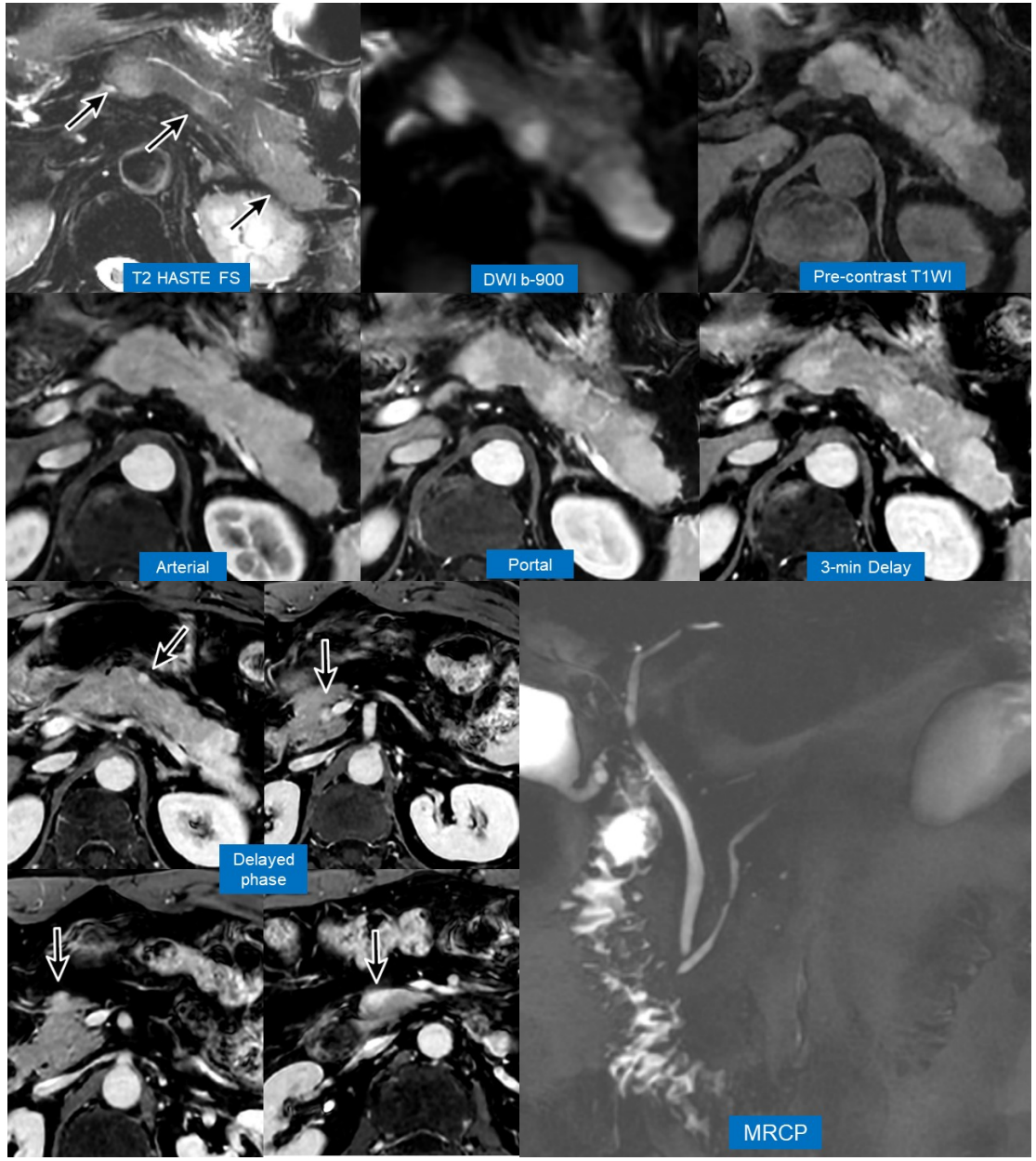
Case of the Day: Answer and Explanation

Q1. What is your diagnosis?



