Craniostenosis treated by endoscopic assistance: A case report

Craneostenosis tratada con asistencia endoscópica

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Resumen

Recién nacido a término, de sexo femenino, con displasia tanatofírica tipo II, acondroplasia, hipertelorismo, implantación baja de la oreja, múltiples malformaciones osteoarticulares, macrocefalia, craneoestenosis y otras anomalías. Tras la evaluación por el equipo de neurocirugía, se constató el rostro sindrómico y signos de hipertensión intracraneal, debida a hidrocefalia, asociada a disyunción bilateral de la sutura temporal y craneoestenosis de la sutura sagital. Se realizó cirugía de compensación de la HIC a los 10 días del nacimiento. La paciente evolucionó con fontanela anterior normotensa y mejoría de la disyunción de la sutura. A los 45 días de la primera cirugía, la paciente volvió a presentar signos progresivos de HIC. Anticipando cirugía para corrección de la craneoestenosis sagital. Realizada con ayuda de neuroendoscopia para resección ósea de la sutura fusionada. La craniostenosis es una enfermedad rara caracterizada por la fusión prematura de las suturas craneales. Su resultado son anomalías del desarrollo cerebral, HIC y disminución de la función cognitiva. Se han identificado patrones de herencia genética y mutaciones como causantes de esta patología. El primer y más común signo de craneoestenosis es la forma anormal del cráneo. La tomografía computarizada 3D del cráneo es el método estándar para el diagnóstico. La cirugía abierta es la más común en la craneoestenosis sindrómica. Sin embargo, deben considerarse enfoques más conservadores en espera de la cirugía. La intervención quirúrgica endoscópica es la más adecuada hasta los 6 meses de edad. El paciente del caso descrito tiene una craneostenosis sindrómica, y el tratamiento endoscópico no es frecuente. En los casos quirúrgicos en los que la enfermedad se identificó precozmente, el pronóstico tiende a ser positivo.

Palabras clave: Sutura craneal, craniostenosis sindrómica, craneostenosis sagital, tratamiento endoscópico, neurocirugía, técnicas.

Abstract

Full-term newborn, female, with tanatophyric dysplasia type II, achondroplasia, hypertelorism, low ear implantation, multiple osteoarticular malformations, macrocephaly, craniostenosis and other anomalies. Upon evaluation by the neurosurgery team, the syndromic face and signs of Intracranial Hypertension were found, due to hydrocephalus, associated with bilateral temporal suture disjunction and craniostenosis of the sagittal suture. Surgery was performed for ICH compensation 10 days after birth. The patient evolved with normotensive anterior fontanel and improvement of the suture dysjunction. On the 45th day after the first surgery, the patient again presented progressive ICH signs. Anticipating surgery for correction of the sagittal craniostenosis. Performed with the aid of neuroendoscopy for bone resection of the fused suture. Craniostenosis is a rare

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condition characterized by premature fusion of cranial sutures. Its result is abnormalities of brain development, ICH, and decreased cognitive function. Patterns of genetic inheritance and mutations are identified as causing this pathology. The first and most common sign of craniostenosis is the abnormal shape of the skull. 3D Computed Tomography of the skull is the standard method for diagnosis. Open surgery is most common in syndromic craniostenosis. However, more conservative approaches should be considered pending surgery. Endoscopic surgical intervention is most appropriate until 6 months of age. The patient in the reported case has syndromic craniostenosis, and endoscopic treatment is not common. In surgical cases in which the disease was identified early, the prognosis tends to be positive.

Key words: Cranial suture, syndromic craniostenosis, sagittal craniostenosis, endoscopic management.

Introduction

Craniostenosis or craniosynostosis is a rare medical condition featured by the premature fusion of one or more cranial sutures, its incindence is 1 in 2,000-2,500 live births per year, less than 30% of cases are syndormic. Tanatophoric dysplasia incindence is 1 in 35,000-50,000 live births per year. This premature closure of a major suture can result in cranial deformity and overall cranial growth restriction, resulting in increased intracranial pressure[1,2].

