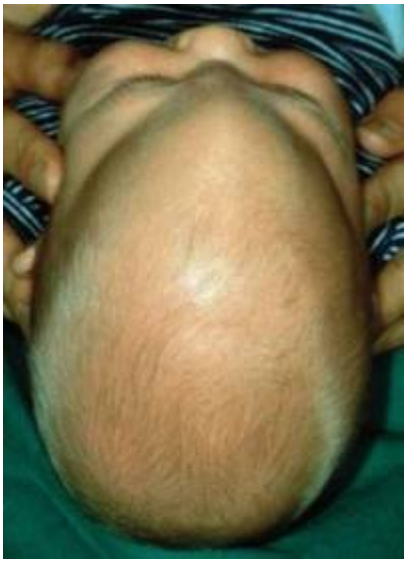
Trigonocephaly



At birth, the frontal bone consists of two halves separated by the frontal or metopic suture. Abnormal closure produces a trigonocephaly (pointed or triangular shaped) forehead with a midline ridge and hypotelorism.

Cranial dysmorphology observed in patients with metopic synostosis varies along a spectrum of severity including varying degrees of metopic ridge, bitemporal narrowing, and trigonocephaly.

Classification

Trigonocephaly Classification.

Epidemiology

75% are male.

The incidence of metopic synostosis is roughly between 1:700 and 1:15,000 newborns globally (differs per country). Trigonocephaly is seen more in males than females ranging from 2:1 to 6.5:1. Hereditary relations in metopic synostosis have been found of which 5.5% were well defined syndromic.

Trigonocephaly is a relatively uncommon form of craniosynostosis, with an incidence of 0.3 per 1000 live births.

The prevalence of trigonocephaly increased during the last two decades both in Europe and in the United States, but no clear contributing factors have yet been identified [1].

Etiology

The cause of trigonocephaly is attributed to premature closure of the metopic suture.

Cytogenetic alterations are probably underestimated in this craniosynostosis, considering the high rate of neurodevelopmental retardation compared to other single-suture synostoses. Genetic counseling is, therefore, more and more effective in this pathology. [2)].

Many of these have a 19p chromosome abnormality and are mentally retarded.

Chromosomal abnormalities described in metopic synostosis comprised deletion of chromosome 11q24, deletion or trisomy of 9p and deletion of 7p, deletions of 3q, 13q, 12pter, 22q11, and duplication of 15q25. SMAD6 mutations should be systematically screened for in familial cases. [3)].

Diagnosis

Trigonocephaly diagnosis.

Diferential Diagnosis

Metopic suture closure can manifest as a benign metopic ridge (BMR), a variant of normal, to “true” metopic craniosynostosis (MCS), which is associated with severe trigonocephaly. Currently, there is no gold standard for how much associated orbitofrontal dysmorphology should trigger surgical intervention.

Cho et al. used three-dimensional (3D) curvature analysis to separate the phenotypes along the spectrum, and to compare surgeons' thresholds for operation. Three-dimensional curvature analyses on 43 subject patients revealed that the mean curvature of mid-forehead vertical ridge was higher for patients who underwent operation than those who did not undergo operation by 1.3 m-1 ($p < 0.0001$). In addition, these patients had more retruded supraorbital areas by -16.1 m-1 ($p < 0.0001$). K-means clustering classified patients into two different severity groups, and with the exception of 2 patients, the algorithm's classification of deformity completely agreed with the surgeons' decisions to offer either conservative or operative therapy (i.e. 96% agreement). The described methods are effective in classifying severity of deformity and in our experience closely approximate surgeon therapeutic decision making. These methods offer the possibility to consistently determine when surgical intervention may be beneficial and to avoid unnecessary surgeries on children with benign metopic ridge and associated minimal orbitofrontal deformity [4)].

It should be distinguished from the benign metopic ridge where hypotelorism and the bitemporal narrowing are absent and have no other clinical features. 3D computed tomographic (CT) scans can be used to distinguish them [5)].

Review

The aim of a review of Mocquard et al. was to report on recent advances in trigonocephaly since the last report on craniosynostosis published in 2006.

