Welcome
To
Central Clinicopathological Seminar

Department of Paediatrics
Dhaka Medical College & Hospital

A 7-year-old girl with fever & jaundice

Presenter

Dr. Ferdous-E-Zannat Natasha
FCPS Trainee
Department of Paediatrics, DMCH

Particulars of the patient

Name	:	Morium
Age	:	7 year
Gender	:	Female
Address	:	Kushtia
Date of admission	:	11/11/2024
Date of examination	:	11/11/2024
Informant	:	Mother

Presenting complaints

1. Fever for 12 days
2. Yellowish discoloration of skin and sclera for 7 days

History of present illness

According to the statement of the mother, her child was reasonably well 12 days back. Then she developed fever which was low grade, intermittent in nature, highest recorded temperature was 101°F. Fever was not associated with chills or rigor and subsided after taking anti-pyretic.

History of present illness cont.

She also developed yellowish discoloration of skin, sclera and dark urine for last 7 days. She complained of generalized weakness, anorexia and vomiting for the same duration. The vomiting was non-projectile, non-bilious, not blood mixed and occurred 1-2 times a day containing partially digested food particles.

History of present illness cont.

On query, mother gave history of consuming street food occasionally but no history of previous jaundice, abdominal pain, abnormal behavior, altered sleep pattern, convulsion, blood-mixed vomiting, black stool or any other bleeding manifestations. There were no history of itching, pale stool, skin rash, joint pain, taking any known hepatotoxic or any other significant drugs.

History of present illness cont.

Her bowel-bladder habits were normal. With these complaints 4 days prior to admission, she was admitted to a Govt. hospital where she received supportive care including 2 units of blood transfusion and albumin infusion with some injectable medications. As her condition did not improve she was referred to Dhaka Medical College Hospital (DMCH) for further evaluation and management.

History of past illness

No history of any significant past illness

Birth history

Antenatal	: Nothing significant
Natal	
Mode of delivery	: LUCS
Gestational age	: Term
Birth weight	: Average
Post natal	: Uneventful

Feeding history

She is on family diet

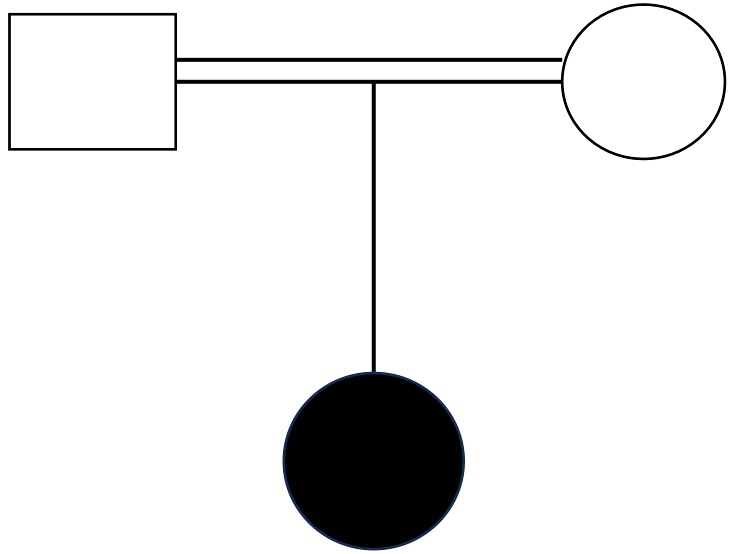
Immunization history

Completed as per EPI schedule

Developmental history

Age appropriate

Family history



Socio-economic history

Socio-economic status	:	Low socio-economic status
Father	:	Garments worker
Father’s income	:	25000 tk/month
Mother	:	Homemaker
House	:	Brick built house
Sanitation	:	Sanitary latrine
Water source	:	Tube well water

Treatment history

Initially treated by a local physician with some oral medications. Then she was admitted to Govt. hospital 4 days prior to admission in our hospital where she received 2 units of blood transfusion and albumin infusion with some injectable medications.

Physical examination

General examination

Appearance : Ill looking
Pallor : Mild
Jaundice : Present
Cyanosis :
Clubbing :
Koilonychia :
Leukonychia :

Absent

General examination cont.

Edema	:	Absent
Dehydration	:	Absent
Bony tenderness	:	Absent
Lymph node	:	Not palpable

General examination cont.

