January-April, 2012/Vol 32/Issue 1
doi: http://dx.doi.org/10.3126/jnps.v32i1.5434

Case Report

Bilateral Congenital Cataract Associated with Multiple Epiphyseal Dysplasia: A case Report

Adhikari S[1], Shrestha U[2], Nepal P[3], Singh JL[4]

[1]Dr. Srijana Adhikari, MBBS, MD, Associate Professor, Paediatric Ophthalmology Service, Tilganga Institute of Ophthalmology, Kathmandu, [2]Dr. Ujjowala Shrestha, MBBS, MD, Assistant Professor, Paediatric Ophthalmology Service, Tilganga Institute of Ophthalmology, Kathmandu, [3]Dr. Pravin Nepal, MBBS, MS. Head, Department of Orthopedic, Norvic International Hospital, Kathamndu, [4]Dr. Janith Lal Singh, MBBS, MD. Department of Orthopaedic, Norvic International Hospital, Kathmandu, Nepal.

Address for correspondence: Dr. Srijana Adhikari, E-mail: srij_a@yahoo.com

Abstract

Cataract in children has varied etiology. It may be associated with systemic diseases including skeletal dysplasias. However cataract in Multiple Epiphyseal Dysplasia is a rare association. A child presented with bilateral dense posterior sub capsular cataract and multiple bony abnormalities. Clinical and radiographic findings suggested the disease to be Multiple Epiphyseal Dysplasia. The aim of presenting this case is to report a case of congenital cataract having a rare association with the Multipe Epiphyseal Dyspalsia.

Key words: Congenital cataract, Multiple Epiphyseal Dysplasia, Skeletal dysplasia

Introduction

Multiple Epiphyseal Dysplasia (MED) is a type of osteochondrodysplasia with several different subtypes. It has autosomal dominant inheritance pattern[1]. It was first described by Swedish radiologist Ribbing in 1937. It has characteristic features of delayed and irregular ossification of epiphysis and the early onset osteoporosis which is diagnosed clinically and radiologically[2]. Congenital and developmental cataracts can be divided etiologically into three groups. About a third is hereditary, other third idiopathic and rest third is associated with different ocular and systemic diseases and syndromes. Skeletal dysplasia is one of the systemic diseases associated with congenital cataracts[3,4]. There are few reported cases of congenital cataracts associated with the skeletal dysplasia[5,6,7]. However there is no reported publication on the ocular abnormalities associated with any sub type of MED. We report a case of a child with skeletal abnormalities and bilateral developmental cataract.

The Case

A 12 year old boy presented in our OPD with the gradual painless and progressive diminution of vision in both the eyes for last two years. There was no positive family history of similar disease however one sibling had died at the age of two years with cause unknown. There was no history of any type of treatment received in the past for his ocular or systemic abnormalities. The Orthopedic and Radiology consultation was done. On general examination, the child had slightly short stature, with multiple joint deformities. The height of the child was 3 feet and 8 inch. and the weight was 25 kg. He had bilateral genu valgum. The radiological finding was suggestive of Multiple Epiphyseal Dysplasias. There were dysplastic and osteoporotic changes in the bone. On ocular examination, we found that child had best corrected visual acuity in both the eyes of 2/60. Anterior segment examination revealed dense posterior sub capsular cataracts in both the eyes. The disc was normal in both eyes; macula could not be commented because of media opacity due to cataract. The child underwent bilateral cataract surgery with intraocular lens implantation under general anesthesia. His best corrected visual acuity in the 6 weeks post operative period was 6/6 in both the eyes.

