Neurofibromatosis in Pregnancy

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ABSTRACT
Neurofibromatosis (NF) is one of the most frequent genetic diseases in humans. It is less common to find pregnancy in NF patients. It may not reflect the real situation as most current information on pregnancy and NF is derived from case reports. In the past 15 years only two series of pregnant NF patients were reported in English language literature.

Neurofibromatosis is a genetic disorder which increases the risk of obstetric complications as well as aggravation of maternal disease. However, here is a case of NF in which transmission to baby has also occurred, which is very rare.

Keywords: Neurofibromatosis, Obstetric complications, Pregnancy.

INTRODUCTION
Neurofibromatosis (NF) is a distinct genetic disorder with a multitude of clinical manifestations. Its incidence in pregnancy varies from 1/5,000 to 1/18,500 deliveries. Most of the current obstetric literature indicates that pregnant women with NF type 1 (NFI) have increased risk of complications like spontaneous miscarriage, preterm delivery, preeclampsia, intrauterine growth restriction, stillbirths, and maternal disease aggravation.

The present case study describes a case of NF in pregnancy in Rohilkhand Medical College and Hospital, Bareilly, Uttar Pradesh, India, in which transmission to baby also occurred at postoperative day 3.

CASE REPORT
A 28-year-old Gravida 3, Para 2, Live 1 woman, who had been married for 8 years, came to OPD at 30 weeks of gestation with a complaint of bleeding per vaginum (p/v). On ultrasonography (USG), placenta praevia was diagnosed and patient was managed conservatively at that time and was put on Mcafee Johnson regimen. Skin lesions were noticed all over the body, and the patient had history of development of nodular lesions on her body since the age of 5 years (Figs 1A and B). With age, these have gradually increased in number and size. Skin reference was done and the patient was diagnosed as a case of NF and discharged under satisfactory condition after 2 weeks of hospitalization, with the advice to report to hospital immediately if she has any bleeding p/v. After that, the patient was presented in obstetrics ward with complaint of bleeding p/v at 38 weeks of gestation. A repeat USG was done, and in view of placenta praevia, her lower segment cesarian section was done and a male baby, weighing 3 kg, was delivered. Neurofibromatosis lesions were noticed on the newborn on the 3rd day of delivery (Fig. 2).

Figs 1A and B: Neurofibromatosis lesions on the back and abdomen of the patient

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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 less frequent NF2 caused by mutation in NF2 gene on chromosome 22. The condition could occur de novo as a result of spontaneous mutations or could be inherited from the parents. The management of these women with NF during pregnancy remains controversial, as some authors recommend early termination of pregnancy in view of the pregnancy complications, listed earlier, and possible transmission to the fetus. But then, there are others, who suggest that there may not be associated significant obstetric complications and therefore may have a normal pregnancy outcome.

The present case bears testimony to the later view.

An increased rate of cesarean section is also reported, which could be due to fetal distress, malpresentation and cephalopelvic disproportion due to undiagnosed pelvic NF and pelvic contractures, including cases of kyphoscoliosis, affecting the lower spine (sequelae of NF1).²

REFERENCES