



A PREGNANT WOMAN WITH NEUROFIBROMATOSIS

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ABSTRACT

Neurofibromatosis-1(NF-1) is an autosomal dominant disorder with variable clinical manifestations. we report a case of woman with neurofibromatosis type 1 who had conceived spontaneously and her pregnancy outcome. A 33 year woman G2P1L1 with previous LSCS, with neurofibromatosis type 1 having neurofibromas all over her body. She had underwent LSCS with tubal ligation at term pregnancy and had delivered healthy 2.8 kg baby. We obstetricians should be aware of these rare disorders can have variable progression and outcome of

pregnancy.

KEYWORDS: neurofibromatosis, pregnancy, café au lait, lower segment caesarean section.

INTRODUCTION

NF-1 is an autosomal dominant disorder with variable clinical manifestations. NF-1 is characterised by neurocutaneous signs, such as café-au-lait spots, axillary freckling, cutaneous neurofibromas and iris hamartomas (Lisch nodules), which occur in most patients. The prevalence is estimated at 1:4500.^[1,2] Approximately 50% of NF-1 cases result from de novo mutations.^[1,3,4] we report a case of woman with neurofibromatosis type 1 of sporadic mutation who had conceived spontaneously and her pregnancy outcome.

Case

A 33 year woman G2P1L1 with previous LSCS, with neurofibromatosis type 1 having neurofibromas all over her body. She had subnormal IQ, on dermatological examination, she had café au lait spots all over the body and ophthalmological examination was normal. No family history of similar lesions or disease was found. She had come for regular antenatal check ups in our hospital. She had no history of development of new lesions during pregnancy. She had not developed preeclampsia or fetal growth restriction during either of her pregnancies nor she had any history of abortions. She had underwent LSCS with tubal ligation at term pregnancy and had delivered healthy 2.8 kg baby. Her both babies are normal and have no signs of neurofibromatosis.

DISCUSSION

Many authors have suggested that pregnancy complications were more common in women with NF 1. case reports we have reviewed were associated with complications like eclampsia, IUGR, pregnancy induced hypertension, preterm labor, oligohydramnios, still birth, few reported regression of neurofibromas while majority observed development of new growths during pregnancy or increase in size of existing lesions.^[5] In a study by Terry et al,^[6] NF1 was associated with increased maternal morbidity in pregnancy (including hypertensive and cerebrovascular complications) but not increased maternal mortality. They have also found increased incidence of caesarean section delivery in these women.^[6,7] Obstetricians should be aware of the potential for increased antenatal and peripartum complications among women with NF1. Some neurofibromas contain oestrogen receptors, which suggests a relationship between the increase in number and size of neurofibromas and the increase in serum oestrogen levels during pregnancy,^[8] as posma et al have reported even a malignant schwannoma leading to lethal outcome of pregnant woman.

In our case we have not come across change in the existing neurofibromas, nor the woman had developed any complications during both her pregnancies and had completed family uneventfully. Obstetricians should be aware of the potential for increased antenatal and peripartum complications among women with NF1.

Detection of disease causing mutations in the NF1 gene allows presymptomatic and prenatal diagnosis, but is complex and time-consuming due to the large size of the gene, the existence of pseudogenes, the lack of clustering of the mutations in a particular region of the gene, and the variability of clinical findings. Because the time for investigations in prenatal diagnosis is

restricted, detection of disease-associated NF1 alleles is more rapid and useful especially for familial cases. Therefore, genetic diagnosis of NF1 is frequently performed by linkage analysis.^[9]

CONCLUSION

We obstetricians should be aware of these rare disorders can have variable progression and outcome of pregnancy. Hence regular follow up of woman and timely evaluation of development of complications can improve the outcome.

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