Khanal, J Clin Case Rep 2016, 6:2
DOI: 10.4172/2165-7920.1000726

Journal of Clinical Case Reports

Case Report Open Access

Metaphyseal Dysplasia: A Rare Case Report

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Abstract

Metaphyseal dysplasia is a very rare inherited bone disorder. Here is a case report and possible treatment options for 11 years old child, detected by Karuna foundation Nepal.

Keywords: Metaphyseal dysplasia; Pyle; Therapeutic rehabilitation; Karuna foundation Nepal

Background

Metaphyseal dysplasia also known as Pyle disease is a heterogeneous group of disorders, characterized by the metaphyseal changes of the tubular bones with normal epiphyses. The disease was described briefly by Pyle in 1931 [1,2]. Incidence occurs at a rate of two to three newborns per 10,000 births involving the proliferative and hypertrophic zone of the physis (epiphysis is normal). Jansen, Schmid and McKusick are the three sub-types with a few reports worldwide [3-9].

Karuna foundation Nepal (KFN) is a non-governmental organization which believes in a world in which each individual, with or without disabilities, has equal access to good quality health care, can lead a dignified life, and can participate as much as possible in community life. KFN approach is entrepreneurial and action oriented, working towards setting up and strengthening existing local health care system, stimulating community participation and responsibility-including health promotion, prevention and rehabilitation through empowerment of communities. Below is a description of a child with such a rare case who was identified during one of KFN project [10].

Case Report

Father of 11 years old male child, with the complain of bending of bilateral legs and pain in bilateral knee after walking for short period of time for three years approach to KFN. As a support for medical intervention he was taken to tertiary hospital by KFN. According to his father, problem was started at the age of three and half years when he noticed both his legs are bending and was having difficulty in walking. Child milestones were normal and he was studying at fourth standard.

Family History-There was no history of consanguinity. Together they had four children, of whom the patient was the second. All other children were normal.

Physical examination-The child weight was 20 kilogram (36.9 kg normal) with the height of 106 centimeters (144 cm normal). His was having bilateral flat foot and genu varum on bilateral knees (Figures 1 and 2). Bilateral upper and lower extremities are short and he was having waddling gait. Motor, sensory and reflexes examinations were normal with no limb length discrepancy.

Radio-graphic examination-X-ray of elbow and wrist joint revealed fraying, splaying and cupping of distal radial and ulnar metaphysis. X-ray of pelvis and knee joint concealed splaying, fraying and cupping of metaphysis of bilateral femur and knee joint respectively. Chest x-ray of dorso-lumbar spine is grossly normal (Figures 3 and 4).

Following investigations were normal: Blood Counts, Haemoglobin, Erythrocyte Sedimentation Rate, Serum Alkaline Phosphates, Blood calcium, Urea and SGPT

Based on clinical features and radio-logical findings, the child was diagnosed as Metaphyseal dysplasia with Schmid type.

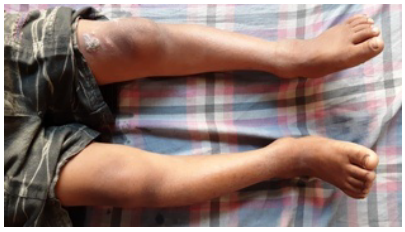

Figure 1: Bilateral genu varum deformity.

Figure 2: Flat foot.

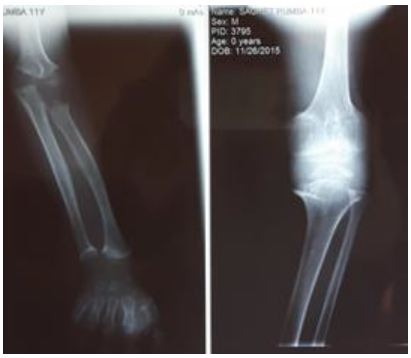

Figure 3: X-ray of Wrist and Elbow shows fraying, splaying and cupping of distal radial and ulnar metaphysis.

Discussion

Metaphyseal dysplasia, schmid type (MDS), is a very rare inherited disorder characterized by short-limbed dwarfism and genu varum.

