CASE PRESENTATION

DR. Muhammad Waqqas Akram

PGT-I
DEPTT OF PAEDIATRICS
BBH Rawalpindi
PROFILE

- Name: Ali Haider
- Age: 3 1/2 Months
- Sex: Male
- Address: Fateh Jang
- DOA: 15-04-2009
PRESENTING COMPLAINTS

- Failure to thrive since birth
- Progressive pallor 01 month
- Fever 01 week
HISTORY OF PRESENT ILLNESS

- SVD at full term, home delivery
- Birth weight was 2.5kg
- Poor feeding
- Poor weight gain
- Present weight 3.4 kg
- Progressive pallor 1 month
- Abdominal Distention
- Fever 1 week
- Breathing difficulty
Systemic Review

No H/O

Cyanosis, wheeze, Stridor, Convulsions, jaundice, 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
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
• 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
– TEMP 101 F

Pulse rate 130/min
B.P 60/40 mmHg
• 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
    • Respiritory distress with sub costal recessions
  – PALPATION
    • Central trachea, Apex beat in 5\textsuperscript{th} ICS in \textit{(midclavicular line)}
    • B/L Normal chest expansion
  – PERCUSSION
    • Resonant & equal on both sides
  – AUSCULTATION
    • B/L crepts and ronchi.
ABDOMINAL EXAMINATION

- **INSPECTION** - Protuberant, Central umbilicus

- **PALPATION** - soft and non tender
  - **HEPATOMEGALY** - 5 cm BRCM, Total span 13 cm
    - Firm, smooth surface, diffuse margins
  - **Splenomegaly** - Palpable 4cm BLCM, Firm

- **PERCUSSION** - No evidence of free fluid

- **AUSCULTATION** - Normal bowel sounds
CNS EXAMINATION

• Higher mental functions
  Conscious  Normal behavior

• Cranial nerves – Intact

• Motor system - Reduced muscle bulk
  Normal tone and power
  Intact deep and superficial reflexes

• Sensor system - normal
SUMMARY

• Ali Haider, 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.
DIFFERENTIAL DIAGNOSES

- Chronic Hemolytic Anemia
  - 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%
Liver Function Tests

- ALT: 21 u/dl
- Bilirubin: 1.3 gm/dl
- ALK. Phophatase: 602 u/dl
Renal Function Tests

- Serum Urea: 32mg/dl
- Creatinine: 0.6mg/dl
• **BSR** – 133mg/dl

• **Serum Calcium** – 7.6mg/dl
- Mantoux test - Negative
- ICT- MP - Negative
- Serum immunoglobulins - Normal
- Urine -
BONE MARROW ASPIRATION

• Hyperactive bone marrow with normal cell lineages

• Giant osteoclasts seen

• No LD bodies seen

• No malarial parasite seen
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INVESTIGATIONS....RADIOLOGY

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

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

• LONG BONES
  – Splayed metaphysis
  – Bone within bone appearance
  – Longitudinal metaphysial striations

• PHALANGES
  – Alternating dense and light bands
INVESTIGATIONS....RADIOLOGY

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FINAL DIAGNOSIS

OSTEOPETROSIS
(MALIGNANT-INFANTILE OSTEOPETROSIS)