



CASE PRESENTATION

DR. Muhammad Waqqas Akram

PGT-I

DEPTT OF PAEDIATRICS


BBH Rawalpindi

PROFILE

- Name Ali Haider
- Age 3¹/₂ Months
- Sex Male
- Address Fateh Jang
- DOA 15-04-2009

HISTORY OF PRESENT ILLNESS

- SVD, full term, at home
- Birth weight was 2.5kg
- Poor feeding
- Poor weight gain
- present weight 3.4 kg

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- Progressive pallor 01 month
 - Abdominal Distention
 - High grade intermittent fever 1 week
 - Breathing difficulty

Systemic Review

No H/O

Cyanosis , Jaundice, Convulsions, Stridor,
Vomiting, Diarrhea, Polyurea, Oligurea,
Petechiae & Bruises, Bleeding from any site

Treatment History:

- Treatment from local doctors
- Received multiple oral antibiotics, antipyretics, bronchodilators and nasal drops
- No favourable response



Birth History

Antenatal History

Perinatal History

Postnatal History

Family history

- First issue of a consanguineous marriage
- No history of Abortions
- Thalassemia
- Contact with TB

FEEDING HISTORY

- Exclusively breast fed

IMMUNIZATION

- Vaccination up to date
- BCG scar +ve

DEVELOPMENTAL HISTORY

- NORMAL DEVELOPMENTAL MILESTONE
 - GROSS MOTOR
 - No head lag
 - Head raised in prone position
 - FINE MOTOR AND VISION
 - Follows light through arc of 180
 - Defensive blink present
 - Grasps the cube

DEVELOPMENTAL HISTORY

- HEARING AND SPEECH
 - Turns to nearby voice
 - Vocalizes
- SOCIAL BEHAVIOUR
 - Happy response to mother's face
 - Social smile present

SOCIOECONOMIC HISTORY

- Lower socioeconomic class

EXAMINATION

- **General Physical Examination**
 - Sick-looking markedly pale young infant having severe respiratory distress with protuberant abdomen.
 - No cyanosis, jaundice, lymphadenopathy, edema, petechiae & bruises



- VITALS

- RR 65/MIN
- Pulse rate 130/min
- TEMP 101 F
- B.P 60/40 mmHg

- ANTHROPOMETRY

- LENGTH 60 cm (50TH PERCENTILE)
- WEIGHT 3.4 Kg
(BELOW 5TH PERCENTILE)
- OFC 42 cm (90TH PERCENTILE)

SYSTEMIC EXAMINATION

● RESPIRATORY SYSTEM

○ INSPECTION

- Normal shaped chest
- Respiratory distress with sub costal recessions

○ PALPATION

- Central trachea, Apex beat in 5th ICS in midclavicular line
- B/L Normal chest expansion

○ PERCUSSION

- Resonant & equal on both sides

○ AUSCULTATION

- Harsh vesicular breathing with B/L crepts and ronchi

ABDOMINAL EXAMINATION

- INSPECTION
 - Protuberant, Central umbilicus of normal shape
- PALPATION
 - **HEPATOMEGALY-** Palpable 5 cm BRCM
Total span 13 cm
Firm, smooth surface, diffuse margins
 - **SPLENOMEGALY-** Palpable 6cm BLCM, Firm, non-tender
- PERCUSSION
 - No evidence of free fluid
- AUSCULTATION
 - Normal bowel sounds

CNS EXAMINATION

- Higher mental functions
 - Fully conscious
 - Normal behaviour
- Cranial nerves
 - Intact
- Motor system
 - Reduced bulk and nutrition of muscles
 - Normal tone and power
 - Intact deep and superficial reflexes
- Sensor system
 - normal

SUMMARY

- Haider ali, 3 1/2 months, MC product of consanguineous marriage, admitted with failure to thrive, progressive pallor & high grade intermittent fever. On admission he was markedly pale, febrile, in severe respiratory distress with gross hepatosplenomegaly.

DIFFERENTIALDIAGNOSES

- Chronic hemolytic Anaemias
 - Hereditary spherocytosis
 - Thalassemia
- Osteopetrosis
- Congenital leukemia

INVESTIGATIONS.... BLOOD CP

- Hb 6.1 g/dl
- PLATELETS 87000 /ul
- TLC 36020 /ul
 - POLMORHS 36%
 - LYMPHOCYTES 40%
 - MONOCYTES 4%
 - EOSINOPHILS 2%
 - MYELOCYTES 8%
 - METAMYELOCYTES 9%
 - BAND FORM 1%

INVESTIGATIONS.... PERIPHERAL FILM

- ANISOCYTOSIS ++
- POIKILOCYTOSIS ++
- MICROCYTOSIS ++
- MACROCYTOSIS ++
- HYPOCHROMIA ++
- NUCLEATED RBC 97/100 WBC
- RETIC COUNT 10%

INVESTIGATIONS.... CHEMISTRY

INVESTIGATION	RESULT
ALT	21 units/Dl
BILIRUBIN	1.3 mg/dL
ALK. PHOSPHATASE	602 units/dL
UREA	32 mg/dL
CREATININE	0.6 mg/dL
BSR	133 mg/dL
CALCIUM	7.3 mg/dL

INVESTIGATIONS-

- Mantoux test - negative
- ICT- MP - negative
- Serum immunoglobulins - normal

INVESTIGATIONS....RADIOLOGY

- CXR
 - Clear lung fields
 - Normal cardiac shadow
 - Rugger-jersey Bones
 - Splayed osteochondral junctions

INVESTIGATIONS....RADIOLOGY

- X-RAY SKULL
 - Sclerosis of the base of skull sparing the culvaria

INVESTIGATIONS....RADIOLOGY

- **LONG BONES**
 - Splayed metaphysis
 - Bone within bone appearance
 - Longitudinal metaphysial striations

- **PHALANGES**
 - Alternating dense and light bands

INVESTIGATIONS....

BONE MARROW ASPIRATION

- Hyperactive bone marrow with normal cell lineages
- Giant osteoclasts seen
- No LD bodies seen
- No malarial parasite seen



FINAL DIAGNOSIS

OSTEOPETROSIS

(MALIGNANT INFANTILE OSTEOPETROSIS)

