Austin Gynecology Case Reports

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Case Report

Sequence Acrania Exencephaly Anencephaly Report of a Case in the San Vicente De Paul Hospital in Ibarra Ecuador

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Received: November 22, 2020; **Accepted:** December 10, 2020; **Published:** December 17, 2020

Abstract

The acrania - exencephaly - anencephaly sequence together with spina bifida are the two most common neural tube defects worldwide with a prevalence of 1.86 per 1,000 live births. The acrania is not really an alteration of the iso *In vivo* lated neural tube belongs to a sequence called acrania - exencephaly - anencephaly, since the lack of bones that make up the cranial vault will cause a protrusion of the cerebral parenchyma (exencephaly) and with the sudden movements of the fetus along with the chemical irritation of the amniotic fluid to the unprotected brain structure causes degeneration and destruction of it causing in the absence of brain mass (*anencephaly*).

Purpose: The present article intends to present a clinical case of young patient with fetus with acrania and anencephaly with a review of its etiology, differential diagnosis and management.

Methods/Search strategy: Review of the patient's medical history in the gynecology and obstetrics service of the San Vicente de Paul Hospital in Ibarra – Ecuador.

Results: Patient 21 years old, born and resident in Lita province of Imbabura. A detailed ultrasound was performed, finding a fetus in a neutral position with very active movements, placental outline of adequate implantation without signs of detachment, deepest vertical pocket of 2.5 cm amniotic fluid (normal 2 - 8 cm). Head with skull base with conserved structures, interorbital diameter of 8 mm, anechoic crystals, absence of cranial vault with mass that protrudes from the base of the skull compatible with acrania and exencephaly. Misoprostol 400 mcg intravaginal is placed for cervical ripening, the product is expelled after 10 hours under asepsis and antisepsis rules, obtaining fetus with acrania.

Implications for practice: Detect with ultrasound of the first trimester (11-13.6 weeks) and early morphological ultrasound the neural tube alterations that are incompatible with life to inform parents in a timely manner about their management and life expectancy.

Implications for research: Statistically quantify the cases of congenital malformations of our community to define prevalent cases and correlate risk factors for the development of them.

Keywords: Acrania; Exencephaly; Anencephaly; Neural tube defects; Acalvaria

Introduction

The acrania exencephaly anencephaly sequence together with spina bifida are the two most common neural tube defects worldwide with a prevalence of 1.86 per 1000 live births [1].Among the risk factors for these defects, the geographical location together with the socioeconomic level stands out, since in the same country there may be a greater risk of congenital nervous disorders in a given region, while the lack of education due to extreme poverty leads to inadequate family planning and low intake of foods rich in folates [5]. Access to food enriched with folates, which are public norm in some countries, are the protective factor to avoid malformations [6], in the same way the genetic origin has been demonstrated as a risk factor for acrania, as is the case of the polymorphism of the methylenetetrahydrofolate reductase found in 2% of Afro-descendants and more than 35% in Chinese and Mexicans [7, 8]. A clinical case of a patient with a product affected by acrania and anencephaly is described, this comprises a pathology that follows a basic sequence of acrania (partial or total absence of the bones that form the cranial vault), exencephaly (exposure of the brain parenchyma to fluid amniotic) and anencephaly (destruction of the brain by chemical irritation and trauma) [9].The diagnosis can be made as early as in the first trimester echo from 11 to 13.6 weeks showing absence of the skull [10], generally the acrania has a 100% mortality and is not compatible with life. Here we present a young patient with an acranic fetus.

Citation: Samaniego Haro VJ. Sequence Acrania Exencephaly Anencephaly Report of a Case in the San Vicente De Paul Hospital in Ibarra Ecuador. Austin Gynecol Case Rep. 2020; 5(1): 1024.

Table 1: Comparison between congenital cranial defects.