Skin survey:

BCG mark	: Present
Bleeding/ scratch mark	: Absent
Palmar erythema/ spider angioma	: Absent
Thenar or hypothenar wasting	: Absent

General examination cont.

Eye examination	: Normal
Examination of Ear, Nose and Throat	: Normal
Signs of meningeal irritation	: Absent
Back and spine	: Normal
Bed side urine albumin	: Nil

Vital parameters:

Temperature	: 100°F
Respiratory rate	: 24 breaths/min
Pulse	: 108 beats/min
Blood pressure	: 100/60 mm of Hg (both SBP & DBP lie in between 50th to 90th centile)

Anthropometry:

Weight : 21 kg
(lies on 50th centile)

Height :121 cm
(lies on 50th centile)

BMI :14.5 kg/m^2
(lies on 25th centile)

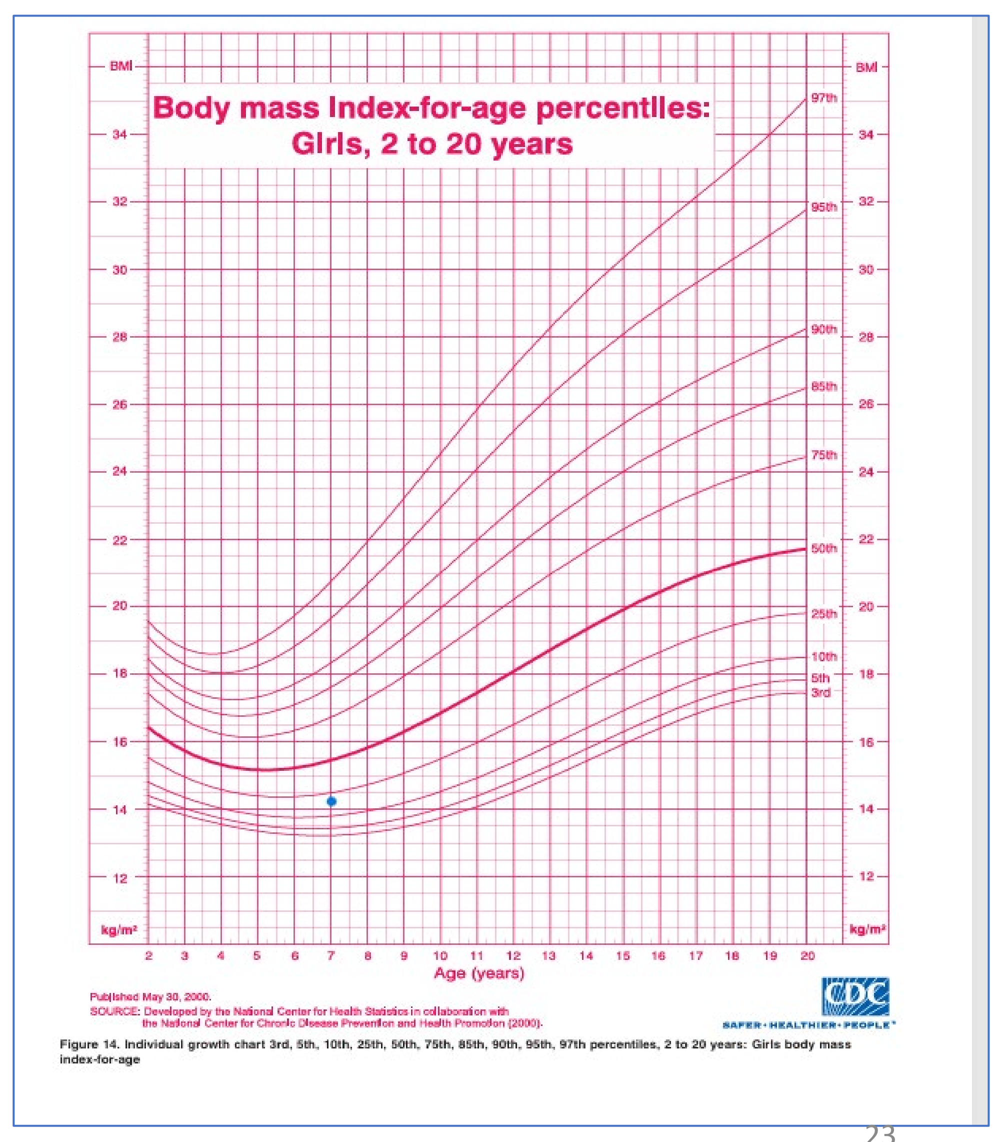


Figure 14. Individual growth chart 3rd, 5th, 10th, 25th, 50th, 75th, 85th, 90th, 95th, 97th percentiles, 2 to 20 years: Girls body mass index-for-age

