Conversation: First Visit 22 September 2022

Doctor: Hello! What brings you here today?

Mother: This is my son, Muhammad Umer. He's 2.5 years old, weighs 12 kg, and is 87 cm tall. We came in for a follow-up on his kidney disease.

Doctor: Alright. Any significant family or birth history?

Mother: Yes, we are a consanguineous couple. Umer has two siblings who passed away from polycystic kidney disease. Two other siblings are healthy. He was born full-term and had no complications during birth.

Doctor: Got it. Has he been meeting his developmental milestones?

Mother: Yes, developmentally he's normal. His vaccinations are up to date as per schedule.

Doctor: When was he diagnosed with polycystic kidney disease?

Mother: He was fine until 6 months of age. We got some tests done during a minor illness and discovered he had PKD. His belly was also swollen around that time.

Doctor: Any history of GI bleeding?

Mother: Yes, he had around 45 episodes of hematemesis and melena. Hes undergone band ligation through endoscopy seven times. The last episode of GI bleeding was in March 2022, and the most recent endoscopy was on May 21, 2022.

Doctor: Let me check the records. The last EGD shows one column of Grade I varices and mild gastropathy. Lets also look at his labs.

Lab Results (20202022)

Doctor (reading labs):

Bilirubin: 0.4

ALT: 19

ALP: 70

INR: 1.07

HB (Mar 2022): 10.0

WBC: 9.4

Platelets: 151

Doctor: His labs seem stable for now. Imaging shows bilateral cystic kidneys with hepatic fibrosis, but no signs of progressive portal hypertension.

Plan (by Dr. Aneeqa Adnan)

Repeat CBC, LFTs, RFTs, S/E, INR

Blood group typing

Urine R/E and C/S

Ultrasound abdomen and pelvis

Start Tab Inderal 5mg TDS

Consider liver biopsy (as EGD is due)

Follow-up in one month with Pediatric Nephrology

Conversation: 22 May 2023 Paeds Gastro

Doctor: Hello again. How is Umer doing?

Mother: Hes been having abdominal pain for about 15 days. Hes still on Inderal and D-Max drops.

Doctor: Any new symptoms or GI bleeding?

Mother: No recent bleeding. The last one was in March 2022. He's had endoscopies regularly. The most recent was September 2022, showing Grade I varices.

Doctor: Let's examine him.

Examination

Weight/Height: 12 kg / 89 cm (Below 5th centile)

GIT: Soft, non-tender abdomen, liver 3 cm BCM, spleen 2 cm BCM

CVS, CNS, Respiratory: Normal

No jaundice, pallor, or edema

Doctor: Based on his history and imaging, this is consistent with Autosomal Recessive Polycystic Kidney Disease (ARPKD) with suspected Congenital Hepatic Fibrosis.

Plan

Labs: CBC, LFT, RFT, S/E, INR

Liver biopsy and EGD planned for tomorrow

Continue current medications

Salt-free diet advised

Anesthesia and OR booking completed

Conversation: Follow-up 17 July 2023

Doctor (Dr. Aneeqa Adnan and Dr. Iqra): Good to see you again. How is Umer now?

Mother: Hes stable and doesnt have any active complaints.

Doctor: Great. Let's go over his labs from May 23:

Lab Results 23 May 2023

ALT: 74

AST: 39

ALP: 138

Urea: 24

S/Cr: 0.7

Na+: 136

K+: 3.29

HB: 7.9

TLC: 4.3

Platelets: 180

Ca++: 9.36

Phosphate: 4.8

PTH: 107

Vitamin D: 27.24

Urine C/E: LE +++

Urine C/S: No growth

PT/APTT: Normal

Doctor: His hemoglobin is low. Urinalysis also shows leukocyte esterase. No infection grew on culture though.

Plan

Repeat labs: CBC, RFTs, LFTs, S/E, Ca++, PO4, HCO3, PT/APTT, INR

Urine R/E and C/S

Continue current medications

Follow-up after 1 month

Summary Final Diagnosis (as of 17 July 2023)

Diagnosis:

Autosomal Recessive Polycystic Kidney Disease (ARPKD) with Congenital Hepatic Fibrosis (confirmed by liver biopsy)

Current status: Not on dialysis

Liver biopsy (24 May 2023):

Ductal plate malformation

Fibrosis involving 6070% of biopsy volume

No cirrhosis

Endoscopy (24 May 2023): Grade II varices (12 o'clock position)