EUROPEAN JOURNAL OF BIOMEDICAL AND PHARMACEUTICAL SCIENCES
http://www.ejbps.com

ISSN 2349-8870
Volume: 7
Issue: 5
730-733
Year: 2020

RADIOLOGICAL FEATURES OF MORQUIO'S SYNDROME- A RARE CASE REPORT

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Article Received on 18/03/2020 **Article Revised on 08/04/2020** **Article Accepted on 28/04/2020**

ABSTRACT
Purpose: To present a rare case of Morquio's syndrome (Mucopolysaccharidosis Type IV). **Methods:** A 4 yr 7 month old male child with complains of progressive widening of wrist and knees and progressive deviation of lower limbs since 1 yr of age was referred. **Results:** There was progressive widening of chest with episodes of dyspnea. His height and weight were – 84 cm (< 3rd percentile) and 10 kg (< 3rd percentile) respectively. The other positive clinical findings were: presence of Frontal bossing; depressed nasal bridge; Broken teeth; Pectus carinatum; Genu valgum and Kyphoscoliosis. **Conclusion:** Mucopolysaccharidoses Type IV is a rare disease entity and appropriate clinical and imaging features are essential for diagnosis.

KEYWORDS: Morquio syndrome, Mucopolysaccharidoses, Skeletal deformities.

INTRODUCTION
Mucopolysaccharidoses (MPS) are a group of inherited metabolic disorders that result in widespread skeletal, visceral, and mental abnormalities. A defect in metabolic degradation leads to the storage of *mucopolysaccharide* macromolecules in the nervous system and other body tissues; there is also excessive urinary excretion of *mucopolysaccharides*.[1] Morquio's syndrome (MPS IV) is a rare autosomal recessive disorder of *mucopolysaccharide* metabolism. It was first described by Morquio, a Paediatrician in Uruguay in the year 1929. It occurs in approximately 1/100,000 births. There is deficiency of enzyme *N-acetyl-Galactosamine 6 sulfatase* (GALNS) in type IV A and Beta-*galactosidase* in type IV B. This leads to accumulation of *keratan sulfate* in various tissues of the body.[2]

Here, we present a case report of Morquio's syndrome in a 4 year old child with typical radiological features.

CASE REPORT
A 4 yr 7 month old male child presented with complains of progressive widening of wrist and knees and progressive deviation of lower limbs since 1 yr of age with no associated pain and tenderness. He was apparently normal till 1 yr of age. There was also progressive broadening of chest with episodes of dyspnoea on exertion and repeated chest infections. He had developmental delay as his parents noticed him growing slower than peers. He was able to walk without support at the age of around 18-24 months and his primary teeth eruption was at the age of 8 months. He also had auditory dysfunction, as his parents noticed reduced responsiveness to voice since 1 yr of age with repeated pus discharge from both the ears.

On examination: His height and weight were – 84 cm (< 3rd percentile) and 10 kg (< 3rd percentile) respectively. The other positive clinical findings were: presence of Frontal bossing; depressed nasal bridge; Broken teeth; Pectus carinatum; Genu valgum and Kyphoscoliosis.

Skeletal Survey was also done which revealed the following findings as depicted in Figuresc.[3,4,5]

