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ANENCEPHALY: A CASE REPORT

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ABSTRACT

Anencephaly is a lethal foetal congenital malformation. This malformation accounts for 40% of neural tube malformations. It is characterized by congenital absence of the majority of brain, skull and scalp. Thus, the cranial neural tissue is exposed. The diagnosis is based on the ultrasound of the 1st trimester by the discovery of an exencephaly. This report is being made to highlight the importance of an early ultrasonic diagnosis of anencephaly, which could help in a thorough evaluation and active management.

INTRODUCTION

Anencephaly is a congenital malformation of the central nervous system that results to the failure of closure of the cranial end of the embryologic neural tube, usually occurs between the 23rd to 26th days after conception. 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]. Neural tube defects are the most common connatural malformation which will affect the natural development of the foetus. These neural tube defects are categorized into spinal and cranial defects. The cranial defects are classified into anencephaly, iniencephaly, and encephalocele. The anencephaly is further divided into holoanencephaly and meroanencephaly[2]. The diagnosis can be made as early as in the first trimester ultrasound from 11th to 14th weeks showing absence of the skull[3]. It is, usually, associated with polyhydramnios. Generally, the acrania has a 100% mortality and is not compatible with life. The causes are multifactorial (iatrogenic, toxic, metabolic, nutritional and exceptionally chromosomal).

CASE REPORT

23-year-old patient, rural geographical origin, incomplete primary school, housewife, unknown blood type, no previous significant pathologies, no allergies, no surgical procedures, no notion of consanguinity, her first menstruation at 15 years of age, a previous pregnancy with vaginal delivery without complications with a live child, upon admission to our hospital on April 10, 2018 with the date of the last menstruation on September 12, 2017, during pregnancy of 30 weeks, sent from another health facility due to foetal malformation.

On general examination, the patient was clinically stable, height at 147cm, weight at 68 kg, no pedal oedema, blood pressure at 110/80 mmHg, 82bpm, 98% saturation and temperature at 36.7 degree Celsius. Obstetric examination: Active uterine contractions, uterine fundus for 32 weeks, the foetus was in longitudinal position, an active foetal heartbeat. Sonography reveals the foetus with active movements, a posterior placenta without signs of detachment, a maximum vertical pocket of 6 cm amniotic fluid (normal 2-8 cm). Absence of cranial vault with mass protruding from the skull base compatible with acrania and exencephaly.

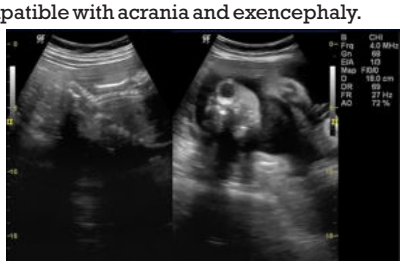

Figure 1: Anencephaly resulting in the "Frog Face sign"

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 foetus 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)[4].

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

In a normal human embryo, the neural plate is formed approximately 18th days after fertilization. During the 4th week of development, the neural plate invaginates to form the neural groove. The neural tube is formed due to closure of the neural groove by fusion of neural folds. The process is initiated at a single site and extends towards the rostral and caudal neuropores. Closure completed by day 24 for the cranial end and day 26 for the caudal end[5]. Anencephaly results from the failure of neural tube closure at the cranial end of the developing embryo leading to incomplete development of calvaria and brain. Babies with anencephaly are either stillborn or die shortly after birth. The incidence of

anencephaly shows a multifactorial pattern of inheritance, with interaction of multiple genetic and environmental factors. The specific genes which cause the neural tube defects are not been identified still. One such gene methylene tetrahydrofolate reductase has been shown to be associated with the rise of neural tube defects[6].

The initial screening for anencephaly and other neural tube defects are performed by testing for high levels of maternal serum alpha-fetoprotein in the second trimester of pregnancy and by ultrasonography in the first trimester of pregnancy[7]. The preventive measures include diet supplementation with folic acid at least 400 µg/day before pregnancy and in the 1st month[8,9]. This can decrease both the frequency and severity of the condition.

CONCLUSION

Anencephaly is a lethal congenital malformation and it may be diagnosed by ultrasound as early as 12 weeks. In the absence of curative treatment, prevention is therefore essential and creating awareness among the people about the preventable causes of nutritional deficiency.

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