Systemic Examination

Alimentary system

Mouth and oral cavity : Healthy

Abdomen proper

Inspection:

Abdomen	:	Distended, flanks were full
Umbilicus	:	Centrally placed, everted
Visible peristalsis	:	Absent
Engorged vein	:	Absent
Scar mark	:	Absent

Alimentary system cont.

Palpation

Local temperature	:	Normal
Tenderness	:	Absent
Palpable mass	:	Absent
Liver	:	Palpable, 4 cm from right costal margin in mid-clavicular line, firm, non tender, surface smooth. Upper border of liver dullness was on right 5th intercostal space. Liver span was 11cm

Alimentary system cont.

Spleen	: Not palpable
Kidneys	: Not ballotable
Bladder	: Not palpable
Renal angle tenderness	: Absent

Alimentary system cont.

Percussion:

Shifting dullness : Present

Fluid thrill : Absent

Auscultation:

Bowel sound : Normal

Nervous system

Higher psychic function:

Level of consciousness	:	Conscious
Orientation	:	Oriented to time, place, person
Memory	:	Intact
Intelligence	:	Normal
Behaviour and speech	:	Normal

Nervous system cont.

Cranial nerves : Intact

Motor functions :

Upper limb

Parameters	Right	Left
Muscle bulk	Normal	Normal
Tone	Normal	Normal
Muscle power	5/5	5/5
Reflexes	Normal	Normal
Co-ordination	Normal	Normal
Involuntary movement	Absent	Absent

Nervous system Cont.

Lower limb

Parameters	Right	Left
Muscle bulk	Normal	Normal
Tone	Normal	Normal
Muscle power	5/5	5/5
Reflexes	Normal	Normal
Plantar response	Flexor	Flexor
Clonus	Absent	Absent
Co-ordination	Normal	Normal
Involuntary movement	Absent	Absent
Gait	Normal	

Nervous system cont.

Sensory function	: Intact
Cerebellar function	: Intact

Respiratory system

Inspection:

Shape of the chest	: Normal
Respiratory rate	: 24 breaths/min
Scar mark	: Absent

No sign of respiratory distress

Respiratory system cont.

Palpation:

Position of the trachea	: Central
Apex beat	: At left 5th intercostal space medial to midclavicular line
Vocal fremitus	: Normal
Chest expansibility	: Bilaterally symmetrical

Respiratory system cont.

Percussion:

Resonant over both lung fields

Auscultation:

Breath sound : Vesicular

Vocal resonance : Normal

Added sound : Absent

Cardiovascular system

Inspection:

No visible apical impulse

Palpation:

Apex beat	: At left 5th Intercostal space medial to midclavicular line
Thrill	: Absent

Cardiovascular system cont.

Left parasternal heave : Absent

Palpable P2 : Not palpable

Auscultation:

S1 and S2 : Audible in all four cardiac areas

: No added sound

Locomotor system

Look : No joint swelling, redness, deformity or periarticular muscle wasting

Feel : No local rise of temperature, no tenderness

Move : No restriction of movement

Salient Feature

Morium, a 7-year-old girl, only issue of her consanguineous parents, developmentally age appropriate, immunized as per EPI schedule, hailing from Kushtia was admitted to DMCH with the complaints of low grade intermittent fever for 12 days and jaundice for 7 days. She also had history of generalized weakness, anorexia and vomiting.

Salient Feature cont.

She had no history of previous jaundice, abdominal pain, abnormal behavior, altered sleep pattern, convulsion, hematemesis, melena or any bleeding manifestations. There were no history of pruritus, pale stool, skin rash, joint pain, family history of similar illness or taking any offending drugs. Her bowel-bladder habits were normal.

Salient Feature cont.

