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

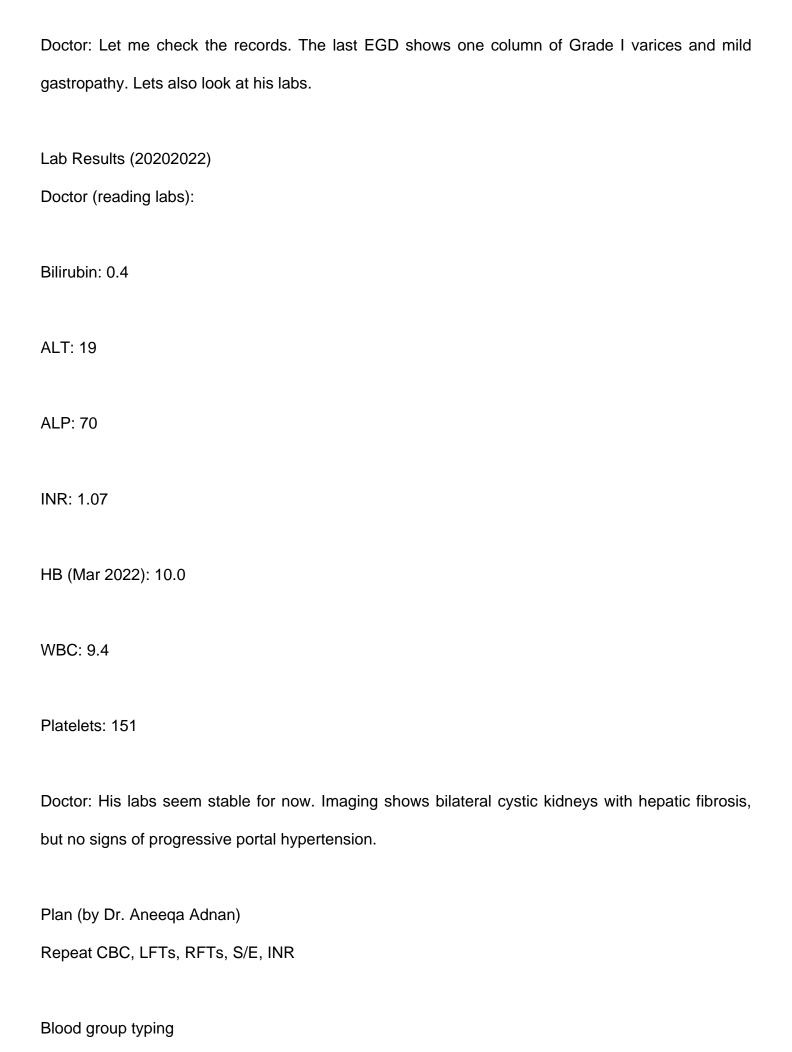
discovered he had PKD. His belly was also swollen around that time.

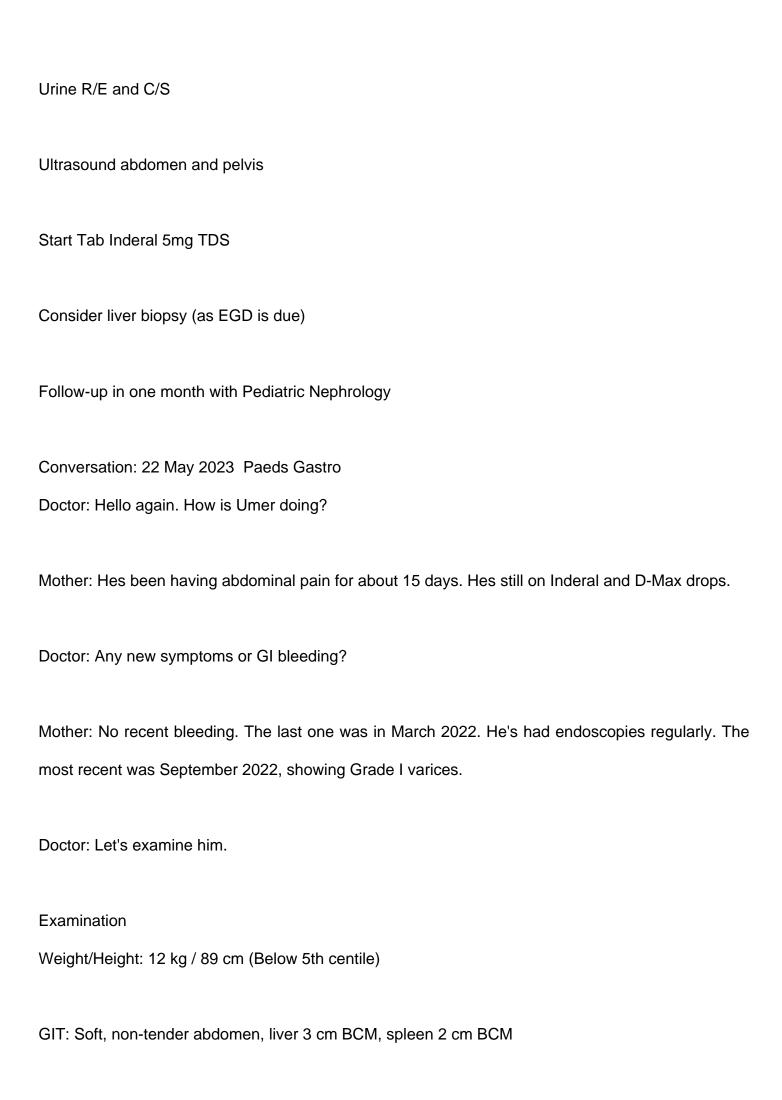
Doctor: Any history of GI bleeding?

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.





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.

Lab Results 23 May 2023

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

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)