Craniosynostosis can be classified according to the sutures involved, the number of structures fused, the anatomical changes of the skull and face, and as syndromic or non-syndromic. When craniosynostosis is suspected, its necessary to perform physical and imaging examination to assess the shape and the movement of the calvarial bones, the 3D Computed Tomography (CT) is considered the standard method to diagnose craniosynostosis[3,4].

The treatment is the surgical correction as early as possible, to avoid compensatory deformations and prevent intracranial hypertension (IH), and visual and neurological impairments. Better prognosis is observed in patients undergoing surgery between 3 and 9 months of age, due to the bigger malleability of the calvarial bones[4,5].

In this study, the authors present the case of a 2-month-old patient with syndromic sagittal craniosynostosis subjected to minimally invasive endoscopic treatment.

Case report

Newborn (NB), female, 4 days old, with thanatophoric dysplasia type II, diagnosed with achondroplasia, hypertelorism, low ear implantation, multiple osteoarticular malformations, macrocephaly, craniostenosis, platyspondyly, evidenced by shortening of the neck (winged) and hypoplasia of the rib cage and limbs. Born by c-section pelvic delivery on May 16, 2021, alive with other congenital malformations, strong crying, showing herself active and reactive, APGAR 07/09, weighing 2,685 kg and height of 37 cm. Subsequently, the newborn was referred to the neonatal Intensive Care Unit (ICU) in a transport incubator with inhaled O_2.

Upon evaluation by the neurosurgery team, the patient's syndromic face and signs of ICH due to hydrocephalus were noted, associated with bilateral temporal sutures disjunction and craniostenosis of the sagittal suture. Given the picture presented, it was concluded that the therapeutic route would be divided into 2 stages. In the first one, surgery for ICH compensation was performed through a ventriculo-peritoneal shunt (VPS) with a neonatal medium pressure valve, which was performed 10 days after birth. While in the second time, surgery for endoscopy-assisted correction of craniostenosis was performed.

On the 1st postoperative day of the VPS placement, the patient evolved with normotensive anterior fontanel and improvement of suture disjunction. Concomitantly, antibiotic therapy (ATB) was started, in which oxacillin together with cefepime was administered for 10 days, as a local protocol for shunt infection prophylaxis.

After 13 days, the patient remained in serious condition. Despite this, she evolved with an improved picture, hemodynamically stable, and was responsive to management. On physical examination, it could be noted that she was ruddy, euthermic (36.6°C) and hydrated, however, still with signs of cyanosis and residual jaundice. The pupils were isochoric and photo reagent. Throughout, an occluded operative wound was noted, with a clean dressing.

On post-operative day 45 of VPS, the patient again showed progressive signs of ICH, such as diastasis of the temporal bone, due to reduced brain compliance, despite there was no sign of shunt malfunction. Due to the progression of the ICH, surgery to correct the sagittal craniostenosis had to be anticipated, and it was performed 57 days after the PVD surgery and 67 days after his birth.

To perform the surgery, neuroendoscopy with a child's working channel was used for resection of 3 cm bilaterally, in the midline, of the parietal bone, from the previous fontanelle to the posterior fontanelle (Figure 1). During the procedure, after incision and separation of planes, the neuroendoscope was coupled to perform the cautious dural and superior sagittal sinus dissections along with the bone resection (Figures 2 and 3).

Discussion

Craniostenosis is a disorder of early closure of the cranial sutures. This phenomenon makes it impossible to expand the cranial box at the suture junction points. Thus, the brain does not grow perpendicularly, and through a compensatory mechanism, it develops into the areas of least resistance. The result is abnormalities of brain development, increased intracranial pressure, respiratory dysfunction, and decreased cognitive function and intelligence quotients[4,5].

Figure 1. Anterior and posterior incision for the approach of the endoscope. Where it was accomplished the 3 cm resection bilaterally, in the midline, of the parietal bone, from the anterior fontanelle to the posterior fontanelle.

Figure 2. Bone resection for the treatment of craniosynostosis.

Figure 3. Resected bone from cranioestenosis surgery Seven days after the second surgery, the patient had an improvement in symptoms and signs of ICH, which was enough to be discharged from the neurosurgery team.

