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RESEARCH ARTICLE

DIAGNOSIS OF NEUROFIBROMATOSIS TYPE 1 WITH DIABETIC KETOSIS

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Abstract

Introduction:-

Neurofibromatosis is a genetic disease, any organ or system of the body may be primarily or secondarily involved , Variable expression results in two major types: Von Recklinghausen's Neurofibromatosis (also known as peripheral NF, or NF1), and Bilateral Acoustic Neurofibromatosis (also known as central NF, or NF2).

The Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare autosomal dominant genetic disorder characterized by the development of multiple noncancerous (benign) tumors of nerves and skin (neurofibromas) and areas of abnormally decreased or increased coloration of the skin. The earliest historical evidence first appeared in the 13th Century, but it was not until Friedrich Daniel von Recklinghausen published his landmark paper in 1882. Beginning in early childhood, patients with NF1 have multiple café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area. These spots increase in size and we report here a case of neurofibromatosis type 1.

A 66 year old woman; followed for hypertension last five years, admitted to the hospital for an inaugural diabetic ketosis, have widespread cutaneous neurofibromas **[FIGURE 1]** with multiple café au lait spots with diameter >1.5 cm **[FIGURE 2]**, and Freckling of the axillaryregion **[FIGURE 3] ,**without skeletal abnormalities, and The mucous membranes were not affected.The disease started in childhood but she was never consulted, her mother has also the same lesions except in the face and hands, the ophthalmological examination shows the lisch nodules in the iris,without clinical visual involvement.**[FIGURE 4]**.

The standard laboratory tests values were in the Normal range. X-ray photography of the long bones was within the normal too.

The diagnosis NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference [our patient presents]:

1. Six or more café-au-lait spots measuring at least 5 mm before puberty or 15 mm after puberty.
2. multiple cutaneous neurofibromas.
3. Axillary freckling.
4. Two or more Lisch's nodule.
5. First degree relative with an NF1 diagnosis.

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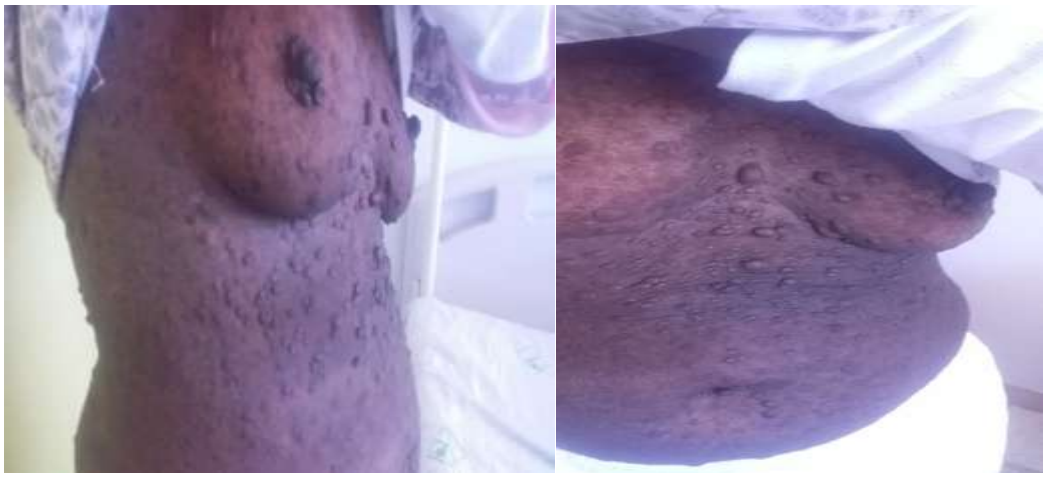

Figure 1:- Multiple Cutaneous Neurofibromas.

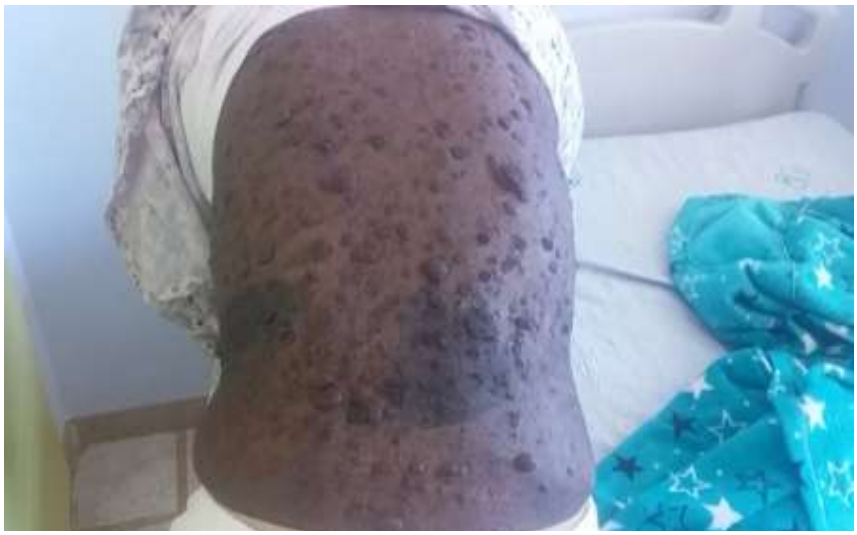

Figure 2:- Café Au Lait Spots.

