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CASE REPORT

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ABSTRACT: The metabolic condition known as mucopolysaccharidosis (MPS) is marked by a lack of specific enzymes that affect the lysosomal glycosaminoglycan (GAG) metabolism. A number of variables, such as the particular enzyme deficit, the afflicted target organ, and the degree of glycoprotein accumulation, determine how MPS manifests in clinical settings. A thorough clinical evaluation, as well as the examination of urine, blood, and/or peripheral cell samples, can be used to determine the diagnosis. We report that a 3-year-old boy youngster who had been suffering generalised weakness for six months was brought to the emergency room. The person also showed indicators of exhaustion and had a history of experiencing dyspnea following strenuous exercise or ascending stairs. In this publication, a case study of a person with mucopolysaccharidoses, a genetic lysosomal storage illness, is presented.

INTRODUCTION:

Mucopolysaccharidosis (MPS) is a metabolic disorder characterised by a deficiency in enzymes that impact the lysosomal glycosaminoglycan (GAG) metabolism. The occurrence rate of any type of MPS is one in every 20,000 births[1-2]. Throughout the 20th century, there were numerous distinct descriptions of different types of MPS. The manifestation of MPS in clinical settings is contingent upon various factors, including the specific enzyme deficiency, the affected target organ, and the extent of glycoprotein accumulation. The primary glycosaminoglycans (GAGs) comprise hyaluronic acid, dermatan sulphate, chondroitin sulphate, chondroitin-4-sulfate, and chondroitin-6-sulfate[3-4].The classification of MPS encompasses numerous subtypes and is organised into seven distinct clinical groups[5].

The diagnosis can be established through a comprehensive clinical assessment, as well as the analysis of urine, blood, and/or peripheral cell samples. The timely diagnosis is of utmost importance due to the limited efficacy of therapy in patients aged 2.5 years and younger[6-7]. Significant advancements have been achieved on a global scale in the treatment of MPS. The task of establishing a conclusive diagnosis for any of the MPS is challenging because to the limited availability of enzyme testing in low- and middle-income nations. Our objective is to present a case study of a patient with MPS in our medical facility. The utilisation of clinical, radiological, and biochemical data in investigations has facilitated the attainment of a definitive diagnosis in these particular circumstances.This paper presents a case study of an individual diagnosed with a hereditary lysosomal storage disorder known as mucopolysaccharidoses.

Case Report: A 3-year-old male child was presented to the emergency department with a chief complaint of widespread weakness persisting for a duration of six months. Additionally, the individual exhibited signs of weariness and had a past medical record of having dyspnoea after physical exertion or stair climbing. The observed phenomenon consists of a single episode lasting a duration of 15 minutes, which is thereafter accompanied by the upward movement of the eyes in conjunction with a deviation of the lips, the presence of drooling from the mouth, the rigidity of the body, and the occurrence of significant spasms in the hands, resembling symptoms commonly associated with epilepsy. The child had treatment with antiepileptic medications. The biochemical analysis yielded the following results: haemoglobin (Hb) concentration of 12.2 grams per decilitre (g/dL), packed cell volume (PCV) of 36%, and platelet count (PLT) of 250 x 1000 per microliter (µL). The serum calcium level was measured to be 9.9 mg/dl, while the alkaline phosphatase level was found to be 442 U/L.

Additionally, the phosphorus level was determined to be 5.4 mg/dl, the creatinine level was measured at 0.2 mg/dl, and the uric acid level was found to be 4.6 mg/dl. The radiological findings

indicate the results obtained from the examination of medical images, such as X-rays, CT scans, or MRIs the radiograph of the lateral view of the skull revealed the presence of scaphocephaly and an abnormal J-shaped sella turcica (Figure 1). The radiograph of the anteroposterior view of the wrist and forearm exhibited the classic features of MPS, including shortened and widened metacarpals with proximal pointing (Figure 2). The radiograph of the lateral view of the spine displayed central beaking of the vertebrae and posterior vertebral scalloping (Figure 3). The radiograph of the anteroposterior view of the pelvis demonstrated rounded iliac wings, inferior tapering of the ilia with a poorly developed acetabulum, underdeveloped medial portion of the proximal femoral epiphysis, increased coro-femoral joint space, and coxo-valga (Figure 4). Lastly, the radiograph of the anteroposterior view of the thorax revealed cardiomegaly, thick ribs, and a short thick clavicle.