64/M

*** C.C: Incidentally detected pancreatic mass**

*** Past medical history: NS**

*** Lab: CA 19-9, 2.5 U/mL (normal < 37)**

Answer

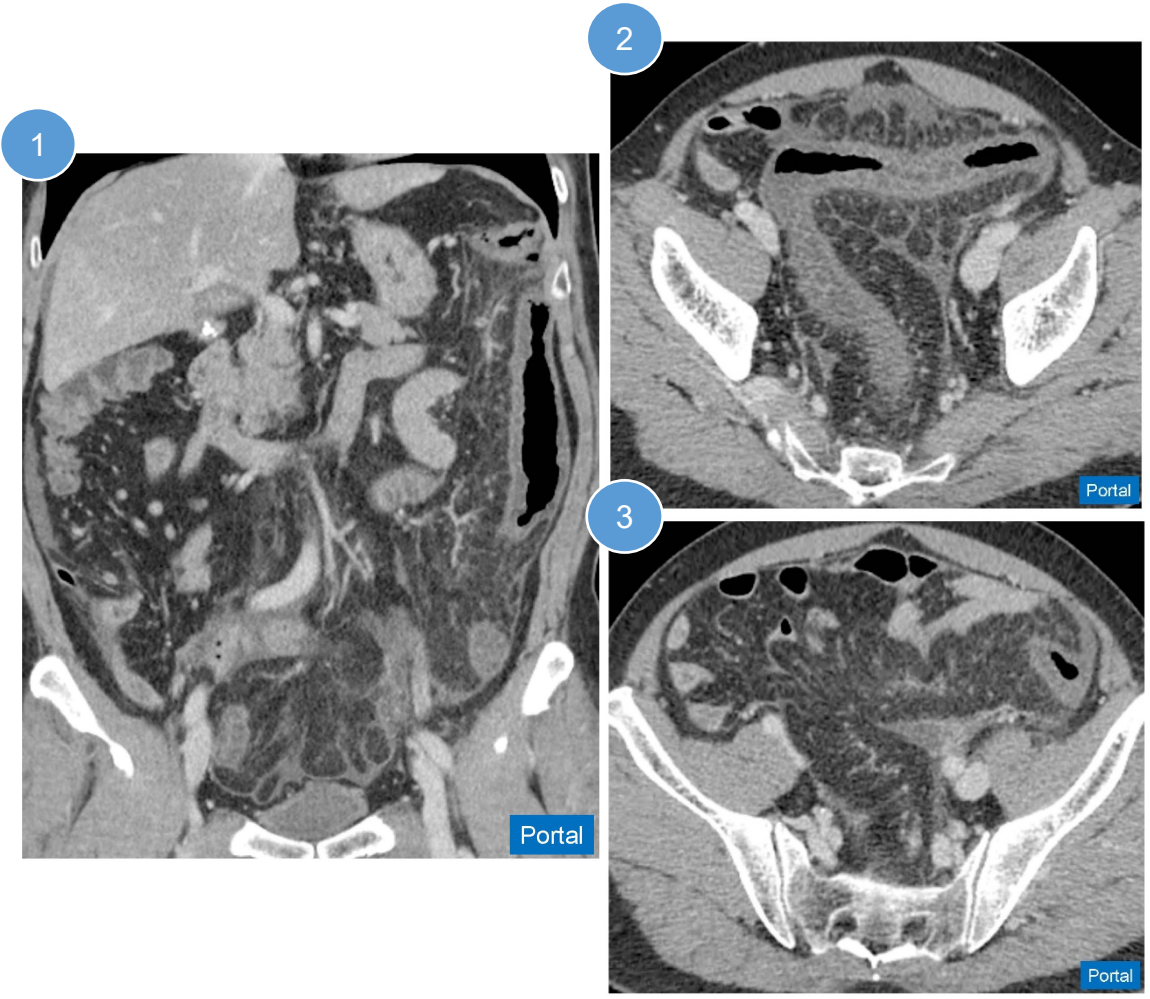
- AIP (or Type 1 AIP)
- Autoimmune pancreatitis
- IgG4-related AIP
- IgG4-related autoimmune pancreatitis
- IgG4-related disease

Explanation

Autoimmune pancreatitis (AIP)

- **Type 1 AIP**: Pancreatic manifestation of IgG4-related disease
- **Histopathology**
  - Dense lymphoplasmacytic infiltrate predominantly involving lobules
  - Storiform fibrosis and obliterative phlebitis
  - Diffuse infiltration of IgG4+ plasma cells
- **Focal AIP (33-41%)**: difficult to differentiated from pancreas tumors
  - **Radiologic features favoring AIP**
    - Homogeneous delayed enhancement
    - Multiple pancreatic masses
    - Multiple MPD strictures & penetrating duct sign
    - Speckled enhancement
    - Other organ involvement

Q2. What is your diagnosis?



67/M
*** Chief complaint: hematochezia, abdominal pain**

Answer

- Idiopathic myointimal hyperplasia of the mesenteric veins
- IMHV
- Myointimal hyperplasia of the mesenteric vein

Explanation

Idiopathic myointimal hyperplasia of the mesenteric veins (IMHMV)

