Optic nerve hypoplasia associated with chromosome 9 inversion

Pericentric inversion of chromosome 9 [inv(9)(p12q13)] is frequently associated with dysmorphic features and congenital anomalies, but ophthalmic findings are exceedingly rare. We report a case of chromosome 9 inversion associated with optic nerve hypoplasia.

A 3-month-old Caucasian girl was referred to the emergency department at the Hospital for Sick Children in Toronto by her pediatrician for nystagmus present from 4 weeks of age and a cystic lesion in the brain discovered through ultrasonography. The patient was born to a teenage primigravida mother through spontaneous vaginal delivery with no complications. It was an unplanned pregnancy. The patient showed psychomotor delay and moderate growth retardation from 6 weeks of age. On examination, she had distinctive facial features, including frontal bossing, cupped ears, and prominent deep-set eyes. A large-angle esotropia and horizontal, jerk, high-frequency nystagmus were present. Fundus examination revealed severe bilateral optic nerve hypoplasia (Fig. 1). CT and MRI of the head confirmed the presence of small optic nerves and an arachnoid cyst in the left perimesencephalic cistern with no other brain abnormalities. Genetic testing showed pericentric chromosome 9 inversion [inv(9)(p12q13)].

A pericentric inversion is a chromosome rearrangement in which a segment of a chromosome, including the centromere, is reversed end to end. Pericentric inversion of chromosome 9 [inv(9)(p12q13)] is estimated to be present in 1%–3% of the population. It is inherited in Mendelian fashion and may occur spontaneously. A number of abnor-
Correspondence

malities have been associated with chromosome 9 inversion, including dysmorphic features (short stature, depressed nasal bridge, down-slanting eyes, low-set ears, hypertelorism), microcephaly, deafness, ectodermal dysplasia (absent sweat glands, hypotrichosis, hypodontia), repeated spontaneous abortion, infertility (absence of ovaries, hypoplastic uterus, oligozoospermia), developmental and psychomotor delay, as well as psychiatric disorders. To the best of our knowledge, this is the first report of optic nerve hypoplasia associated with chromosome 9 inversion. Whether there is a true association between these 2 conditions or whether they are coincidental findings in our patient remains to be elucidated.

The authors have no proprietary or commercial interest in any materials discussed in this article.

REFERENCES


Roderick Ting Fung Cheung, Agnes M.F. Wong
University of Toronto and Hospital for Sick Children, Toronto, Ont.
Correspondence to Agnes Wong, MD, PhD, FRCSC:
agnesw@uhnres.utoronto.ca

Can J Ophthalmol 2009;44:610–1
doi:10.3129/i09-119