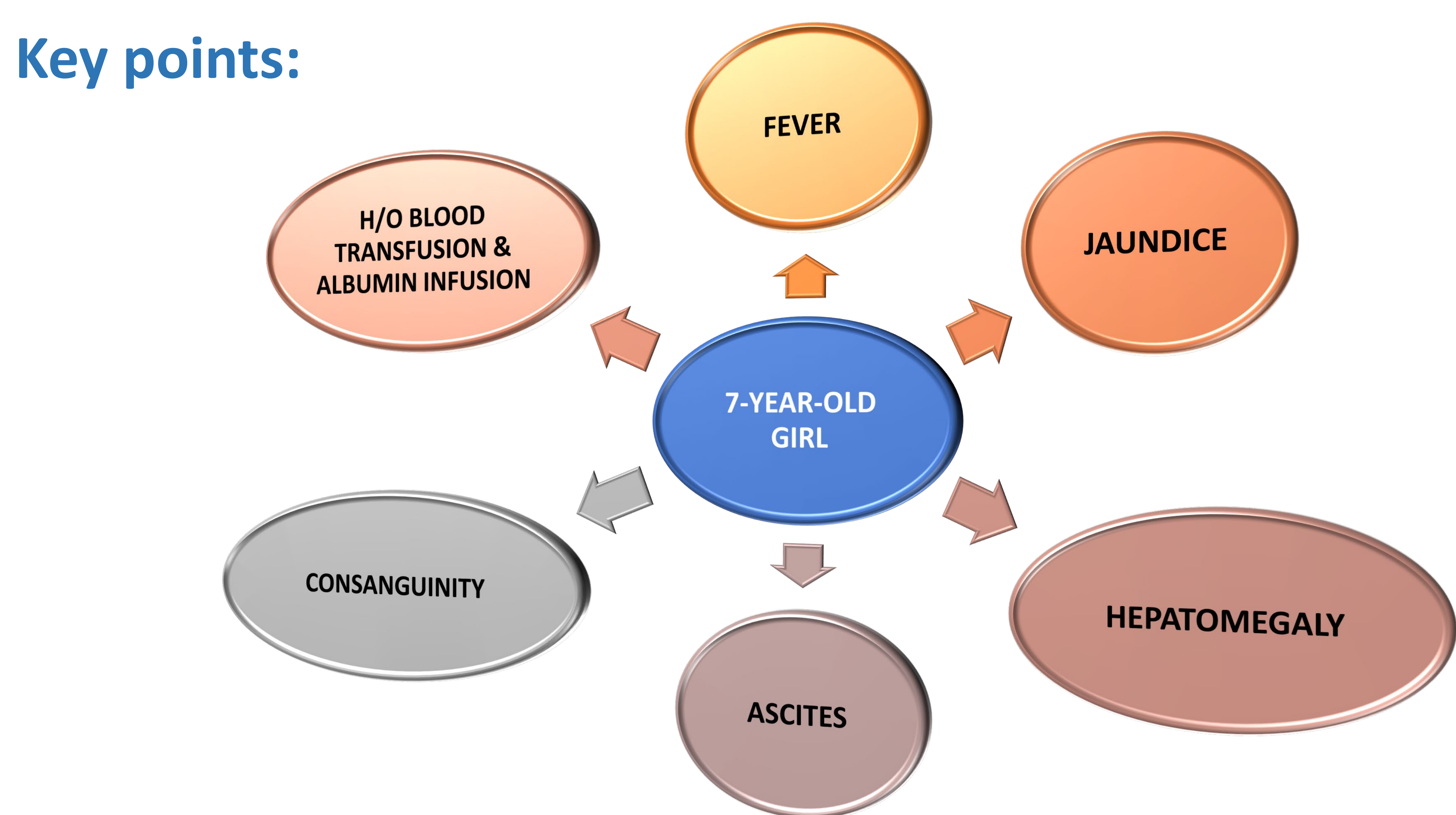
She received 2 units of blood transfusion and albumin infusion with some injectable medications in the past 4 days, prior to this admission. On examination, patient was ill looking, febrile, mildly pale, icteric, other vital parameters were within normal limit. She was anthropometrically well-thrived.

Salient Feature cont.

Non-tender hepatomegaly and ascites were found with no other features of chronic liver disease. No signs of altered consciousness, abnormal behavior or neurological deficits were present. Other systemic examinations revealed no abnormality.

