Case Report

Metaphyseal Dysplasia - A Case Report

Md. Shakibur Rahman[1], Md. Sultan Uddin[2]

[1]Medical Officer, Department of Paediatric, Dhaka National Medical Institute Hospital, [2]Professor, Department of Paediatric, Dhaka National Medical College

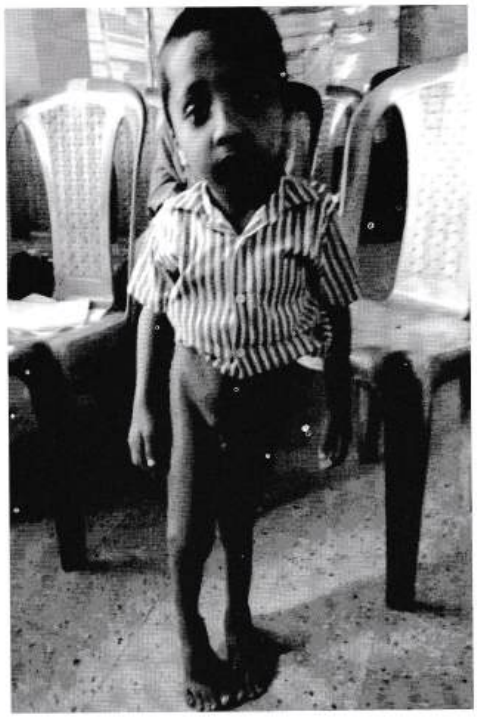
Introduction :

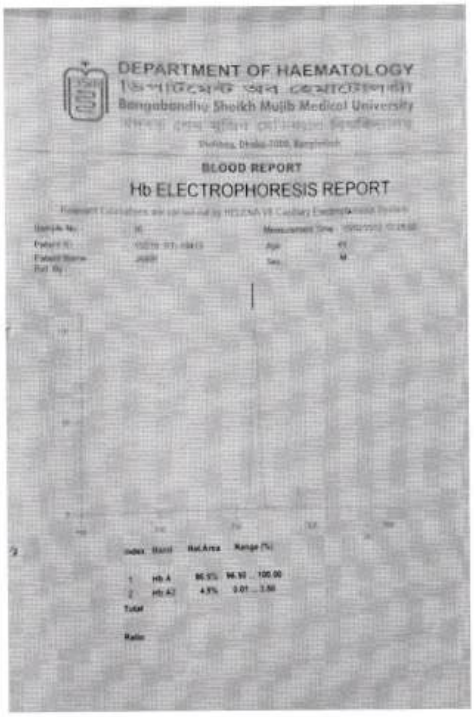
Skeletal dysplasias are congenital and hereditary conditions that infers generalized involvement of epiphyses, metaphyses, or diaphyses and is associated with disproportionate short stature. Metaphyseal dysplasia is also knodwn as Pyle's disease,[1] Pyle's syndrome, Pyle-Cohn syndrome, and Bakwin-Krida syndrome[2].Metaphysial dysplasia is a heterogenous group of disorders characterized by abnormalites of the size and shape of the bones due to defective metaphyses. Prevalence is estimated to be 1 in 4000 births. It is important to understand metaphyseal dysplasia to differentiate it from rickets which is more common and treatable.

Case report:

Jabir, a 7 years 2 month old boy, weighing only 12 kg, immunized as per EPI schedule, 2nd issue of consanguineous parents (1st degree), came from middle class socio-economic family, hailing from Rayshaheb Bazar , Dhaka presented with not growing well in height and weight since birth, widening of both ankle, wrist joints, widening and bending of both elbow joints.

He had no history of joint pain, bowing of legs, headache, vomiting, convulsion, visual disturbance, ear problem recurrent cough, respiratory distress, jaundice, recurrent diarrhea, constipation or any urinary problem. His bowel and bladder habit is normal. He used to live in area where there was adequate sunshine. He was delivered at term at home per vaginally with average birth weight and bilateral clubfoot. Meconeum passed within 24 hrs after birth. The child was treated by Orthopedic surgeon and advised to gentle manipulation of the affected feet then serial plastering and finally tenotomy was done in CRP centre, Savar. He visited endocrinologist due to not gaining height and did some investigations and then referred to a hemato-oncologist and diagnosed as a case of Thalassaemia trait and treated with folic acid. He was on exclusive breast feeding for 5 months. Then started mixed feeding from 6 month of age. His milestones of development were age appropriate. His father is a businessman and mother house wife. His elder sister and other family members are healthy. He is a student of KG-1 with good academic performance. On examination, Jabir looked healthy, cooperative, no facial dysmorphism, mildly pale, not icteric, not cyanosed; edema, dehydration, clubbing koilonychia, leuconychia were absent. Vitals were within normal limit. Weight: 12 kg, (corresponding to about 2 ½ years of age), height: 89 cm, (corresponding around 2 years of age) OFC: 51 cm, WAZ: -4.4 HAZ: -6.6, BMI: 14.8 (between 5th -10th centile), US:LS= 1.1:1, Mid Parental height: 160 cm, his projected height is below target height (151.5-168.5cm). There was widening of both elbow, knee and ankle joints, pectus carinatum present and no rachitic rosary and bowing legs but flexon deformity present both upper limbs and Equino-varous deformity present in right foot. Muscle power, tone and strength was normal. Elbow, wrist and ankle joint is swollen but non tender. Range of movement is normal except both elbow and right ankle joints. Extension is restricted in both elbow joints and dorsi flexion is restricted in right ankle joints. Leg-length discrepancy (2cm) was present. Spine was normal. Limping gait with right sided toe walking present. There was no organomegaly. Higher psychic funtion , cranial nerves and reflexs were normal. Other systemic examination reveals no abnormalities.





