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Craniorachischisis totalis: A rare and severe neural tube defect

Vinodhini K[1]*; Sweta Singh[2]; Parthasarathy R[3]; Naimisha Priya[4]; Kamla Jamra[4]

[1]Senior Resident, Department of Obstetrics and Gynaecology, AIIMS Bhubaneswar, India.

[2]Professor and Head, Department of Obstetrics and Gynaecology, AIIMS Bhubaneswar, India.

[3]MCh Neurosurgery, AIIMS Bhubaneswar, India.

[4]Junior Resident, Department of Obstetrics and Gynaecology, AIIMS Bhubaneswar, India.

***Corresponding Author: Vinodhini Kadirvel**
Senior Resident, Department of Obstetrics and Gynaecology, AIIMS Bhubaneswar, India.
Email: vinokadir@gmail.com

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Abstract

Craniorachischisis is a very rare congenital anomaly characterised by anencephaly and total spina-bifida. We present a case of a woman in her 30s, second gravida and a 20 week pregnancy, who presented with vaginal bleeding to the obstetrics department. An ultrasound scan confirmed the foetal anencephaly of a single live foetus. The pregnancy was terminated, and the expelled abortus showed non-formation of the cranial vault with hypoplastic neural tissue. The history revealed that the mother has not taken folic acid supplementation during her second pregnancy, which confirms the importance of preventive folate therapy for neural tube defects.

Background

Neural Tube Defects (NTDs) are a heterogeneous group of diseases of the central nervous system with multifactorial origin and a prevalence ranging from 0.5 to 10 per 1000 pregnancies. The primary source of these defects is a malfunction in the neural tube closure processes, which affects the formation of skin, paravertebral muscles, connective tissue, bone, and spinal cord [1]. NTDs are caused by a variety of factors, including folic acid deficiency, valproic acid exposure, maternal type 1 or pregestational insulin-dependent diabetes mellitus, consanguineous marriage, environmental factors, polymorphism mutations in the folate metabolism genes, and maternal fever in the early months of gestation [2]. This abnormality can range in severity and complexity from an occult anomaly to a serious, potentially fatal issue [3].

Anencephaly is one of the most prevalent and deadly neural tube disorders, which is brought about by a defective closure of the rostral pore. Conventionally, a process is thought to begin at a single location and extend in both directions, rostrally and caudally, to the neuropores located rostrally and caudally [4]. Day 24 for the cranial end and Day 26 for the caudal end mark the completion of closure. The anomaly is caused by inadequate development of the brain's tissue and the skull's vault, although the facial region is normal. The pituitary gland is frequently hypoplastic or non-existent [5]. Among NTDs in India, the highest reported prevalence of anencephaly was 2 per 1000 infants. Lower socioeconomic class groups are typically more likely to experience anencephaly and spina bifida, and at least half of neural-tube defects can be averted if women take adequate folic acid supplementation before conception and during the early stages of pregnancy [6]. Here we report a very rare case of craniorachischisis totalis, which includes both anencephaly and spina bifida.

Case presentation

A female in her 30s presented to the obstetrics department at 20 weeks pregnant, with complaints of vaginal bleeding for a day. The patient was married for 10 years, and her first pregnancy was at 27 years old. She had delivered a normal female baby vaginally at term.

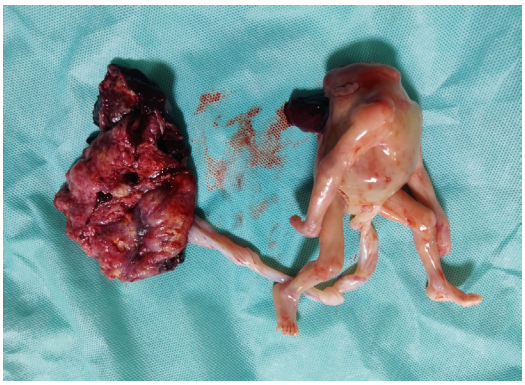

Figure 1: Expelled abortus with placenta.

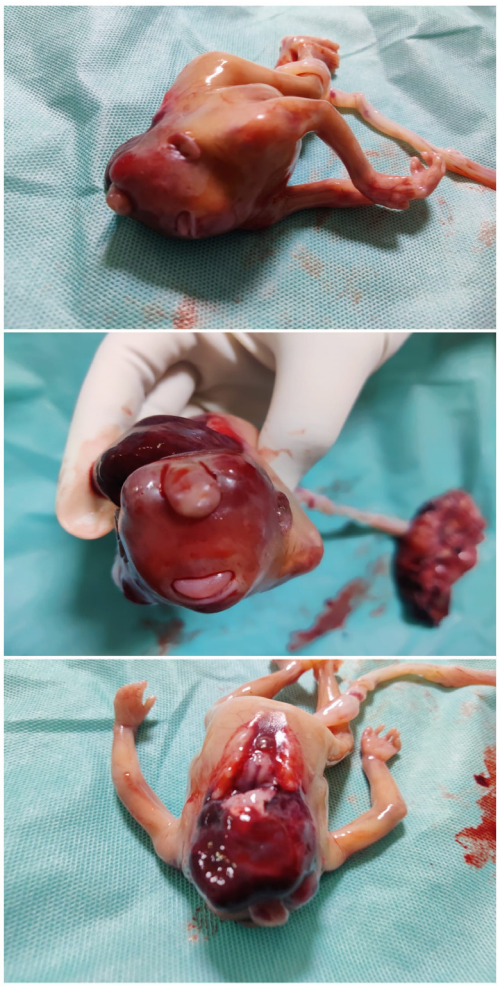

Figure 2: Anencephaly with rachischisis.

