International Journal of Research in Medical Sciences
Rabarikoto HF et al. Int J Res Med Sci. 2018 Apr;6(4):1454-1456
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012
DOI: http://dx.doi.org/10.18203/2320-6012.ijrms20181044

Case Report

Craniorachischisis: first case reported in Madagascar

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Received: 04 March 2018
Accepted: 10 March 2018

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ABSTRACT

Craniorachischisis is the most severe type of neural tube defect in which almost the entire brain and spinal cord remain open. We report a case in a female fetus born at gestational week 38, with both anencephaly and open spina bifida. It was the second pregnancy of a 26-year-old woman. The first pregnancy had to be interrupted by a medical termination at 18th gestational week because of an anencephaly. We aim to report the first case documented in Madagascar.

Keywords: Anencephaly, Craniorachischisis, Neural tube defects, Spina bifida

INTRODUCTION

Craniorachischisis is a very rare congenital anomaly characterized by anencephaly and total spina bifida.[1,2] The entire brain and spinal cord is open. It is the most severe form of neural tube defects (NTDs) and is incompatible with life. The word prevalence is unknown, Johnson KM and al reported 0.51 per 10,000 live births in a Texas-Mexico border population.[1] We aim to report a case, the first documented in Madagascar.

CASE REPORT

This is a newborn baby from a second gestation of a Malagasy 26-year-old woman. Her first pregnancy ended with a medical termination of pregnancy for anencephaly at 18th weeks of gestation, diagnosed in ultrasound at the Soavinandriana Hospital Center Antananarivo, Madagascar. The second pregnancy started 22 months after the first, it was a spontaneous conception. The diagnosis of recurrence of anencephaly had been made by ultrasound at gestational week 13 in the same hospital. The prognosis was explained to the parents, and they chose to continue the pregnancy.

Parents were not consanguineous. There was no family or personal history of diabetes or congenital anomalies. The father had no particular antecedent; the mother had a BMI at 20.23kg/m^2, was not diabetic or epileptic and did not take any particular drugs or toxicants before and during the conception period until the time of diagnosis, apart from folic acid.

The pregnancy was scheduled, she took folic acid 5mg per day 3 months before conception and during the two first months of pregnancy to prevent recurrence. There was no particular event during pregnancy. After discussion with the parents, the birth was performed by caesarean section at 38 weeks of gestation. The newborn female presented a craniorachischisis: absence of a major portion of scalp and cranial vault, a cervical myelomeningocele, a spina bifida open from the cervical to the thoracic region (Figure 1). The neck was short, the

ears low inserted and orbits were prominent (Figure 2). Any other associated external deformities were observed. Anencephaly was diagnosed at 13th week and rachischisis at birth because the parents did not want to continue ultrasound surveillance. The baby died two hours after extra-uterine life. Radiological examination, autopsy and histological examination were refused by the parents.

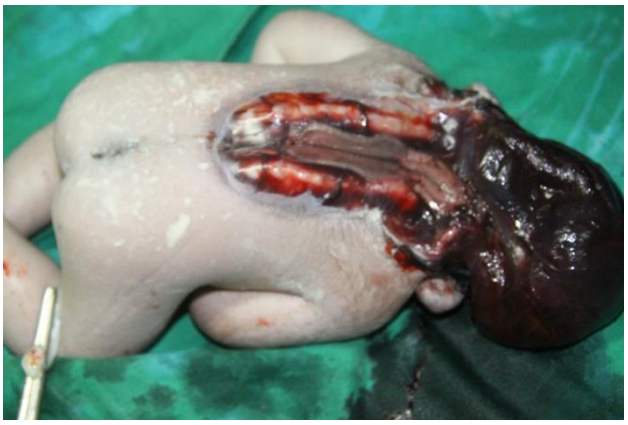

Figure 1: Absence of cranial vault, cervical myelomeningocele, spina bifida.

Figure 2: Short neck, ears low and orbits were prominent.