CLINICAL PICTURES AND XRAY FINDINGS

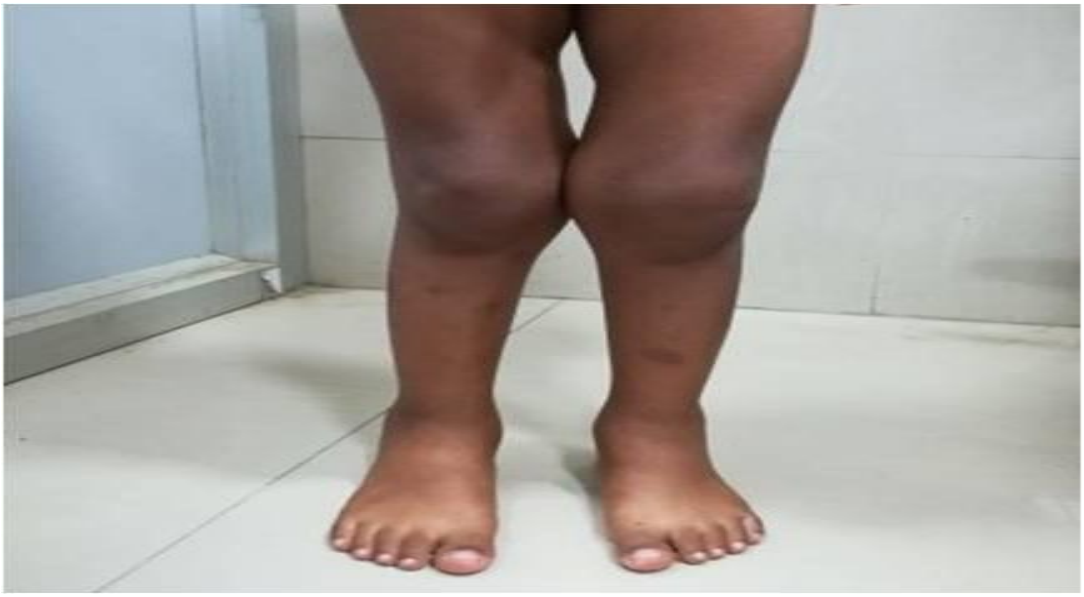

Fig. 1: Knock Knees and Genu Valgum Deformity.

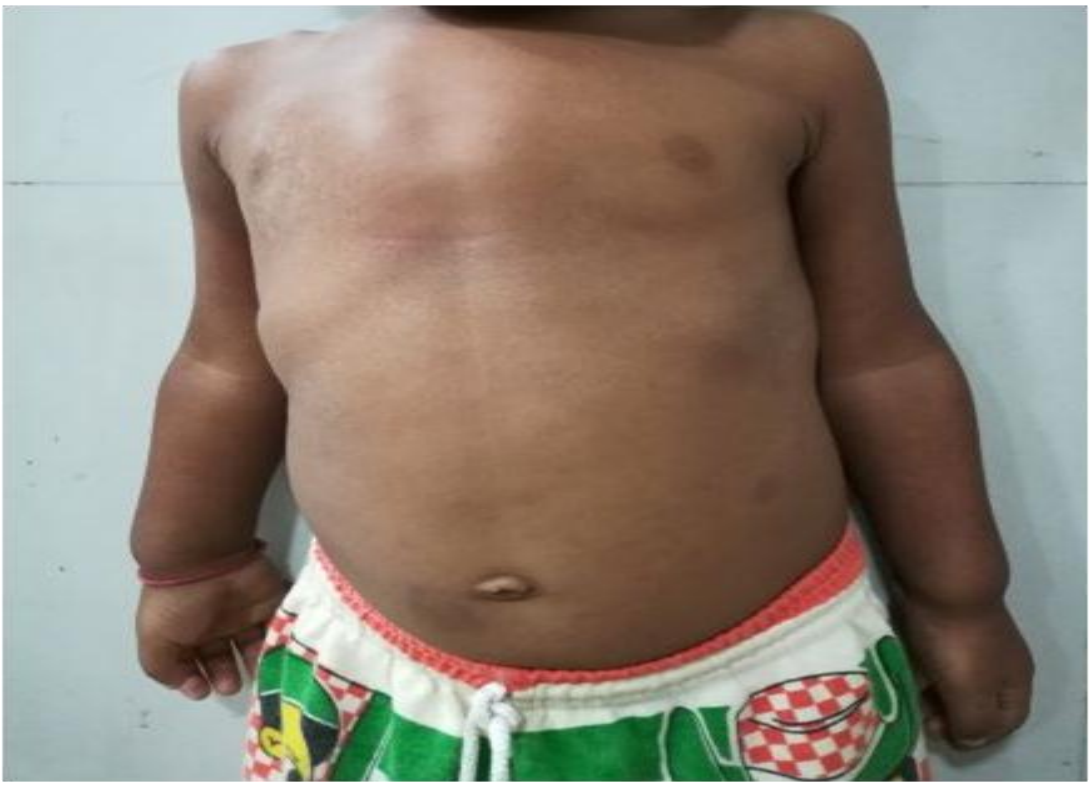

Fig. 2: Pectus Carinatum and Abdominal Distension.