Pathophysiology

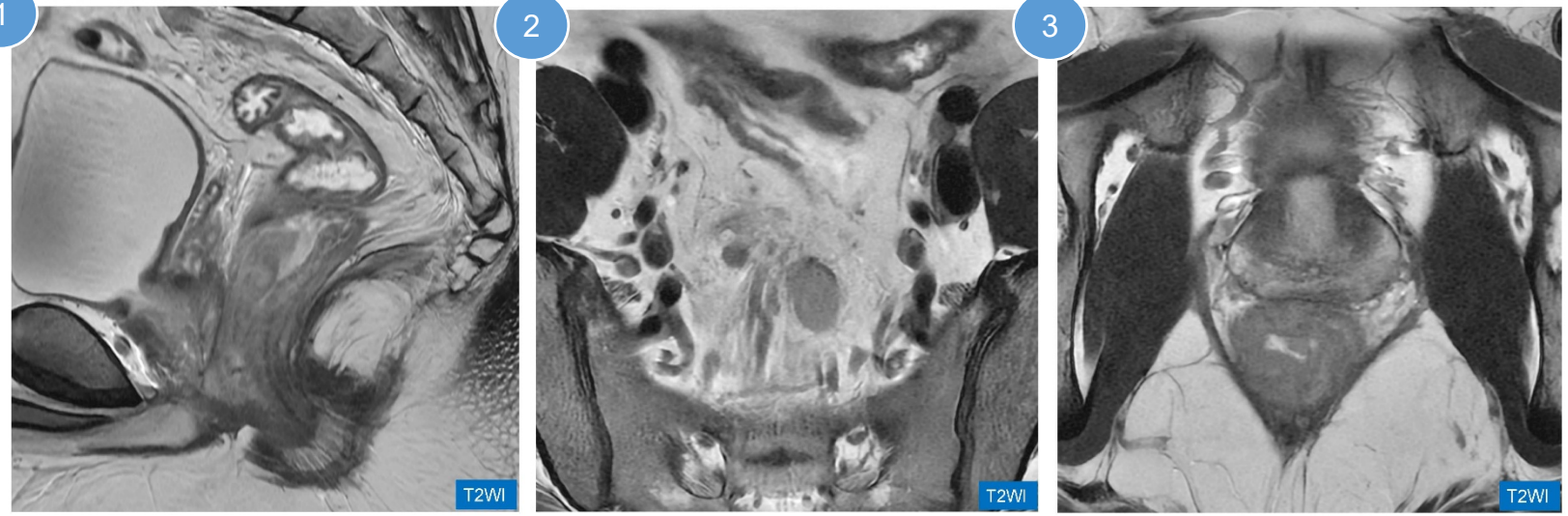
- Non-thrombotic, non-inflammatory disease Histopathology
- Intimal smooth muscle hyperplasia of large- to medium-sized mesenteric veins
- Results in chronic venous outflow obstruction
- Predominant involvement of the rectum and sigmoid colon

Imaging findings

- Findings of venous ischemia, such as low-attenuating wall thickening of rectosigmoid colon
- Characteristic vascular findings:
    - Severely narrowed, or absent inferior mesenteric vein, with subsequent collaterals
    - Tiny dysmorphic veins around the affected bowel, with some showing arterial enhancement (possibly due to arterio-venous fistula) or aneurysmal dilatation

Q3. What is your diagnosis?

34/M
*** Chief complaint: hematochezia**



Answer
• Any answer containing **'Syphilis'** or **'Chancre'** will be marked as correct.

Fig 1 T2-weighted image in sagittal plane showing rectal wall thickening and multiple enlarged lymph nodes
Fig 2 T2-weighted image in axial plane showing enlarged lymph node in the mesorectum
Fig 3 T2-weighted image in axial plane showing mass-like wall thickening of the low rectum

Explanation

Rectal syphilis
• Rectal manifestation of Treponema pallidum infection
• Can affect any sexually active individual, but occurs most commonly in patients with receptive anal intercourse

Clinical features
• Rectal pain, bleeding, tenesmus, mucus discharge, or mass-like lesion
• May accompany lymphadenopathy

Imaging findings
• Findings that mimic rectal cancer, inflammatory bowel disease, or ischemic proctitis
• Rectal wall thickening, ulceration, or infiltrative mass
• Associated lymphadenopathy

Diagnosis and treatment
• Histologic findings are nonspecific, and definitive diagnosis relies on serologic testing (RPR/VDRL with treponemal confirmation)
• Symptoms typically resolve rapidly with penicillin therapy, making recognition crucial to avoid unnecessary surgery or oncologic treatment

Q4. What is the diagnosis of gastric lesions before and after surgery? The gastric mass was completely resected surgically withnegative margins. Please submit the diagnoses of each one.

