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Case Report

Morquios- Brailsford Disease: A Case Report

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ABSTRACT

Morquios Brailsford disease is a rare condition with an incidence of 0.04-0.3% and is characterized by dwarfism, kyphosis, defective ossification of many epiphyses, and normal intelligence with familial tendency and occurs by autosomal recessive inheritance. Though the clinical presentation is usually typical, conventional radiological and biochemical investigation helps in confirming the diagnosis. We present a rare case of Morquios Brailsford disease with most of the clinical and radiological features as pictorial assay.
Key words: Dwarfism, kyphosis, Morquios

INTRODUCTION

Morquios Brailsford disease is a rare condition with manifestation of dwarfism, [1] kyphosis, normal intelligence, familial tendency common with early fatal outcome and occurs by autosomal recessive pattern with reported incidence of 0.04-0.3% and M: F ratio of 1:1.

Case report: A 4 year old male child presented with progressive swelling and deformity over the back since 2 years, failure to thrive, and difficulty in walking. He was apparently normal till the age of one and a half year. Then the parents started noting the above features. There was family history of similar complaints in his elder brother. Perinatal history was uneventful. On General examination the child is of short stature with a height of 85cm, weighing 11 kgs, body ratio 1.17, malnourished, sunken head between the shoulders (Fig 1[A]) facial appearance and intelligence was normal. pectus carinatum, barrel shaped chest, gibbus over the dorsolumbar (Fig 1[B]) region, hands were broad blunt short and stubby, wrist enlarged, MCP joint extension more than 90^0 knees in valgus (Fig 2[A]) stands with hip and knee flexed with a waddling gait, Intoeing fore foot adduction,

hallux valgus. Provisional diagnosis of Morquios Brailsford disease was made with the above clinical features. Investigation reported mild neutrophilia and mild thrombocytosis, rest all investigation were normal including ultrasound abdomen.

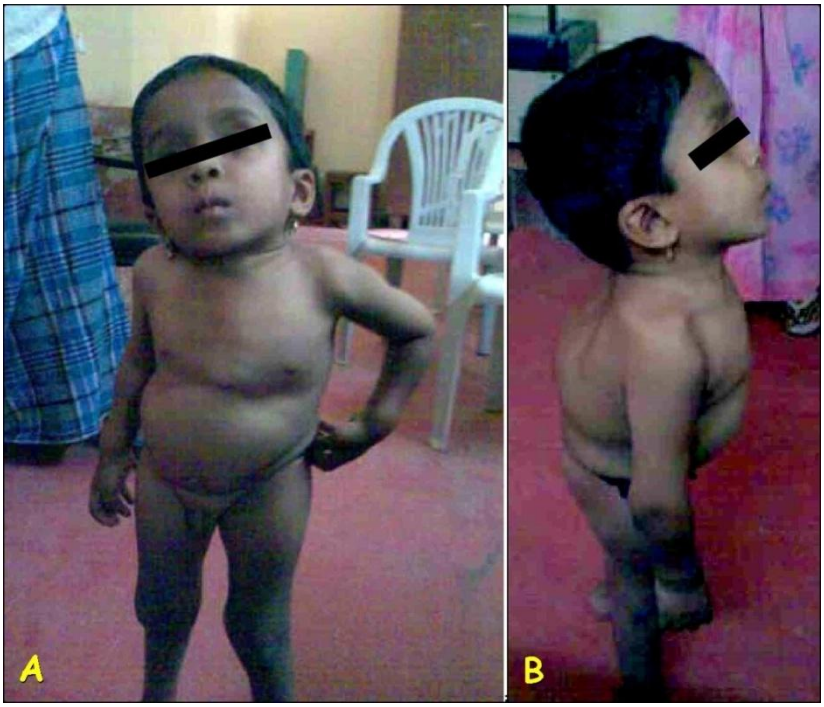

Figure 1:
A. Malnourished, sunken head between the shoulders.
B: Gibbus over the dorsolumbar spine.

On skeletal survey radiograph of the chest shows barrel shaped chest (Fig 3[B]), increased Manubrio sternal angle (Fig 3[D]). DL spine shows kyphosis with characteristic central anterior beaking and tongue like process extending forward from the anterior aspect with posterior scalloping. Disc space are wide, ossification of the epiphyseal rings is delayed and irregular (Fig 3[D]). Pelvis shows widened femoral neck, deformed femoral head, coxa valga associated with wide joint space and shallow acetabular roof (Fig 3[A]). Radiographs of the hand shows pointed metacarpal bases with thick undertubulated shaft of 2, 3, 4, 5^{th} metacarpals (Fig 3[C]). Knee shows genu valgum, irregular epiphysis, sclerosed metaphysis with widened growth plate [2] (Fig 2[B]).