Key points

Key points:



Provisional Diagnosis

Provisional Diagnosis

Acute hepatitis most probably due to Wilson disease

Differential Diagnosis

1. Acute viral hepatitis
2. Autoimmune hepatitis

Investigations

Investigations

Investigation	07/11/24 (Before admission)	12/11/24 (After blood transfusion)
CBC	Hb- 5.8 gm/dl (↓) TC- 6300/cumm Neutrophil- 38% Lymphocyte- 56% Platelet count-1,05,000/cumm (↓)	Hb-10.2 gm/dl TC-8370/cumm Neutrophil-63.4% Lymphocyte-30.2% Platelet count-1,50,000/cumm

Investigations cont.

Date	Investigation	Result	Reference range
07/11/24	PBF	RBC-predominance of spherocytes, polychromatic cells, few nucleated cells WBC-mature with above count and distribution Platelet- reduced in number Suggestive of hemolytic anemia with thrombocytopenia	
09/11/24	Reticulocyte count	16.4 %(↑)	0.5- 2.5%
12/11/24	Coomb's test	Negative	

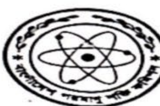
Investigations cont.

Investigation	O7/11/24 (Before admission)	14/11/24 (After admission)
S. Bilirubin	Total: 2.1 mg/dl (↑)	Total : 4.2mg/dl (↑) Indirect : 2.6mg/dl Direct : 1.6mg/dl
ALT	156 U/L (↑)	160 U/L (↑)
AST		193 U/L (↑)
S. Albumin	2.2 gm/dl (↓)	3 gm/dl (after albumin infusion)
Prothrombin time		25.9 sec (↑)
International Normalized Ratio		1.93 (↑)

Investigations cont.

Date	Invetigation	Result	Reference range
14/11/24	S. Ceruloplasmin	10 mg/dl(↓)	20-40 mg/dl
	Slit lamp examination	No KF ring or sunflower cataract	
20/11/24	24 hours urinary Cu	3528 microgram/day (↑)	<100mcg/day
14/11/24	Anti HAV IgM	Negative	
	Anti HEV IgM	Negative	
16/11/24	ANA	Negative	

24 hours urinary Cu report:



ANALYTICAL CHEMISTRY LABORATORY (ACL)
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Fax: 880-2-5861 79 46, Telephone: 880-2-223 36 38 37

Test Report

Batch No 24-093

Patient Details

Name of Patient : Morium
Gender : Female
Age : 7 y
Referred by : MO, Dhaka Medical College Hospital (DMCH)

Sample Details

Type of Sample : Morning Urine
Color of Sample : Straw
Received Date : 20-11-2024
Reporting Date : 20-11-2024

Test Result

Customer ID	Lab ID	Parameter(s) Tested	Technique Used	Test Method Used	Result	Unit
Morium	24112378	Copper (Cu)	FAAS	ACL-TMSOP-15	3528 ± 135	µg/L

Comments:
- The amount of copper per litre in the morning urine is equivalent to the 24 hour urinary copper (Ref. Biological Trace Element Research, 190 **(2019)** 283–288)
- Concentration above **100 µg/L (1.6 µmol/L)** confirms Wilson's Disease

Assuring our best service

Analyst:

Md. Azizul Maksud, SEO	
Md. Nur E Alam, SSO	
Md. Shahidur Rahman Khan, SEO	
Lutfun Naher Lutfa, EO	[signature]
Sarmin Sultana, EO	

An amount of Tk. 800.00 has been paid in cash to the Atomic Energy Centre Dhaka.

FAAS = Flame Atomic Absorption Spectrometry

N.B. This report is valid only for particular sample tested as received in the laboratory cannot be used for publicity.

Prepared by: *Dr. Tasrina Rabia Choudhury*	*Reviewed by:* *Md Azizul Maksud, DQM*	*Approved by:* *Dr. Tasrina Rabia Choudhury, QM*

ACL-QF-42 *Page 1 of 1* *Rev. Date: 07-08-2024* *Rev. 16*

Investigations cont.

Investigation	07/11/24 (Before admission)	24/11/24
Urine R/M/E	Protein: Nil RBC: Nil Pus cell: 1-3/HPF	Protein: Trace RBC: Nil Pus cell: 0-2/HPF
USG of whole abdomen	Mild hepato-splenomegaly, moderate ascites	Moderately coarse liver with mild hepato-splenomegaly

Investigations cont.

