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Expanding the ocular phenotype of 14q terminal deletions: A novel presentation of microphthalmia and coloboma in ring 14 syndrome with associated 14q32.31 deletion and review of the literature.

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Abstract

A variety of ocular anomalies have been described in the rare ring 14 and 14q terminal deletion syndromes, yet the character, prevalence, and extent of these anomalies are not well defined. Identification of these ocular anomalies can be central to providing diagnoses and facilitating optimal individual patient management. We report a child with a 14q32.31 terminal deletion and ring chromosome formation, presenting with severe visual impairment secondary to significant bilateral coloboma and microphthalmia. This patient is compared to previously reported patients with similar ocular findings and deletion sizes to further refine a locus for coloboma in the 14q terminal region. Those with ring formation and linear deletions are compared and the possibility of ring formation affecting the proximal 14q region is discussed. This report highlights the severity of ocular anomalies that can be associated with ring 14 and 14q terminal deletion syndromes and reveals the limited documentation of ocular examination in these two related syndromes. This suggests that many children with these genetic changes do not undergo an ophthalmology examination as part of their clinical assessment, yet it is only when this evaluation becomes routine that the true prevalence and extent of ocular involvement can be defined. This report therefore advocates for a thorough ophthalmological exam in children with ring 14 or 14q terminal deletion syndrome. © 2016 Wiley Periodicals, Inc.

KEYWORDS: 14q terminal deletion syndrome; chromosomes; coloboma; human; microphthalamos; ophthalmology; pair 14; ring 14 syndrome; ring chromosomes

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