The review was conducted in accordance with the PRISMA guidelines. Research focused on four main topics: epidemiology, neurodevelopmental disorders, genetics and surgical techniques.

Forty reports were included. The prevalence of trigonocephaly increased during the last two decades both in Europe and in the United States, but no clear contributing factors have yet been identified. Neurodevelopmental disorders are frequent in syndromic trigonocephaly and not particularly rare in nonsyndromic cases (up to 34%). Developmental retardation (speech, motor or global) was almost always present in children exposed to valproic acid. Chromosomal abnormalities described in metopic synostosis comprised deletion of chromosome 11q24, deletion or trisomy of 9p and deletion of 7p, deletions of 3q, 13q, 12pter, 22q11, and duplication of 15q25. SMAD6 mutations should be systematically screened for in familial cases. Recent advances in surgical techniques have mainly concerned endoscopic-assisted procedures, as they significantly reduce perioperative morbidity.

Neurosurgeons, maxillofacial and plastic surgeons will be increasingly concerned with trigonocephaly because of the increase in prevalence observed over the last two decades. Cytogenetic alterations are probably underestimated in this craniosynostosis, considering the high rate of neurodevelopmental retardation compared to other single-suture synostoses. Genetic counselling is therefore more and more effective in this pathology. An objective method to evaluate the cosmetic results of both endoscopic and open surgeries is necessary, as some under-corrections have been reported with minimally invasive surgery [6].

Treatment

Trigonocephaly treatment.

Outcome

Severity of cranial dysmorphology does not predict the occurrence or severity of associated abnormal neurodevelopment, as children with mild-to-moderate trigonocephaly may also experience developmental delays.

Applegren et al. sought to determine the relationship between mild-to-moderate trigonocephaly and anterior cranial volume using a noninvasive laser shape digitizer (STARscanner) in patients with abnormal head shape.An IRB-approved retrospective review of a prospectively maintained database and medical records was performed. Two hundred three patients less than 1 year of age with abnormal head shape were categorized as having a metopic ridge with mild-to-moderate trigonocephaly, metopic ridge without trigonocephaly, or no ridge. Measurements of cranial volume, circumference, and symmetry were calculated by the STARscanner, which quantifies three-dimensional shape of the cranial surface. Measures were analyzed using a series of analyses of variance and post-hoc Tukey honest significant difference.The authors results showed ACV was significantly reduced in patients with mild-to-moderate trigonocephaly compared with those without metopic ridge ($P = 0.009$), and trended toward significance compared with those with a ridge but

without trigonocephaly (P=0.072). The ratio of anterior-to-posterior cranial volume was significantly reduced in those with mild-to-moderate trigonocephaly compared with those without metopic ridge (P=0.036).In conclusion, patients with milder anterior cranial deformities demonstrated an association between a metopic ridge with mild-to-moderate trigonocephaly and reduced anterior cranial volume [7].

An objective method to evaluate the cosmetic results of both endoscopic and open surgeries is necessary, as some under-corrections have been reported with minimally invasive surgery [8].

Case series

Trigonocephaly case series.

Case reports

A patient with microcephaly and trigonocephaly, moderate intellectual disability, speech and language delay, and poor social interaction in addition to minor but atypical dysmorphic features. This report provides further insight into the pathogenicity of the Xp22.31 duplication by extending knowledge of its clinical features. This case, in association with those reported in the literature, indicates that the Xp22.31 duplication may contribute to cause pathological phenotypes with minor facial dysmorphisms, microcephaly, and intellectual disability as main features [9].

The diagnosis of a combination of both Sturge-Weber syndrome and trigonocephaly has been reported. Ristow et al., presents a patient with the unusual findings of a Sturge-Weber syndrome and simultaneous trigonocephaly induced by premature metopic synostosis. Thus, the rare combination of a port-wine stain involving the first division of the trigeminal nerve with the diagnosis of a craniosynostosis justifies the indication of a prophylactic magnetic resonance imaging acquisition before craniofacial surgeries, in order to prevent seizures and stroke-like episodes triggered by the surgical intervention [10].

References

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