44/F

*** Chief complaint: incidental gastric mass**

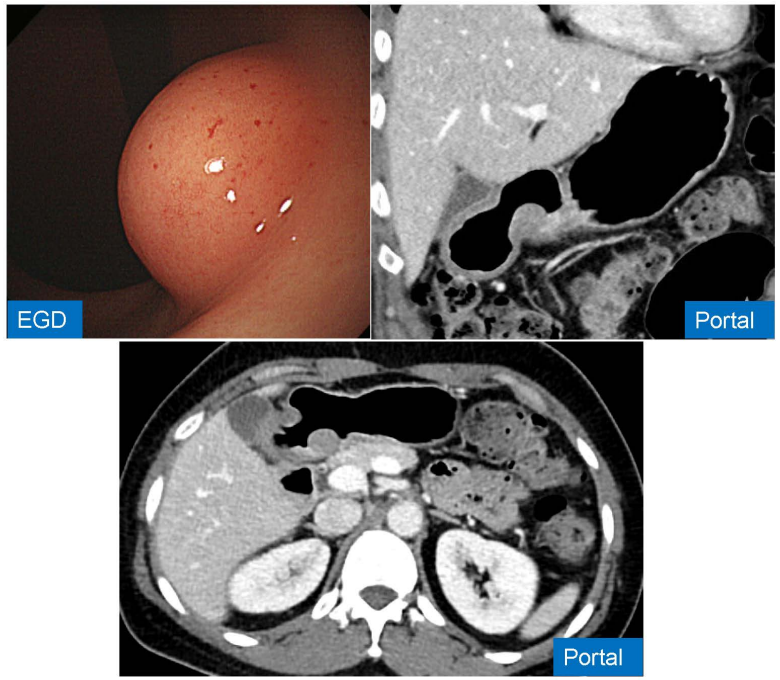


Figure 1. Before surgery

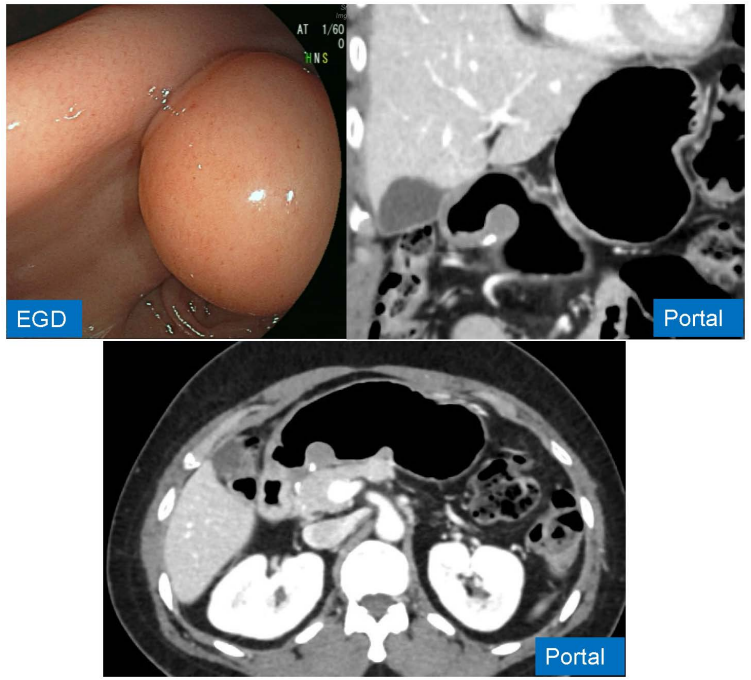


Figure 2. 7 months after surgery

Answer

1) Before surgery: gastric leiomyoma
2) After surgery: desmoid tumor
(= fibromatosis, desmoid type fibromatosis).

Explanation

1. Summary of this case

• **Preoperative images**

✓ CT: well-defined mass in the gastric antrum with intact overlying mucosa, suggesting a subepithelial tumor.

✓ The gastric tumor was confirmed to be a leiomyoma after wedge resection

• **After gastric wedge resection,**

✓ CT: well-defined low attenuation gastric mass at the surgical site with a surgical clip.

✓ Upon subsequent surgery, it was confirmed to be a desmoid tumor.

