CASE PRESENTATION
THALASSEMIA

DEMOGRAPHIC DETAILS

- Name-Ms. ABC
- Age-12 years (6th standard)
- Sex –female
- Address- Vidyanagar, Hubli
- Religion- Hindu
- Informant – Mrs, XYZ (information is reliant and consistent)
- Date of admission-19/02/2020
- Date of examination-21/02/2020

- Nutritional anaemia is not common under the age of 6 months in breastfed infants.
- Thalassaemia presents in late infancy (after 6 months) due to the presence of foetal haemoglobin (HbF).
- Fanconi's anaemia usually presents at the age of 6–8 years.
- Sickle-cell anaemia presents by 2–4 months of age.
    - Sickle-cell anaemia is common in hilly areas and tribal communities such as those in Chhatisgarh.
    - Thalassaemia is more common in Gujarat and Maharashtra.

X-linked disorders such as glucose-6-phosphate dehydrogenase (G6PD) deficiency and haemophilia are common in males.

CHIEF COMPLAINTS

- Easy fatigability since 7 days
- Increased Pallor since 5 days.

TABLE 13.1 Mechanism by Which Drugs Cause Anaemia

Mechanism	Drugs
Bone marrow depression	Chloramphenicol Phenylbutazone Cyclophosphamide 5-FU
Increased destruction of RBCs	Penicillin Cephalosporins
Folate antagonists	Phenytoin

HISTORY OF CHIEF COMPLAINTS

- Patient is a known case of haematological disorder came for regular transfusion with complaints of Easy fatigability on doing the daily activities which is relieved on taking rest. This is not associated With any other Complaints
- Patient's mother noticed pallor since 5 days which is insidious onset and progressive to present stage in Eyes and skin of patient
- No history of yellowish discoloration of skin ,yellow colored urine, Hematuria
- No history easy bruising, bleeding gums
- No history of pain in legs , abdominal pain, headache, convulsions

- No history of worms in stool
- No history of bone pain , weight loss
- No history of blood loss in stool, in vomitus
- No history of abdominal pain , passing worms in stools or vomitus
- No history pica
- No history of diarrhoea or constipation
- No history of fever with chills and rigors, dark colored urine
- No history of oliguria or anuria
- No history of dyspnoea, tachypnoea, palpitations.

PAST HISTORY

- Known case of haematological disorder diagnosed at the age of 6 months when child presented with fever ,loose stools, irritability and decreased frequency of Micturition for 4 days
- History of blood transfusion Started At 6th month once in 3 months for the first 2 years ,then it is given once a month, last blood transfusion was given 3 months back
- Total number of transfusion-109
- No history TB, chronic UTI ,Jaundice
- No history of previous surgery

TREATMENT HISTORY

- On tablet Defarasirox,started since 5 years of age,40mg/kg/day
- Not on any other drugs.

BIRTH HISTORY

Antenatal history

- Booked case uneventful
- Anomaly scan was done
- Iron folic acid tablet taken
- No history of fever with rashes
- No history of antepartum haemorrhage
- No exposure to radiation
- No history of multiple pregnancy.
- Inter pregnancy interval – 4 years

Natal history

- Cesarean section at KIMS At 10 months 16 days
- Birth weight-3.5kg Cried immediately after birth
- No history of admission to NICU
- No history from bleeding from cord
- Breast feeding started within an hour after birth

Postnatal history

- No history of neonatal jaundice
- Exclusively breastfed for 6months and continued for 1year
- Supplementary food was given after 6 months

Developmental history

- Developmental milestones achieved regularly
- Good scholastic performance

Immunization history

- Immunized upto 5 years of age [hepatitis B ,pentavalent were given]

