VON RECKLINGHAUSEN DISEASE. CASE PRESENTATION

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Abstract

Neurofibromatosis (NF) 1 or von Recklinghausen disease is one of the most common autosomal dominant genetic diseases and is characterized by "café-au-lait" spots and multiple tumors starting from the central and peripheric nervous system.

Clinical case: We present the case of an eight-year-old patient from rural environment first diagnosed with von Recklinghausen disease, based on the presence of three out of the seven diagnostic criteria.

Discussions: The diagnosis is determined on two out of seven criteria: 1) six or more light brown spots larger than 5 mm diameter – pre-puberty or 15 mm diameter – post-puberty; 2) two or more neurofibromas or one plexiform neurofibroma; 3) axillary or inguinal freckling; 4) optic glioma; 5) two or more Lisch nodules; 6) bone injuries: tibia pseudarthrosis or dysplasia of sphenoid wing; and 7) a relative of first degree having NF 1 diagnosis.

Conclusion: The evolution may be different in patients, some having only "café-au-lait" spots and usually considering themselves as being healthy. Interdisciplinary medical examination is essential for a complete diagnosis and setting up a treatment.

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Keywords:

von Recklinghausen disease, neurofibromatosis (NF) 1, café au lait spots, diagnostic criteria.

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Introduction

Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare 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 (1). Beginning in early childhood, patients with neurofibromatosis type 1 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 number as the individual grows older (2).

Clinical case

An eight-year-old girl living in rural area is presented in the Department of Neuropshychometric Rehabilitation of the Emergency Clinical Hospital for Children, Galați, for the screening of neurofibromatosis, after we evaluated two brothers diagnosed with NF 1 and from the anamnesis we found out that the sister has "café-au-lait" spots.

Hereditary-collateral background: mother and two maternal uncles, also showing NF-1

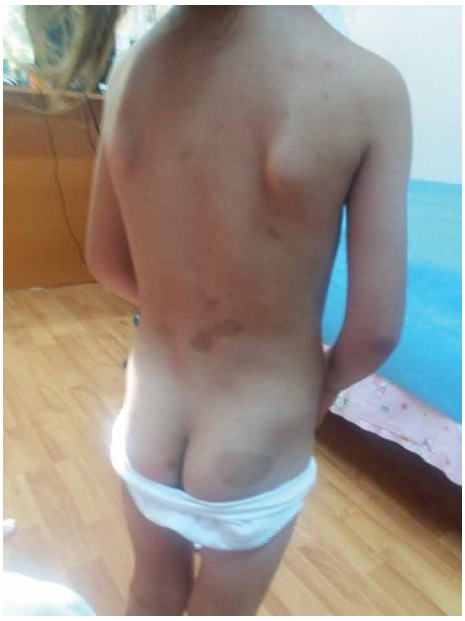

Figure 1. Clinical manifestation of NF 1

cutaneous markers, uninvestigated and undiagnosed; a 13-year-old brother diagnosed in 2015 (10-year-old) with NF 1; a 17-year-old brother diagnosed in 2005 (four-year-old) with NF 1.

Personal pathological history: insignificant, "café-au-lait" spots appeared, affirmatively, after the age of 1 and progressively multiplied, with no other clinical manifestations of the disease.

Clinical exam: humeral imbalance, with right upright shoulder, spine deviation in frontal plane: dorsal – right, lumbar – left; multiple "café-au-lait" spots (over six in number), the largest on the right buttock, 5/4 cm in diameter (Figure 1); axillary freckles; oral breathing.

Based on the presence of three out of the seven diagnostic criteria: 1) multiple "café-au-lait" spots larger than 5 mm diameter (prepubertary); 2) axillary freckles; and 3) two relatives of first degree diagnosed with NF 1 we have diagnosed her with von Recklinghausen disease.

Recommendations: for the detection of possible complications we guided her to neurology, ophthalmology, cerebral MRI, endocrinology and after to return to begin a customized kinetic program.

MRI: focal areas with hypersignal T2 and FLAIR located in the midbrain and internal capsule, bilateral and with a diameter between 7 mm and 13 mm – bilateral hamartomas under and up of tentorium (Figure 2). The right optic nerve is thickened and has a sinuous tract – optic glioma.

Discussions

The disease manifests with an equal frequency and severity for men and women, and the mutant gene located on an autosome can be inherited and transmitted with an equal probability by both

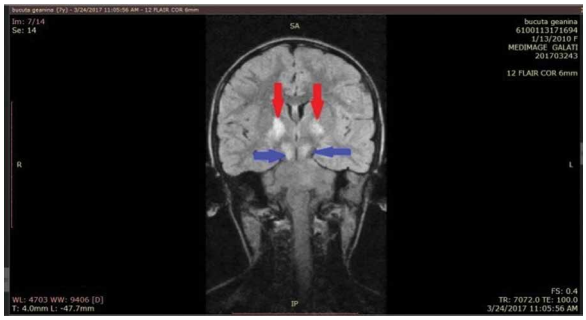

Figure 2. MRI image – Hypersignal Flair midbrain and internal capsule

sexes and independent of race. The disease is determined by a mutation in the NF1 gene, a suppressor gene of growth located on 17q11.2 (3-7). It is one of the genes with the greatest potential to generate mutations in humans. Following this increased susceptibility, more than half of patients diagnosed with neurofibromatosis have no family history of disease, most of the cases being induced by a spontaneous mutation (8).

The diagnosis is determined based on two out of seven criteria (9, 10): 1) six or more light brown spots larger than 5 mm diameter – pre-puberty or 15 mm diameter – post-puberty; 2) two or more neurofibromas or one plexiform neurofibroma; 3) axillary or inguinal freckling; 4) optic glioma; 5) two or more Lisch nodules; 6) bone injuries: tibia pseudarthrosis or dysplasia of sphenoid wing; and 7) a relative of first degree having NF 1 diagnosis (4, 11, 12).

Conclusions

1. The presence of "café-au-lait" spots represents a warning point and allow the easier diagnosis, supported by the presence of other cases in family.
2. The evolution may be different in patients, some having only "café-au-lait" spots and usually considering themselves as being healthy.
3. Interdisciplinary medical examination is essential for a complete diagnosis and setting up a treatment.
4. The complications of this disease may have serious functional consequences and a reserved prognosis.

Conflicts of interest: none declared.
Financial disclosure: none.
Informed consent obtained.

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