ORIGINAL RESEARCH PAPER

Volume-8 | Issue-5 | May-2019 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

CRANIORACHISCHISIS – A CASE REPORT OF RARE CONGENITAL MALFORMATION.

Gynaecology

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ABSTRACT

Craniorachischisis, a lethal congenital malformation, is an extremely rare and severe type of open neural tube defects arising from defective morphogenesis of central nervous system during embryonic stage of life resulting in fissure of the skull and vertebral column. We report the case of a newborn female baby exhibiting features of craniorachischisis, which was detected after birth following assisted breech vaginal delivery of a 36year old woman who presented to us at 30 weeks of gestational age, in second stage of labor. Through this case report, our objective is to reemphasize that interventions like health education, parental counselling, use of folic acid along with additional micronutrients in peri-conceptional period and throughout the pregnancy are crucial, especially in low resource settings, substantially decreasing the risk of neural tube defects.

KEYWORDS

Neural tube defects, Craniorachischisis, Anencephaly, Congenital malformations, spina bifida, folic acid

INTRODUCTION

Neural tube defects (NTD) are one amongst the most prevalent congenital fetal anomalies accountable for significant global health care burden and have wider geographic prevalence . Its incidence ranges from 0.6/1000 births in Africa to 10/1000 births in middle east [1]. The condition in low-income countries is even more formidable due to increased prevalence, delay in diagnosis and improper management of mother and neonates. In India, these disorders have a reported incidence of around 0.5 – 11/1000 births with higher rates documented from North India than from the southern states of India [2]. Human NTD's have a multifactorial aetiology, including various environmental and genetic factors.

Craniorachischisis is an extreme example of a rare, open neural tube defect characterised by combination of anencephaly along with open spina bifida with meningomyelocele and such malformations are incompatible with life. Though the prognosis is grave mostly leading to intrauterine deaths, nevertheless live births are possible. The data on its exact world-wide prevalence is not available. Johnson KM et al, reported its prevalence to be 0.51/10,000 live births in Texas-Mexico border population, with highest incidence found in china [3].

CASE REPORT

We report the case of a newborn baby with craniorachischisis, which was diagnosed after birth.

A 36year old, gravida 3 para 2 presented to us in advanced labor, at 30 weeks period of gestation with complaints of abdominal pain for 5hrs and leaking per vagina for 2hrs. On clinical examination, patient had a normal vital data. On obstetric examination, uterine contractions were regular, fetal heart rate was not localised on stethoscope. Local examination revealed clear draining of liquor from introitus with breech climbing the perineum. While conducting assisted breech vaginal delivery, deficiency of skin over vertebral spine was observed followed by delivery of head with absence of cranial vault. Further examination of the female new born weighing 1200grams demonstrated following features of craniorachischisis (as shown in figure 1 and 2) - deficiency of major part of the scalp and cranial vault, a cervical myelomeningocele, open spina bifida from cervical to thoracic region, prominent orbits, flat nasal bridge, low set folded ears, fish mouth apperance of mouth, short neck, broad shoulders with extreme extension of head, club hand with brachydactyly on the right side. The baby expired 30 minutes after birth. Postpartum period was uneventful. Radiological examination, genetic analysis, autopsy and pathological examination were denied by the couple.

Reviewing obstetric history, patient had her previous two pregnancies inadequately booked and supervised with full term vaginal deliveries conducted at home. Her last child birth was 3years back. Her menstrual periods were regular. Her index pregnancy was also inadequately booked and supervised by a local registered medical practioner (RMP). She had a single ultrasonography done at 8weeks demonstrating intrauterine viable fetus. Further scans were not available. Couple had a non-consanguineous marriage, both of them were not educated and daily wage workers belonging to lower socioeconomic status. There was neither family nor personal history of congenital anomalies or diabetes. The mother was not an epileptic. She was neither using any medicines nor toxicants preconceptionally or during pregnancy except for folic acid tablets which she took irregularly during her index pregnancy. Her blood pressure and blood sugar records were normal.

DISCUSSION

From fertilisation to birth, the embryo and fetus have to acclimatize, at a transcriptional and molecular level, to numerous changes in their cellular milieu. At the time of fertilisation, the milieu depends on maternal micronutritional status and paternal germ cells. After fertilisation maternal metabolism, nutritional status, lifestyle holds paramount importance. Any alteration in this process of acclimatization during morphogenesis period results in congenital anomalies.

