Residência RP Pediátrica
Publicação Oficial da Sociedade Brasileira de Pediatria

Submitted on: 08/29/2022
Approved on: 03/18/2023

CASE REPORT

The importance of anthropometry and cranial semiology for the early diagnosis of Trigonocephaly: Case report

Carolina Fernandes Correa Leite[1], Júlia Buzzato Rainer[1], Cecília Pereira Silva[1,2], Camilla Kalil Laviola[2]

Keywords:
Craniosynostoses,
Cephalometry,
Early Diagnosis.

Abstract

Trigonocephaly is a type of craniosynostosis in which there is premature closure of the metopic suture, in order to determine an abnormal growth of the skull. In a minority of cases, trigonocephaly can cause increased intracranial pressure and the onset of neurological disorders. When present, these complications can be prevented or mitigated if the diagnosis and treatment occur between 6 and 12 months of life. The objective of the study is to report a case of craniosynostosis in an infant and reaffirm the importance of anthropometry and cranial semiology. On physical examination, a 4-month-old infant had a head circumference: 41 cm, metopic suture mounted, biparietal diameter: 23 cm, fronto-occipital diameter: 28 cm. Cranial computed tomography with 3D reconstruction confirmed the diagnosis of trigonocephaly, and he was referred to pediatric surgery. The study was submitted to the Human Research Ethics Committee approved under CAAE number 53607821.7.0000.5237. The diagnosis of trigonocephaly is made through the analysis of anthropometric measurements of the skull, represented by the head circumference, anteroposterior distance and binaural distance, and confirmed through computed tomography with 3D reconstruction. In pediatric practice, it is common to only measure the head circumference, which is often normal, with changes only in the anteroposterior and binaural distances. Consequently, there is a delay in the diagnosis of craniosynostosis. Thus, the importance of routinely measuring the three anthropometric measurements of the skull is emphasized for the early diagnosis of craniosynostosis and for the prevention and reduction of its complications, even though they are rare.

[1] UniFOA, Curso de Medicina - Volta Redonda - RJ - Brazil.
[2] Hospital Santa Cecília - Volta Redonda - RJ - Brazil.

Correspondence to:
Carolina Fernandes Correa Leite.
UniFOA. Av. Dauro Peixoto Aragão, 1325, Três Poços, Volta Redonda - RJ, Brazil. CEP: 27240-560. E-mail: carolinasfcorrea@gmail.com

Resid Pediatr. 2024;14(2). DOI: 10.25060/residpediatr-2024.v14n2-944

This work is licensed under a Creative Commons Attribution 4.0 International License.

INTRODUCTION

Craniosynostosis is a set of congenital cranial anomalies defined by the premature closure of one or more cranial sutures[1,2]. This alteration is seen in 0.6 people per 1,000 live births[3], with a prevalence increase in recent times[1,4,5].

According to its etiology, this pathology is classified as primary or non-syndromic, and secondary or syndromic. Primary craniosynostosis, seen in 85% of patients, is due to a primary defect in cranial ossification; while the secondary craniosynostosis is the result of genetic diseases that lead to hematological or metabolic dysfunction, such as rickets and congenital hypothyroidism. Furthermore, this condition is also defined as simple, when only one cranial suture is affected, or complex, when two or more sutures are prematurely closed[6,7].

Early closure of the metopic suture, called trigonocephaly, is the second most common cause of craniosynostosis, being found in 10 to 31% of those with this condition[1,3,4] and in 1 in every 5,000 to 15,000 live births[3]. It has not yet been established a specific etiology for trigonocephaly, but it is believed that its development is related to genetic abnormalities, changes in cell signaling pathways, environmental factors, metabolic disorders and restrictions on intrauterine fetal movement[3,8]. The genetic abnormalities most frequently associated with trigonocephaly include the deletion of chromosome 11q24, 7p, 3q, 13q, 12pter or 22pter, the deletion or trisomy of chromosome 15q25 and mutations in the ERF and SMAD63 genes. Restriction of intrauterine fetal movement is a condition in which fetal movements are significantly reduced, so that the fetus cannot move freely, which can cause craniofacial abnormalities[3].

Trigonocephaly is clinically characterized by the presence of the following craniofacial anomalies: narrow forehead, hypotelorism, arching of the eyebrows and lateral orbital retrusion with associated bilateral frontotemporal narrowing and compensatory bilateral occipitoparietal widening. In cases where these craniofacial anomalies are absent and there are no other associated clinical conditions, premature closure of this suture is called benign metopic suture tying[8].