Langer and Carry have presented a comprehensive analysis of the radiographic findings pertaining to MPS[8], which aligns with the MPS observed in our particular case. The family underwent counselling and was subsequently referred to a specialised facility for the purpose of conducting mutation analysis, whole exome sequencing, and enzyme replacement treatment.

Parents were provided with an explanation regarding the necessity of bone marrow transplantation as the sole therapeutic measure, along with an understanding of the associated risks and long-term advantages. They were further recommended to consider genetic counselling prior to making any decisions regarding future pregnancies.

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Conflicts of interest:
There are no conflicts of interest.

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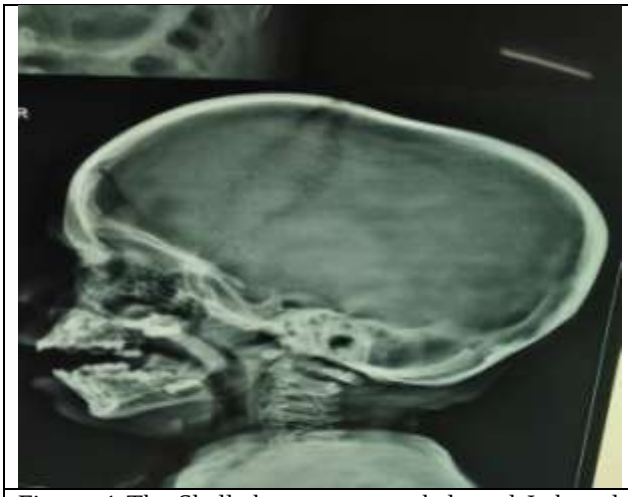

Figure: 1 The Skull shows macrocephaly and J shaped sella.

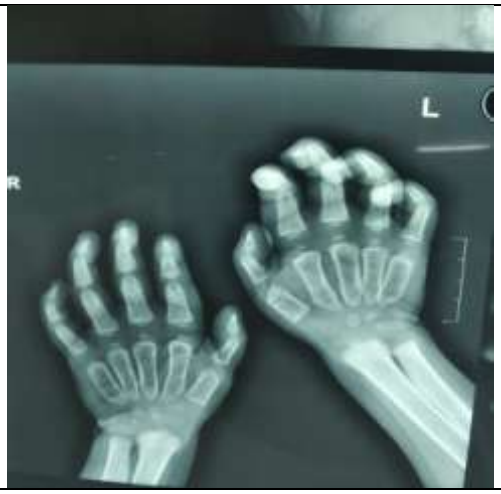

Figure: 2 the hand radiograph shows proximal pointing metacarpals.

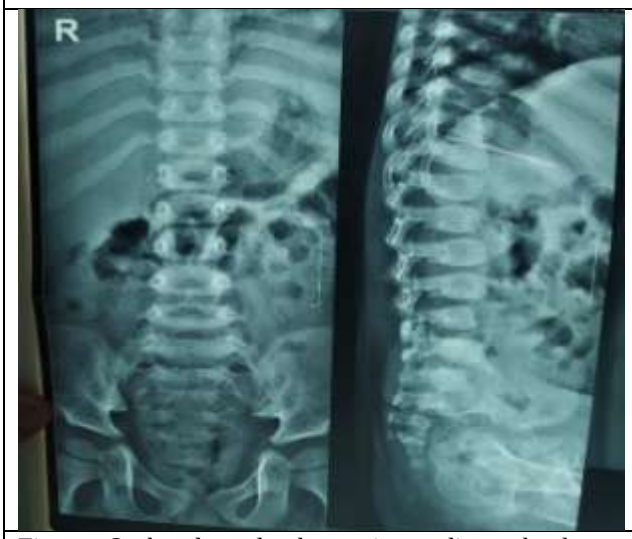

Figure: 3 the dorso-lumbar spine radiograph shows hypoplastic dorso-lumbar vertebrae with anterior breaking of the inferior end plates.