NUTRITIONAL HISTORY

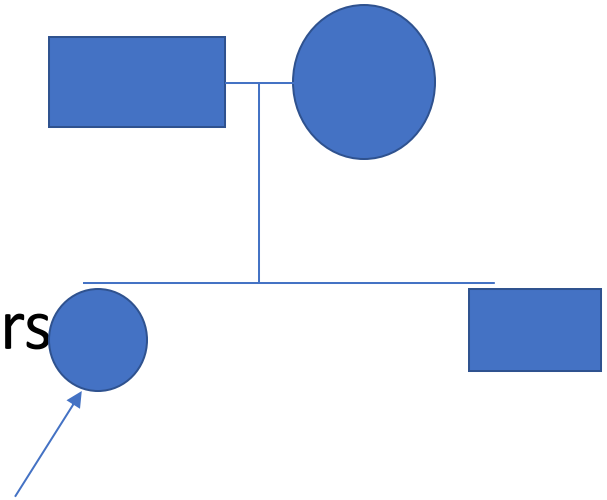
- Morning -2 idli + sambar – 300k cal ,5gm
- 1 glass of milk – 110 k cal , 3g
- After noon- 1 chapati +2 bowl rice+sambar-530kcal,10g
- Evening- 1 cup tea- 80k cal ,2g
- Dinner -1chapati+1bowl rice+sambar -360k cal, 6g Total- 1380k cal ,26g
- Required – 1900 k cal ,32 Deficient- 520 k cal ,6g Percentage – 27%in calories, 18% in protein
- Tea was given with meals

PERSONAL HISTORY

- Diet- mixed
- Appetite- normal
- Sleep- sound
- Bowel and bladder- normal and regular
- Patient denied any having habbits

FAMILY HISTORY

- 2nd degree consanguineous marriage
- Married life -15 years



- No history of similar illness in other family members
- No history of surgeries in family members[splenectomy,cholecystectomy]

TABLE 13.2 Mode of Inheritance of Blood Disorders

Inheritance	Conditions
Autosomal dominant	Hereditary spherocytosis
Autosomal recessive	Thalassaemia Sickle-cell anaemia
X-linked	G6PD deficiency

SOCIOECONOMIC STATUS

- Father farmer studied till SSLC
- Belongs to upper lower class according to kuppuswamy classification
.
- Lives in a Kucha house with overcrowding and with good ventilation.
- No open defication , drink filtered water.

SUMMARY

- Here is a 12 year old female child who is born to a 2nd degree consanguineous marriage belonging to upper lower socioeconomic status who consumes a diet of 27% deficient in calories and 18% deficient in protein who is immunized upto date with normal developmental milestones and with past history of Known case of haematological disorder came with complaints of Easy fatigability since 1 week and progressive pallor since 5 days.

GENERAL PHYSICAL EXAMINATION

- Here is a 12 years old female child conscious cooperative and well Oriented to time place and person
- Examined in supine position

- Pulse rate increased in severe anaemia, bounding pulses due to hyperdynamic circulation
- Fever may be present in leukaemia
- Respiratory rate—tachypnoea in severe anaemia, CCF

Vital signs

- Pulse -98beats per minute, in right radial artery, good volume ,non collapsing, no radioradial , no radio femoral delay
- Respiratory rate-22 breaths per minute
- Blood pressure-106/82mm Hg , measured in right brachial artery in sitting position
- Temperature- 98° F , measured in axilla

TABLE 13.4 Normal Range of Haemoglobin According to Age

Age Group	Normal Range (g/dL)
Cord blood	13.7–20.1
2 weeks	13.0–20.0
3 months	9.5–14.5
6 months–6 years	10.5–14.0
7–12 years	11.0–16.0
Adult female	12.0–16.0
Adult male	14.0–18.0

HEAD TO TOE EXAMINATION

- Hair –normal
- Frontal bossing- present
- Parietal bossing-present
- Malar prominence- present
- Eye- pallor is present
- Nose- flat nasal bridge
- Teeth –normal
- Oral cavity- Good Hygiene no features of anaemia
- Nails – normal

TABLE 13.3 Clinical Findings and Associated Blood Disorders

Clinical Finding		Associated Condition
Short stature		Fanconi anaemia
Head size	Microcephaly	Fanconi anaemia
	Macrocephaly	Chronic haemolytic anaemia
Facies	Frontal and parietal bossing with malar prominence	Haemolytic anaemia (thalassaemia)
	Box-like face, high arched palate, tripha-langeal thumb	Diamond Blackfan syndrome
Skin	Hyperpigmentation	Megaloblastic anaemia, Fanconi anaemia, congenital dyserythro-poietic anaemia
	Petechiae	Aplastic anaemia
Upper limbs	Dactylitis	Sickle-cell anaemia
	absent radius, absent or bifid humerus, absent thumb	Fanconi anaemia
	Triphalangeal thumb	Pure red cell aplasia
Eyes	Blue sclera	IDA—severe
	Microcornea	Fanconi anaemia
Tongue	Smooth, red, painful tongue	Pernicious anaemia
Nails	Koilonychia	Iron deficiency
Leg ulcers		Sickle-cell anaemia
Hepatosplenomegaly		Chronic haemolytic anaemia