Figure 3:- Axillary freckling.

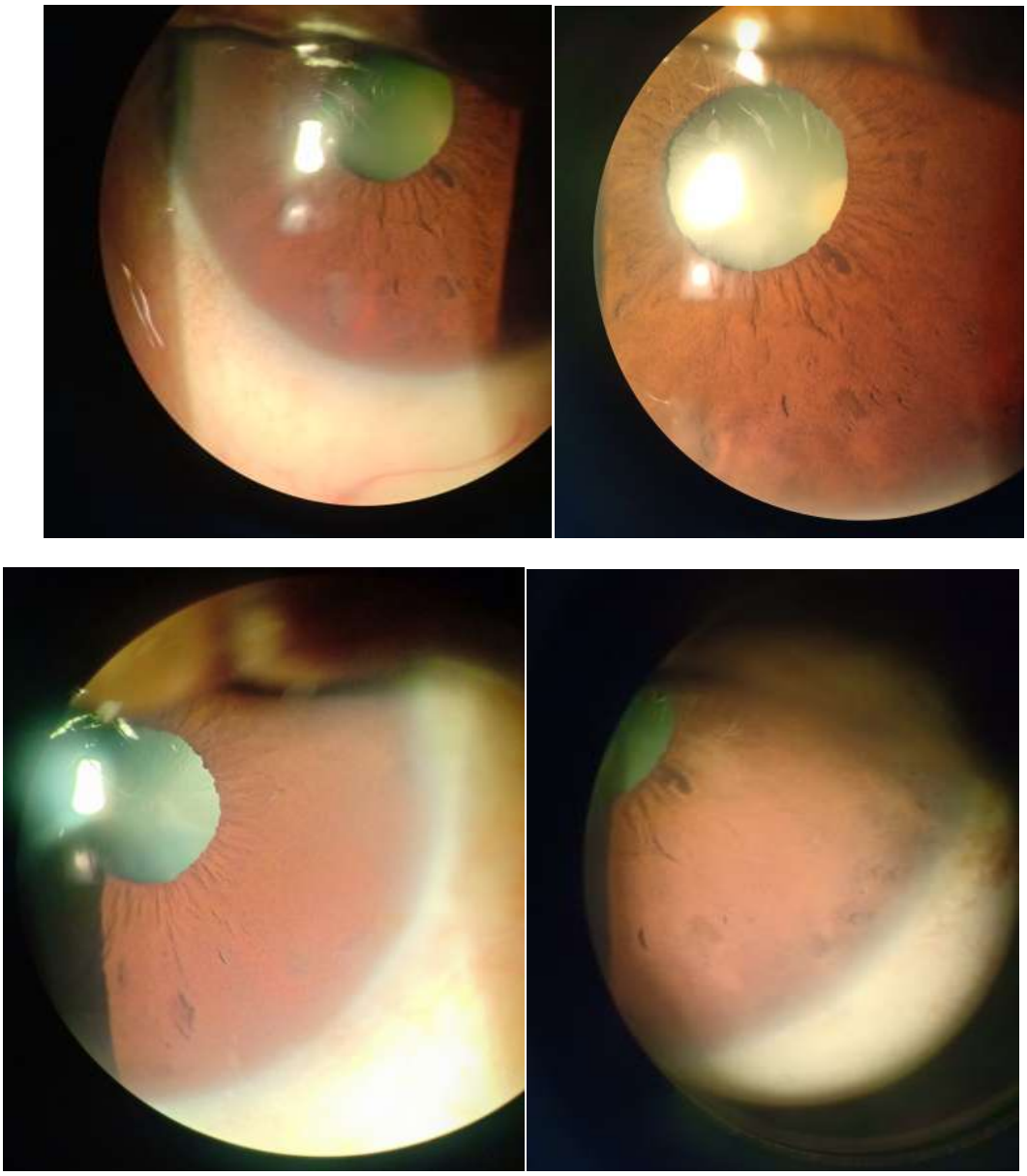

Figure 4:- The lisch nodules in the iris.