	Chronic pelvic pain	No	HT, DM	Sibling with RCC	Adenomyosis	TAH+BSO
5	34	Abnormal uterine bleeding	No	None	None	None	Myomectomy (laparoscopic)
6	45	Chronic pelvic pain	No	None	None	None	TAH+BS
7	41	Secondary infertility	Yes	HT, TIA	None	LAMN	TAH+right USO+left salpingectomy+appendec-
8	43	Abnormal uterine bleeding	No	None	None	None	tomy
9	29	Abnormal uterine	No	None	None	None	Myomectomy (abdominal)
10	52	bleeding	No	HT, VHD	Sibling with endometrial cancer	None	Myomectomy (abdominal) TAH+BS

TAH: Total abdominal hysterectomy; BSO: Bilateral salpingo-oophorectomy; BS: Bilateral salpingectomy; USO: Unilateral salpingo-oophorectomy; HT: Hypertension; DM: Diabetes mellitus; VHD: Valvular heart disease; TIA: Transient ischemic attack; RCC: Renal cell carcinoma; LAMN: Low-grade appendiceal mucinous neoplasm

sequently lost to follow-up due to loss of contact information. The remaining seven patients have been followed for 2 to 5 years, and no recurrences, reoperations, or renal pathologies have been detected to date; however, the relatively short and heterogeneous follow-up period should be taken into consideration. In Case 3, a pancreatic lesion was identified at 7 months postoperatively, and the patient was subsequently referred for genetic counseling and further follow-up.

Discussion

FH-deficient leiomyomas represent a rare and distinct subtype of uterine fibroids, frequently linked to HLRCC syndrome, an autosomal dominant disorder caused by germline mutations in the FH gene (1). Although many cases occur sporadically, the recognition of this variant has gained importance because of its potential association with aggressive renal cell carcinoma and familial cancer syndromes (2,5).

In our series of ten patients with confirmed FH-deficient leiomyomas, the clinical features were consistent with previous reports, most commonly involving younger women presenting with large, symptomatic, or multiple fibroids (9). The mean age was 40.2 years, and all patients required surgical intervention. Myomectomy was performed in five cases, including one laparoscopic procedure, while the remaining five underwent hysterectomy, mainly via Pfannenstiel incision. Histopathological evaluation supported by IHC demonstrated complete loss of FH expression in tumor cells, which remains the cornerstone of diagnosis (10).

Two patients exhibited coexisting gynecological pathologies: adenomyosis in one and a LAMN in another (11,12). The latter finding is regarded as incidental, since no association between FH deficiency and appendiceal mucinous tumors has been documented in the literature. Additionally, one patient was found to have a pancreatic lesion during follow-up, which was later identified as a pseudocyst and is currently under clinical surveillance. No established association with FH deficiency was identified.

Family history findings also highlighted potential syndromic relevance: one patient had a sibling diagnosed with renal cell carcinoma, while another had several first-degree relatives with uterine and endometrial pathology. Although molecular testing was not routinely performed throughout the study period, genetic referral was offered to five patients in the most recent year, reflecting an evolving clinical approach. However, in the absence of systematic germline FH testing, a definitive diagnosis of HLRCC could not be established.

Follow-up data were heterogeneous. Three patients were monitored for approximately five years before being lost to follow-up due to loss of contact information. The remaining seven have continued surveillance for 2-5 years, with no recurrences, reoperations, or renal malignancies detected to date. These findings suggest favorable short- to mid-term outcomes, but they also emphasize the necessity of prolonged monitoring in this patient population.

Fertility preservation is an important consideration in FH-deficient leiomyomas. In our study, myomectomy was selected in reproductive-age women, consistent with the literature suggesting that these patients often undergo surgery nearly a decade earlier than those with sporadic fibroids because of larger size, multiplicity, and more severe symptoms (3,6). While myomectomy supports fertility potential, it carries a theoretical risk of uterine rupture in subsequent pregnancies. Nevertheless, available studies, including that of La Verde et al., have not demonstrated an increased risk of rupture in women who delivered vaginally after myomectomy (13). In our cohort, five patients underwent uterus-sparing surgery, but no pregnancies were recorded during follow-up, preventing clinical conclusions on obstetric outcomes.

