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Multiple Epiphyseal Dysplasia: Uncommon Diagnosis in a Patient with Swelling over Pinna.

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ABSTRACT

Multiple Epiphyseal Disorder (MED) is a congenital birth disorder presenting with abnormalities of bone and cartilage and is of two types – dominant and recessive. Nearly 50% of individuals with recessive multiple epiphyseal dysplasia (rMED) are born with at least one abnormal feature, including clubfoot, cleft palate, clinodactyly, or ear swelling. A case report of 6-year-old male child with left ear swelling and deafness is presented to highlight the need for maintaining a high index of suspicion for this disorder. A missed or delayed diagnosis of this diagnosis may lead to an erroneous treatment plan.

Keywords: Multiple epiphyseal dysplasia, Pure tone Audiometry, Pinna swelling.

INTRODUCTION

Multiple Epiphyseal Dysplasia (MED) is a rare genetic disorder of cartilage and bone development, primarily affecting the ends of the long bones in the arms and legs (epiphyses). Bones usually elongate by a process that involves the deposition of cartilage at their ends, called ossification. This gradually mineralizes and hardens to become bone.[1] In MED, this process is defective. There are two types of multiple epiphyseal dysplasia – dominant and recessive. Both types have relatively similar signs and symptoms, including joint pain that most commonly affects the hips and knees, early-onset arthritis, and a waddling walk. The recessive type may also present with aural swelling. Studies suggest a prevalence of 10-15 cases per 100,000 births. In general, MED is inherited in an autosomal dominant pattern; however, recessive forms are also seen.[2] This diagnosis is to be considered in patients with pinna swelling, joint abnormalities and hearing loss.

CASE REPORT

A 6-year-old male, who is the second child of healthy non- consanguinous parents with normal history presented with complaints of left pinna swelling for 5 years. The swelling was initially soft and cystic. There was a history of aspiration, followed by incision and drainage at two years of age, after which it turned bony hard. An excision was made subsequently soon, but the swelling soon recurred.

Figure 1: Picture depicting swelling over pinna.

There was history of progressive hearing deficit with the child being initially responsive to verbal commands, but less so over the years. On further enquiry, the mother revealed she had first noticed swelling over the child's joints at 3 months of age. Following this, the child developed a waddling gait, with difficulty in walking and bending forwards (restricted movements).

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Figure 2: Picture depicting Pure Tone Audiometry shows left mild conductive hearing loss.

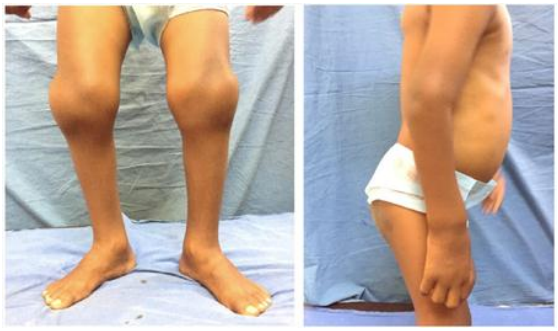

Figure 3A: Picture depicting genu varum of knees and swelling of bilateral ankles. 3B: Picture depicting swelling of wrist joint and abnormal curvature of spine.

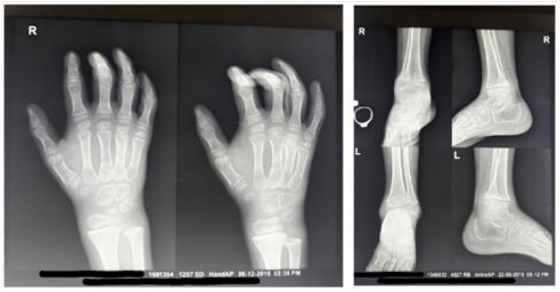

Figure 4A: Picture of X-ray Bilateral Hands AP shows irregular epiphyses and clinodactyly. 4B:Picture of X-ray Bilateral Ankles AP shows demineralisation of bones.

An inspection of the limbs showed swelling over bilateral elbows, ankles and knees. While his stature was normal for his age, he had laterally rotated feet, bowed knees (genu varum) and short fingers (brachydactyly). His developmental milestones were delayed in relation to walking and also dentition. This atypical presentation led to the suspicion of a systemic disorder. He was referred to the departments of Paediatrics and Orthopaedics.

Routine blood investigations were done and found to be well within normal limits. Audiological examination was done which revealed mild conductive hearing loss. X-ray of the spine showed scoliotic changes. X-rays of bilateral wrists, knees and hips revealed demineralization of bones.