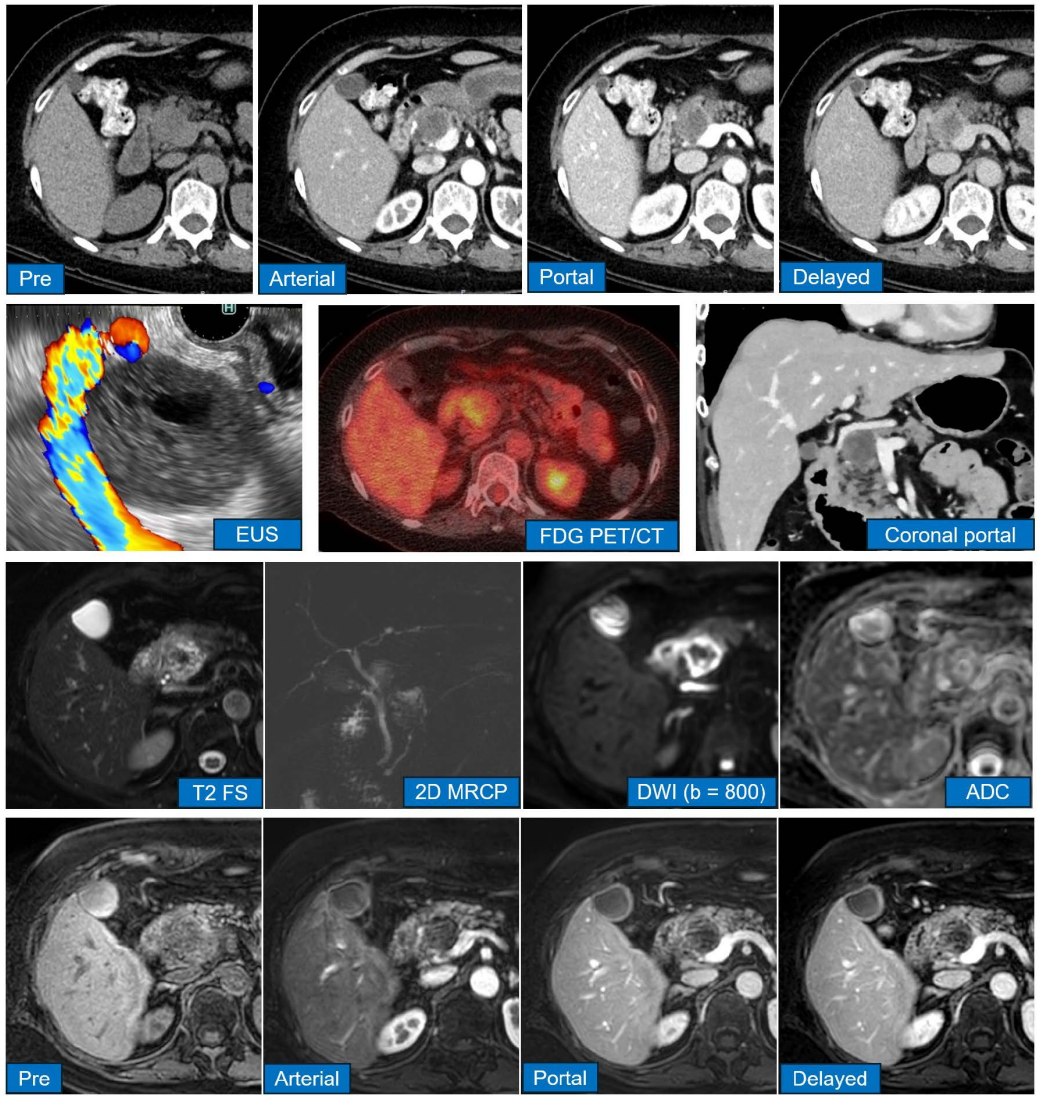
2. Review

• Although rare, desmoid tumors arising at the anastomotic site after gastric cancer surgery have been reported. (https://doi.org/10.1186/s13256-022-03635-w)

Q5. What is the diagnosis of the pancreatic mass?

65/F

*** Chief complaint: incidental pancreatic mass without laboratory abnormality**



Answer

• pancreatic schwannoma

Explanation

Pancreatic schwannoma

• An extremely rare tumor that typically occurs in middle-aged adults, with no clear sex predilection.

• **CT and MRI:**

✓ Well-defined pancreatic mass with cystic degeneration

✓ Heterogeneous T2 hyperintensity

✓ Gradual delayed enhancement

✓ Typically without main pancreatic duct dilatation.

• **FDG PET/CT:**

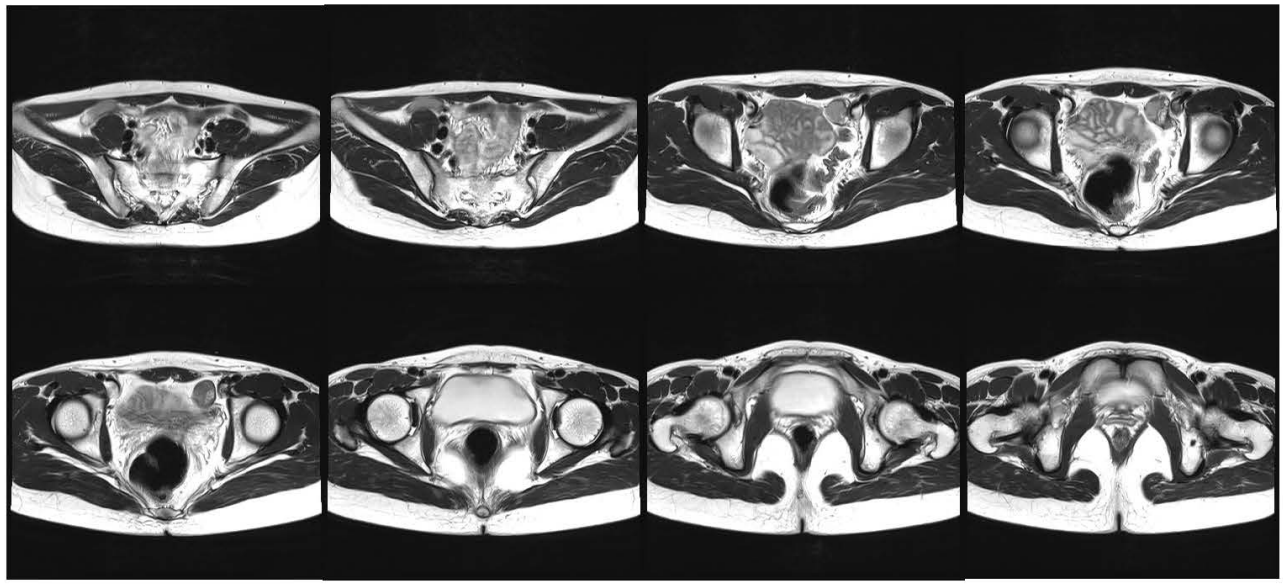
✓ Frequently exhibit increased FDG uptake on PET/CT, likely related to glucose transporter expression in schwannoma cells

Q6. What is the most likely diagnosis?