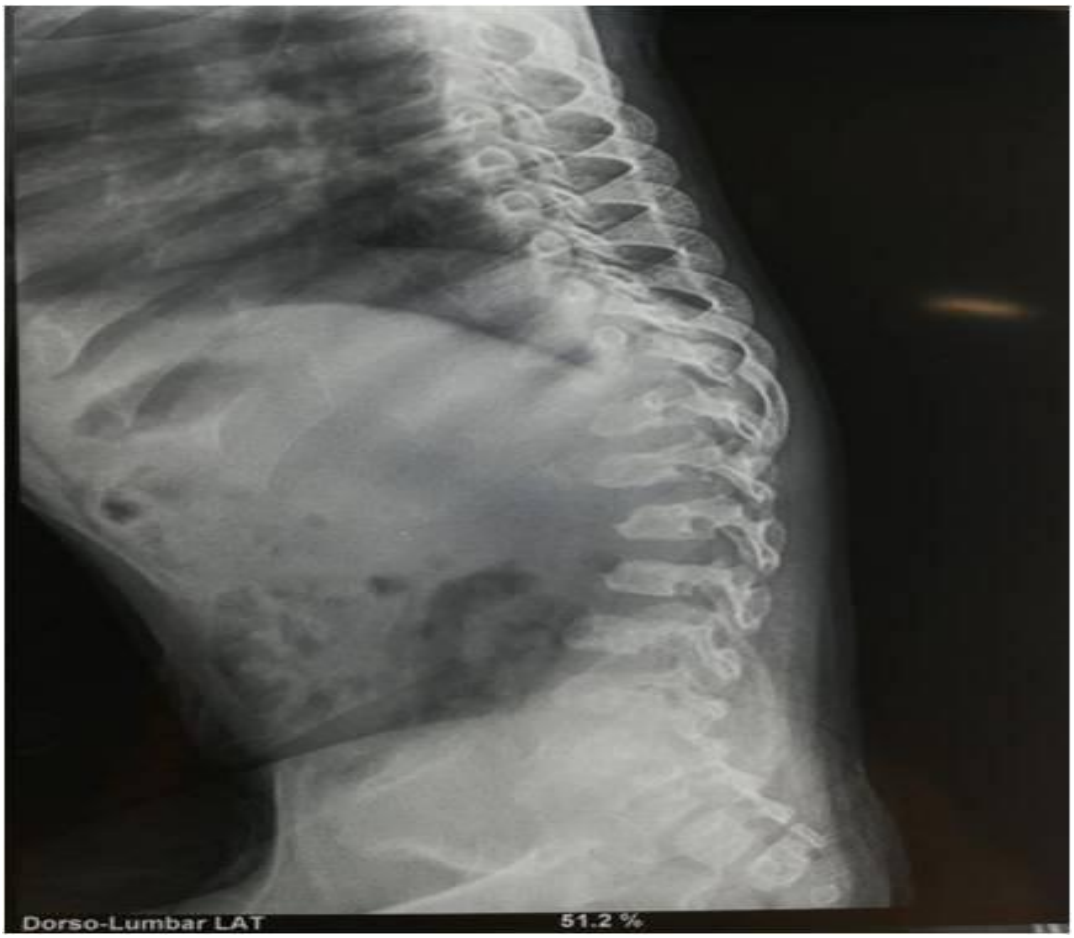

Fig. 2: X RAY Dorso-Lumbar Spine (Lateral View) Hypoplastic Ovoid, Hook-Shaped Vertebral Bodies with Central Beaking and Gibbus At Thoracolumbar Junction.

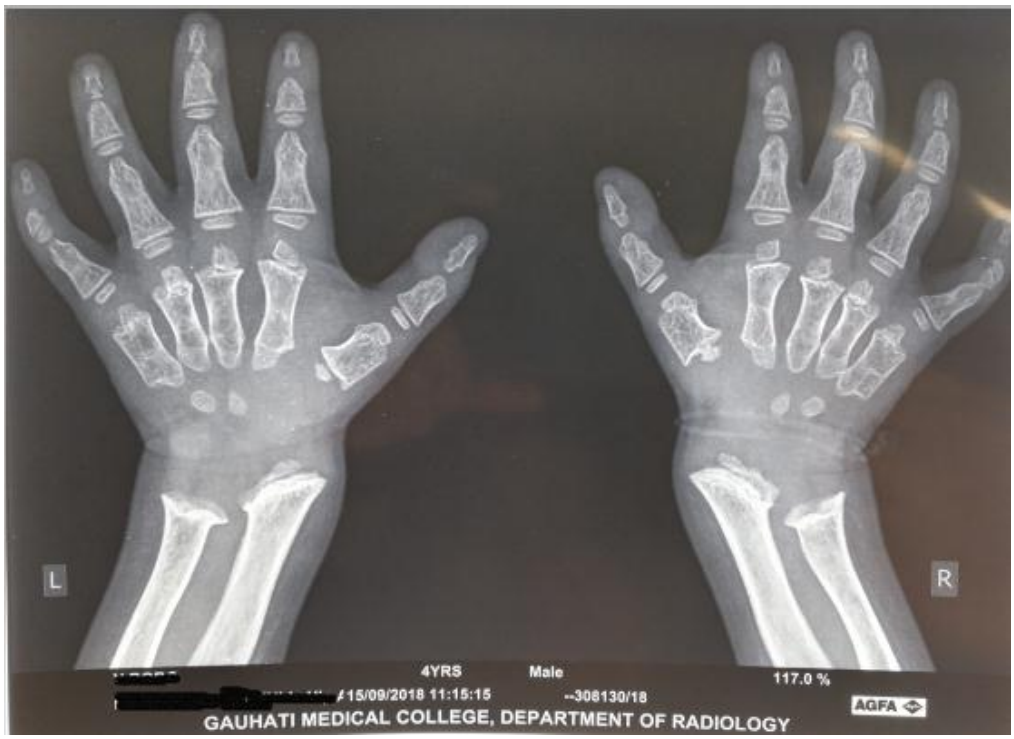

Fig. 3: Antero-Posterior Radiograph of Both Hands and Wrist: Short Wide Phalanges With Characteristic Proximal Pointing Of 2nd To 5th Metacarpals. Metaphyseal Expansion With V Shaped Distal Radioulnar Joint.