Neural tube develops and closes normally within 28 days post conception, that is even before clinical diagnosis of pregnancy is made. In the normal human embryos, neural plate develops around 18th day post fertilisation. Neural groove is formed by its invagination, during the 4th week of development. Fusion of neural folds results in neural groove closure and there by formation of neural tube. Historically, this process is initiated at a single site, and extends bidirectionally both to the rostral and the caudal neuropores. Complete closure at the cranial end takes place at around day 24 and at the caudal end by day 26 (4). In animal models, multiple sites of neural tube fusion have been demonstrated (5). Craniorachischisis is a severe type of birth defect resulting from defective embryonic neural groove closure during this period of gestation, in which virtually entire brain and spinal cord remains open.

Campbell LR et al., proposed that secondary reopening of the closed tube may explain certain patterns of NTD's(6). Van Allen et al., through his multisite closure model of neural tube with five sites of closure in humans, proposed that error in closure at sites 2, 4 and 1 results in craniorachischisis(5). In our reported case, absence of NTD's at the lumbosacral region can be explained by last two theories.

Multifactorial etiology of NTD's includes- nutritional defieciency (vitamins B6, B12, Folate, zinc), low socioeconomic status (SES), consanguineous marriage, maternal pregestational or type 1 insulin dependent diabetes mellitus, maternal fever during early pregnancy, obesity, use of anticonvulsant drugs, insulin, (MTHFRC677T), oxidative stress, folate antimetabolites, other toxins (arsenic, tetracholoroethylene), genetic mutations encoding 5, 10-methylenetetrahydrofolate reductase (2,7). In 2002, Vangl 2 gene was the first gene of craniorachischisis to be found. Mutations in the CELSR1, SCRIB, ptk7 genes have been demonstrated to be associated with this malformation. Few genetic and malformative syndromes were reported to be associated with craniorachischisis like pentology of Cantrell, Fryns syndrome, trisomy 18 etc (2).

In our case low SES, noncompliance to folic acid intake, advanced maternal age are the possible risk factors. Possible association of genetic abnormality or internal malformation could not be established. Folic acid deficiency in the peri-conceptional period is strongly attributed to many cases of NTD as evident from various studies. There is valuable data from western countries that peri-conceptional folate supplementation prevents the first occurrence of NTD (with the role of 0.4 mg FA resulting in 60% reduction in NTD's) and its recurrence (with use of 4mg FA) (2,7). Collins JS et al demonstrated that the recurrence rate of NTD's was 0.2% with periconceptional folate supplementation and 6.1% without any supplementation (8). Though peri-conceptional and antenatal folic acid supplementation have decreased the risk of NTD's in women with or without a previous history of pregnancies affected by NTD, still there are 0.7-0.8 per 1000 pregnancies which were affected by NTD despite FA supplementation(9).

This condition can be diagnosed by biochemical tests, ultrasound (~ 13 weeks of gestational age) and magnetic resonance imaging at 22weeks (10). In our case, as the woman has not undergone any 2nd or 3rd trimester ultrasonography scans, the condition was diagnosed only at the birth. There is neither cure nor therapy for craniorachischisis. Hence, parental counselling and elective termination of pregnancy for fetal anomalies (ETOPFA) should be considered if diagnosed timely.

CONCLUSION:

NTD's originate from complex interplay between various genetic and environmental factors. Nonetheless, multifarious aetiology of NTD's leads us to reduce further risk by primary prevention through multiple interventions like health education, parental counselling, use of folic acid along with additional micronutrients in peri-conceptional period and antenatal period, even in low resource settings.

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

Abbreviations:
NTD: Neural tube Defect
SES: Socio economic Status
FA: Folic acid

Figure 1: Illustration showing craniorachischisis with cervical meningomyelocele and extension of defect into thoracic region with exposed spinal cord and nerve rootlets

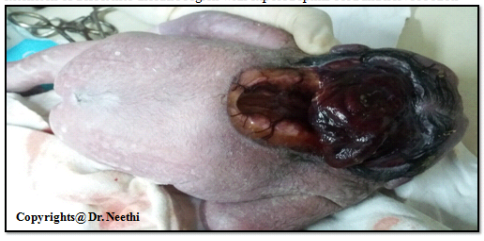

Figure 2: Illustration showing prominent orbits, flat nasal bridge, fish mouth like appearance of mouth, short neck, right club hand with brachydactyly.

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