18/F

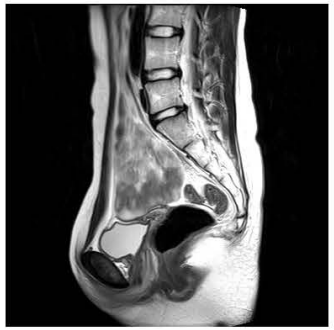
*** Chief complaint: Primary amenorrhea**

Axial



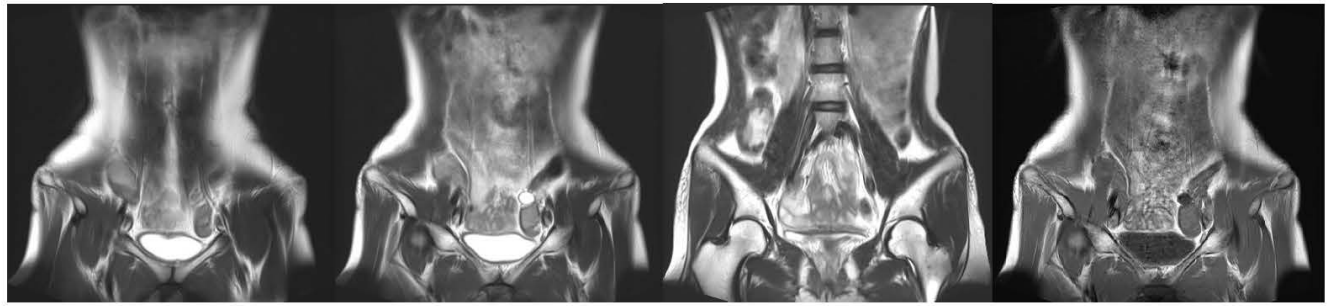
T2wi axial

Sagittal



T2wi sagittal

Coronal



T2wi coronal

T1wi coronal

Answer

- (Complete) Androgen insensitivity syndrome (CAIS)
- Androgen insensitivity syndrome – complete type
- Androgen insensitivity syndrome
- Testicular feminization syndrome

→ Explanation continues on the next page →

Q6. What is the most likely diagnosis?

Explanation

This patient presented with a female phenotype, but had no pubic or axillary hair, suggesting complete androgen resistance despite adequate or even elevated levels of circulating androgens. Chromosome analysis revealed a 46, XY karyotype, confirming a genotypic male with a disorder of sex development rather than primary ovarian failure.

A pelvic MRI showed an absent or rudimentary uterine development, with no normal ovaries identified, while bilateral round, homogeneous structures were seen in the pelvis consistent with undescended testis or ovo-testis; on the left side, a cord-like paratesticular structure was also noted. These findings reflect Müllerian duct regression under the influence of testicular anti-Müllerian hormone (AMH), combined with a failure of androgen action on the Wolffian duct and external genital structures due to androgen receptor dysfunction.

The patient subsequently underwent bilateral gonadectomy, and histopathology confirmed as unclassified testicular tissue, supporting the diagnosis of 46, XY complete androgen insensitivity syndrome (CAIS). In CAIS, gonadectomy is usually recommended after the completion of spontaneous puberty to reduce the long-term risk of gonadal malignancy, while preserving estrogen-mediated secondary sexual development. This case illustrates the classic presentation of CAIS: a 46,XY karyotype; a female external phenotype, sparse or absent body hair, an absent uterus and ovaries, intra-pelvic testis, and confirmatory histology after gonadectomy.

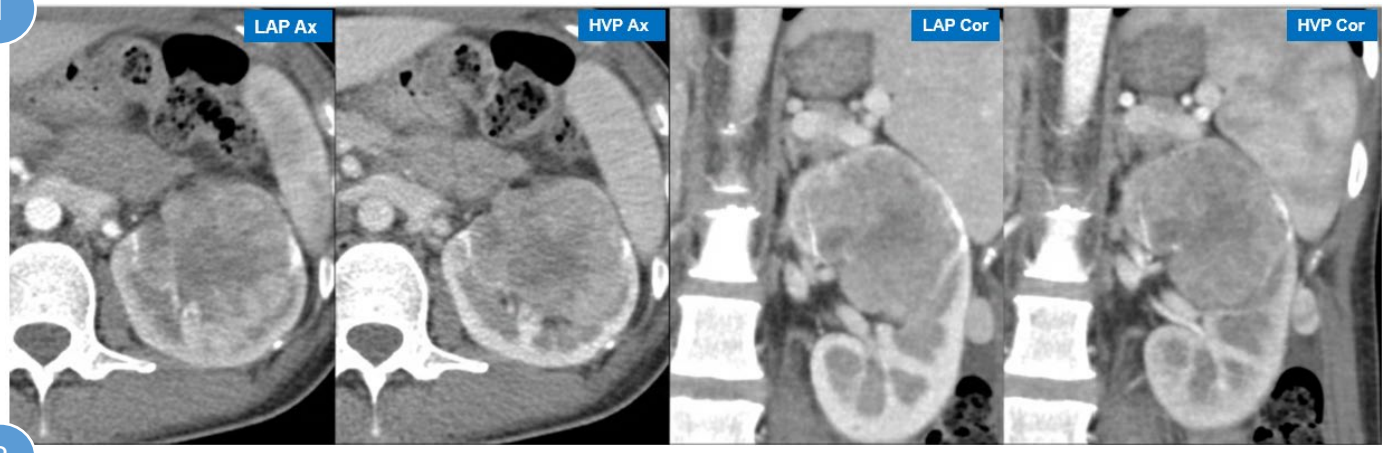
	Findings	Typical features of CAIS
Karyotype	46,XY	Genetically male (46,XY) despite female external phenotype.
Body hair	Absence of pubic and axillary hair	Markedly reduced or absent pubic/axillary hair due to androgen resistance.
Uterus and ovaries	MRI shows absence or rudimentary uterus; no normal ovaries identified	Regression of Müllerian structures and absence of ovaries.
Gonads	Bilateral round homogeneous pelvic structures suggesting testes or ovotestes; left-sided cord-like paratesticular structure	Undescended intra-abdominal or inguinal testes, sometimes mistaken for ovaries.
Histopathology	Bilateral gonadectomy; pathology confirmed unclassified testis	Testicular tissue with impaired spermatogenesis and variable stromal changes.
Clinical implication	46,XY phenotypic female with primary amenorrhea and intra-pelvic testes	Classic CAIS constellation; gonadectomy recommended after puberty because of tumor risk.

