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Achondroplasia Associated with Bilateral Developmental Cataract

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***Abstract**: Achondroplasia is an autosomal dominant congenital disorder of enchondral ossification. It is clinically characterized by low stature, craniofacial deformity, and vertebral malformation. Associated ophthalmic features include telecanthus, exotropia, angle anomalies, and cone-rod dystrophy. A 15 year old boy presented with diminution of vision in right eye and typical clinical features of achondroplasia. Two years back, he was diagnosed to have cataract in both eyes and his left eye was operated, after which vision improved. Anterior segment examination showed cataract in RE. As developmental cataract and achondroplasia are developmental disorders, regular ophthalmological examination is required in children with achondroplasia.*

***Keywords:** Achondroplasia, Developmental cataract, Congenital.*

I. Introduction

Achondroplasia is an autosomal dominant congenital disorder of enchondra lossification.[1] Achondroplasia develops as a result of dysplasia of enchondral formation due to the mutation of fibroblast growth factor receptor 3 (FGFR3). The disorder is clinically characterized by short stature, an embossed frontal bone, craniofacial deformity and vertebralma lformation Reported ophthalmic features associated with achondroplasia include simple microphthalmos[4], Crouzon syndrome[5], telecanthus, exotropia, inferior oblique overaction, angle anomalies , Duane retraction syndrome, cone-rod dystrophy , and chorioretinal coloboma . We report a case of bilateral developmental cataract in association with achondroplasia.

II. Case Report

A 15 year old boy presented with diminution of vision in right eye since 3 months. Two years back, he was diagnosed to have cataract in both eyes and his left eye was operated, after which vision improved . On examination, he had short staure, distorted skull, rhizomelic short limbs andincreased spinal curvature. He had normal intelligence and hearing. A thorough orthopaedic examination was done and the patient was diagnosed to have achondroplasia. (figure 1). Family history was negative. His best corrected visual acuity in right eye was hand movements and 6/6 in left eye. Anterior segment examination revealed a total cataract in the right eye and pseudophakiain left eye. (figure 2). B scan of right eye was normal and fundus of left eye was normal. Axial lengthwas 22mm and average keratometry was 43.5D in both eyes. Patient underwent cataract surgery with posterior chamber intraocular lens implantation. On postoperative day one the best corrected visual acuity in right eye was 6/6 and intraocular lens was well centered .

(Figure 3)

Figure 1- Clinical Appearance Of 15year Old Male, Presenting With Typical Achindroplasia.

Figure 2- anterior segment showing cataract in RE.

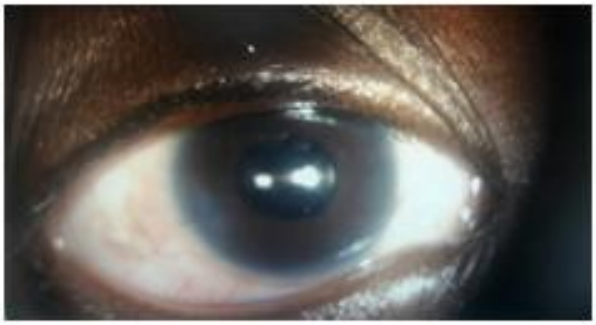

Figure 3- postoperative, Day 1.

Preoperative and postoperative pictures

III. Discussion

Achondroplasia is an autosomal dominant trait that occurs as a result of the mutation of genes encoding fibroblast growth factor- FGFR3. Rosenthal et al. [6] described ophthalmologic characteristics in patients with achondroplasia. According to these authors, telecanthus was found in 26 (50%) of 52 patients and V type exotropia and bilateral inferior oblique muscle overaction were seen in 10 (20%) patients. Five patients presented with the tortousity of retinal blood vessels. In 26 of 46 patients, there were angle abnormalities, such as the definite presence of the iris process, and an incomplete sequestration and abnormal tissue at the anterior angle. Guirgis et al. [7] reported that Duane retraction syndrome and cone-rod retinal dystrophy were concurrently present in children with achondroplasia. Garg et al. [8] reported cases in which the fundus albipunctatus was identified in pediatric patients with achondroplasia. Maumenee and Mitchell [9] proposed the hypothesis that this disease entity occurs due to the genetic defects of a single gene. These authors noted that it occurs in the early embryonic stage and features a high incidence of systemic and central organ malformation.

No previous association between achondroplasia and developmental cataract has been reported. Such concurrence of achondroplasia and developmental cataract raises the possibility of a genetic linkage, although a chance association cannot be excluded. Developmental cataracts occur in about 3 in 10000 live births. The most common cause is genetic mutation, usually autosomal dominant, other causes include chromosomal abnormalities, metabolic disorders and intrauterine infections. Developmental cataract also occur in association with skeletal syndromes like Hallermann Streiff Francois syndrome andNance Horan syndrome. So, developmental cataract can also be associated with achondroplasia, which is also skeletal disorder.

IV. Conclusion

In this case report, we presented the correlation between bilateral developmental cataract and achondroplasia. It is, therefore, important for early and regular ophthalmologic examination of pediatric patients with achondroplasia

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