www.jmscr.igmpublication.org
Impact Factor (SJIF): 6.379
Index Copernicus Value: 71.58
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v6i6.97

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Pregnancy with Neurofibromatosis

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Abstract
The report presents one case of neurofibromatosis type 1 who was detected during pregnancy. It describes how the disease was detected and diagnosed, and what was the outcome of the pregnancy with a positive family history of the disease. This is the first case of prenatal neurofibromatosis type 1 diagnosed at our OPD.
Keywords: *Neurofibromatosis, Obstetric complication in NF1, pregnancy, genetic testing, genetic transmission.*

Introduction

A 37 year old Gravida 4, Para 2+1, living issue 2 twice post CS women , who had been married for 16 years, came to OPD at 40 weeks of gestation with scar tenderness in early labour. She had earlier two deliveries at private set up for cephalopelvic disproportion (CPD). On examination general condition was average; she was afebrile with pallor and icterus not present. There were multiple big and small fibromas all over the body (figs 1A and 1B). In her past obstetrical history her eldest daughter aged 10 years having NF skin lesion since age 5years while her youngest daughter aged 4 years has not been noticed any sign of clinical lesion suspicion of NF. In her family history her mother too had NF and delivered 2 babies via caesarean section for CPD and died during antenatal period of third child due to cerebrovascular disease. On general examination PR 76/min, BP – 120/80 mm hg, pallor and icterus absent. On per abdomen examination uterus 40 weeks, FHR 132 beat/min, scar tenderness +. On PV examination os one finger, cervix long, early effaced, on lateral position. Thereby, lower segment caesarean section and bilateral tubectomy was done and a healthy male baby, weighing 2.7 kilograms was delivered with apgar score 8.

Case Report

A 37 year old Gravida 4, Para 2+1, Live 2 twice post CS women, who had been married for 16 years, came to OPD at 38 weeks of gestation with scar tenderness in early labour. She had earlier

two deliveries at private set up for cephalopelvic disproportion.

On examination general condition was average; she was afebrile with pallor and icterus not present. There were multiple big and small fibromas all over the body (Fig 1A and 1B). In her past obstetrical history her eldest daughter aged 10 years too suffering from neurofibromatosis since age of five years while her youngest daughter aged 4 years has not been noticed any sign of clinical lesion suspicion of NF. Moreover in her family history her mother too had NF and deliverd 2 babies via caesarean section for CPD and died during antenatal period of third child due to cerebrovascular disease.

On general examination PR 76/min , BP – 120/80 mm hg, pallor and icterus absent. On per abdomen examination uterus 36+ weeks, FHR 132 beat/min, scar tenderness +. On PV examination os one finger, cervix long, early effaced, on lateral position. Thereby, lower segment caesarean section and bilateral tubectomy was done on 07/08/2017 and a male baby, weighing 2.7kg was delivered.

Fig 1A

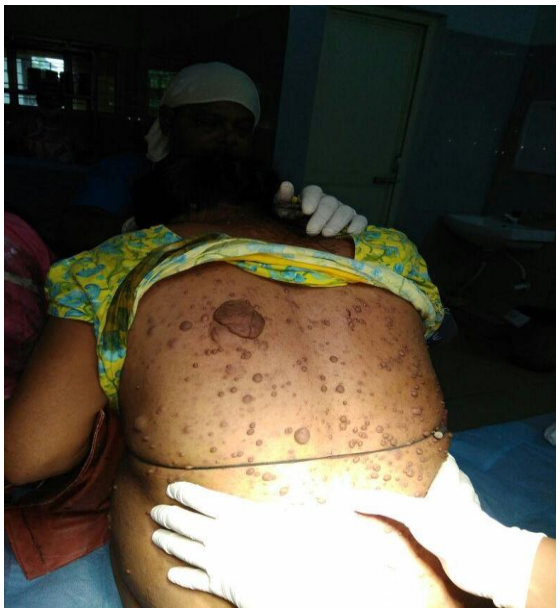

Fig 1B

Discussion

Neurofibromatosis 1 is commonly associated with pregnancy complication. Neurofibromatosis type 1 (NF1, peripheral neurofibromatosis, Von Recklinghausen's disease) is one of the most common genetic disorders occurring once in every 4,000 births. The condition is characterised by markedly variable clinical expressions, with manifestations ranging from mild cutaneous lesions like café-au-lait spots and axillary freckling to plexiform neurofibromas, optic gliomas, bony abnormalities, pseudoarthrosis and malignancies.[1]

NF has increased risk of complications like spontaneous miscarriages, preterm delivery, pre-eclampsia, IUGR, still birth & HELLP syndrome as well as maternal disease aggravation.[2]

It has increased frequency of obstetric disease there by placing these women and their fetus at risk. An increased incidence of cesarean section is also reported which could be due to fetal distress, malpresentation and CPD. Some recommend early termination of pregnancy because of adverse effect of pregnancy on the course of the disease & poor pregnancy outcome.[3] Measures should be taken before and during pregnancy to prevent transmission of gene to future generation. Before and during pregnancy: Couples with family history of NF1 who are thinking of baby

should be referred to a genetic specialist for advice. As in this case we have seen that NF affected gene is being transferred from one generation to the other. To avoid further transmission of the affected gene couples can opt for adoption or artificial insemination.

There are also a number of tests that can be carried out during pregnancy to check if a baby will develop NF1 or not: Chorionic villus sampling (CVS) – 11 to 14 week of pregnancy, amniocentesis – 15 to 20 week of pregnancy. Preimplantation genetic testing – here during cleavage stage or blastocyst stage of embryo, embryo carrying the affected be detected and discarded and moreover unaffected embryo can be transferred to the mother and also through linkage analysis of family member & direct characterisation of gene mutations.

Here in our case neurofibromatosis has been transmitted to two generation and the patient's mother died of cerebrovascular stroke during antepartum period which raises the suspicion that NF1 might have complicated the pregnancy. Both the patient and her mother had CPD and many authors recommended CPD complicating pregnancy in patients with known NF as said earlier.

Whether she is either willing to continue or terminate pregnancy, it is our obligation, as an obstetrical team to help her in the best possible way. On the other hand, we are also obliged to make as accurate prenatal diagnosis as possible.[4] Since the disease is causing mutations that are dispersed throughout the gene, prenatal diagnosis usually relies on linkage analysis of family members and rarely on direct characterisation of the mutation. The extreme variability of the phenotypic expression of the NF1 gene makes it very difficult for NF1 families to decide whether to have children or not, as molecular diagnosis cannot predict clinical expression of the disease.[5] Hence the role of genetic testing may be inconclusive in decision making in these cases.

Conclusion

The psychological management of the would-be parents is therefore very sensitive. With this case report, we wanted to describe the diagnostic possibilities and complications related to NF in pregnancy and dilemmas in everyday clinical practice of a gynaecologist teaming up with the physicians in critical decision making for the benefit of both the mother and the new born.

Source of support: None
Conflict of interest: None
Compliance with ethical standards
Human and animal rights – this article does not contain any studies with human participants or animals by any of the authors.

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