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Case Report (Open Access)

NEUROFIBROMATOSIS TYPE 1: A FAMILY CASE FROM NIGER REPUBLIC

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ABSTRACT
We present a case of a 47 years old lady affected by Neurofibromatosis type 1. Her family story find that her mother and her grandmother are affected by this pathology. This case raised the issue of treatment and prevention of neurofibromatosis type 1.

Keywords: *Neurofibromatosis, Family, Prevention*

INTRODUCTION

Neurofibromatosis type 1(NF1) or Von Recklinghausen disease is a dominantly inherited disorder which occurs with a frequency of between 1 in 3000 and 1 in 4000. It is characterized by the presence of café au lait spots, peripheral neurofibromas, Lisch nodules, axillary freckling, skeletal dysplasia, and optic gliomas (Ricardi, 1992; Gaughran *et al.*, 1999). The diagnosis is raised on clinical findings, and confirmed by genetic examination. The consensus on the diagnosis criteria is revised on 2020; the treatment is difficult even if some researches are trying to solve the issue.

CASE

The Authors report the case of a lady aged of 47 years with medical history of hyper blood pressure who get married at the age of 24 years old receive at our department for generalized cutaneous nodules. The family story found that her mother and her grandmother have generalized cutaneous nodules. One of her sisters has multiple café-au lait macules. The story is summarized in the family pedigree and our patient is showed by the arrow (figure 1).

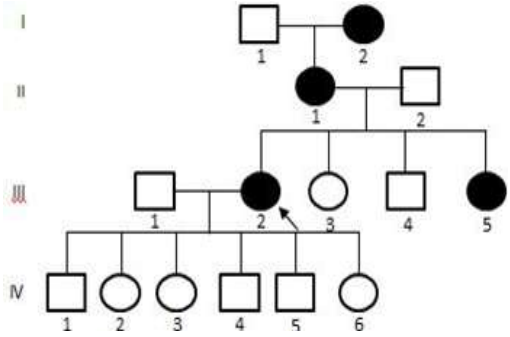

Figure 1: Pedigree of the family

The first nodule appeared on our patient's skin when she was at twenty years old and after the first delivery she noticed the appearance of multiple nodules on the hall body. She consulted a dermatologist who told her that it is a genetic issue and there is no treatment to date.
At the admission in our department the clinical exam found a lady in a good general health status; with multiples nodules on here skin mostly at the back, the face and the chest (figure 2). She has no orthopedic or spine deformities, the neurological exam trying to find cerebral tumors was normal and there were no iris Lisch nodules on eye exams. We schedule to follow her any six months to make sure we don't misdiagnose any complication.

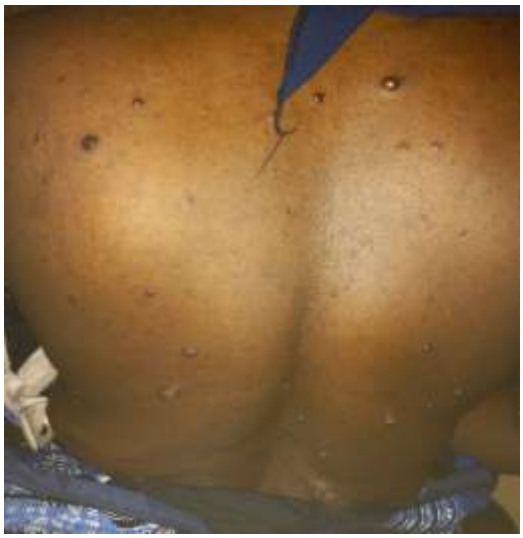

Figure 2: Multiples nodules all over the skin

DISCUSSION

Neurofibromatosis type 1 is an autosomal dominant affection and the penetrance is almost complete at the age of eight years. The NF1 gene, responsible of the disease is localized on the long arm of the 17 chromosome on 17q11.2 (isolated in 1990); its code for neurofibrine which is a protein that has an onco suppression role (Ricardi 1992; Matthias *et al.,* 1998). The inactivation of this gene explains the occurrence of many types of tumors with the disease; For our patient the diagnosis of NF1 is retained because of the presence of more than 15 café au lait spots, nodules and the same lesions in the family with axillary freckling which confirm the variability of the disease inside the same family. The pregnancy is followed by many hormonal modifications responsible of the growth of neurofibromas (Roth *et al.,* 2008). The blood level of steroid hormones, 17bestradiol, testosterone, progesterone increases significantly and decreases since the first days after delivery (Rodriguez-Cuenca *et al.,* 2006; Gardner *et al.,* 2007). A study reported a risk of perinatal complications in NF1, with a higher rate of born death, an intra uterine growth delay and a high rate of caesarean section (Lorraine *et al.,* 1996).
During pregnancy, neurofibromas could grow in size and in number and physicians must be aware of the risk of cord compression in case of development of plexiform neurofibroma.
The treatment is multidisciplinary, mostly by Laser for skin neurofibromas but always disappointing because to date there is no treatment that can stop the disease evolution
The genetic advice before the conception is important for all affected by NF1.

CONCLUSION

The type 1 neurofibromatosis is a genetic disease with dominance autosomal transmission
A good Clinical and radiological evaluation is mandatory to determine the value of potential therapeutic agents.

Conflict of interest: none

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