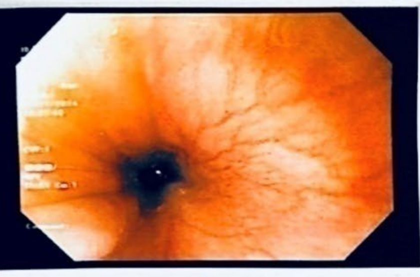
Investigation	Result	Reference range
S. electrolytes	Na^+ : 141 mmol/L K^+ : 3.5 mmol/L Cl^- : 109 mmol/L	135 - 145 mmol/L 3.5 - 5.5 mmol/L 95 - 105 mmol/L
S. Creatinine	0.31 mg/dl	0.5 - 1.1mg/dl
Blood culture	No growth	
Urine culture	No growth	

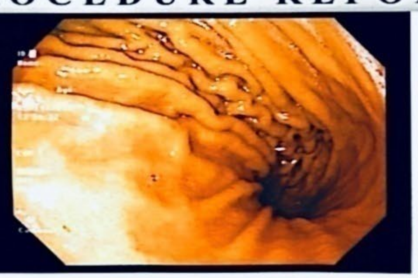
Upper GI endoscopy: (25/11/25)

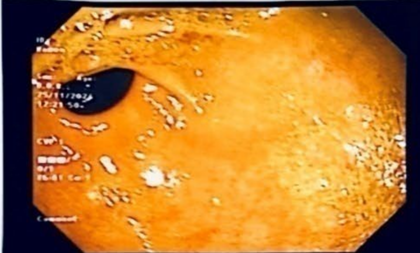
Department of Paediatric Gastroenterology & Nutrition
Dhaka Medical College Hospital

Identity No.	306	MRN	99651\241	Visit Date	25-11-2024
Patient Name	Moriom	Bed		Age/Sex	7yrs / Female
Referrer	207			Instrument	N/A

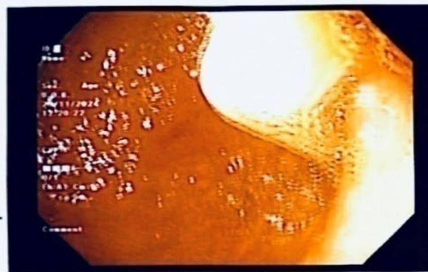
PROCEDURE REPORT







Procedure : UPPER G I ENDOSCOPY
Indication : CLD
Medication : Inj Midazolum



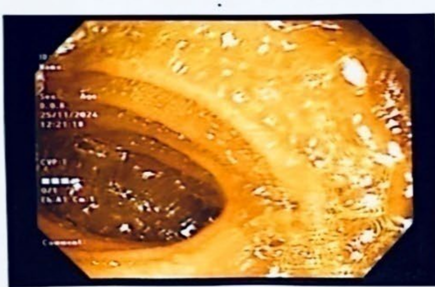
FINDINGS :

•***Oesophagus:-***
The mucosa, vascular pattern, lumen and peristalsis appear normal. Both upper and lower oesophageal sphincters are normal in position and function.

•***Stomach:-***
The mucosa of the cardia, fundus, body and antrum appear normal.

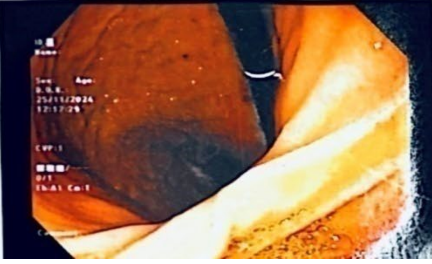
•***Duodenum:-***
The bulb, post-bulbar area and second part appear normal.

•***Biopsy:-*** Not taken.



COMMENTS :

Normal Upper G.I.T. at Endoscopy.