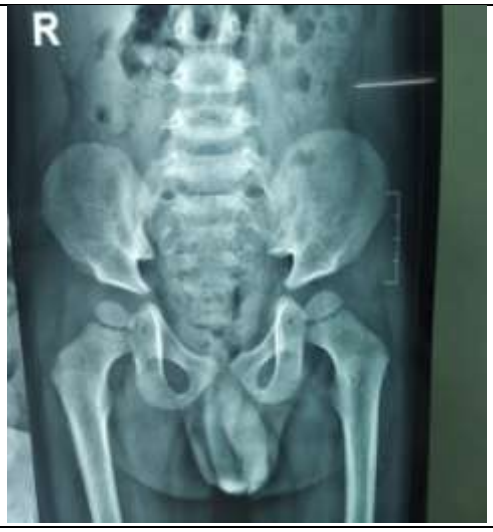

Figure: 4 the pelvis radiograph shows hypoplasia of the base of ilia with poorly developed acetabulum & coxa valga

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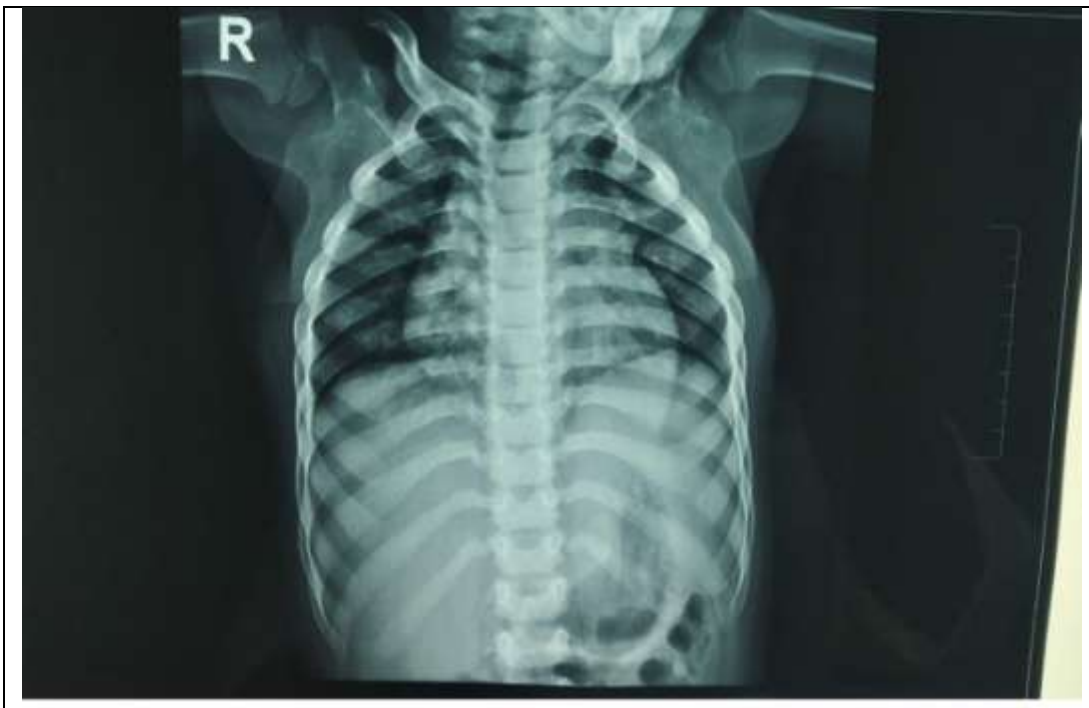

Figure:5 Chest radiograph shows Paddle shaped widened ribs and cardiomegaly.