FUMARATE HYDRATASE-DEFICIENT RENAL CELL CARCINOMA (FH-RCC)

A Case of an Infiltrative Left Renal Mass with Multiorgan Invasion & Distant Metastases

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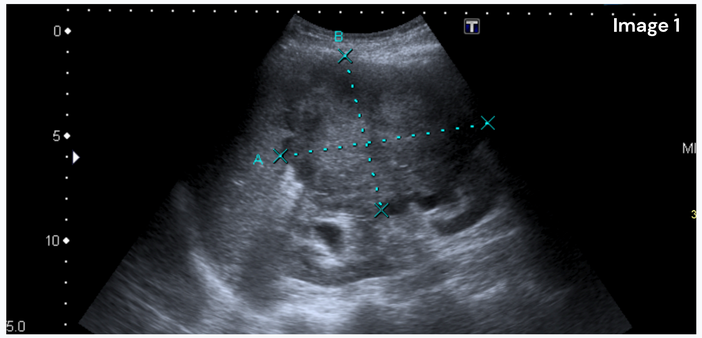
- **FUMARATE HYDRATASE DEFICIENT RENAL CELL CARCINOMA (FH-RCC)** is an **exceedingly rare, aggressive subtype of renal cell carcinoma** (RCC) characterized by the loss of mitochondrial enzyme fumarate hydratase activity.
- Strongly linked to autosomal-dominant hereditary leiomyomatosis (HLRCC) syndrome, but may occur sporadically.
- Seen in **younger patients** and shows more aggressive behaviour, with frequent **early metastatic spread** compared to other RCC subtypes.
- Classified in the 2025 EAU guidelines as a **distinct, syndrome-linked aggressive entity.**

CLINICAL CASE:

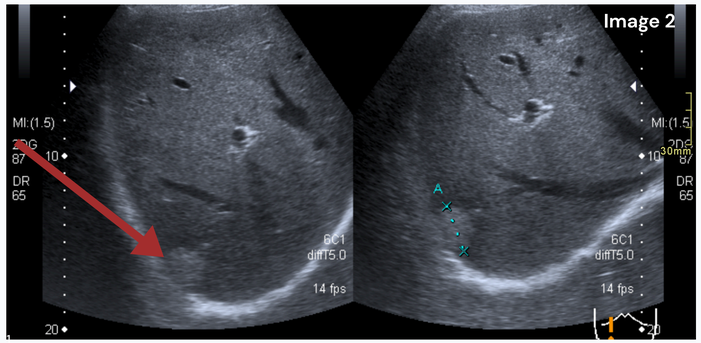
- The presented case illustrates the main imaging features of an advanced renal tumour in a 53-year-old patient.
- Ultrasound demonstrated a **large heterogeneous tumour formation of the left kidney** and liver lesions suspicious for **metastases**.
- Contrast-enhanced CT of the abdomen and chest confirmed an aggressive, **hypovascular left renal tumour invading** the spleen and colon, accompanied by pathological **lymph nodes** as well as **hepatic and pulmonary metastases**.
- The diagnosis was established histologically via **ultrasound-guided tumour biopsy**, invasion of the colon was confirmed by colonoscopy.

ABDOMINAL ULTRASOUND (US)

Left Kidney: US identified a massive heterogeneous solid renal tumour measuring up to 10 cm, **protruding into perirenal fat and infiltrating the caudal pole of the spleen.** Moderate hydronephrosis of the collecting system (Image 1).



Liver: Two distinct subcapsular hyperechoic lesions in segments 6 and 7 with discrete hypoechoic halos, highly suspicious for metastases (red arrow, Image 2).