- Skin – pallor, no hyper pigmentation or petechiae
- Upper Limb – Normal
- Eye – Normal, no microcornea
- Tongue - Normal
- Lower Limb – Normal, no leg ulcer
- Chest ,Spine and back normal
- No icterus, cyanosis, clubbing, lymphadenopathy
- External genitals normal
- Tanner grade-stage 2

TABLE 13.5 Morphological Classification of Anaemia

Type	Investigation	Conditions
Microcytic	Ferritin decreased	IDA
	Ferritin normal/ increased	Thalassaemia Haemoglobinopathies Sideroblastic anaemia
Normocytic	Reticulocyte count decreased	Bone marrow hypoplasia Red cell dysplasia
	Reticulocyte count normal/ increased	Haemorrhage/acute blood loss Haemolytic anaemias—congenital or acquired Secondary anaemia (anaemia due to acute infections such as malaria)
Macrocytic	Megaloblastic	Vitamin B_{12} deficiency Folate deficiency
	Non-megaloblastic	Liver disorders Thyroid disorders Congenital dyserythropoietic anaemia

ANTHROPEMETRY

		CENTILES
HEIGHT	142 CMS	Within 90th centile
WEIGHT	40KGs	Within 50th Centile

SYSTEMIC EXAMINATION

Per Abdomen examination

INSPECTION

- Abdomen is distended on left hypochondrium
- Umbilicus central and everted
- Corresponding quadrants move equally with respiration
- Dilated veins Present ,no scars
- Hernial orifices normal.

PALPATION

- No local rise of temperate,no tenderness
- Deep palpation Mass in right hypochondrim moves with respiration ,firm in consistency smooth surface, sharp boarder, 3cm below right costal margins suggestive of liver
- A mass in left hypochondrim moves with respiration,non tender ,firm in consistency,smooth surface sharp boarder of 8cm below costal margin suggestive of spleen splenic notch appreciated

PERCUSSION

- Liver span – 14 cm
- No free fluids

AUSCULTATION

- Bowel sounds heard

OTHER SYSTEMS

- CVS- No raised JVP, S1 S2 heard no murmur
- RS – Normal vesicular breath sounds heard Bilateral equal air entry no added sounds
- CNS - Higher mental function normal no sensory or motor deficit

DIAGNOSIS

- Case of haemolytic anaemia probably Thalassemia major undertransfused well chelated, not in heart failure.

THALASSAEMIA

It is an autosomal recessive haemolytic anaemia that results from defective production of Hb due to mutation in the genes which direct the production of Hb (a protein that carries oxygen to the cells and tissues of the body).

Beta-thalassaemia is the most common single-gene disorder. Patients with thalassaemia produce abnormal Hb that cannot transport enough oxygen to tissues. This produces symptoms ranging from fatigue to organ damage.

TABLE 13.10 Clinical Variants of Thalassaemia

Features	Thalassaemia Major (β^0/β^0)	Thalassaemia Intermedia (β^+/β^+ or β^0/β^+)	Thalassaemia Minor (β^0 or β^+)
Genetics	Homozygous form—two mutated genes	Homozygous or heterozygous form; two mutated genes	Heterozygous form—one normal and one mutated gene
Age of presentation	Early infancy (6–18 months)	Presents by second year of life	Adolescence
Clinical features	Progressive pallor Hepatosplenomegaly Bony changes	Anaemia Hepatosplenomegaly Bony changes	Mild persistent anaemia
Requirement of blood transfusion	By the age of 6 months	Rarely needed. By 2 years Hb level may fall to about 7 g/dL	By adolescence
Complications	Hypersplenism Iron overload	Iron overload Cholelithiasis	Iron overload
Prognosis	Fatal in first few years of life	Normal lifespan	Normal lifespan