Clinical and genetic aspects of trigonocephaly: A study of 25 cases

Abstract

We reviewed 25 patients ascertained through the finding of trigonocephaly/metopic synostosis as a prominent manifestation. In 16 patients, trigonocephaly/metopic synostosis was the only significant finding (64%); 2 patients had metopic/sagittal synostosis (8%) and in 7 patients the trigonocephaly was part of a syndrome (28%). Among the nonsyndromic cases, 12 were males and 6 were females and the sex ratio was 2 M:1 F. Only one patient with isolated trigonocephaly had an affected parent (5.6%). All nonsyndromic patients had normal psychomotor development. In 2 patients with isolated metopic/sagittal synostosis, FGFR2 and FGFR3 mutations were studied and none were detected. Among the syndromic cases, two had Jacobsen syndrome associated with deletion of chromosome 11q 23 (28.5%). Of the remaining five syndromic cases, different conditions were found including Say-Meyer syndrome, multiple congenital anomalies and bilateral retinoblastoma with no detectable deletion in chromosome 13q14.2 by G-banding chromosomal analysis and FISH, I-cell disease, a new acrocraniofacial dysostosis syndrome, and Opitz C trigonocephaly syndrome. The last two patients were studied for cryptic chromosomal rearrangements, with SKY and subtelomeric FISH probes. Also FGFR2 and FGFR3 mutations were studied in two syndromic cases, but none were found. This study demonstrates that the majority of cases with nonsyndromic trigonocephaly are sporadic and benign, apart from the associated cosmetic implications. Syndromic trigonocephaly cases are causally heterogeneous and associated with chromosomal as well as single gene disorders. An investigation to delineate the underlying cause of trigonocephaly is indicated because of its important implications on medical management for the patient and the reproductive plans for the family. © 2003 Wiley-Liss, Inc.

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