DEPARTMENT OF HAEMATOLOGY

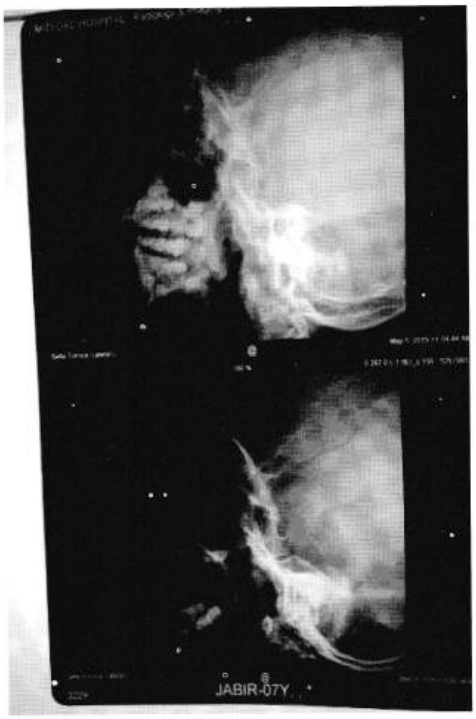
Bangabandhu Sheikh Mujib Medical University

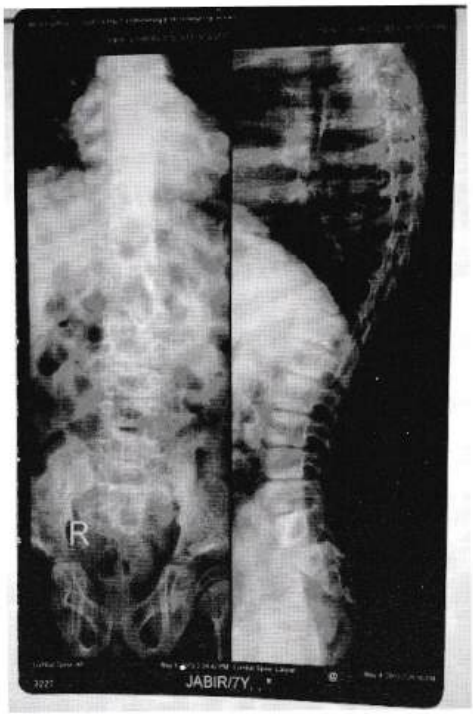
BLOOD REPORT

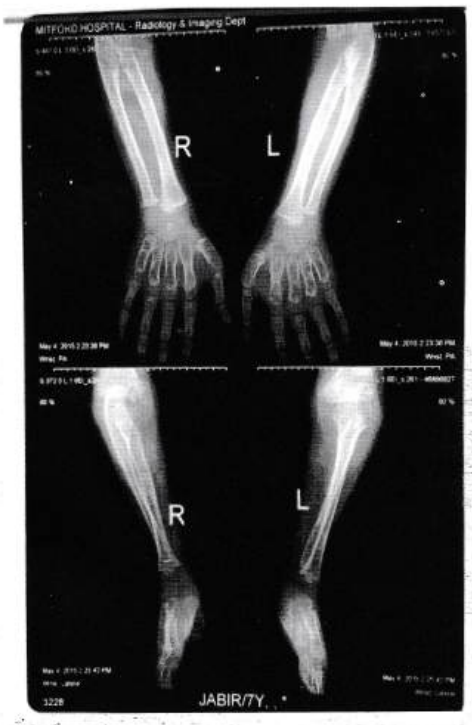
Hb ELECTROPHORESIS REPORT

Investigation: CBC- Hb= 12.5 gm/dl, ESR= 11mm in 1st hr, TC= 9000/cmm, Platelet =2,90000/cmm, PBF= normal. TSH= 2.32 mIU/L, T4= 107.7 nmol/L(normal), S.ferritin= 49 micro gm/L (normal), Hb electrophoresis= Hb A-95.5%, Hb A2- 4.5%(BetaThalassaemia trait) . S. creatinine:1, S.sodium: 136 mmol/l.S. potasium:5.2 mmol/l, S.chloride110 mmol/l. S. Alkaline phophatase-188U/L, S. Calcium- 10.1 mg/dl, S. inorganic phosphate- 5.4 mg/dl, Growth hormone (basal) 0.212ng/ml(ref: 0.06-5.0ng/ml), GH(provocative) 3.24 ng/ml, Karyotype-46 XY.

Skeletal survey: **USG(intrauterine at 35 weeks)** rhizomelic short humerus and femur. **X ray skull**- Calveria is enlarged, sella turtica normal. **X ray hand**- bone age 4 years, widening of lower end of radius, **X ray ankle**- widening of lower end of tibia, mildly ragged metaphysis. **X ray spine** - partial collapse at







Investigation: CBC- Hb= 12.5 gm/dl, ESR= 11mm in 1st hr, TC= 9000/cmm, Platelet =2,90000/cmm, PBF= normal. TSH= 2.32 mIU/L, T4= 107.7 nmol/L(normal), S.ferritin= 49 micro gm/L (normal), Hb electrophoresis= Hb A-95.5%, Hb A2- 4.5%(BetaThalassaemia trait) . S. creatinine:1, S.sodium: 136 mmol/l,S. potasium:5.2 mmol/l, S.chloride110 mmol/l. S. Alkaline phophatase-188U/L, S. Calcium- 10.1 mg/dl, S. inorganic phosphate- 5.4 mg/dl, Growth hormone (basal) 0.212ng/ml(ref: 0.06-5.0ng/ml), GH(provocative) 3.24 ng/ml, Karyotype-46 XY.

Skeletal survey: **USG(intrauterine at 35 weeks)** rhizomelic short humerus and femur. **X ray skull**- Calveria is enlarged, sella turtica normal. **X ray hand**- bone age 4 years, widening of lower end of radius, **X ray ankle**- widening of lower end of tibia, mildly ragged metaphysis. **X ray spine** - partial collapse at D11 & D12. End plate irregularities are seen in lower dorsal vertebrae, widened interpeduncular distance caudally is seen. X ray pelvis- Epiphysis of both femur is reduced in height, Iliac bones appear to be widen. ***Comment- Feature are consistent with dysplastic change possibly Achondroplasia.***

