MME-Rare Can Be There

Dr. Anubhav Chauhan* (M.S Ophthalmology), Medical Officer (Specialist), Dr. Deepak Kumar Sharma (M.S Ophthalmology), Assistant Professor and Dr. Pankaj Kumar Thakur (M.S Ophthalmology), Assistant Professor

Deptt. of Ophthalmology, Shri Lal Bahadur Shastri Government Medical College and Hospital, Nerchowk, Distt. Mandi, Himachal Pradesh, India
*E-mail: chauhan.anubhav2@gmail.com

Abstract. We present a 10-year-old male with mental retardation & megalocornea (Neuhauser syndrome). It is an uncommon condition, with only a few occurrences documented in the literature. As several disorders are commonly connected with megalocornea, these individuals also require a full systemic assessment.

INTRODUCTION

Neuhauser syndrome is an exceedingly uncommon hereditary condition with no known aetiology and no diagnostic test. In most cases, oculo-neurological criteria are used to make the diagnosis in children [1].

CASE

A 10-year-old boy was sent to us for regular eye examination by the department of paediatrics. He was diagnosed as a case of mild mental retardation. There was no substantial medical, traumatic, familial, surgical or drug misuse history. An ocular examination was carried out & his visual acuity is excellent, in both eyes was 6/6. Ocular motions, intraocular pressure & fundus were all normal on both sides. Slit lamp/torch an investigation revealed bilateral corneal diameter of 13 mm (megalocornea) (Figure 1). Keratometry, optical coherence tomography and B scan ultrasonography were within normal limits. A diagnosis of Megalocornea-Mental retardation(MMR) Syndrome was made. No further intervention was done from ophthalmology side.

DISCUSSION

Megalocornea can be seen as a standalone aberration inherited by an X-linked pathway, or it can be coupled with other entities [2]. Megalocornea (corneal diameter more than or equal to 13 mm) is related to mental and neurological disability, as well as minor defects in Neuhauser syndrome (megalocornea-mental retardation syndrome) [3]. Neuhauser syndrome is distinguished by megalocornea. This syndrome’s genetic cause is still unclear. The majority of documented instances are autosomal recessive in
nature [4]. The main symptoms for diagnosis are megalocornea, mental retardation, hypotonia and probably [5].

Various conditions which can be associated with megalocornea are Buphthalmus, Congenital hereditary endothelial dystrophy, Aniridia, Peters anomaly, Axenfeld-Rieger syndrome, Marfan syndrome, Primary congenital glaucoma, Sclerocornea, Frank-Ter Haar syndrome, Lamellar ichthyosis, Crouzon syndrome, Albinism, Ritscher-Schinzel syndrome, Wolfram-like syndrome and Osteogenesis imperfecta [6].

SOURCE OF SUPPORT-NONE

The article submitted has never been published in any journal before, nor has it been concurrently forwarded or approved for publishing elsewhere.

CONFLICTS OF INTEREST

The writers claim to have no conflicting interests.

FINANCIAL DISCLOSURE(S)

The writers had no financial or proprietary interest in any of the materials mentioned in this study.

REFERENCES