

RARE CASE OF RECURRENT NEURAL TUBE DEFECTS IN A OBESE WOMAN

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Abstract-

Introduction: Increasing trend of pre-conceptional obesity has also led to adverse reproductive outcomes like subfertility, miscarriage, gestational diabetes, gestational hypertension, preeclampsia, macrosomia, preterm birth, and sudden fetal demise. Furthermore, studies on maternal obesity and birth defects have reported that maternal pre conceptional obesity was associated with an increased risk of neural tube defects. Etiology of NTDs in maternal obesity could be explained by the body distribution of folic acid in women with obesity. A study has showed obesity could influence folate pharmacokinetics in women of childbearing age by reducing serum folate concentration. Spina bifida is the most common neural tube defect.

Case report: A 29 year old G4P1021 at POG 19 weeks 2 days with BMI OF 34.6kg/m² came to show her level 2 scan. She is married for 5 years and her first two conceptions were aborted spontaneously. 1 year later she again conceived and was booked and supervised throughout her pregnancy and told normal, she has spontaneous onset of labour around term and delivered a female child weighing 2400 grams with suspicious looking lesion in lumbosacral region in midline. Baby was referred to tertiary care center and finally was diagnosed and treated as spina bifida occulta at PGI. This time she has conceived after 3 years of previous childbirth she took folic acid post conception and level 1 scan was told to be normal. Dual screen was not done in this patient. Level 2 scan done at 19 weeks showing disappearance of posterior line and soft tissue overlapping in the lumbosacral lesion, but continuity of skin over the lesion maintained. Quadruple test yielded raised maternal serum Alpha Feto Protein level. She choose her further treatment at PGI.

Discussion: When a spine defect is detected, it is necessary a neurosurgical counseling with the patient; the risk of recurrence is 2 to 5%. The children with SB may have walking problems, sphincter dysfunction, sexual dysfunctions, skeletal deformities, cognitive disease. The degree of handicap and the survival rate depend on the level of injury, the size of the defect, and the presence of associated anomalies. The lower the injury level, the better the prognosis. The cognitive outcome is related by the presence or absence of hydrocephaly. Altogether about 70% of children with SB will have IQ>80.

Conclusion: Most studies conducted on NTDs did not highlight the maternal characteristics of women, as primary focus was on neonatal characteristics. There is still a lack of understanding on maternal characteristics, in particular maternal obesity, in women with NTD-affected pregnancy. There is a need to conduct more studies on the relationship between maternal obesity and neural tube defects in Indian population.

INTRODUCTION:

As per NFHS-5 survey 2019-2021, prevalence of obesity (defined as a body mass index [BMI] >30 kg/m²) in India was 39.6% among women compared to 34% of the women among then fall in reproductive age group¹.

In recent years, this increasing trend in pre conceptional obesity has also become a matter of of adverse reproductive outcomes like subfertility, miscarriage, gestational diabetes, gestational hypertension, preeclampsia, macrosomia, preterm birth, and fetal death^{2,3}. Furthermore, one meta-analysis on maternal obesity and birth defects published in 2008⁴ reported that maternal pre conceptional obesity was associated with an increased risk of neural tube defects. A second meta-analysis published in 2009 also concluded that maternal pre pregnancy obesity was associated with neural tube defects and possibly congenital heart defects⁵.

One of the recently proposed etiology of NTDs is maternal obesity. This association could be explained by the body distribution of folic acid in women with obesity. A study has showed obesity could influence folate pharmacokinetics in women of childbearing age by reducing serum folate concentration, which could be explained by increased body adiposity or volumetric dilution of the blood⁶.

Spina bifida is the most common malformation of the central nervous system. It could be defined as a vertebral median defect, with external exposure of the spinal cord. In most cases, the defect could be found in the dorsal part of the vertebral arch, while the splitting of the vertebral body represents only rarely the defect. Its incidence changes according

to several factors: Geographical area, ethnic differences, and seasonal variations; it is more common in Caucasians than Eastern, and such a difference persists after migrations, suggesting therefore, a genetic background more than an environmental one. Its incidence shows a 20-fold increase in diabetics, a 10- to 20-fold increase in patients taking valproic acid during their first trimester of pregnancy, a 30-fold increase in case of a previous affected son, and a 60-fold increase with two sons affected. The lesions characterizing the SB could be distinguished in ventral and dorsal lesions⁷.

Closed SB represents about 15% of cases, and it consists of a small defect completely covered by normal skin. Such condition is often asymptomatic and is diagnosed incidentally or after a radiological scan of the column. In other cases, hypertrichosis or a pigmented area, or a subcutaneous lipoma, could be found over the defect. The clinical relevance of this lesion is its frequent association with spinal cord infections. The open SB, instead, is the most common lesion and it is found in 85% of the cases. The spinal cord may be exposed or the defect can be covered by the meningeal membranes⁷.