DISCUSSION

In the normal human embryo, the neural tube develops and closes during the 3rd and 4th weeks after conception. The neural plate is formed approximately 18th days after fertilization. During the fourth week of development, the neural plate invaginates to form the neural groove. The neural tube development is due to closure of the neural groove by fusion of neural folds. Traditionally, a process is considered to be initiated at a single site, and extending bi-directionally, rostrally and caudally, to the rostral and caudal neuropores. Closure completes by day 24 for the cranial end and day 26 for the caudal end.[3,4] A multiple site of fusion like in animal models can be considered after NTDs reviewed.[5-7] In any case, it is normally completed by 28 days post conception.[8]

Craniorachischisis is a defect that appeared when the neural tube fails to close during this period of gestation. Van Allen MI and al hypothesized that craniorachischisis results from failure of closures of 2, 4 and 1 among the four separated initiation sites for neural tube.[5] Campbell LR and al had suggested that a closed tube may reopen in some cases of NTDs.[9] These two last hypotheses seem plausible to explain the absence of NTDs at the lumbosacral region in the case that we report.

The cause of NTDs is multifactorial: exposure to valproic acid, maternal type 1 or pregestational insulin-dependent diabetes mellitus, consanguineous marriage, environmental influences, folate deficiency and polymorphism mutation in gene encoding 5, 10-methylenetetrahydrofolate reductase (MTHFRC677T), antimetabolites of folic acid, other toxins with the same effects as folic acid, maternal fever in early gestation and amniotic band disruption during pregnancy.[10-14] Cellular and molecular mechanisms in the occurrence of cracniorachischisis is poorly understood and difficult to study in humans. Some studies have suggested that mutations in the CELSR1 and SCRIB genes may be associated with this malformation.[15] Effects of genetic and association with some malformatives syndromes including craniorachischisis were reported: case with trisomy 18, pentalogy of Cantrell, Fryns syndrome, sirenomelia, diprosopus twin.[2,10,16,17] Possibility of association with internal malformation was not known in our case, nor genetic anomaly.

Because of the antecedent of anencephaly, the mother took folic acid supplementation with a dose of 5mg/day before conception and during organogenesis to prevent risk of recurrence. Folic acid supplementation is one of the most significant preventative interventions. It has been shown to be an effective means of lowering recurrence risks for future pregnancies.[12,18,19] But this supplementation was not effective in our patient because anencephaly had recurred in the second child, associated with a rachischisis. Depending on the sex of the affected fetus, Lalouel JM and al evaluated that the risk of recurrence of NTDs is multiplied by 0.051 after one female affected, by 0.058 after one male affected. After two affected fetus, the risk for a third one increase, it varies from 0.09 for a male birth after two affected females to 0.16 for a female birth after two males affected.[20] Collins JS and al found that the recurrence rate of NTDs was 0.2% with folic acid and 6.1% without any prevention. They reported one recurrence of anencephaly with a 13q deletion in group with folic acid supplementation. One case of NTDs recurrence in group without prevention was similar with our case report, the first child had a spina bifida and the second a craniorachischisis.[19]

Concerning the diagnostic means, craniorachischisis can be detected on ultrasound at gestational age of 13 weeks and magnetic resonance imaging at 22 weeks.[2,11] In our case, the rachischisis was diagnosed at birth. After the

first ultrasound examination at 13th weeks, and explanation to the parents of anencephaly recurrence, no other investigation was realized.

There is no cure or standard treatment for craniorachischisis. It is a lethal congenital malformation. Fetuses often miscarry during the pregnancy or shortly after birth.[1,21] In our case, the newborn baby died two hours after birth.
Before another conception, genetic counselling and folic acid supplementation are recommended as preventive measures for parents who had one or more affected child.[14,21]

CONCLUSION

Even if the recurrence remains possible despite prevention, parental counseling and folic acid supplement before conception and during early pregnancy is still the best preventive measures for NTDs. Termination of pregnancy should be realized as soon as the diagnosis is made.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Rabarikoto HF, Rakotozanany PS, Tsifiregna RL, Ratovondrainy W, Randriambolona DMA. Craniorachischisis: first case reported in Madagascar. Int J Res Med Sci 2018;6:1454-6.