Structure	Exencephaly	Anencephaly	Acalvaria	Acrania
Brain	Mass of elongated, disorganized, and deformed brain tissue	Flattened remnant of disorganized forebrain tissue mixed with ependymals, choroid plexus, and meningothelial cells	Present but deformed	Present but deformed
Brain hemispheres	Present but deformed	Absent	Present	Present
Cerebellum	Present but deformed	Absent	Present	Present
Extracranial brain tissue covering	Vascular layer of epithelium	Cerebrovascular area	Dura mater and skin	Thin membrane
Calvarium	Absent over the orbits	Absent over the orbits	Absent over the orbits	Completely absent
Base of skull	Normal	Normal	Normal	Absent
Face	Normal	Deformed	Normal	Normal

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21-year-old patient, born and residing in Lita, Imbabura province, incomplete primary school, housework, free union, unknown blood type, no previous significant pathologies, no allergies, no surgical procedures, her first menstruation at 15 years of age , beginning of sexual life 18 years, a sexual partner, a previous pregnancy with cephalovaginal delivery without complications with a live child, upon admission to our hospital on April 9, 2018 with the date of the last menstruation on December 16, 2017, during pregnancy of 16.2 weeks, sent from another health facility due to fetal malformation. On physical examination, hemodynamic parameters were normal with a pregnant abdomen with a uterine fundus for 16 weeks, inguinogenital region without alterations. A detailed ultrasound is performed, finding the fetus in a neutral position with very active movements, a future placenta with adequate implantation without signs of detachment, a maximum vertical pocket of 2.5 cm amniotic fluid (normal 2-8 cm). Head with skull base with preserved structures, 8 mm interorbital diameter, anechoic lenses, absence of cranial vault with mass protruding from the skull base compatible with acrania and exencephaly. (Figure 1&2) Normal chest, 4-chamber heart, adequate outflow tracts, intact diaphragm, visible bladder. (Figure 3) A clinical case is presented to a group of doctors from the Gynecology and Obstetrics Service and it is decided due to high mortality of this pathology close to 100% the termination of pregnancy with the consent of the mother, intravaginal misoprostol 400 mcg is placed for cervical ripening , the product is expelled after 10 hours under the standards of asepsis and antisepsis, obtaining a fetus with acrania (figure 4) without a heartbeat without any other obvious major malformation (figures 5&6), instrumental curettage is performed with an approximate bleeding of 200 cc. Patient passes hospitalization without complications and is discharged in 48 hours after evaluation by the Department of Psychology and Genetics.

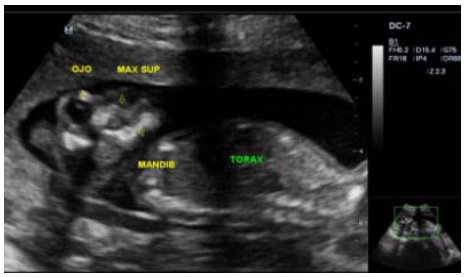

Figure 1: Fetus with acrania parasagittal section, note that there are no bones of the cranial vault.

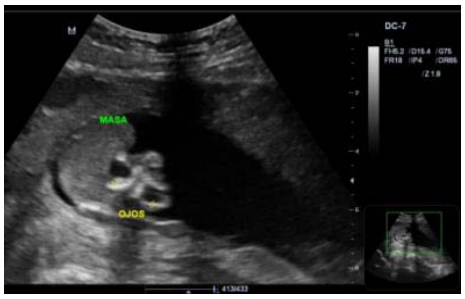

Figure 2: Coronal section of the fetal skull, observing mass that protrudes over the base of the skull with isoechoic content (exencephaly).

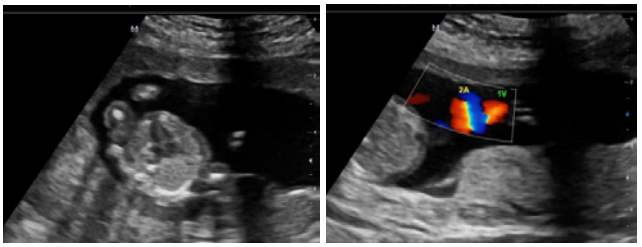

Figure 3: Axial section of the 4-chamber heart (left image), Doppler view of the umbilical cord with two arteries and 1 vein.

