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

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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 Dysplasia.

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 pattern¹. 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 radiologically². 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 cataracts³.⁴. There are few reported cases of congenital cataracts associated with the skeletal dysplasia⁵.⁶.⁷. 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.
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. 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. Similarly, another study showed the spondyloepiphyseal dysplasia associated with the vitreoretinal abnormalities but not the cataracts.

The delayed presentation of this case at the age of 12 years could be because the lens opacity became visually significant only later, even though 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


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