In a minority of cases, trigonocephaly is associated with a number of serious complications, including the presence of sensory, respiratory and neurological dysfunctions and the development of intracranial hypertension (ICH)[5-7]. In these situations, there is a greater risk of cognitive impairment, due to restricted brain growth and increased cranial pressure[1]. Due to these complications and the possible association of trigonocephaly with certain genetic anomalies, early diagnosis and treatment is extremely important, preferably within the first 6 to 12 months of life[6].

The diagnosis of craniosynostosis is clinical, based on morphology, measurement and cranial palpation[1,2], and confirmed through computed tomography with 3D reconstruction, which evaluates the permeability of all sutures and possible structural abnormalities[1]. Ultrasonography can be used as an early screening method in suspected cases, assessing the permeability of sutures without the use of ionizing radiation, reserving computed tomography for pre-surgical mapping[9]. Thus, measuring anthropometric measurements of the skull, such as head circumference, anteroposterior distance (APD) and binaural distance (BAD) and determining the head index (APD/BAD) allow early identification of this condition[2].

In pediatric practice, it is common to simply measure the head circumference, with the understanding that, due to the presence of craniosynostosis, this measurement will be reduced, which would enable the identification of these conditions. However, in most cases, the head circumference is within normal limits for the age, with changes only in other anthropometric measurements of the skull. Consequently, there is a delay in diagnosis, which is very harmful to the child's healthy development. Therefore, although little used, the routine measurement of anteroposterior and binaural distances and also the analysis of the cephalic index are extremely important for diagnosing this pathology[2].

CASE REPORT

This is a 4-month-old male Infant, seen in a pediatric medical appointment for childcare. During the medical interview, there were no signs of alarm. Upon clinical examination, there were no changes in vital signs or other systems. Ectoscopy: keeled forehead (figure 1). On cranial examination, head circumference: 41 cm, anterior fontanelle: 0.5 cm, metopic suture, anteroposterior diameter: 28 cm, binaural diameter: 23 cm, cephalic index (DAP/DAB): 1.2.



Figure 1. Frontal view of the face, showing the keel aspect on the forehead.

Neuropsychomotor development was appropriate for age. A computed tomography of the skull with 3D reconstruction confirmed the diagnosis of trigonocephaly (figure 2), and he was referred to pediatric neurosurgery and operated after two months.

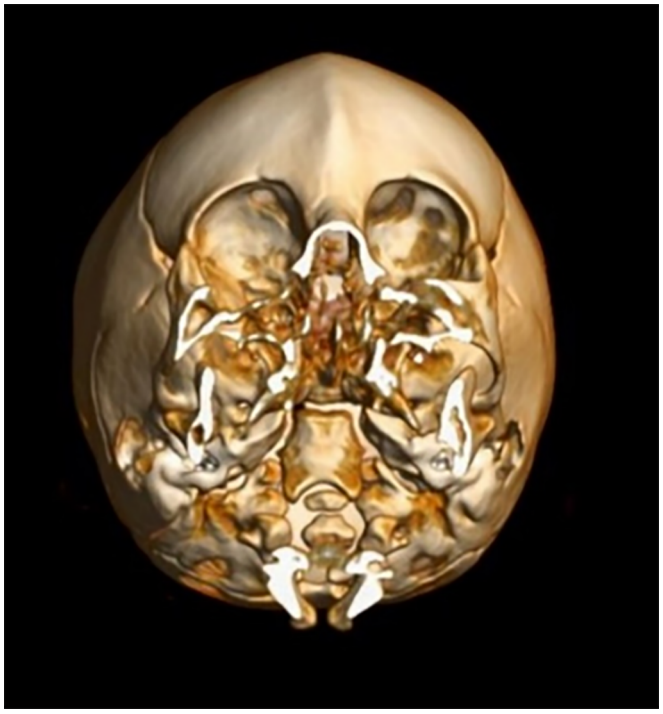


Figure 2. skull CT-Scan with three-dimensional reconstruction, demonstrating early closure of the metopic suture, giving a triangular, "keel" appearance to the frontal bone.

DISCUSSION

The diagnosis of craniosynostosis is clinical and, ideally, should be carried out in the first months of life. The first step towards its early identification is to carry out a detailed clinical interview, seeking to identify the existence of conditions predisposing to this pathology[1]. These conditions are multifactorial and include environmental factors, such as restricted intrauterine growth, oligohydramnia, abnormal fetal position, pregnancy complications, prenatal exposure to teratogens, maternal smoking and use of antiepileptic drugs, and genetic factors, which encompass single genetic mutations, abnormalities chromosomal and polygenic environments[1,10].

