Clinical case: radiological

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ABSTRACT
Craniosynostosis is a congenital defect of unknown cause in which one or more sutures of the baby's head close partially or totally earlier than normal, it has an incidence of 1 in 1,000 to 3,000 live births, with higher prevalence in males. The evolution of this disease is different in each case and can be mild or severe. They present symptoms such as: irritability or inconsolable crying and very little activity. As your skull grows, it can become more and more deformed and give different neurological symptoms. It may be due to certain genetic syndromes such as Apert, Pfeiffer or Croizon syndrome affecting the development of the baby's skull. We present a case of a newborn with this rare pathology.

Keywords: craniosynostosis, infant.

1 CLINIC HISTORY
AGA term newborn, female, birth weight 3190 grams, height 47 cm, skull circumference 32.5 cm. Without any complications resulting from the pregnancy. Born by cesarean section due to acute fetal distress. At birth, the physical examination was overall normal, with an APGAR score of 9-9-10-10. Small cranial size with brachycephalic appearance is noted, with no palpable fontanelles, thus prompting a request for anteroposterior and latero-lateral radiography of the skull (Figure 1 and 2).
2 RADIOLOGICAL FINDINGS

Figures 1 and 2 shows sclerosis of the coronal suture with less development of the anterior cranial fossa.

3 DIAGNOSIS

Craniosynostosis.

4 DISCUSSION

Craniosynostosis consists of the premature closure of the cranial sutures. The incidence of primary craniosynostosis is described as 0.4 per thousand to 1 per thousand newborns. The cause is unknown in most children. In some cases, between 10% to 20% is associated with genetic syndromes. The premature closure of the sagittal suture produces an elongated and narrow skull known as Scaphocephalea, this being the most frequent of the Craniosynostoses. It is accompanied by a prominent occiput, a broad forehead, and a small or absent anterior fontanel. This disorder is sporadic and more common in males. It is not associated with intracranial hypertension or hydrocephalus, and the children are neurologically normal. Brachycephaly or Plagiocephaly, the second most frequent, is more frequent in females and is clinically characterized by unilateral flattening of the forehead, the elevation of the orbit, the elevation of the eyebrow and ipsilaterally prominent ear, due to premature closure of the coronal and sphenofrontal sutures. Surgical management achieves aesthetically good results. Occipital plagiocephaly is generally a postural deformity acquired during infancy, being more frequent in immobile or disabled children. Closure of the lambdoid suture causes unilateral occipital flattening and bulging of the ipsilateral frontal bone. Trigonocephaly, a rare form of Craniosynostosis, is caused by premature closure of the metopic suture. On examination, children present with a keel-shaped forehead, hypotelorism, and risk of presenting abnormalities associated with brain development. Turricephaly presents with a conical-shaped head caused by premature closure of the coronal suture and often the sphenofrontal and frontoethmoidal sutures. Neurological complications, either as a deficit, hydrocephalus, and intracranial hypertension, occur more frequently when the premature closure of two or more sutures occurs, making surgical intervention essential. The genetic disorders most frequently associated with Craniosynostosis are Crouzon, Apert, Carpenter, Chotzen, and Pfeiffer Syndromes. Some teratogens have been associated with craniosynostosis, such as diphenylhydantoin, valproic acid, aminopterin, methotrexate, retinoic acids, and oxymetazoline. It can be suspected in the fetal period when there is an absence of the oval shape of the skull, and a trilobed skull or narrowing of the skull can be found. It is important in cases that require surgery, prior to it, to rule out alterations that may be associated with craniosynostosis, such as midface retrusion, septo-optic dysplasia, or...
ventriculomegaly\textsuperscript{3,4}. For this, neuroimaging studies, such as CT and MRI, play an important role, which usually helps to plan the intervention. Surgical treatment has a good prognosis, with relatively low morbidity and mortality, especially in non-syndromatic newborns. It is known that to obtain good cosmetic results, surgery must be performed at a relatively early stage, probably in the first weeks of life\textsuperscript{5,6}.

It has been proposed that surgical management consists of two main objectives: the release of the sutures to allow the correct growth of the brain and the reconstruction of all the dysmorphic skeletal components. The classic management of craniofacial surgery is performed by a team made up of a pediatric neurosurgeon, who is in charge of the formal craniotomy, and a craniofacial surgeon, in charge of simultaneous skeletal reconstruction. During the procedure, work is done to replace the dysmorphic bones, according to the desired anatomical shape based on the prototype of the planned patient\textsuperscript{7}.

Despite all this, the literature does not identify a surgical technique that is superior to others, and there are discrepancies in the treatment of non-syndromic craniosynostosis among craniofacial surgeons. Currently, the most popular treatments are the aforementioned open surgery and treatment using endoscopic techniques (Endoscopic strip craniectomy). In patients under three months, a minimally invasive procedure has obtained great results\textsuperscript{8}.

Endoscopic techniques focus on restoring the cranial anatomy to normal, relying on subsequent brain and skull growth for correction, unlike open surgeries, which seek cranial reconstruction at the time of surgery. The advantages of these endoscopic techniques are based on the low level of stress to which the baby is subjected, allowing better tolerance to the process, smaller surgical scars, and a decrease in blood loss and the appearance of hemorrhages. Given the accelerated growth of the brain between the first 3 to 6 months of age, the endoscopic technique needs to be performed early to ensure an optimal recovery period, losing prominence after 6 months of age, due to this, open surgery is still used, as surgical treatment, allowing the pediatrician to maintain recurrent observation of the deformity for a few months and thus ensure that it is indeed a craniosynostosis before referring the patient to a craniofacial expert\textsuperscript{9}.

As previously mentioned, there are craniosynostosis associated with genetic syndromes, which commonly involve multiple sutures. In most of these genetic syndromes, mutations have been identified in the genes that code for members of the fibroblast growth factor receptor (FGFR) family mutations that can also be found in patients with non-syndromic craniosynostosis\textsuperscript{10,11}.

For this reason, early detection of genetic mutations can be performed in newborn patients with suspected syndromic craniosynostosis, it being more common to identify mutations in patients with coronal or bicoronal non-syndromic craniosynostosis, rather than sagittal or metopic craniosynostosis.
The genes to be examined correspond, among others, to the aforementioned gene encoding members of the FGFR family together with TWIST (helix-loop-helix transcriptional regulator).
REFERENCES


APPENDIX

Figure 1: Anteroposterior radiograph of the skull showing early closure of the coronal suture.

Source: radiography of the skull.

Figure 2: Lateral skull radiograph showing less development of the posterior fossa.

Source: radiography of the skull.