Case Report

Achondroplasia Associated with Bilateral Keratoconus

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1. Introduction

Achondroplasia is a rare genetic disorder which affects the skeletal system. It is the result of increased signal transduction from a mutated fibroblast growth factor Receptor 3 (FGFR3) which causes an abnormality of cartilage formation. This disorder is characterized by frontal bossing, midface hypoplasia, otolaryngeal system dysfunction, and rhizomelic short stature with normal intellect [1]. Reported ophthalmic features associated with achondroplasia include simple microphthalmos [2], Crouzon syndrome [3], telecanthus, exotropia, inferior oblique overaction, angle anomalies [4], Duane retraction syndrome, cone-rod dystrophy [5], and chorioretinal coloboma [6]. We report a rare case of bilateral keratoconus in association with achondroplasia.

2. Case Report

A 26-year-old male presented with history of gradual deterioration in vision for the past few years. Ophthalmic evaluation revealed uncorrected visual acuity (UCVA) of 20/40 in the right eye (RE) and 20/400 in the left eye (LE) improving with pin hole to 20/30 and 20/50 in the RE and LE, respectively. His refraction was $-2.75 + 1.75 \times 125$ in RE and $-22.00 + 7.75 \times 70$ in LE. Slit lamp biomicroscopy showed bilateral central corneal protrusion and stromal thinning at the apex consistent with keratoconus. A trial of hard contact lens fitting failed to improve VA in the left eye (LE). Right eye (RE) improved to 20/25. The patient underwent penetrating keratoplasty (PKP) in his LE. Twenty-seven months postoperatively, UCVA was 20/30.

3. Discussion

No previous association between achondroplasia and keratoconus has been previously reported. Such concurrence of achondroplasia and keratoconus raises the possibility of a genetic linkage, although a chance association cannot be
excluded. Reports implicate gross structural changes in the gene encoding type II collagen (COL2A1) as the basic defect in achondroplasia [7, 8]. Other reports could not reach the same conclusion [9]. Although type II collagen is not found in the cornea, the presence of a defect in a type of collagen my lead us to think of the possibility that other types of collagen are affected as well. This could explain the association between keratoconus and achondroplasia since corneal stroma contains collagen.

4. Conclusion

To the best of our knowledge, no previous association of bilateral keratoconus with achondroplasia was reported. Ophthalmologists should be aware that patients with this syndrome who complain of poor vision should be suspected of having keratoconus once other more common conditions are ruled out.

Disclosure

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References


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