NEUROFIBROMATOSIS TYPE 1 AND PREGNANCY: A CASE REPORT

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ABSTRACT

Neurofibromatosis 1 or Recklinghausen’s disease is an autosomal dominant genetic disease that can cause serious maternal and fetal complications, hence the need for good monitoring during pregnancy and postpartum.

KEYWORDS: Neurofibromatosis, Pregnancy, Complications.

INTRODUCTION

Neurofibromatosis 1 or Recklinghausen's disease (NF1, MIM 162200) has been known for more than a century from the name of the German physician, Friedrich Daniel von Recklinghausen, who is the first, described this disease in 1881. It is a common genetic disease that affects 1/4000 to 1/3000 individuals with homogeneous worldwide distribution and an estimated incidence of 1/2500 births.\(^1\) It is a monogenic neurodevelopmental disease, characterized by multisystem symptoms including an increased risk of cognitive impairment (50-70%) and a predisposition to tumor formation.\(^2\) It is one of the most frequent genetic diseases with the autosomal dominant transmission, which induces multivisceral involvement. The NF1 gene encodes a protein, neurofibromin, made up of more than 2,800 amino acids and which is said to be part of the family of enzymes, which hydrolyze GTP (Guanosine triphosphate). The NF1 gene is located on chromosome 17 at position 17q11.2.\(^3\) The incidence during pregnancy ranges from 1/5000 to 1/18500 deliveries.\(^4\) Most of the current obstetric literature indicates that pregnant women with NF1 have an increased risk of complications such as premature labour, spontaneous miscarriage, HTA, preeclampsia, intrauterine growth retardation. However, we
report a case of normal obstetric recurrence despite worsening dermatologic lesions of the condition during pregnancy.

**CASE REPORT**

A 30-year-old gravida 1, para 0 women who had been married for 2 years, presented in the labour ward with 39 weeks pregnancy and labour pains. Having as history the appearance at the age of 14 years of "café au lait" spots, which gradually increased in size and number during pregnancy. His mother and brother also had the same skin lesions. The Patient did not followed who presented to the obstetric emergency room with a clinical examination as follows: Height is 162 cm, weight 80 kg, pulse 86 beats per minute and blood pressure 120/80 mm Hg. No edema of the lower limbs and a negative urine test. On inspection, we note the presence of several rounded "café au lait" spots of varying sizes characteristic of NF1 presents all over the body as well as several nodular lesions. Cardiovascular and respiratory examinations were without abnormalities. Obstetric examination reveals uterine height at 29 cm, positive fetal heart sounds, uterine contractions present. The fetal heart rate is 141 beats per minute. A vaginal examination; the flexible neck in the middle position, dilated to 2cm, 60% erased with intact membranes, mobile cephalic presentation. The emergency obstetric ultrasound showing an evolving mono-fetal pregnancy, positive cardiac activity, in cephalic presentation, postero-fundic placenta, biparietal diameter and length of the femur at term, and an estimate of the fetal weight at 2886g. The labour proceeded under optimal conditions resulting in the vaginal birth of a newborn living at term, birth weight of 2902g female with an Apgar of 10/10. Postpartum follow-up was without abnormalities.

**Figure 1:** multiple small neurofibromas in the skin and a "café au lait" spot.
DISCUSSION

Neurofibromatosis 1 (NF1) or Von Recklinghausen's disease is one of the most common genetic diseases. It is an autosomal dominant disorder and the risk of an individual with the disease passing the disease on to their children is 50%. NF1 is characterized by an extreme variability in its clinical expression, which is also found within the same family. The clinical picture of NF1 most often associates many “café au lait” spots, axillary and inguinal lentigines, skin neurofibromas and Lisch nodules. “Café au lait” spots are among the first manifestations of NF1. Their numbers and sizes change during pregnancy under the influence of hormones. The diagnosis of NF1[6] is made in an individual if two or more of the following criteria are met:

- At least six TCL, greater than 5 mm in their largest diameter in prepubertal individuals, and more than 15 mm in pubescent individuals;
- Two or more neurofibromas of any type or one plexiform neurofibroma;
- Axillary or inguinal “ephelides”;
- A glioma of the optic tract;
- Two or more Lisch nodules (iris hamartomas);
- A characteristic bone lesion such as sphenoid dysplasia, thinning of the cortex of long bones with or without pseudarthrosis;
- First degree relative with NF1 according to the above criteria many authors describe that pregnant women with neurofibromatosis 1 are more likely to have maternal and fetal complications. Such as spontaneous miscarriage, premature labour, pre eclampsia, HELLP Syndrome, and intrauterine growth retardation.[5] Pregnancy, due to increased blood flow, hormonal secretions, and distensions can be complicated by arterial ruptures, bleeding and haemorrhage of the more frequent delivery.

An increased rate of cesarean delivery is also reported, which could be due to acute fetal distress, obstructed presentation, and fetopelvic disproportion due to undiagnosed pelvic neurofibromas, including cases of kyphoscoliosis affecting the lower spine (sequelae of NF1).[7]

Because of poor pregnancy outcome and possibility of transmission to the fetus, Ansari and Nagamani are recommended early termination of pregnancy and sterilization in these women.[8]
However, a literature search found only three studies that suggested that NF1 may not be associated with significant obstetric complications and may result in a normal pregnancy. Clinicians should keep in mind that any woman with neurofibromatosis could have a normal pregnancy and a eutocic delivery.

CONCLUSION

Neurofibromatosis type 1 was associated with increased maternal morbidity in pregnancy. Obstetricians should be aware of the potential for increased antenatal and peripartum complications among women with NF1.

REFERENCES