Based on the clinical examination and investigations, a diagnosis of Multiple Epiphyseal Dysplasia was made.[3,4] Molecular genetic testing revealed a pathogenic variant in the sequence analysis of SCLA26A2 gene confirming a diagnosis of Recessive Multiple Epiphyseal Dysplasia.[5]

Parents were counselled about the etiology, progression and outcome of the condition. He was adviced to maintain regular follow up.

DISCUSSION

Multiple epiphyseal dysplasia is a broad term for a group of skeletal disorders. It is believed to represent a group of at least six disorders, classified as per the specific genetic mutation that causes each subtype. Most subtypes are inherited in an autosomal dominant manner. One form, multiple epiphyseal dysplasia type 4, is inherited in an autosomal recessive manner.

The five separate genes that cause dominant MED are COMP gene, Collagen type IX alpha-2 (COL9A2) gene, Collagen type IX alpha-3 (COL9A3) gene, Matrilin 3 (MATN3) gene, Collagen type IX alpha-1 (COL9A1) gene.[5] Recessive MED is a rare form of the disease, and is caused by mutations in the SLC26A2 gene. This gene is also known as the diastrophic dysplasia sulphate transport gene (DTDST gene). The disorder is also known as multiple epiphyseal dysplasia type 4.[6] These various genes are responsible in the production of extracellular matrix or formation of collagen in hyaline cartilage. Mutations can lead to pseudo achondroplasia or early onset arthritis.[7] While COMP gene mutations are responsible for nearly 70% of MED cases, SLC26A2 gene mutations are much rarer.[8]

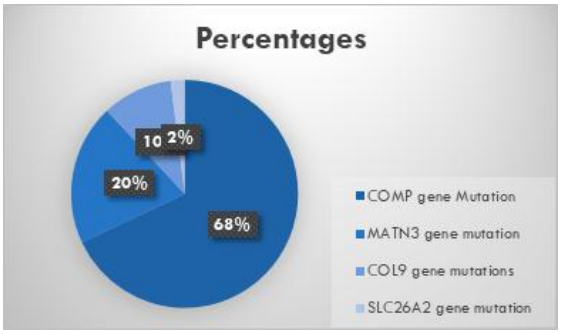

Figure 5: Graph depicting percentages of mutations responsible for MED cases.

Patients with MED present with some or all of the following features. Signs and symptoms can vary.

Onset is usually in early childhood and progressively worsens with age leading to significant disability in the late 30s. Pain in the hips and knees following exercise is usually the first sign that is noticed. Some children develop a characteristic waddling manner of walking. Because of growth deficiency, affected children may be short for their age. Adult height is usually normal, but in the shorter range. The child's arms and legs may be short in comparison to the torso.[1,9]

Some may develop restriction in movement of joints – elbows, back. Others present with low muscle tone and strength making it difficult to lift heavy objects.

In rare cases, additional symptoms may occur including a hip deformity in which the thigh bone is angled toward the centre of the body (cox vara), bowed legs (genu varum), and 'knock knees' (genu valgum).[10]

Our patient presented with a characteristic swelling over the left pinna associated with mild conductive hearing loss. MED can cause hearing loss by the deposition of ossified cartilage over the pinna or external auditory canal; or, by the abnormal ossification of middle ear ossicles, which can progressively worsen in later decades. So regular follow up is essential.

Diagnosis

This is based upon a detailed patient history, clinical evaluation, radiological features and a variety of specialized tests.

This disorder should be suspected in individuals with joint pain, particularly in the hips and knees, skeletal malformation of the hands, feet and knees, and scoliosis along with history of unusual pinna swellings.

Molecular genetic testing confirms clinical diagnosis.[9]

Treatment

Misdiagnosis is a major concern in such patients. Management of MED involves providing symptomatic relief. NSAIDS are prescribed to alleviate pain, and physiotherapy to improve mobility of restricted joints, and to delay the onset of early osteoarthritis. Surgical intervention is done in severe cases – for the correction of angular deformities and joint contractures.[9]

A combined treatment approach with pediatricians and orthopaedic surgeons, physical therapists and other healthcare professionals may need to comprehensively plan the child's treatment.[11]

Genetic counselling, and providing psychological aid to the patient and family members is also essential.

There is no cure for the hearing loss that may occur in some cases, and chronic excision of pinna swelling may cause further development of ossified cartilage.

CONCLUSION

Genetic disorders such as Multiple Epiphyseal Dysplasia are rare, yet must be taken into consideration as a differential diagnosis for unexplained pinna swellings. Several advances have been made into the study of MED and its underlying genetic disorders, yet treatment still involves only treating the symptoms. The combined effort of the various departments can help alleviate symptoms and help improve the quality of life for the patient. Meanwhile, further research and knowledge of these disorders is sorely required.

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