Final Diagnosis

Chronic Liver Disease (CLD) due to Wilson Disease

Management

Treatment

- Counselling
- Copper restricted diet
- Inj. Vitamin K1

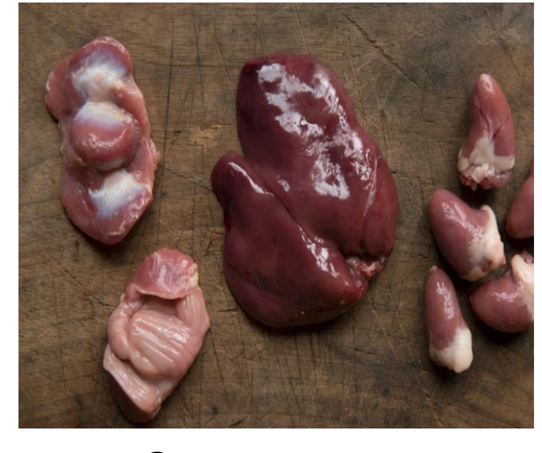
  0.2mg/kg/day OD for 3days
- Tab. Paracetamol

  10mg/kg/dose 3 times daily
- Syp. Lactulose

  3ml/kg/day in 2 divided doses

Copper rich foods that should be avoided

Dark chocolate


Organ meat

Shell fish

Mushroom

Nuts

Cola drinks

Treatment Cont.

- Cap. D- penicillamine (250mg)

  1+0+0, 2 hours before meal for 7 days (12mg/kg/day)

  After 7 days 20mg/kg/day

- Syp. Zinc

  $2^1/_2$ tsf 3 times daily, 2 hours after meal

- Tab. Pyridoxine (20mg)

  $1^1/_4$ +0+0

Follow up

Follow up

Clinical parameters	After 7 days of treatment	After 1 month of treatment
Pallor	Mild	Mild
Jaundice	Absent	Absent
Any bleeding manifestation	No	No
Liver	Size same as before	Size decreased
Spleen	Not palpable	Not palpable
Ascites	Absent	Absent
Neurological sign symptoms	Absent	Absent
Adverse drug reaction	None	None

Follow up cont.

Biochemical parameters	After 7 days of treatment	After 1 month of treatment
CBC	Hb- 8.8 gm/dl TC- 4110/cumm Platelet count-1,42,000/cumm	Hb- 9.2 gm/dl TC- 5300/cumm Platelet count-1,42,000/cumm
ALT	106 U/L	79 U/L
PT	16.7 sec	16.8 sec
INR	1.41	1.42
Urine R/M/E	Protein: Nil RBC: 0-1/HPF	Protein: Nil RBC:Nil

Further follow up plan

- Weekly for 1st month
- 1 to 3 monthly for the first year
- 6 to 12 monthly thereafter
- 24 hours urinary Cu yearly

Clinical improvement of the child after treatment

Wilson's Disease Presenting with Severe Hemolytic Anemia

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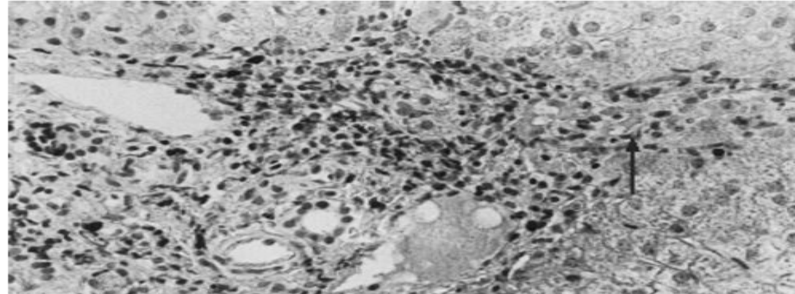
Key words: Hemolytic anemia, Wilson's disease.

Wilson' s disease or progressive hepatolenticular degeneration, originally described by Wilson in 1912, is a disorder of copper metabolism generally characterized by hepatic and / or neurological symptoms. Other findings include Kayser-Fleischer rings, hypoceruloplasminemia, hypocupremia, and hypercupriuria. Initial hematological manifestations are rare (1-3). We herein report a case of Wilson' s disease presented with severe hemolytic anemia as an uncommon initial manifestation.



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Case Report

Hemolytic anemia as first presentation of Wilson's disease with uncommon ATP7B mutation

