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Case Report

Gastrointestinal hemorrhage complicating Recklinghausen neurofibromatosis: a case report

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Abstract: Von Recklinghausen's disease is the most common neuroectodermosis. It is an autosomal dominant disease that affects approximately 1 in 3,000 individuals. It is secondary to the alteration of the NF-1 gene, located on chromosome 17q11.2. Its clinical manifestations are polymorphous in adulthood, with a predominance of cutaneous localization. Digestive localization is rare. This particular location often goes unnoticed, while in extreme cases it can mimic the picture of an abdominal emergency. We report a case of digestive localizations of Von Recklinghausen disease diagnosed after a lightning digestive hemorrhage. This location was intestinal. It seems appropriate to carry out profound digestive explorations during Von Recklinghausen's disease of the adult in order to avoid multiple complications.

Keywords: Gastrointestinal hemorrhage, Recklinghausen neurofibromatosis, rectal bleeding, intestinal neurofibroma.

INTRODUCTION

Von Recklingausen disease or neurofibromatosis type I (NF1) is the most common neuroectodermosis. It is an autosomal dominant disease that affects about 1 in 3,000 individuals, may be sporadic. It involves an abnormality on chromosome 17 (Huson, S.M. 1994). The clinical manifestations are huge and polymorphous, dominated by cutaneous lesions. Lesions of the digestive system are rare, but not exceptional (Sørensen, S. A. et al., 1986; Fuller, C.E., & Williams, G.T. 1991; Cavallaro, G. et al., 2010). We report a case of digestive localization of Von Recklingausen disease revealed by a digestive hemorrhage of great abundance.

OBSERVATION

We report the case of a 63-year-old patient, active chronic smoker with 10 packs / year, followed for a neurofibromatosis type I since the age of 25, without any other particular pathological antecedent, admitted to the emergency for abundant rectal bleeding evolving in a context of hemodynamic instability, without other associated signs. The patient initially required hospitalization in intensive care or received a transfusion of more than 14 packed red blood cells. After hemodynamic stabilization, our patient received a general clinical examination. The abdominal examination is without abnormality. On the other hand, the dermatological examination objectified diffuse milky coffee spots with multiple cutaneous nodules in relation to NF type 1. Upper gastrointestinal endoscopy does not aim for significant abnormality. Colonoscopy found bright red blood without macroscopically visible lesions. The etiologic assessment is completed by a CT angiography which showed a right lateral pelvic mass measuring 65 mm, continuing with the terminal ileum, in the form of a circumferential intestinal thickening, intensely enhanced by the contrast product and containing a large parietal angiogenesis. Due to the persistence of the bleeding, our patient underwent an emergency laparotomy with excision of a 6cm mass of the small bowel, hyper vascularized and prolapsed in the pelvis.The histological study of the surgical specimen concluded that a neurofibroma was part of NF1.

COMMENT

Von Recklingausen disease or neurofibromatosis type I (NF-1) is among the most common autosomal dominant hereditary diseases (Ferner, R.E. 2007). It is secondary to the alteration of the NF-1 gene, a tumor suppressor gene, located on chromosome 17q11.2. The NF-1 gene encodes a cytoplasmic protein that controls cell proliferation

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(Gottfried, O.N. *et al.*, 2010). The diagnosis of NF-1 is based on the presence of at least two of the key clinical criteria established by the National Health Institute (NIH) Consensus Development Conference in 1988 (Ferner, R.E. 2007). The clinical manifestations are often cutaneous, it is the nodular lesions often multiple and sometimes grouped.

Neurofibromatosis type 1 is associated with gastrointestinal involvement in 12 to 60% of cases depending on the series (Melin, M. *et al.*, 1994). The digestive disorders of this disease occur in the middle age of life, usually later than the cutaneous lesions, and can be divided into four entities (Fuller, C.E., & Williams, G.T. 1991): the lesions of the intrinsic digestive nervous system and its supporting tissues; stromal tumors; endocrine tumors of the duodenum and peri-ampullary region; various tumors not classifiable in the preceding categories. Besides the digestive tract, which is most often involved, the liver and pancreas can also be affected (Dewailly, A. *et al.*, 197; Walsh, M.M., & Brandspigal, K. 1989) .

The frequency of intra-abdominal (gastrointestinal) or retroperitoneal manifestations of NF-1 varied considerably in previous studies ranging from 5% to 25%. This variation probably reflects the poor symptoms associated with intra-abdominal manifestations of NF-1 (only 5% are symptomatic). Gastrointestinal manifestations of NF-1 usually occur during midlife or later; usually later than the appearance of cutaneous manifestations of the disease. The main symptoms are pain, palpable abdominal masses, hemoperitoneum, gastrointestinal bleeding and symptoms associated with digestive obstruction (Fuller, C.E., & Williams, G.T. 1991; Basile, U. *et al.*, 2010; Cavallaro, G. *et al.*, 2010).

In the case of gastrointestinal hemorrhage complicating NF-1, hail and jejunum are most often affected (Devereux, R. B. *et al.*, 1975). Degeneration of neurofibromas is estimated at 13%. It is most often sarcomas or neuro-sarcomas (Katz, R. N. *et al.*, 1990). In the case of an acute clinical presentation such as gastrointestinal hemorrhage or obstruction, surgery remains an effective therapeutic method, the only limit is the multiple localization of gastrointestinal involvement, which may require only excision of symptomatic or complicated locations (Fuller, C.E., & Williams, G.T. 1991).

CONCLUSION

The clinical manifestations of type 1 neurofibromatosis are multiple and the severity depends on the timing, extent and number of mutations in the NF-1 gene. The diagnosis of gastrointestinal manifestations of NF-1 is very important in order to avoid complications such as gastrointestinal bleeding; which can be life-threatening.

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