

Trisomy 4q with morning glory disc anomaly

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ABSTRACT. The authors describe a case of trisomy 4q with a unilateral morning glory disc anomaly, a previously unreported ocular manifestation. Previous ocular involvements are summarized.

Key words: trisomy 4q; morning glory disc; optic disc coloboma; chromosomal anomaly; trisomy

INTRODUCTION

Partial trisomy 4q is a rare genetic abnormality first identified in 1971¹. Approximately 30 cases have been described with highly variable ophthalmic and systemic manifestations. We report a case of trisomy 4q with a unilateral morning glory disc anomaly and review previously described ocular findings. The most frequently reported systemic manifestations of this disorder are craniofacial malformations, cardiac and renal defects, cryptorchidism and mental retardation. Recently, an association with acute lymphoblastic leukemia (ALL) and acute nonlymphocytic leukemia (ANLL) has been reported^{2,3}.

CASE REPORT

A 9-year-old black girl with partial trisomy 4q (q12 qter) was referred for the evaluation of strabismus. She was the product of a full-term, uncomplicated

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pregnancy and has no family history of genetic disease.

Her vision was central, steady, and maintained, in both eyes and she followed a near target normally. Her intercanthal distance was 45 mm, and her pupillary distance was 65 mm. Her refractions and versions were full. She was orthophoric at near and had a distance exophoria of 10 diopters. Her conjunctiva, cornea, anterior chamber, iris, and lens, were entirely within normal limits. The pupillary examination was normal without afferent defect. Dilated retinoscopy showed -2.00 sphere OD and -1.00 sphere OS. Fundus examination was entirely within normal limits OD. In the left eye, the optic nerve was enlarged with a central coloboma and had peripheral, radial exiting of vessels. In addition, there was a tuft of glial tissue centrally and a pigmented, raised annulus of subretinal tissue (see Fig. 1). These findings were consistent with a morning glory disc anomaly.

DISCUSSION

Ocular involvement has been described in 12 cases of trisomy 4q, current case included, and is summarized in Table 1⁴⁻¹². This involvement appears to be rather nonspecific. The most typical findings are antimongoloid slant (seven out of

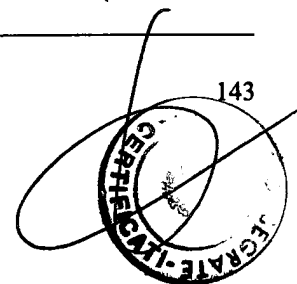




Fig. 1. This drawing is of the left optic nerve of our patient. The disc is enlarged and has a central coloboma with peripheral, radial exiting of vessels. Centrally, there is an overlying tuft of glial tissue. Surrounding the disc is a raised, pigmented annulus.

12), hypertelorism (four out of 12), epicanthus (four out of 12), and esotropia (three out of 12). Microphthalmos has been reported in a significant percentage of cases, but whether all cases were examined by ophthalmologists is unclear. Some cases called microphthalmic (three out of 12) or 'small' eyes may well be normal or nanophthalmic eyes.

The appearance of the optic nerve in the left eye of our patient is consistent with the diagnosis of morning glory disc anomaly¹³. In 1982 Apple and colleagues, in a very thorough review of the congenital anomalies of the optic disc, classified the morning glory disc anomaly as a specific type of optic nerve coloboma, putting forward that this condition might be created when the most superior aspect of the embryonic fissure fails to close, allowing posterior prolapse of the tissues at

TABLE 1. Ocular findings in trisomy 4q

Ocular Findings	Dutrillaux 4q(q25-q34)	Baccichetti 4q(q32-qter)	Biederman 4q(q21-q35)	Cervenka 4q(q25-qter)	Andrie 4q(q25-qter)	Stella 4q(q27-qter)**	Stella 4q(q27-qter)**	Bonfante 4q(q27-qter)*	Bonfante 4q(q27-qter)*	Kelly 4q(q25-q21)	Fryns 4q(q25-q32)	Nucci 4q(q12-qter)
Antimongoloid slant	-	-	-	+	+	+	+	+	+	-	-	+
Hypertelorism	+	-	-	-	-	-	+	-	+	-	-	+
Epicanthus	+	-	-	-	-	-	+	-	+	+	-	-
Esotropic	-	-	-	+	+	-	-	+	-	-	-	-
Microphthalmia	+	-	-	-	-	-	-	+	+	-	-	-
Small and deeply set eyes	-	+	+	-	-	-	-	-	-	-	+	-
Bushy eyebrows	-	-	-	+	-	-	-	+	+	-	-	-
Ptosis	-	-	-	-	+	-	-	-	-	-	+	-
Tapeto degeneration of the macula	-	-	-	-	-	-	+	-	-	-	-	-
Nystagmus	-	-	-	+	-	-	-	-	-	-	-	-
Narrow palpebral fissures	-	-	-	-	-	-	-	-	+	-	-	-
Blepharophimosis	+	-	-	-	-	-	-	-	-	-	-	-
Morning glory disc anomaly	-	-	-	-	-	-	-	-	-	-	-	+

* Same paper; ** Sisters, same paper.



the disc and peripapillary region¹⁴.

This description of morning glory disc anomaly represents the first report of coloboma in isolated trisomy 4q. This new finding appears, however, in a setting one might expect to see it. Colobomatous malformations are frequent in many gross chromosomal anomalies (trisomy 13, 13q-, 4p-, etc.)¹⁵. Furthermore, microphthalmia and colobomatous defects are frequently seen in association, and there is some evidence that microphthalmia is common in trisomy 4q¹⁶.

The variability of both ophthalmic and system-

ic phenotypes reported in partial trisomy 4q may reflect the involvement of different segments of the long arm of chromosome four in the trisomy. Analysis of the small number of reported cases, however, reveals no characteristic phenotypic involvement of any particular 4q trisomy.

We believe that ophthalmic evaluation should be performed in this and other rare genetic conditions both to rule out treatable ocular disease and to augment our frequently scanty understanding of potential ocular involvement.

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