



Normal Obstetric Outcome in Neurofibromatosis-1 Complicating Pregnancy

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Abstract

Neurofibromatosis (NF), a genetic disorder, has increased risk of obstetric complications as well as aggravation of maternal disease. However, here is a case of Neurofibromatosis associated with normal obstetric outcome despite the aggravation of dermatological lesions.

Key Words

Neurofibromatosis, Pregnancy, Obstetric Complication

Introduction

Neurofibromatosis (NF) is a distinct genetic disorder with a multitude of clinical manifestations. The incidence in pregnancy varies from 1/5000 to 1/18500 deliveries (1). Most of the current obstetric literature indicates that pregnant women with NF1 have increased risk of complications like spontaneous miscarriage, preterm delivery, preeclampsia, intrauterine growth retardation, stillbirths as well as maternal disease aggravation. However, we report a case with repeated normal obstetric outcome despite the aggravation of dermatological lesions of the condition during pregnancy.

Case Report

A 30 year old gravida 3, para 2 woman who had been married for 6 years, presented in the labor ward with 39 weeks pregnancy and labor pains. After an uncomplicated first pregnancy, she had delivered a baby girl of 2.75 kg birth weight by caesarean section done 4 years ago for breech presentation at 40 weeks gestation. After another 2 years, she had a second uneventful pregnancy whereby a male baby of 2.7 kg birth weight was born per vaginam. Both the children are currently healthy and doing well.

She had history of development of nodular lesions on her body at the age of 10 years. These had gradually increased in number and size with age and pregnancy events. Similar lesions were present in her mother as well as in her brother and sister.

Examination revealed a height of 150 cm, a pulse rate of 84 beats per minute and a blood pressure of 120/80 mm Hg along with mild anemia. There was no pedal edema or varicose veins. Multiple rounded, nontender, nodular lesions of variable sizes characteristic of NF1 were present on the entire body. The cardiovascular system and respiratory system were normal on clinical examination. Abdominal examination revealed a term sized uterus with single fetus in cephalic presentation, adequate liquor and FHR of 132 beats per minute. The cervix was 2 cm dilated, soft, uneffaced and midposition with intact membranes, vertex presenting at - 2 station in the gynecoid pelvis. Labor progressed normally and full term baby girl weighing 2.6 kg was delivered per vaginam. Both the mother and baby fared well in postnatal period.

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Fig-1 Clinical Photograph of Lower Abdomen Showing Multiple Lesions of Neurofibromatosis of Variable Sizes During Pregnancy

Discussion

Neurofibromatosis is an autosomal dominant condition which includes the more common NF1 caused by mutation in NF1 tumor suppressor gene on chromosome 17 and the less frequent NF2 caused by mutation in NF2 gene on chromosome 22. The condition could be inherited from the parents or could occur de novo as a result of spontaneous mutations.

Many authors have suggested that pregnant women with NF1 have increased frequency of obstetric complications as well as aggravation of maternal disease thereby placing these women and their fetuses at risk. Spontaneous miscarriage, preterm delivery, preeclampsia, HELLP syndrome, intrauterine growth restriction and stillbirths have been reported (2). An increased rate of caesarean section is also reported which could be due to fetal distress, malpresentations and cephalopelvic disproportion due to undiagnosed pelvic neurofibromas and pelvic contractures including cases of kyphoscoliosis affecting the lower spine (sequelae of NF1) (3). Moreover,

neurofibromas, the clinical hallmark of NF1 enlarge and many new lesions appear for the first time during pregnancy in many cases as was seen in the present case also. This could be due to hemorrhage within the masses or lysophosphatidic acid (LPA) mediated promotion of F-actin polymerization with increased migration and survival of Schwann cells (4). Because of poor pregnancy outcome and possibility of transmission to fetus, Ansari & Nagamani recommended early termination of pregnancy & sterilization in these women (5).

However, a literature search revealed only three studies (1-3) which suggested that NF1 may not be associated with significant obstetric complications and may have normal pregnancy outcome. The present case too, corroborates this view, which in spite of having a positive family history and fulminant disease had a normal obstetric career. Clinicians should bear in mind during counseling as well as management that a normal obstetric outcome could be expected in these women also.

References

1. Oh YM, Koh MS, Yeu JE, Park CS, Cho SJ. A case report of vertebral artery aneurysm and pre eclampsia complicating a pregnancy with neurofibromatosis. *Korean J Obstet Gynecol* 2000 ; 43(6):1114-18.
2. Weissman A, Jakobi P, Zaidise I, Drugan A. Neurofibromatosis and pregnancy. An update. *J Reprod Med* 1993; 38(11):890-96.
3. Dugoff L and Sujansky E. Neurofibromatosis Type 1 and pregnancy. *Am J Med Genetics* 1996; 66:7-10.
4. Todd DN, Wenyu M, Shi C *et al.* Neurofibromin deficient Schwann cells have increased lysophosphatidic acid dependent survival and migration – implications for increased neurofibroma during pregnancy. *Glia* 2007; 55(5):527-36.
5. Ansari AH, Nagamani M. Pregnancy and neurofibromatosis (Von Recklinghausen's disease). *Obst & Gynecology* 1976; 47:30.

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