Q7. What is the most likely diagnosis of the renal mass in this patient?

31/F

C.C : Progressive weight loss (4kg/month)
Past medical history: NS
Physical examination: Multiple skin nodules.
Initial imaging : Abdominopelvic CT revealed a large left renal mass with aggressive imaging features.
Treatment and clinical course: Radical left nephrectomy was performed. The patient subsequently received targeted therapy (sorafenib and sunitinib) and radiotherapy for aggressive metastases.

1



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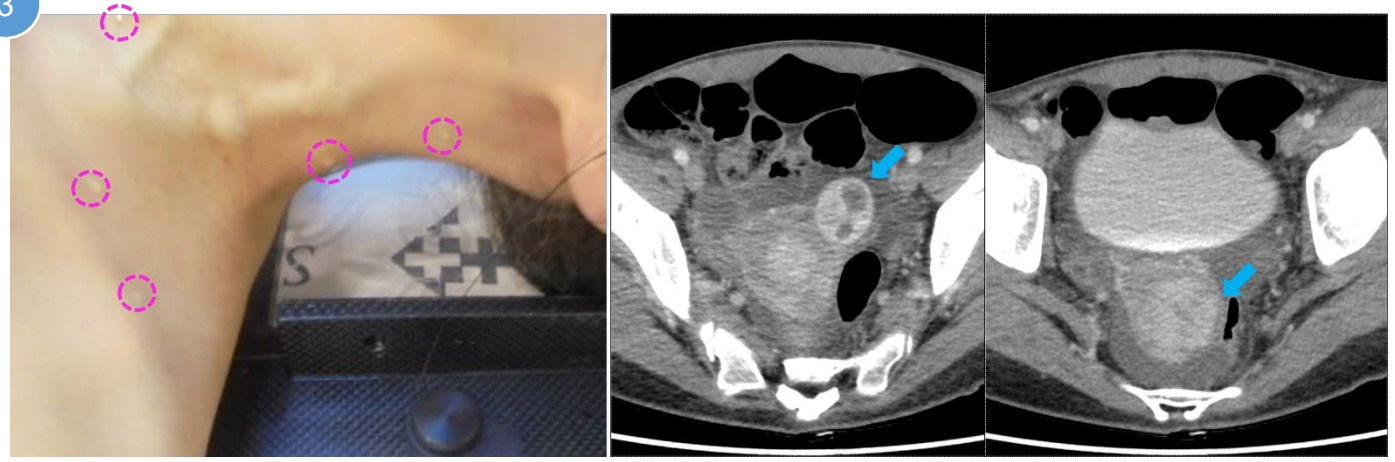

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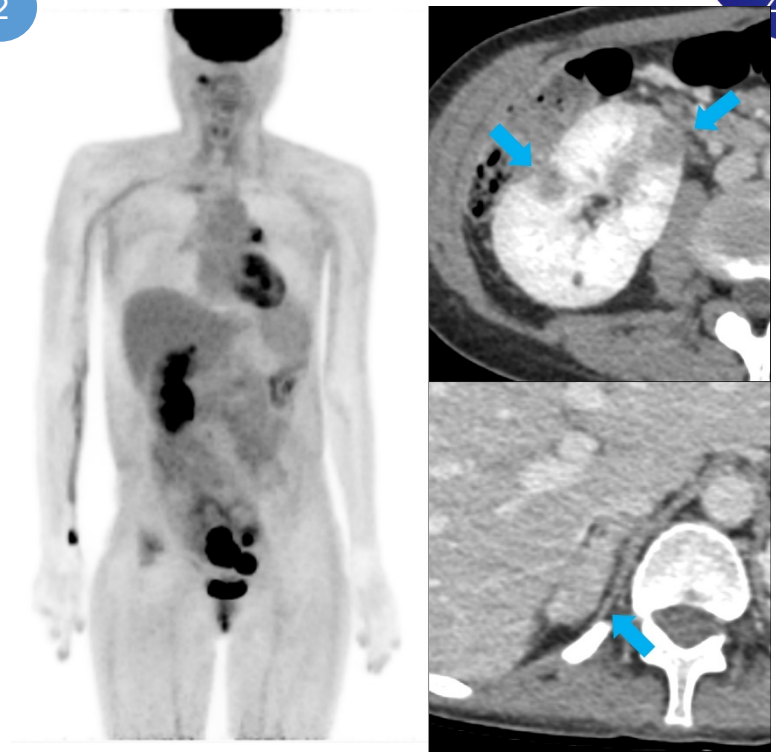

Fig1 : Abdominopelvic CT
(Lt renal mass with retroperitoneal LN metastasis)
Fig2 : Follow-up PET-CT and APCT images
Fig3: Uterine leiomyomas and cutaneous leiomyomatosis.