Her current pregnancy was a spontaneous conception, and there was no intake of Folic Acid tablets during the entire pregnancy. An ultrasound scan showed a single live intrauterine foetus with anencephaly. The estimated foetal weight was 195 grams, with a crown rump length of 3.96 cm, and a foetal heart rate of 160 bpm. A decision was taken for the medical termination of pregnancy. She was given Mifepristone, followed by misoprostol as per the FIGO protocol. She expelled an abortus of 180 grams along with the placenta in toto (Figure 1). The gross examination showed an anomaly of anencephaly with rachischisis, an open neural tube defect (Figure 2). There was non-formation of the cranial vault, with exposed neural tissues on gross observation.

The patient and her attendants were not willing to undergo a foetal autopsy, which could have really been an asset in establishing the diagnosis. And Molecular genetic testing which could not be done in this case due to financial constraints, is much recommended in this current era. Genetic counselling, advice regarding the preconceptional folate regimen, and early booking on planning next pregnancy were suggested to the mother at discharge.

Discussion/conclusion

Neural tube defects are one of the most prevalent structural congenital anomalies, which include a spectrum of diseases divided into primary neurulation defects, which include craniorachischisis, anencephaly,spina bifida, and secondary neurulation disorders, including spinal dysraphism, encephalocele, and meningocele. The current case reported here is a severe form of NTD, craniorachischisis totalis, which happens due to the failure of the initiation of neural tube closure [7]. The neural plate develops in a typical human embryo about 18 days after fertilisation. The neural groove is formed when the neural plate invades during the fourth week of development. When the neural folds fuse together, the neural groove closes, forming the neural tube. Failure of neural tube closure at the cranial end of the developing embryo results in anencephaly [8].

It is challenging to investigate the cellular and molecular mechanisms underlying craniorachischisis in humans, and these mechanisms are not well known. The majority of mouse craniorachischisis cases are thought to be caused by a disruption in the molecular signalling cascade Planar Cell Polarity Pathway (PCP). Human PCP gene sequencing has shown potential mutations in CELSR1, VANGL1, VANGL2, FZD6, SCRIB1, and DVL2 in some patients with craniorachischisis, spina bifida, anencephaly, or closed forms of spina bifida. Recent investigations have revealed that craniorachischisis in humans could entail coupled heterozygous mutations of two or more genes regulating PCP signalling.

It was proposed by Campbell LR et al. that a closed tube might reopen in certain NTD situations. Many women are even unaware that they are pregnant since the neural tube generally closes and develops fully within 28 days of conception. According to certain theories, the neural tube closes at multiple points in humans and mice, with the clinical manifestations of neural tube defects varying based on the location of the closure failure [9].

Even though the cause of NTDs is multifactorial, folic acid deficiency is a very well-known factor in their development. During the critical period, which is up to six weeks after the last menstrual cycle, exposure to valproic acid and other antimetabolites of folic acid, as well as other pollutants like lead, etc., can interfere with normal folate metabolism and raise the risk of anencephaly [10]. In our case study, the mother did not take folic acid during the first trimester of her pregnancy. This reaffirms the importance of folic acid in the prevention of NTD. Twenty NTD recurrence rates, according to Collins JS and colleagues, were 6.1% in the absence of prophylaxis and 0.2% in the presence of folic acid. Supplementing with folic acid has been demonstrated to be a successful method for reducing the likelihood of the recurrence of NTDs in subsequent pregnancies [11].

Contrary to our case, another case report by Rabarikoto, the mother took folic acid supplements at a dose of 5 mg/day prior to conception and during organogenesis, in order to reduce the chance of a recurrence because of the antecedent history of anencephaly. But in his report, there was a recurrence of the NTD with craniorachischisis, proving the existence of a multifactorial origin [12].

In terms of diagnostic tools, ultrasound and magnetic resonance imaging can identify craniorachischisis as early as 13 weeks of gestation. The current case was diagnosed only at 20 weeks through ultrasound since the patient was from a uneducated poor background.

Craniorachischisis has no known treatment or cure. This congenital deformity is fatal. The miscarriage rate is increased in such pregnancies [13]. In our case report, the patient presented with bleeding per vaginum, and the foetus was alive until it was expelled. If detected early, medical termination of the pregnancy is indicated because survival is not possible. The suggested preventive interventions include folic acid supplements and parental counselling, which should begin before conception and continue throughout the pregnancy. It is recommended that women who are capable of getting pregnant take 400 micrograms of folic acid daily, starting at least one month before conception [14].

Learning points/take home message

To lessen the occurrence of such severe defects, pre-conceptional folate supplementation, first trimester screening, which includes ultrasound and serum marker testing at 11-13+6 weeks, and alpha-fetoprotein levels in the early detection of NTD are crucial.

Adequate folic acid intake before and throughout pregnancy can prevent craniorachischisis by up to 75%.

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