This pathology has some classifications influenced by its contained mechanism. First, craniostenosis that occurs due to ossification defect of the sutures are called primary; while those that result from systemic disorders are known as secondary, which can also occur in NBs with microcephaly[4]. There are also criteria based on the anatomy of the skull. As an example, according to Nagaraja, Cohen brings us a classification system based on the ossified sutures, while Marchac and Renier use the shape of the brain as a reference in the classifications. This one has as terms the scaphocephaly, trigonocephaly, plagiocephaly, oxycephaly, brachycephaly and turricephaly[3]. Another classification is considered when the present pathology is associated to syndromes, such as Crouzon and Apert, or when it is not associated, being called syndromic and non-syndromic, respectively[6,7].

Genetic mutations are present in 20% of cases. Recently, patterns of genetic inheritance and mutations are identified as causing such pathology. This inheritance is autosomal dominant in about 50% of cases, which suggests influence on the number of cases in the family, the number of sutures involved, and the syndromic conditions. This raises the question about the understanding and classification of craniostenosis as to its morphology, since clinical standardizations are no longer adequate[8,9].

Craniostenosis is present in 24% of cases of thanatophoric dwarfism type 1 and 93% of cases of thanatophoric dwarfism type 2. This malformation is caused by mutations in

the FGFR3 gene[2]. The patient in the case was not performed the genetic test to confirm the thanatophoric dwarfism type.

The relationship between ICH and this disorder is still not well established due to its multifactoriality. However, this phenomenon is common in complex craniostenosis, so that increased intracranial pressure is present in 47%-67% of these newborns. Its detection is not simple, since non-invasive methods such as papilledema, increased optic nerve diameter on ultrasound, and dilation of the previous fontanelle are not very specific[10].

Craniosynostosis has a characteristic clinical presentation in newborns. The first and most common sign is the abnormal shape of the skull and the cephalic index in the first year of life. So that the alteration in a single suture is more common than in multiple ones. It is necessary that the shape of the head be well documented by pictures and imaging examinations, such as cranial computed tomography. During observation, the fontanelles and cranial sutures should be palpated. In addition, examination of the face, ear height and body for syndromic features are findings for a possible differential diagnosis of a syndromic craniosynostosis[11,12].

Your clinical history is important in the diagnosis of the anomaly. Data of the baby's morphology in the prenatal period, the history of pregnancy and delivery, and the use of medications are factors to be surveyed by the team. Because flattened and growing deformities that are present from birth and are not affected by positioning are suggestive of craniostenosis[12].

Imaging examinations are necessary to close the pathological diagnosis, search for possible anomalies - such as hydrocephalus and congenital malformation - plan surgery and define the prognosis[13]. Different methods can be performed, such as plain radiography, skull CT with 3D reconstruction and magnetic resonance imaging (MRI). The first method shows signs of primary and secondary craniostenosis. The primary signs are periosteal sclerosis, absence of sutures and located fractures; while the secondary signs are the copper-beaten skull and the presence of fingerprints, which are suggestive of ICH[9]. Skull CT with 3D reconstruction is a richer method for the study of craniosynostosis, because it allows the evaluation of the skull structures as well as other abnormalities; moreover, it is advantageous for the study and preoperative planning and for the postoperative evaluation of the patients. This implements the accuracy of this diagnosis[3,11]. As for MRI, it is essential for cases of syndromic craniostenosis, as well as for the identification of possible intracranial herniations[3].

Similar to the various clinical manifestations of craniostenosis, the treatment possibilities are also varied[4]. Although treatment with open surgery is the most common, more conservative approaches should be considered initially, while awaiting surgery, such as the use of a remodeling helmet in very young patients associated or not with endoscopic intervention[4,13]. The main objective is to ensure the development of the encephalon, control of intracranial pressure and an aesthetically acceptable appearance to the patient[13].

Endoscopic surgical intervention is most appropriate for patients up to 6 months of age, ideally around 3 months of age due to the flexibility of the skull bones, the rapid growth period of the brain and decreased production of fetal hemoglobin. [14] Its advantages are less blood loss, shorter duration of surgery, and faster postoperative recovery. Despite these advantages, open surgery is more common after 6 months of age and in cases of syndromic craniosynostosis. [15] The patient of the reported case presents with sagittal craniostenosis, but in its syndromic form, and endoscopic treatment is not common in this case and before 3 months of age, as reported.