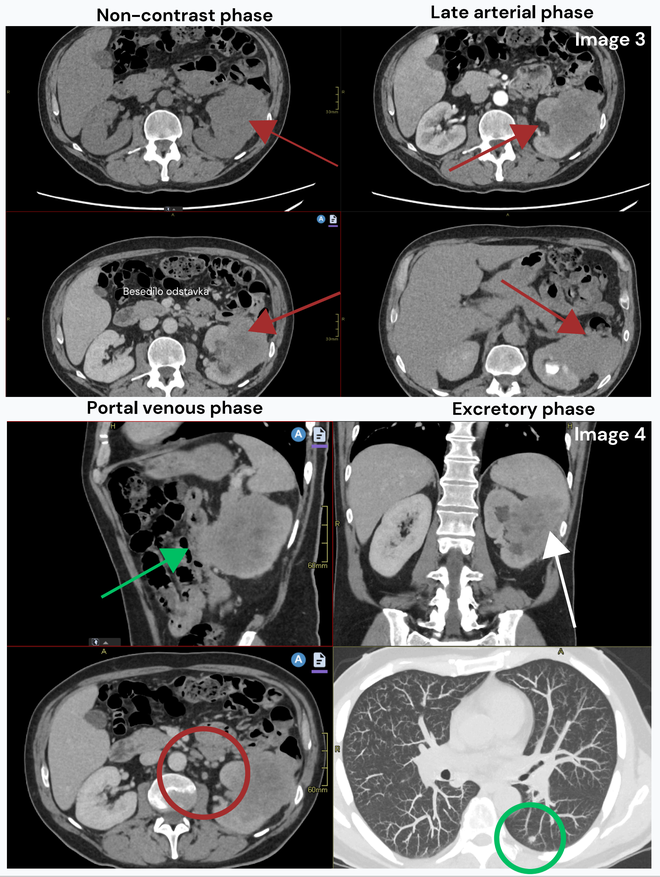
COMPUTED TOMOGRAPHY (CT)

Primary Mass: Infiltrative hypovascular left renal tumour involving the middle calyceal group and the renal pelvis, resulting in hydronephrosis (red arrows, Image 3).

Organ Invasion: Direct extension beyond Gerota's fascia into the pancreatic tail, jejunum, the descending colon (green arrow on sagittal reformat of portal venous phase, Image 4) and the spleen (white arrow on the coronal reformat, Image 4).

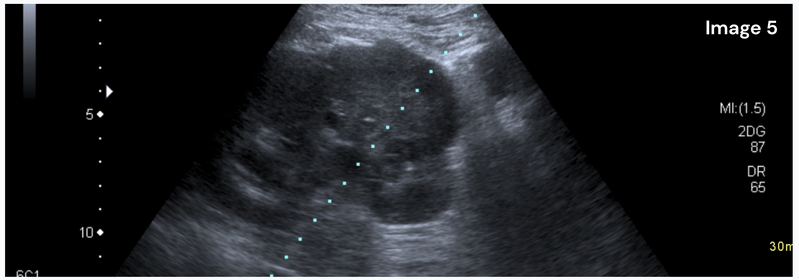
Nodal Status: Multiple pathologically enlarged lymph nodes in the left renal hilum (red circle, axial reformat of portal venous phase, Image 4).

Distant metastases:
Hepatic and pulmonary metastases were confirmed (pulmonary metastasis is indicated by the green circle, axial reformats with lung window, Image 4).



US-GUIDED CORE BIOPSY

Procedure: Targeted, real-time US-guided core needle biopsy using an 18 G biopsy needle. Three white, cylindrical tissue specimens were successfully obtained under local anesthesia.



CONCLUSION & KEY TAKEAWAYS

- FH-RCC is a **very rare** and **aggressive** subtype of RCC occurring in **younger** patients compared to other RCC subtypes.
- The **imaging findings** are **not specific**: FH-RCC most often presents as a **hypovascular, locally aggressive renal tumour** with **central cystic/necrotic components** and distant **metastases**.
- The **hypovascular nature** of the tumour **resembles papillary RCC**, whereas its **aggressive behavior** and **central necrosis** are more characteristic of **clear cell RCC.**
- CT and MRI are the primary imaging modalities, as in the workup of conventional RCC. Ultrasound has a role in initial detection and is the most commonly used modality in image-guided biopsy.
- Because of the **low specificity of imaging findings**, **tissue biopsy** and immunohistochemical confirmation of FH loss are essential for establishing the diagnosis.
- Given its hereditary basis, **genetic screening/testing of the family** should be considered when FH loss is confirmed.

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