

Neural Tube Defect: A Case Report

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ABSTRACT

Neural tube defect is rare congenital defect of fetus. We report a case of 13 weeks period of gestation with meningomyelocele came for termination of pregnancy. The diagnosis was confirmed by ultrasonography. History suggests that patient had previous neural tube defect baby, non-compliance to folic acid supplements. It is concluded that prenatal counseling, adherence to folic acid supplement and regular follow-up can prevent further complication.

Keywords: folic acid; meningomyelocele; neural tube defect.

INTRODUCTION

Neural tube defects (NTDs) are congenital structural abnormalities of the central nervous system and vertebral column affecting 1 in every 1000 pregnancies.¹ It may occur as an isolated malformation, in combination with other malformations, as part of a genetic syndrome, or as a result of teratogenic exposure. Neural tube defects (NTDs) are common major congenital anomalies that result from very early disruption in the development of the brain and spinal cord. They emanate from a failure of neurulation, which occurs around the 28th day after conception. There are three principal forms of neural tube defects: The first is anencephaly, second is encephalocele and the third is spina bifida.²

Neural tube defects are the second-most-common major congenital anomaly^{3,4} after cardiac malformations, and their prevalence varies by geographic region, race, and environmental factors.⁵ The International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) suggest first ultrasound scan when gestational age is thought to be between 11 and 13+6 weeks' gestation, as this provides an opportunity to detect gross fetal abnormalities.⁶ Though second-trimester '18-22-week'scan remain the standard of care for fetal anatomical evaluation in both low-risk and high-risk pregnancies.⁷ First-trimester evaluation of fetal anatomy and detection of anomalies was introduced in the late 1980s and early 1990s with the advent of effective trans-vaginal probes.⁸ Early detection and exclusion of many major anomalies, early reassurance to at-risk mothers, earlier genetic

diagnosis and easier pregnancy termination if appropriate.⁹

CASE REPORT

We report a case of 25 years multigravida at 13 weeks period of gestation with the diagnosis of meningo-myelocele (Neural tube defect) came for termination of pregnancy in outpatient department.



Figure 1. Fetus with Neural tube defect (after termination of pregnancy).

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Her history says that she has 6 years of marital life, non-consanguineous marriage and she had conceived spontaneously. Regarding her obstetrics history, she is G₄P₂A₁L₁D₁, first pregnancy was term vaginal delivery with neural tube defect, it was diagnosed in third trimester because no scan was done before baby died after few days of life, second baby she had neural tube defects diagnosed at 16 weeks period of gestation and pregnancy was terminated, third pregnancy she delivered healthy baby caesarean section was done. On her present (four) pregnancy she had first trimester scan i.e. at 13 weeks period of gestation ultrasound finding suggested a cystic mass of approximately 12*5 mm. apparently splaying of posterior spine at lumbosacral region associated with posterior-caudal displacement of midbrain and its deformation against the occiput as the crash sign and obliterating fourth ventricles (Figure 1). According to patient she was taking folic acid supplements. Genetic counseling was advised for chromosomal analysis but the parents refused and opted for termination of pregnancy. Induction of labor was done with misoprostol as per protocol. She delivered dead fetus weighing 200gms. Couple was counseled for the pre-conceptual care and regular follow up.

DISCUSSION

Since 2009, obliteration of intracranial translucency (IT) continues to be used as an indirect marker of open spina bifida in the first trimester. Occiput distance and crash sign in axial plane; fronto-maxillary facial angle and Maxillo-Occipital line in the sagittal plane is the indirect marker for open spina bifida.

Among the NTDs, anencephaly is not compatible

with life and fetus is still born or dies few hours after birth. While depending upon the severity, the child with spina bifida can survive with the advancement of technology. The neural cord is subsequently covered with a biocellulose patch and the skin closed over the patch with a single running stitch. If the defect is too large, a skin substitute is placed over the biocellulose patch for two-layer closure of the defect. Following coverage of the lesion, the insufflated carbon dioxide is removed with simultaneous refilling of the amniotic cavity.

One study from China reported an estimate of recurrence risk of NTDs, based on a retrospective survey in the early 1990s. The overall recurrence was 6.9%, with differing rates in low, average and high prevalence areas of 3.7%, 4.5% and 8.3%, respectively.¹⁰ A meta-analysis of the randomized trials indicated a 69%-87% reduction with use of folic acid for the prevention of recurrent NTDs and 85-100% reduction in observational studies.¹¹

CONCLUSION

All women of childbearing potential should be receiving folic acid at a dosage of 0.4 mg/day, in accordance with the recommendation of the US Center for Disease Control and Prevention (CDC). Pre-conceptual counseling for importance of folic acid in pregnancy. First trimester ultrasound i.e. from 11 to 13+6 week is recommended, meticulous first trimester scanning to detect major fetal anomalies and direct visualization of the lesion is challenging. Intracranial translucency (IT) and other indirect sonographic marker of open spina bifida can increase our sensitivity to accurate diagnosis.

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