There are several possibilities of complications during the treatment of craniosynostosis. According to Ghizoni et al. 2016, those patients not treated surgically can develop: psychosocial problems - such as social isolation - physical and mental disorders and increased intracranial pressure, 60% in the case of complex craniosynostosis and 20% in the case of simple ones, - although Governale 2015 affirm that most patients do not suffer from ICH before 6 months of age -[1,16]. In our case, due to the progression of ICH, the patient had to anticipate the surgical process for the second month of life.

Furthermore, according to Kajdic et al. 2018, patients treated surgically may present postoperative hyperthermia, complications such as blood loss, rupture of the dura mater, cerebrospinal fluid leakage, subgaleal and subcutaneous bruise, and infections. It is necessary, even after surgery, a follow-up of the patient to observe possible complications[4]. In the postoperative period of our patient, there were no intercurrence and/or complications, so that on the 7th postoperative day the NB evolved to discharge from the specialty.

Surgical cases in which the disease was identified early, the prognosis tends to be positive. Endoscopic surgery, whose patient cited in the text was submitted, allows an intervention with low risk of mortality, according to the case study, in which 139 patients were analyzed, by Jimenez et al. 2004, 87% of the cases analyzed obtained excellent cephalic index results, 8.7% good results and 4.3% bad results, with no cases of death, being shown by Yan et al. 2018, when analyzing 3 studies, that when compared to open surgeries, the endoscopic procedure presents a small percentage of reoperation[17,18].

Conclusion

In short, craniostenosis can develop skull deformities and ICH if left untreated. For its diagnosis, the clinical history, signs of shunt malfunction, ICH signs and imaging exams are used, being the 3D cranial computed tomography the most specific one. About the treatment of sagittal craniostenosis, the decompensated ICH demands surgical intervention, only; the prognosis of surgically treated patients is excellent, due to the appropriate control of ICH and the ability of the brain mass to grow. Endoscope-assisted intervention is less invasive, and therefore, the recovery is faster. So, we report a case of endoscopy-assisted management in a patient with syndromic sagittal craniostenosis, something not common within the daily routine of pediatric neurosurgery, enriching the literature towards the understanding of the pathology.

Declarations

Ethics approval.

Not applicable.

Consent to participate.
Not applicable.
Consent for publication.
Not applicable.
Availability of data and material.
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All authors contributed to the study conception and design. material preparation, data collection and analysis were performed by christian diniz ferreira, matheus felipe henriques brandão and lucas gabriel henriques brandão. the first draft of the manuscript was written by matheus felipe henriques brandão, joão vitor lopes de medeiros gonçalves and josé vitor martins veras. the paper was reviewed by josé lopes sousa filho and christian diniz ferreira. all authors read and approved the final manuscript.