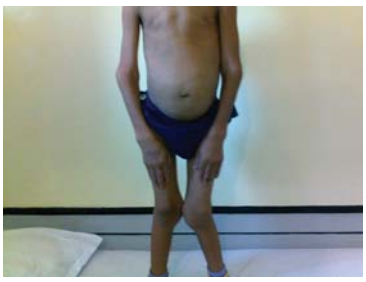

Fig 1: The picture showing genu valgum, deformed elbow and wrist joints

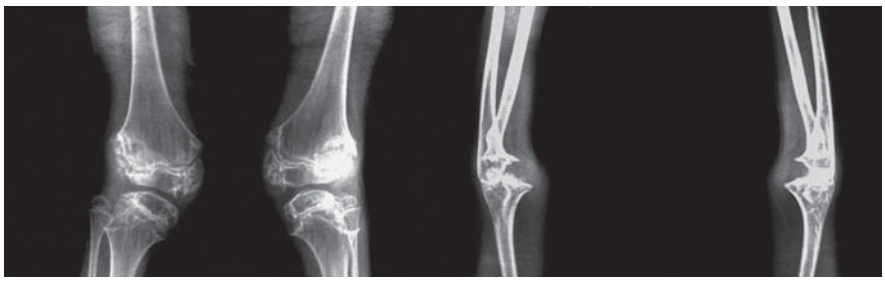

Fig 2: Radiological picture showing dysplastic and osteoporotic changes in the bones and deformities in the elbow and knee joints.

Discussion

The association of cataract with the Multiple Epiphyseal Dysplasia is very rare. On the search of literature, we found few cases of heritable bone diseases associated with congenital cataracts. In the study by Sharma et al cataract and vitiligo was seen in the pseudoachondroplastic variant of Multiple Epiphyseal Dysplasia[5]. Our case was also similar; however there was no skin abnormality, neither was there any clear family history of similar disease. Also, according to the new classification, Pseudoachondroplasia is not a variant of MED. Sarah et al reported ocular disease including two cases of visually insignificant cataract in heritable bone dysplasia associated with the type II collagen defect[7]. Similarly, another study showed the spondyloepiphyseal dysplasia associated with the vitroretinal abnormalities but not the cataracts[8].

The delayed presentation of this case at the age of 12 years could be because the lens opacity became visually significant only later, eventhough it was present since birth. Genes involved in many types of MED have been identified, some are still unknown. There can be some genetic association of developmental cataract and the MED which needs further investigation.

References

1. Andrea Superti-Furga, Sheila Unger, and the Nosology Group of the International Skeletal Dysplasia Society. Nosology and Classification of Genetic Skeletal Disorders: 2006 Revision American Journal of Medical Genetics Part A 143A:1–18 (2007).
2. Johanna Dahlqvist, Hanna Örlén, Hans Matsson, Niklas Dahl, Torsten Lönnerholm, and Karl-Henrik Gustavson. Multiple epiphyseal dysplasia A clinical and genetic study of 12 cases in a Swedish 6-generation family. *Acta Orthopaedica* 2009;80(6):711–15.
3. American Academy of Ophthalmology. Childhood cataract and other pediatric lens disorders. In Basic and Clinical Sscience Course. Section 6 Pediatric Ophthalmology and strabismus. 2009-2010, 285-309.
4. Lorraine Cassidy, David Taylor. Congenital cataract and multisystem disorders Eye (1999) 13, 464-473 © 1999 Royal College of Ophthalmologists.
5. Sharma P, Angra SK, Khurana A Pseudo-achondroplastic variant of multiple epiphyseal dysplasia with cataract and vitiligo. *Ann Ophthalmol* 1987;19(2):56-9.
6. Hugh Ryan.Cataracts of dysplasia epiphysialis Punctata. *Br J Ophthalmol* 1970;54;197.
7. Meredith SP, Richards AJ, Bearcroft A, Pouson AV, Snead MP. Significant ocular findings are a feature of heritable bone dysplasias resulting from defects in type II collagen. *Br J Ophthalmol* 2007;91:1148–151. doi: 10.1136/bjo.2006.112482
8. Toosi SH, Maumenee IH. Vitreoretinal Degenerationin Spondyloepiphyseal Dysplasia. *Congenita Arch Ophthalmol* 1982;100:1104-107.

How to cite this article ?

Adhikari S, Shrestha U, Nepal P, Singh JL. Bilateral Congenital Cataract Associated with Multiple Epiphyseal Dysplasia: A case Report. *J Nepal Paediatr Soc* 2012;32(1):71-72.