Comparative literature suggests that FH-deficient leiomyomas are frequently misclassified as conventional fibroids, particularly in younger patients. Studies have estimated that approximately 0.4% of all uterine leiomyomas are FH-deficient, with most lacking overt syndromic features (10,14). Siegler et al. reviewed 22 IHC-confirmed FH-deficient leiomyomas and found that the majority occurred in women under 40, with frequent multiplicity and large tumor size (9). Similarly, our findings reflect a high proportion of large, symptomatic leiomyomas requiring definitive surgical management.

Of note, one patient in our cohort had a history of TIA. However, no clear association between FH deficiency and cerebrovascular disease has been established, and this finding is likely incidental.

Overall, our study underscores the critical role of histopathology and IHC in identifying FH-deficient leiomyomas, particularly in young patients with multiple or rapidly enlarging fibroids, as recognition of this entity has important clinical implications (2,5,15).

Clinical management may benefit from a multidisciplinary approach, including consideration of genetic counseling and appropriate surveillance strategies, particularly in patients with features suggestive of a syndromic condition.

This study has several limitations that should be acknowledged. First, the small sample size and retrospective design limit the generalizability of the findings. The analysis relied on archived medical records, which may be incomplete or inconsistent. Second, immunohistochemical evaluation for FH was not systematically applied to all leiomyoma specimens, especially before 2021, when awareness and accessibility were limited, potentially leading to underdetection in earlier years. Third, germline FH mutation testing was not routinely

performed, and only five patients were referred for genetic counseling in the most recent year, reducing the molecular contribution of the study. Follow-up was variable, with three patients lost after 5 years and seven patients under ongoing surveillance for 2-5 years. However, no recurrences or renal malignancies have been observed; these data are insufficient to assess long-term outcomes. Finally, as a single-center experience, the results may not be broadly generalizable.

Nevertheless, this study provides meaningful insights into the clinical and pathological spectrum of FH-deficient leiomyomas, an entity that remains underrecognized in gynecologic practice. By detailing patient characteristics, surgical approaches, and medium-term outcomes, our findings underscore the need for heightened awareness, structured surveillance, and integration of genetic evaluation in suspected cases.

Conclusion

FH-deficient uterine leiomyomas are uncommon tumors that may serve as a marker for underlying hereditary syndromes. In our cohort of ten patients, diagnosis was consistently established by histopathology and immunohistochemistry, underscoring their central role in clinical recognition. Fertility-preserving surgery was feasible in selected cases, although long-term reproductive outcomes could not be assessed. Limited genetic testing and variable follow-up highlight the need for greater awareness, routine referral for genetic counseling, and structured long-term surveillance, particularly in young women and those with suggestive family histories. Multidisciplinary collaboration between gynecologists, pathologists, and genetic specialists remains essential for appropriate patient management and familial risk assessment.

Declarations

Ethics approval and consent to participate: The study was approved by the Clinical Research Ethics Committee of the University of Training and Research Hospital (Approval No: 295, Date: 10.11.2023). All procedures were conducted in accordance with the principles of the Declaration of Helsinki. Due to the study's retrospective nature, informed consent was waived; however, patient confidentiality was strictly maintained.

Availability of data and materials: The data supporting this study are available through the corresponding author upon reasonable request.

Competing interests: The authors declare that they have no competing interests.

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Authors' Contribution: Surgical and Medical Practices: TI, NU, ZYY; Concept: TI, SCO, NU, ZYY; Design: TI, SCO, NU, ZYY; Data Collection or Processing: TI, NU, ZYY; Analysis or Interpretation: TI, SCO; Literature Review: TI, SCO, NU, ZYY; Writing: TI; Critical Review: TI, SCO.

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