CASE REPORT:

A 29 year old G4P1021 presented to the OPD at POG 19 weeks 2 days to show level 2 scan. She is a booked patient at PHC. She has obesity with BMI OF 34.6kg/m². She is married for 5 years and she conceived spontaneously after 8months of marriage and booked at nearby PHC but there was spontaneous abortion 2 months later. She again conceived after 6 months and was booked at nearby PHC she got dating scan which was told to be normal, but again she has spontaneous bleeding and abortion was confirmed by sonography. 1 year later she again conceived and was booked and supervised throughout her pregnancy her level1 and level2 ultrasounds also reported normal, she has spontaneous onset of labour around term and delivered a female child weighing 2400 grams with suspicious looking lesion in lumbosacral region in midline. Baby was referred to tertiary care center and finally was diagnosed and treated as spina bifida occulta at PGI. This time she has conceived after 3 years of previous childbirth she took folic acid post conception and level 1 scan was told to be normal. Dual screen was not done in this patient. Level 2 scan done at 19 weeks showing disappearance of posterior line and soft tissue overlapping in the lumbosacral lesion, but continuity of skin over the lesion maintained. Quadruple test yielded raised maternal serum Alpha Feto Protein level. Patient choose for further treatment at PGI.

DISCUSSION:

Global estimation of the prevalence of NTD affected deliveries of 1.9 per 1000 deliveries⁸. Furthermore, after exclusion of stillbirth and termination of pregnancy data, our study showed the incidence of livebirth NTD of 0.34 per 1000 livebirth.

Birth defects are multifactorial in origin, involving complex interplays between genetic and environmental factors. Therefore, in assessments of whether observed associations between an environmental factor such as maternal obesity and birth defects are likely to be causal, it is useful to consider the extent to which such associations meet the Bradford-Hill criteria for causality⁹ particularly in comparison with a well-known teratogen such as pre pregnancy diabetes. The reported associations between maternal pre pregnancy obesity and neural tube defects and congenital heart defects are known to meet some of the seven criteria for causality, namely, temporality, consistency of the association across studies, strength of the association, plausibility/coherence, biological gradient and experimental evidence.

When a spine defect is detected, it is necessary a neurosurgical counseling with the patient; the risk of recurrence is 2 to 5%. The children with SB may have walking problems, sphincter dysfunction, sexual dysfunctions, skeletal deformities, cognitive disease. The degree of handicap and the survival rate depend on the level of injury, the size of the defect, and the presence of associated anomalies. The lower the injury level, the better the prognosis. The cognitive outcome is related by the presence or absence of hydrocephaly. Altogether about 70% of children with SB will have IQ>80.

Accurate and timely prenatal diagnosis of SB is a major goal of modern antenatal care. The diagnosis of SB may be only suspected during I trimester because the detection rate of intracranial traslucency is 50%; the final diagnosis is made in the II trimester by direct visualization of defect of spine or the presence of myelomeningocele or the visualization of indirect signs.

CONCLUSION:

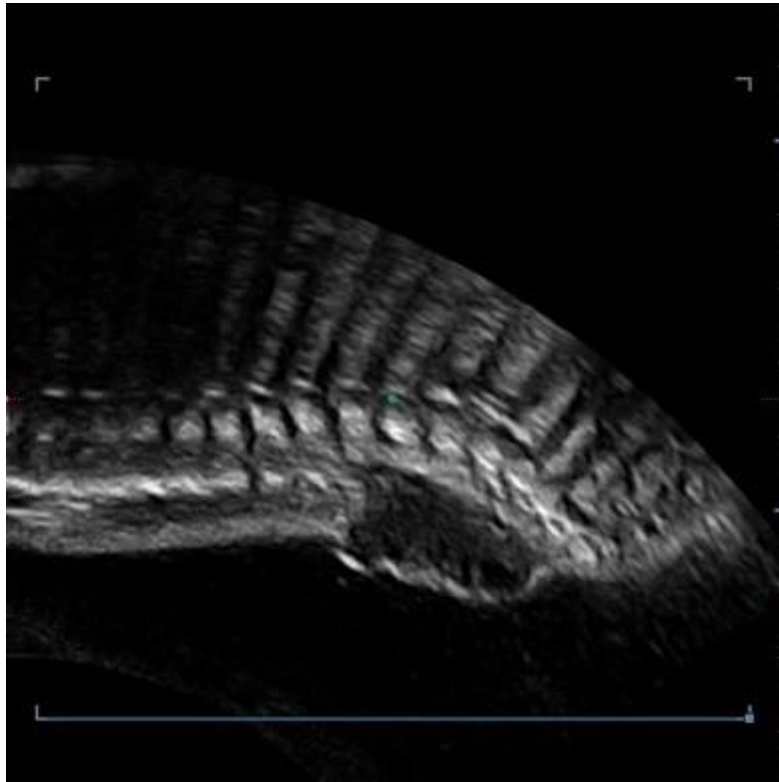
Obesity is increasing globally including among women in the reproductive age group. Obesity has been shown to contribute to certain types of congenital malformations particularly NTD, CHD and orofacial defects. Although absolute numbers of congenital anomalies caused by obesity are probably low, healthcare costs are substantial. Most studies conducted did not highlight the maternal characteristics of women with NTD-affected pregnancies, as primary focus was on neonatal characteristics. This indicates there is still a lack of understanding on maternal characteristics, in particular maternal obesity, in women with NTD-affected pregnancy. There is a need to conduct more studies on the relationship between maternal obesity and neural tube defects in Indian population.

ETHICAL ISSUES: None declared.

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LIST OF FIGURES

Figure1: showing disappearance of posterior line and soft tissue overlapping in the lumbosacral lesion, but continuity of skin over the lesion maintained.



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