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METHODS

Megalocornea-mental retardation syndrome: Rare but can be there

Anubhav Chauhan*, Deepak Kumar Sharma and Pankaj Kumar Thakur

Department of Ophthalmology, Shri Lal Bahadur Shastri Government Medical College and Hospital, Nerchowk, Mandi, Himachal Pradesh, India

***Correspondence:**
Anubhav Chauhan,
chauhan.anubhav2@gmail.com

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We report a case study of a 10-year-old male who had megalocornea with mental retardation (Neuhäuser syndrome). Megalocornea-mental retardation is a rare syndrome with a few cases reported in literature. The patient with megalocornea-mental retardation requires a thorough systemic examination.

Keywords:

Introduction

Neuhäuser syndrome is an extremely rare genetic disease, in which the specific cause is unknown and there is no diagnostic test. The diagnosis in childhood is usually performed by oculo-neurological criteria (1).

Case report

A 10-year-old male was referred to us from department of pediatrics for routine ocular examination. He was diagnosed with mild mental retardation. There was no significant medical, surgical, family, traumatic, or drug abuse history. Ocular examination was carried out, and his visual acuity was 6/6 in both the eyes. Ocular movements, fundus, and intraocular pressure were normal bilaterally. Slit-lamp/torch examination revealed bilateral corneal diameter of 13 mm (megalocornea) (**Figure 1**). Keratometry, optical coherence tomography, and B-scan ultrasonography were within the normal limits. Later, a diagnosis of megalocornea-mental retardation (MMR) syndrome was found. No further intervention was done from ophthalmology side.

Discussion

Megalocornea can be defined as an isolated abnormality inherited by an X-linked mechanism, or it can be associated with other entities (2). Megalocornea (corneal diameter $\geq$ 13 mm) is associated with mental and neurological impairment and minor anomalies in Neuhäuser syndrome (MMR syndrome) (3). Megalocornea is a defining feature of Neuhäuser syndrome. The genetic cause of this syndrome is currently unknown. The majority of reported cases are associated with an autosomal recessive mode of inheritance (4). Megalocornea, mental retardation, and, presumably, hypotonia are the major manifestations for diagnosis (5).

Various conditions associated with megalocornea are Axenfeld-Rieger syndrome, Peters anomaly, primary congenital glaucoma, aniridia, congenital hereditary endothelial dystrophy, sclerocornea, Frank-ter Haar syndrome, buphthalmos, Crouzon syndrome, Marfan syndrome, albinism, Ritscher-Schinzel syndrome, Wolfram-like syndrome, Lamellar ichthyosis, and osteogenesis imperfecta (6).

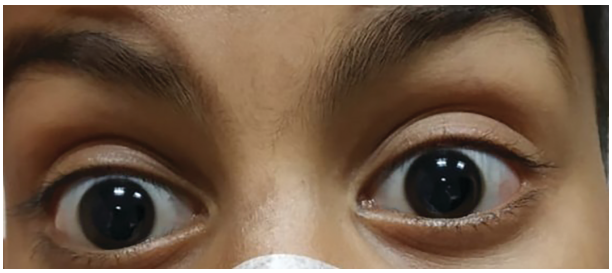

FIGURE 1 |

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