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Received December 15, 2015; **Accepted** February 03, 2016; **Published** February 07, 2016

Citation: Khanal D (2016) Metaphyseal Dysplasia: A Rare Case Report. J Clin Case Rep 6: 726. doi:10.4172/2165-7920.1000726

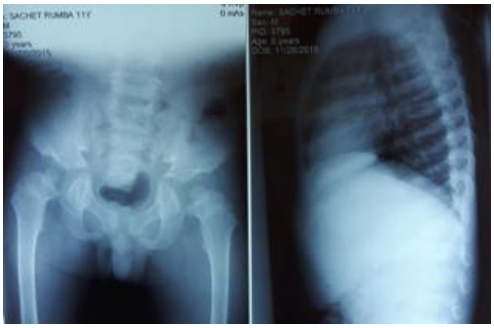

Figure 4: X-ray of Pelvis shows splaying, fraying and cupping of metaphysis of bilateral femur and Dorso-lumbar Spine is grossly normal.

Other characteristics include outward "flaring" of the bones of the lower rib cage, lumbar lordosis, pain in the legs and hip deformities i.e. coxa vara resulting in an unusual waddling gait. It is caused by a mutation of the gene type X collagen called COL10A1. This gene has been mapped to chromosome 6q21-22.3. It affects males and females in equal numbers.

The diagnosis of MDS is alleged during early childhood and may be confirmed by a thorough clinical evaluation, identification of characteristic physical findings, and a variety of specialized tests, particularly advanced imaging techniques. These techniques include x-ray studies that may reveal abnormal development of the large bulbous metaphyseal ends of certain bones of the body, particularly those of the arms and legs, abnormal enlargement of the growing end of the upper portion of the thigh bone i.e. capital femoral epiphysis.

Molecular genetic testing for the COL10A1 gene is available to validate the diagnosis. The test involves sequence of DNA from the COL10A1 gene which detects any disease causing mutations. Prenatal diagnosis can also be made if the specific COL10A1 mutation has been identified in the family. MDS is often mistaken for vitamin D deficiency rickets so it is important that proper diagnosis should be made to avoid redundant vitamin D therapy.

The treatment is symptomatic therefore requiring the coordinated efforts of a team of specialists. Pediatricians, orthopedic surgeons, physical therapists, and other health care professionals may need to analytically and comprehensively plan for the treatment.

Physical therapy and/or orthopedic surgery may help correct certain deformity of the hip and knee. In this case, medial shoe arch support was made for flat foot. For pain management hot moist pack was given and the strengthening exercises for the lower extremities were taught.

Special services that may be beneficial include speech therapy, special social support, physical therapy, and other medical, social and vocational services. Growth hormone therapy is not found to be effective to increase adult height. Genetic counseling will be of benefit for affected individuals and their families [11-13].

References

1. Pyle E (1931) A Case of Unusual Bone Development. Journal of Bone and Joint Surgery 1: 874.
2. David JEA, Palmer PES, Bulawayo, Rhodesia S (1958) Familial Metaphysial Dysplasia. The Journal of Bone and Joint Surgery 40: 1.
3. Percin EF, Percin S, Koptagel E, Demirel H (2003) A case with Pyle type metaphyseal dysplasia: clinical, radiological and histological evaluation. Genet Couns 14: 387-393.
4. Raad MS, Beighton P (1978) Autosomal recessive inheritance of metaphyseal dysplasia (Pyle disease). Clin Genet 14: 251-256.
5. Heselson NG, Raad MS, Hamersma H, Cremin BJ, Beighton P (1979) The radiological manifestations of metaphyseal dysplasia (Pyle disease). Br J Radiol 52: 431-440.
6. Turra S, Gigante C, Pavanini G, Bardi C (2000) Spinal involvement in Pyle's disease. Pediatr Radiol 30: 25-27.
7. Narayananan VS, Ashok L, Mamatha GP, Rajeshwari A, Prasad SS (2006) Pyle's disease: an incidental finding in a routine dental patient. Dentomaxillofac Radiol 35: 50-54.
8. Lindberg EJ, Watts HG (1997) Postosteotomy healing in Pyle's disease (familial metaphyseal dysplasia). A case report. Clin Orthop Relat Res : 215-217.
9. Ferrari D, Magnani M, Donzelli O (2005) Pyle's disease. A description of two clinical cases and a review of the literature. Chir Organi Mov 90: 303-307.
10. http://www.karunafoundation.nl/overons/in_nepal_uk.html
11. https://rarediseases.org/rare-diseases/metaphyseal-chondrodysplasia-schmid-type/
12. Beighton P (1987) Pyle disease (metaphyseal dysplasia). J Med Genet 24: 321-324.
13. Gupta N, Kabra M, Das CJ, Gupta AK (2008) Pyle metaphyseal dysplasia. Indian Pediatr 45: 323-325.