In addition to the anamnesis, it is extremely important to carry out a complete physical examination, seeking to identify characteristics indicative of early closure of the cranial sutures, such as unusual skull shapes, and the existence of characteristics suggestive of genetic syndromes. As described in some studies, people with trigonocephaly have an enlarged occipital portion, a narrow and pointed forehead and a triangular-shaped head. Furthermore, they generally present hypotelorism and a low nasal dorsum with epicanthic folds[1,2,6]. However, these changes may not be present or may be found in isolation, as was seen in the patient in this report, who only had a narrow and pointed forehead, indicative of the metopic suture being overridden.

Due to the possible absence of striking physical characteristics indicative of craniosynostosis, it is extremely important to determine the anthropometric measurements of the skull and palpation and measurement of the fontanelles for clinical diagnosis and for requesting imaging tests[1]. A study demonstrated that a considerable number of pediatricians do not routinely measure the anteroposterior and binaural distances and the cephalic index during routine consultations, which constitutes a major obstacle to the early identification of craniosynostosis[2].

Most people with craniosynostosis do not have a reduced head circumference but have changes in the anteroposterior and binaural distances and in the head index[2]. The patient in this case report had a head circumference within the normal range for his age, having only a change in the anteroposterior diameter of the skull and, consequently, in the head index. This finding corroborates the importance of routinely establishing these anthropometric measurements in routine pediatric consultations for the early diagnosis of craniosynostosis.

Although the minority of patients with trigonocephaly do have some neurological abnormality, they are at greater risk of developing cognitive and behavioral changes[1,6], psychomotor delays, learning difficulties, visual defects and maladaptive behaviors[10]. The choice between surgical or conservative management depends on the neuropsychomotor development assessment, neurological examination and aesthetics desired by the family. Early surgical treatment is usually recommended to provide more adequate brain expansion, as well as better craniofacial aesthetics. However, conservative management can be adopted if the deformity is mild or if the suture placement is benign[3]. In this case, the infant's neuropsychomotor development was appropriate for his age, which reiterates the importance of anthropometric measurements of the skull for identifying craniosynostosis.

CONCLUSION

Since the diagnosis of craniosynostosis is essentially clinical, the importance of measuring the head circumference, anteroposterior distance and binaural distance, and determining the head index in routine pediatric consultations since birth is highlighted. Furthermore, as a considerable number of pediatricians do not routinely measure anteroposterior and binaural distances, it is important to disclose to the scientific community the impact that anthropometric measurements and skull semiology have on the early detection of these conditions and the prevention of complications, even if they are minimal.

REFERENCES

1. Kajdic N, Spazzapan P, Velnar T. Craniosynostosis-Recognition, clinical characteristics and treatment. Bosn J Basic Med Sci. 2018; 18(2):110-116. doi: 10.17305/bjbms.2017.2083.
2. Val Filho JAC. As medias cranianas no diagnóstico das craniossinostoses [Internet]. Faculdade de Medicina da UFMG; 2013[citado em 2022 nov 26]. Disponível em: https://repositorio.ufmg.br/bitstream/1843/BUBD-A2DFW5/1/tese__total_revisadal.pdf
3. Trigonocephaly Ricardo J. Fernández-de Thomas 1, Orlando De Jesus 1 In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan[citado em 2022 nov 26]. Disponível em: https://pubmed.ncbi.nlm.nih.gov/33351432/.
4. Boulet SL, Rasmussen AS, Honein MA. A Population-Based Study of Craniosynostosis in Metropolitan Atlanta, 1989-2003. Am J Med Genet Part A. 2007 Nov 19;146(A):984-991. doi: 0.1002/ajmg.a.32208
5. Johnson D, Wilkie AOM. Craniosynostosis. Eur J Hum Genet. 2011 Jan 19;19: 369-376. doi: 10.1038/ejhg.2010.235.
6. Garza RM, Khosla RK. Nonsyndromic craniosynostosis. Semin Plast Surg. 2012; 26:53-63. doi: 10.1055/s-0032-1320063.
7. Sharma RK. Craniosynostosis. Indian J Plast Surg. 2013 Jan-Apr; 46(1): 18-27. doi: 10.4103/0970-0358.113702.
8. Jaskolka MS. Current Controversies in Metopic Suture Craniosynostosis. Oral Maxillofac Surg Clin North Am. 2017 Nov;29(4):447-463. doi: 10.1016/j.coms.2017.07.003.
9. Soboleski D, Mussari B, McCloskey D, Sauerbrei E, Espinosa F, Fletcher A. High-resolution sonography of the abnormal cranial suture. Pediatr Radiol. 1998 Feb;28(2):79-82. doi: 10.2214/ajr.168.3.9057541.
10. Greenwood J, Flodman P, Osann K, Boyadjiev SA, Kimonis V. Familial incidence and associated symptoms in a population of individuals with nonsundromic craniosynostosis. Genet Med. 2014 Apr;16(4): 302-310. doi: 10.1038/gim.2013.134.