Discussion

Neural tube defects have a prevalence of 1.86 per 1000 live births, among the two most common pathologies are spina bifida and acrania. The latter is not actually an isolated neural tube alteration, it belongs to a sequence called acrania exencephaly anencephaly, since the lack of bones that make up the cranial vault will cause a protrusion of the cerebral parenchyma (exencephaly) and with sudden movements of the fetus and the chemical irritation of the amniotic fluid to the unprotected brain structure causes degeneration and destruction of the brain and causing its absence (*anencephaly*) [11]. There is a confusion between what is acrania and acalvaria, the first is the absence of the scalp and partial or complete cranial vault that inevitably causes anencephaly, while the second is the total or partial absence of the skull bones but with an intact scalp and it will not cause anencephaly [12] therefore, acrania that is incompatible with life, acalvaria could have a survival expectancy. It is described as a postneurulation defect that, after the closure of the cranial neuropore, there is an alteration in the migration of the membranous portion of the neurocranium [14]. The fetal neurocranium has two sections,

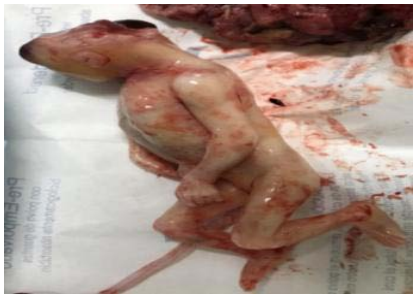

Figure 4: Fetus with acrania - anencephaly.Taken after the expulsion of the fetus with the authorization of the mother.

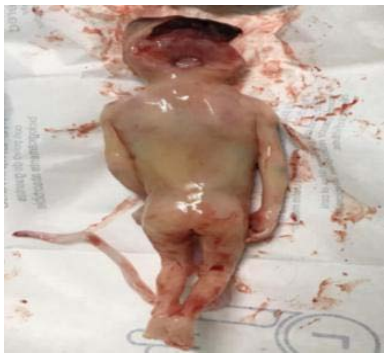

Figure 5: Fetus with acrania, note the absence of occipital bone and cerebral hemispheres. Normal spine.

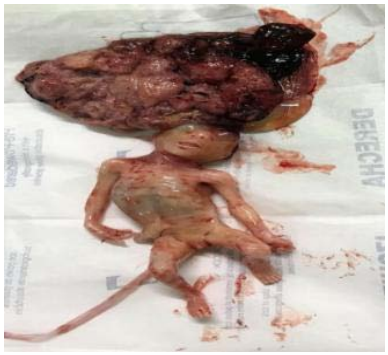

Figure 6: Fetus with acrania next to the placenta, note the absence of alterations in the midface, abdomen, genitals and extremities.

the chondrocranium that forms the base bones and the membranous flat bones that form the cranial vault, acrania is an alteration in said formation and occurs at 4 weeks of gestation when there is a defect in the closure cranial neuropore [13]

The diagnosis is made in the 12-week ultrasound where the bones of the cranial vault are not visualized, which are characterized by being a hyperechoic ring surrounding the brain parenchyma, it is associated with multiple malformations such as heart disease, cleft palate, omphalocele or Pentalogy of Cantrell [13].

Since the 1960s, the relationship of folate deficiency with neural tube defects was studied, it was determined that folate was essential for the transfer of a carbon unit for the transformation of homocysteine to methionine, DNA methylation and others. Cellular reactions are an essential part for rapid tissue growth and cell replication (2, 3, and 4), it is for this reason that our Ecuadorian Legation is obliged to supply folic acid to all pregnant women up to 12 weeks of gestation. Acrania per se has a high mortality rate of almost 100%, in the case presented by the medical board and due to incompatibility with life, it was decided to terminate the pregnancy.

Conclusion

The sequence called acrania exencephaly anencephaly, is the total or partial absence of bones that make up the cranial vault causing a protrusion of the cerebral parenchyma (exencephaly) and with the sudden movements of the fetus together with the chemical irritation of the amniotic fluid to the brain structure Unprotected causes degeneration and destruction of the same resulting in the absence of brain mass (*anencephaly*), the diagnosis is made by ultrasound as early as 12 weeks where the bones of the cranial vault are not visualized, which are characterized by being a hyperechoic ring surrounding the brain parenchyma. Most cases are incompatible with life.

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