ORIGINAL RESEARCH PAPER

Paediatrics

MALIGNANT INFANTILE OSTEOPETROSIS (MIOP) IN INFANT: A RARE CASE

KEY WORDS:

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INTRODUCTION

Osteopetrosis is a rare congenital genetic disorder characterized by increased bone density due to impaired bone resorption by osteoclasts.[1] The autosomal dominant (AD) form of osteopetrosis is usually asymptomatic. It is diagnosed incidentally or may exhibit mild symptoms in late childhood or adult life, but is compatible with long term survival.[2] Autosomal recessive (AR) malignant infantile osteopetrosis (MIOP), incidence rate is 1 in 250,000 newborns [1]. This condition is most commonly diagnosed soon after birth or within the first year of life with severe symptoms of abnormal bone remodeling, including significant hematologic abnormalities with bone marrow failure and extramedullary hematopoiesis, resulting in hepatosplenomegaly, a characteristic macrocephaly with frontal bossing, exophthalmus, bone fractures, and failure to thrive. [1,3] Neurological manifestations of osteopetrosis can also occur due to narrowing of osseous foramina. Here, we report a case of MIOP, presented as pneumonia and visual disturbance at 6 months 20 days child.

CASE REPORT

A 6 months 20 days old, male child born to nonconsanguineous, by LSCS, term gestation with birth weight 3.5 kgs presented with not recognized mother since 1 month, fever, cough and fast breathing since 3 days. There was frontal bossing, depressed nasal bridge, wide opened anterior fontanelle (5×5 cms), prominent wrist joint and posterior fontanelle (0.6×0.5 cms). He was febrile (101.2°.F) and respiratory rate 54/min, Pulse rate 134/min, BP 90/60 mm of Hg, he was pale other skin and mucosa is normal. weight is 5 kgs (< 3 rd centile) length is 60 cms (< 1 st centile), head circumference 41 cms (3rd centile) and abdominal examination revealed hepatomegaly liver Span is 9 cms, splenomegaly and respiratory system revealed crepitations and fundus examination shows Disc pallor and neurosonogaphy show large subarachnoid space and enlarged lateral ventricles other systemic examination is unremarkable. Investigations revealed Hb 6.1g/dl TC 20.5 with differential count is N-10% L-85% M-0.4% E-01% B-0% and ESR 55 mm in firs hour and platelet count was 15,000/mm3, peripheral smear reveling pessary cells, anisopoikilocytosis, nucleated Rbc's phenotype features such as macrocephaly, frontal bossing, (5/100) WBCs, reticulocyte count 1.0%, renal, liver and blood sugars was normal. serum calcium is 8.2, AIK PO4 1528, LDH 841 U/L and INR 1.4, PT-18 , APTT 13.5 sec and other viral screening negative . X ray showing increased bone density in all bones, he was diagnosed as a case of MIOP with anemia and bronchopneumonia, treated with antibiotics and blood transfusion. Suggested bone marrow transplant but not done due to cost.

DISCUSSION

MIOP is AR form which if untreated as a lethal outcome.[3] Most commonly presented with in first year of life. MIOP diagnosis is based on the clinical features and heamatological features and radiological bone changes of increased bone density due to abnormalities in osteoclast differentiation/functions

There are 4 subtypes 1) Malignant / infantile OP 2) Benign / Adult OP 3) Intermediate OP 4) Carbon Anhydrate Type 2 deficiency. our child had symptoms since the age of 5 months. The incidence is 1 in 25000 births Phakde et al conducted a study. In which the age between 3 m to 18 months.[2] The mean age at the time of diagnosis was 3 – 9 m (range 15 days to 9.5 m). IN a study conducted from india out of 8 children 7 were diagnosed after 6 m of age.[3]

The phenotype features such as macrocephaly, frontal bossing and tooth eruption defects due to increased bone mass. Short stature and predisposing to fracture and osteomyelitis due to impairment of longitudual growth of bones.[6,7]

Usually MIOP presented with anemia, thrombocytopenia, hepatomegaly, visual impairment due to optic atrophy, due to over growth of bone and deafness within a year of life.[1,2,5] The most of clinical manifestation due to bony over growth of the marrow and compression of optic and auditory nerves. The commonest clinical presentation was due to optic nerve compression in various studies. our child presented with visual disturbances, anemia, and gross hepatosplenomegaly, which was develop because of the extra medullary hematopoeisis, charecteric radiographic findings are defective metaphyseal remodelling and a bone in bone (fig 1 and fig 2) appearance. Alternative scleroting and lucent bands can give the vertebrae a sandwich appearance.

Computer tomography can be use for diagnosis and to detect the effect of treatment.It also used to asses auditory and optic canal. Increased risk of infection because of unrecognized immunologic abnormalities with MIOP was observed. Infact our child presented to us with bronchopneumonia.

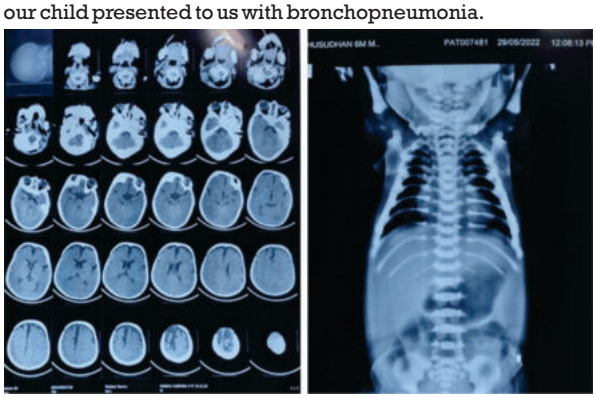

Figure: 1 Figure :2

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