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References

1. Governale LS. Craniosynostosis. Pediatr Neurol. 2015 Nov;53(5):394-401. doi: 10.1016/j.pediatrneurol.2015.07.006. Epub 2015 Jul 22. PMID: 26371995.
2. Wilcox WR, Tavormina PL, Krakow D, et al. Molecular, radiologic and histopathologic correlations in thanatophoric dysplasia. Am J Med Genet 1998;78:274-281.
3. Nagaraja S, Anslow P, Winter B. Craniosynostosis. Clin Radiol. 2013 Mar;68(3):284-92. doi: 10.1016/j.crad.2012.07.005. Epub 2012 Aug 28. PMID: 22939693.
4. Kajdic N, Spazzapan P, Velnar T. Craniosynostosis - Recognition, clinical characteristics, and treatment. Bosn J Basic Med Sci. 2018 May 20;18(2):110-116. doi:10.17305/bjbms.2017.2083. PMID: 28623672; PMCID: PMC5988529.
5. Ursitti F, Fadda T, Papetti L, Pagnoni M, Nicita F, Iannetti G, Spalice A. Evaluation and management of nonsyndromic craniosynostosis. Acta Paediatr. 2011 Sep;100(9):1185-94. doi: 10.1111/j.1651-2227.2011.02299.x. Epub 2011 May 5. PMID: 21457300.
6. Marchac D, Renier D. Classification of the different types of craniosynostosis. In: Marchac D, Renier D, editors. Craniofacial surgery for Craniosynostosis. Boston: Little Brown; 1982. p. 1e8.
7. Wang JC, Nagy L, Jc Demke. Syndromic Craniosynostosis. Facial Plast Surg Clin North Am. 2016 Nov;24(4):531-543. doi: 10.1016/j.fsc.2016.06.008. PMID: 27712819.
8. Brah TK, Thind R, Abel DE. Craniosynostosis: Clinical Presentation, Genetics, and Prenatal Diagnosis. Obstet Gynecol Surv. 2020 Oct;75(10):636-644. doi: 10.1097/OGX.0000000000000830. PMID: 33111964.
9. Sawh-Martínez R, Steinbacher DM. Syndromic Craniosynostosis. Clin Plast Surg. 2019 Apr;46(2):141-155. doi: 10.1016/j.cps.2018.11.009. PMID: 30851747.
10. Tamburrini G, Caldarelli M, Massimi L, Gasparini G, Pelo S, Di Rocco C. Complex craniosynostosis: a review of the prominent clinical features and the related management strategies. Childs Nerv Syst. 2012 Sep;28(9):1511-23. Doi: 10.1007/s00381-012-1819-4. Epub 2012 Aug 8. PMID: 22872268.
11. Nagy L, Jc Demke. Craniofacial anomalies. Facial Plast Surg Clin North Am. 2014 Nov;22(4):523-48. doi: 10.1016/j.fsc.2014.08.002. Epub 2014 Nov 8. PMID: 25444726.
12. Massimi L, Bianchi F, Frassanito P, Calandrelli R, Tamburrini G, Caldarelli M. Imaging in craniosynostosis: when and what? Childs Nerv Syst. 2019 Nov;35(11):2055-2069. Doi: 10.1007/s00381-019-04278-x. Epub 2019 Jul 9. PMID: 31289853.
13. Chong S, Wang KC, Phi JH, Lee JY, Kim SK. Minimally Invasive Suturectomy and Postoperative Helmet Therapy: Advantages and Limitations. J Korean Neurosurg Soc. 2016 May;59(3):227-32. doi: 10.3340/jkns.2016.59.3.227. Epub 2016 May 10. PMID: 27226853; PMCID: PMC4877544.
14. Burokas L. Craniosynostosis: caring for infants and their families. Crit Nurse Care. 2013 Aug;33(4):39-50; quiz 51. doi: 10.4037/ccn2013678. PMID: 23908168.
15. Honeycutt, J. (2014). Endoscopic-Assisted Craniosynostosis Surgery. Seminars in Plastic Surgery, 28(03), 144-149. doi:10.1055/s-0034-1384810.
16. Ghizoni E, Denadai R, Raposo-Amaral CA, Joaquim AF, Tedeschi H, Raposo-Amaral CE. Diagnosis of infant synostotic and nonsynostotic cranial deformities: a review for pediatricians. Rev Paul Pediatr. 2016 Dec;34(4):495-502. doi: 10.1016/j.rpped.2016.01.004. Epub 2016 May 12. PMID: 27256993; PMCID: PMC5176072.
17. Jiménez DF, Barone CM, McGee ME, Cartwright CC, Baker CL. Endoscopy-assisted wide-vertex craniectomy, barrel stave osteotomies, and postoperative helmet molding therapy in the management of sagittal suture craniosynostosis. J Neurosurg. 2004 May;100(5 Suppl Pediatrics):407-17. doi: 10.3171/ped.2004.100.5.0407. PMID: 15287447.
18. Yan H, Abel TJ, Alotaibi NM, Anderson M, Niazi TN, Weil AG, Fallah A, Phillips JH, Forrest CR, Kulkarni AV, Drake JM, Ibrahim GM. A systematic review and meta-analysis of endoscopic versus open treatment of craniosynostosis. Part 1: The sagittal suture. J Neurosurg Pediatr. 2018 Oct;22(4):352-360. doi: 10.3171/2018.4. PEDS17729. Epub 2018 Jul 6. PMID: 29979135.