→ **Answer and Explanation continues on the next page** →

Q7. What is the most likely diagnosis of the renal mass in this patient?

Answer

- Fumarate hydratase–deficient renal cell carcinoma.
- FH-deficient renal cell carcinoma.
- Hereditary leiomyomatosis and renal cell carcinoma–associated renal cell carcinoma.
- HLRCC-associated renal cell carcinoma.

Explanation

This case is most consistent with fumarate hydratase–deficient renal cell carcinoma, a highly aggressive subtype of renal cell carcinoma associated with hereditary leiomyomatosis and renal cell carcinoma syndrome.

Several key features support this diagnosis.
First, the patient's young age at diagnosis strongly suggests a hereditary form of renal cell carcinoma.
Second, the renal mass demonstrates aggressive imaging characteristics, including large tumor size, renal sinus invasion, and early lymph node and lung metastases.
Third, the clinical course is marked by rapid disease progression and poor response to conventional targeted therapies.

Importantly, this patient shows characteristic extra-renal manifestations, including uterine leiomyoma and cutaneous leiomyomatosis. Cutaneous leiomyomas are a hallmark clinical feature of hereditary leiomyomatosis and renal cell carcinoma syndrome and are caused by germline mutations in the fumarate hydratase gene.
The coexistence of aggressive renal cell carcinoma with both uterine and cutaneous leiomyomas is a critical diagnostic clue for FH-deficient renal cell carcinoma.

In addition, FH-deficient RCC typically demonstrates very high glucose metabolism driven by metabolic reprogramming (Warburg effect). Therefore, FDG PET-CT often shows markedly intense FDG uptake in the primary renal tumor and metastatic lesions, which can be a useful supportive imaging clue in the appropriate clinical context.

FH-deficient renal cell carcinoma is now recognized as a distinct molecular entity. It often exhibits type 2 papillary or unclassified histology, loss of fumarate hydratase expression on immunohistochemistry, and aggressive clinical behavior with early metastasis and poor prognosis.
Early recognition of this entity is essential due to its implications for genetic counseling, family screening, and patient management.

Q8. What is your diagnosis?

14/M

*** Cosmetic problem of glans penis**

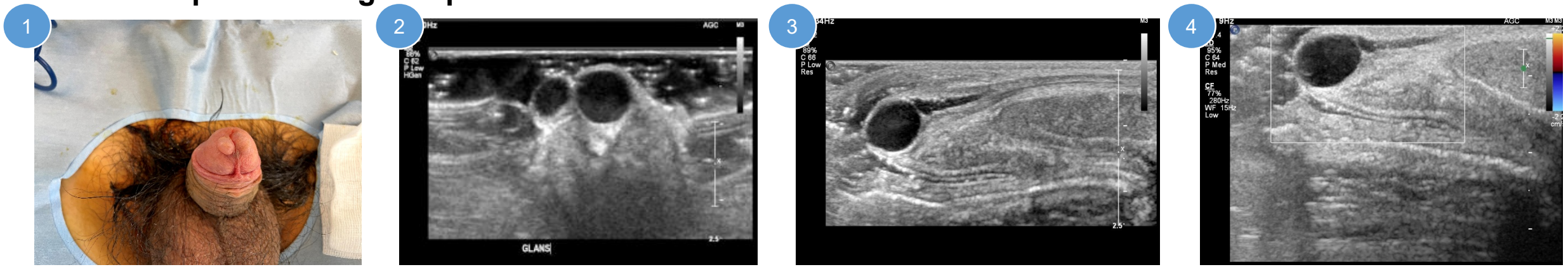

Figure 1: nodular protruding lesion at glans penis
Figure 2: Two lobulated cysts in glans penis arising from Rt lip of urethral meatus
Figure 3: Fine lobulated canaliform anechoic structure is abutting to the cyst
Figure 4: No internal flow on color doppler image

Answer

• Parameatal cyst
(or Medial raphe cyst, penile mucus cyst, hidrocystoma)

Explanation

Parameatal cysts are also referred to as parameatal glans cysts, penile mucus cysts, or hidrocystomas. They are known to be very rare and are predominantly benign.
While considered a congenital condition, they are not known to be associated with any specific comorbidities or syndromes.

Typically, these cysts are observed on the lateral or ventral aspect of the urethral meatus, measuring approximately 0.5–1 cm in size.
In most cases, the size remains constant as the patient grows.

Most patients are asymptomatic. The primary reasons for seeking medical consultation include cosmetic concerns and voiding dysfunction—such as urinary spraying (due to distortion of the meatus), a weak stream, dysuria, or urinary retention. When discovered during the neonatal period, clinical observation is recommended in anticipation of spontaneous resolution. If the cyst does not resolve naturally, it is surgically removed.
(Urology Digest 2(3):115-118 (2021))