Diagnosis: ***Genetic skeletal dysplasia - Hypochondroplasia or Metaphyseal dysplasia with short stature with Thalassaemia trait.***

Management: The parents were counseled that their child is suffering from congenital genetic disorder causing defective bone formation leading to short stature. There is no specific treatment. But he has normal intelligence, can lead independent, productive normal life unless complicated. Genetic counseling- 50% chance of offspring being affected. Advised the child no cousin marriage and screening before marriage as he is a thalassaemia trait.

Psychological support: To increase self esteem & referred to Orthopedic surgeon for skeletal problem.

Discussion

It is an autosomal recessive disorder in which mild clinical manifestations contrast with radiological appearances of gross metaphyseal undermodeling. Most patients present with mild genu valgum. The elbows are unable to extend fully. There may be widening of the lower femora and clavicles.

Bones can sometimes be fragile, but fracturing is usually not common. Patients may present with dental caries, mandibular prognathism, spinal alignment, and disproportionate limb lengthening [4] (upper limbs do not reach the midpelvis in infancy or upper thigh after infancy[3]). Mental development, physical development, and height are usually normal.[4] investigation: X-ray of long bones: striking radiological changes including irregularly frayed, expanded and cup shaped metaphysis with normal epiphysis and diphysis. S.calcium,inorganic phosphorus, alkaline phosphatase, parathormone levels are normal. There is no specipic remedy.Counseling about the nature and future of the disease.[3]

References

1. Jump up Pyle, Edwin (Oct 1, 1931). **"A Case of unusual bone development".** ***Journal of Bone and Joint Surgery*** **(Needham, Massachusetts)** 13 (4): 874-876. Retrieved Aug 18, 2009.
2. ***synd/1140*** at **Who Named It?**
3. Kabir ARML Pediatric practice on parents' presentation 1st edition Lamburg
4. Syndrome of the month. Journal of Medical Genetics, 1987, 24, 321-322
5. The radiological manifestations of metaphyseal dysplasia (Pyle disease). The British Journal of Radiology, 1979, June, 52 (618) 431-40.