NEUROFIBROMATOSIS TYPE 1 AND PREGNANCY

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- Aim:

Neurofibromatosis Type 1 (NF-1) is an autosomal dominant condition which has markedly variable clinical expression, with manifestations ranging from mild cutaneous lesions to severe orthopaedic complications and functional impairment.[1] Aim of this case report is to understand multidisciplinary management of pregnancy in NF-1 women in pregnancy and intrapartum period.

- Case:

This is a case of 38 years old woman, G3P0+2, with spontaneous conception, diagnosed with NF-1 in the past. She suffered from multiorgan involvement with NF-1, with chronic but stable right optic nerve glioma, a suprasellar multiloculated lesion, a benign plexiform neurofibroma of the neck and multiple areolar lesions of breasts.

She was booked under consultant led care and a multidisciplinary plan was made in liaison with fetal medicine, neurosurgery and local anaesthetics. Areolar lesions grew large and became tender during course of pregnancy and required excision at 31 weeks of gestation, however rest of the lesions remained stable as per MRI findings. Serial growth scans revealed normally growing baby. She was deemed suitable for vaginal delivery with no contraindications to active second stage after neurosurgery and anaesthetics review, but regional anaesthesia was advised to be avoided due to potential risk of coning. Amniocentesis revealed baby to be affected. She went into spontaneous labour at 38 weeks and Neville Barnes forceps delivery under pudendal block was achieved due to prolonged second stage. Baby was delivered in good condition and normal Apgars and cord gases. In addition to right mediolateral episiotomy, she needed a second degree vaginal tear repair. Postnatal period was uneventful.

- Discussion:

The current obstetric literature, based on mostly case reports indicates that women with NF-1 have increased complications associated with pregnancy, is probably biased[1], however a few studies have reported higher complication rates. While fertility does not seem to be impaired in neurofibromatosis, these patients experience a higher-than-expected rate of first-trimester spontaneous abortions (20.7%), stillbirths (8.7%) and intrauterine growth retardation (13.0%)[2].Therefore, these pregnancies are high risk and women do need multidisciplinary plan involving maternal fetal medicine, neurosurgery, anaesthetics and other specialities depending upon the location of lesions and will require imaging to assess lesions growth during pregnancy. In view of risk of growth retardation, they should be offered serial growth scans in third trimester. Invasive testing may be required after counselling to assess if fetus is affected. Woman should also be counselled about the option of preimplantation genetic diagnosis (PIGD) as well as in vitro fertilisation (IVF) with donor egg/sperm for future pregnancies.

- References:

1. Dugoff L, Sujansky E. Neurofibromatosis type 1 and pregnancy. Am J Med Genet. 1996 Dec 2;66(1):7-10. doi: 10.1002/(SICI)1096-8628(19961202)66:1<7::AID-AJMG2>3.0.CO;2-R. PMID: 8957502.

2. Weissman A, Jakobi P, Zaidise I, Drugan A. Neurofibromatosis and pregnancy. An update. J Reprod Med. 1993 Nov;38(11):890-6. PMID: 8277488.