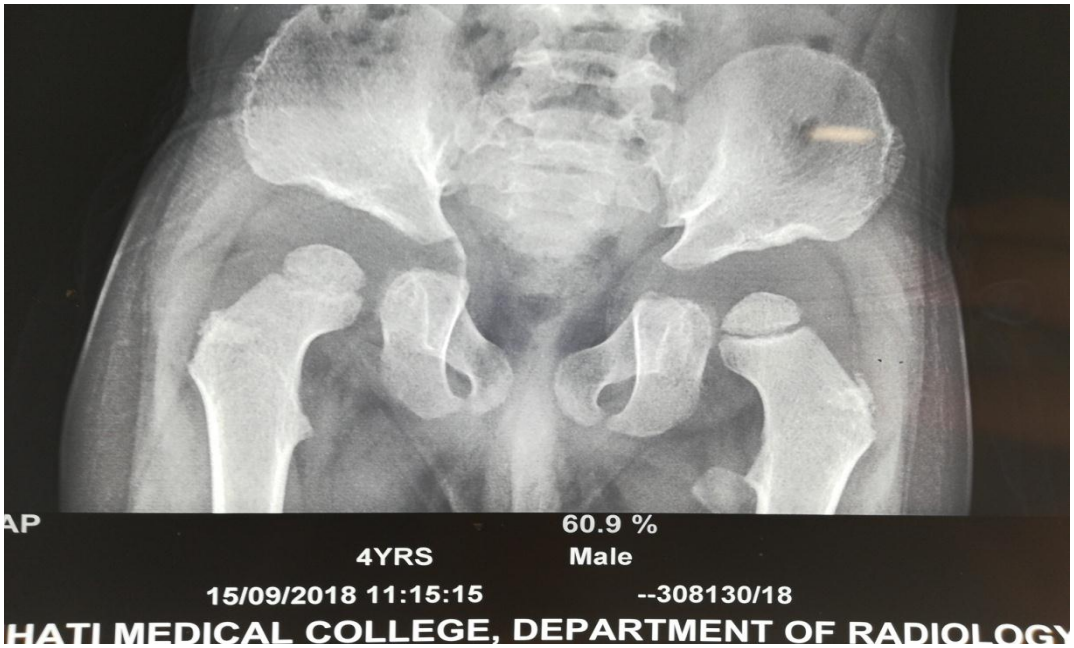

Fig. 4: X RAY Pelvis: Flared Iliac Wings With Inferior Constriction & Horizontal Acetabular Roof. Coxa Valga Deformity of The Femoral Neck.

DISCUSSION

MPS- IV A is a rare autosomal recessive disease affecting males and females equally. Deficieny of the enzyme responsible for the breakdown of the MPS Keratan Sulphate leads to its accumulation along with *Chondroitin 6 Sulfate* leading to various skeletal deformities.

It is suggested by history, physical examinations, skeletal X-rays and urine *Glycosaminoglycans* analysis.

The characteristics clinical findings include: short stature, pectus carinatum, kypho-scoliosis, genu valgum and abnormal gait.[3] They are differentiated from other forms by the preservation of intelligence. They are usually asymptomatic at birth, usually diagnosed at around 2nd year of life for the skeletal deformities.

Roentgenogrpahic findings described by Langer et al distinguishes it from other forms by the presence of greater skeletal manifestations and spine involvement. These may include hyperlordosis, severe gibbus, small or sometimes absent carpal bones, shortening of metacarpals, long pelvis with widening at the acetabula and pubic symphysis widening.

Complications might occur later in life in the form of pulmonary compromise, valvular heart disease, auditory problems, dental problems, visual disturbances and also spinal cord damage leading to possibility of paralysis.

Management is a multidisciplinary approach comprising of medicine specialists, surgeons, physical therapists, a psychiatrist, education professionals and home care professionals.[4] Various modalities of treatment have been used which comprise of Enzyme replacement therapy, gene therapy, haematopoeitic stem cell transplantation but none has shown to be of proven benefit.[4]

CONCLUSION

Patients with MPS IV is a rare disease entity. Clinical findings and appropriate skeletal survey is essential for diagnosis. Therapies for established skeletal defects is a challenge. More insight into the topic is needed for a better quality of life.

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