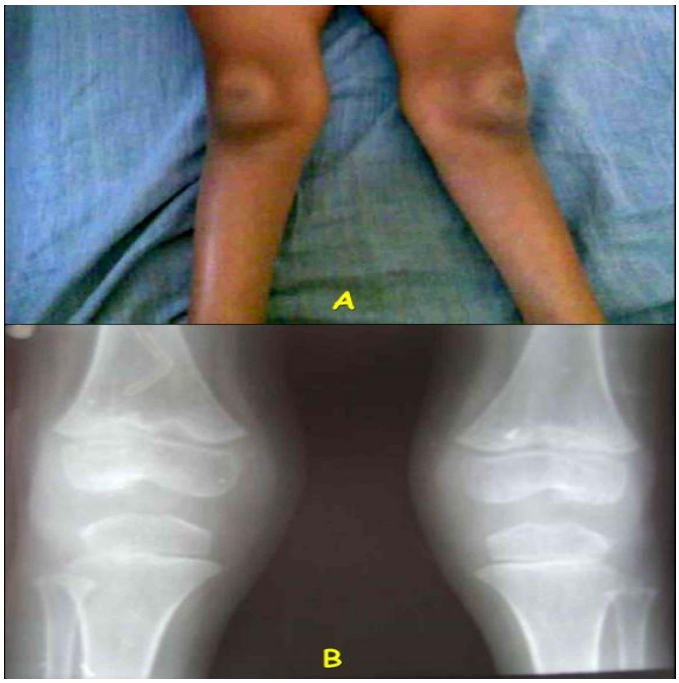

Figure 2:

A. knees in valgus. B. Knee showing genu valgum, irregular epiphysis, sclerosed metaphysis with widened growth plate.

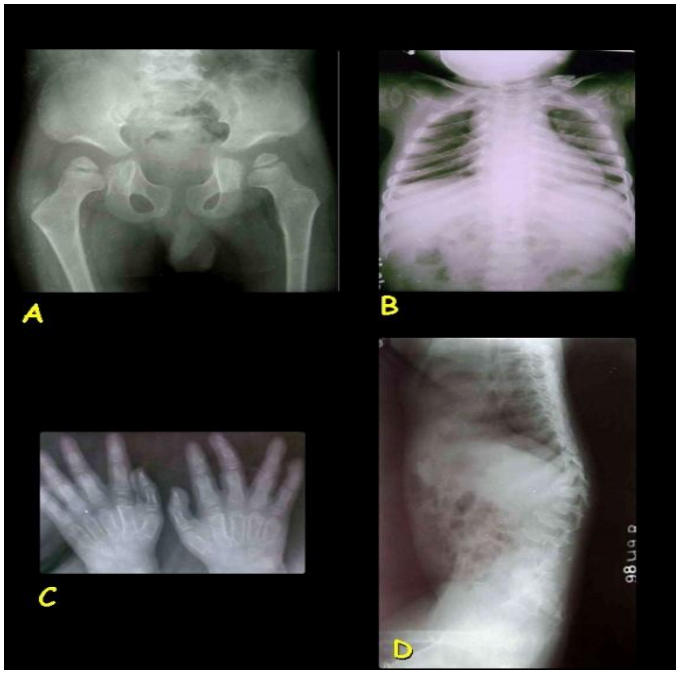

Figure 3:

A. Widened femoral neck, deformed femoral head, coxa valga associated with wide joint space and shallow acetabular roof. B. Barrel shaped chest. C. Pointed metacarpal bases with thick undertubulated shaft of 2, 3, 4, 5th metacarpals. D. Increased Manubrio sternal angle, kyphosis with characteristic central anterior beaking and tongue like process extending forward from the anterior aspect with posterior scalloping. Disc space are wide, ossification of the epiphyseal rings is delayed and irregular.

Above radiological findings supports the diagnosis of Morquios Brailsford disease.

DISCUSSION

This rare condition is MPS type 4 [3] characterized by dwarfism, normal in first and second year although the walking is delayed child looks dwarfed later presents with a kyphotic deformity in the dorsolumbar region, neck shortened and protuberant sternum, there is marked joint laxity and progressive genu valgum. Suitable tests reveal

Conductive hearing loss, unaffected face and normal intelligence. X ray of the spine shows kyphosis, scoliosis, hypoplastic vertebral bodies, platyspondyly with tongue shaped projection anteriorly, odontoid hypoplasia is usual with cervical instability [4,5] which risk the spinal cord should be included in the skeletal suvey. [6] Disc spaces are wide, pelvis shows narrow hourglass inlet, flared iliac wings, coxa valga or coxa vara , hips and other joints dislocation with joint laxity, widened femoral neck, coxa valga deformity associated with widened actabular roof are characteristic. Femoral Head epiphysis is underdeveloped, marked manubrio sternal angle more than 90^0 is pathognomonic. Irregular metaphysis of long bones, proximal pointing of 2 to 5^{th} metacarpal bones which are short and broad, and knee shows genu valgum with medial spur formation at proximal tibial metaphysis, irregular and sclerosed metaphysis at femoral and tibial growth plate margins.

Differential diagnosis: Include other mucopolysaccharoidosis such as 1) Hurler's disease characterized by kyphosis, hepatosplenomegaly and coarse facies, protruded tongue, defective hearing and mental retardation and stiff joints. 2) Hunters syndrome is an x linked recessive with Clinical features similar to hurlers but of lesser severity. 3) Rickets, generalized osteoporosis mainly in diaphysis genu varum more common respond to administration of vitamin D. 4) Gorgylism chondroosteodystrophy type of dwarf characterized by mental deficiency, cloudy cornea, hepatosplenomegaly shape of the vertebra is different. 5) Dysplasia Epiphysealis Multiplexa where stunting of the growth is general not chiefly caused by spine, dwarfism is not as severe as chondrdosteodystrophy. Multiple ossific centers in epiphysis are mulberry like. Muscle power is normal.

Consent: It has been taken from the parents.

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