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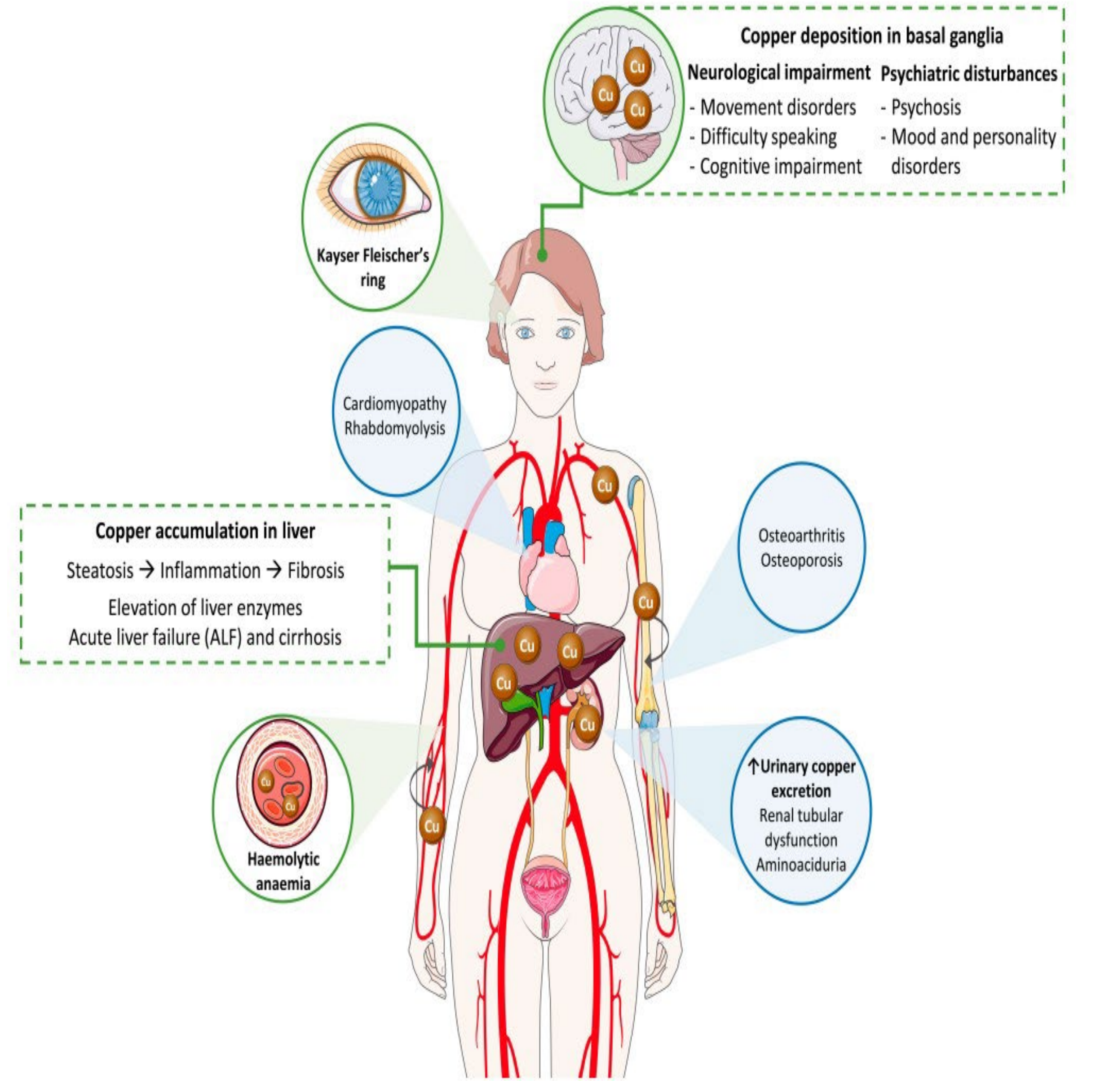
Abstract: Wilson's disease (WD) is a rare inherited disorder of copper metabolism and the main manifestations are liver and brain disorders. Hemolytic anemia is an unusual complication of WD. We describe a 15-year-old girl who developed hemolytic anemia as the first manifestation of Wilson's disease. An Arg952Lys mutation was found in exon 12 of the ATP7B gene, which is uncommon among Chinese Han individuals. From this case and reviews, we can achieve a better understanding of WD. Besides, we may conclude that the probable diagnosis of WD should be considered in young patients with unexplained hemolytic anemia, especially in patients with hepatic and/or neurologic disorder.

Keywords: Hemolytic anemia, Wilson's disease, ATP7B

Take home message

Besides CLD and neurological manifestation, Wilson disease can present with acute hepatitis, fatty liver, gall stone, isolated raised ALT or even haemolytic anaemia



DECEMBER 6TH
WILSON DISEASE
INTERNATIONAL
AWARENESS DAY
leave no one behind

Thank
you