Introduction

Type I Neurofibromatosis (NF1), known as von Recklinghausen disease, is an autosomal-dominant inheritable disorder, with an incidence of 1:3,000, and a prevalence of 1:4,000 to 5,000 (1). The pathogenesis is based on mutations of the *NF1* gene, a tumor suppressor gene encoding a cytoplasmic protein named neurofibromin that controls cellular proliferation (2-7).

Neurofibromas are the most common visceral tumors in NF1 patients. They are composed of Schwann cells, fibroblasts, and myxoid matrix and take origin from a peripheral nerve or a plexus (plexiform neurofibromas). Visceral neurofibromas are often asymptomatic (65%) but when growing in size they may present with pain, palpable abdominal mass, symptoms secondary to bowel obstruction or main vessels compression, and even gastrointestinal bleeding when mucosa or submucosa are involved. In these cases surgery becomes mandatory in order to remove all neoplastic tissue.

The Authors describe a case of a young man affected by NF1 with associated retrocaval abdominal mass with compression and displacement of the inferior vena cava, thus requiring a complex surgical procedure.

**SUMMARY:** Retrocaval mass in patient with von Recklinghausen disease. Case report.

G. CAVALLARO¹, D. CROCETTI², G. PEDULLÀ², S. GIUSTINI², C. LETIZIA³, G. DE TOMA²

**RIASSUNTO:** Massa retrocavale in paziente affetto da malattia di von Recklinghausen. Caso clinico.

G. CAVALLARO, D. CROCETTI, G. PEDULLÀ, S. GIUSTINI, C. LETIZIA, G. DE TOMA

© Copyright 2012, CIC Edizioni Internazionali, Roma
mandatory in order to remove all neoplastic tissue. Plexiform neurofibromas involve paravertebral nervous plexus or many nerve fascicles inside a large-sized nerve, are often bilateral and symmetric, and grow up and invading and displacing surrounding structures, such as paravertebral muscles.

The Authors describe a case of a young man affected by NF1 with associated retrocaval abdominal mass in retroperitoneum, with compression and displacement of the inferior vena cava, thus requiring a complex surgical procedure.

Case report

M.M., 46-year old man, affected by NF1, hypertension and a pelvis bone cyst. Due to chronic low back pain and lower limbs edema, the patient underwent abdominal MRI that revealed a neurogenic tumor about 4.8 cm in diameter, located in right lumbar paravertebral position. The tumor compressed and displaced anteriorly both the inferior vena cava and right renal vein. MRI showed another neurogenic mass (4.6 cm in diameter), located in the pelvis, between internal and external left iliac vessels (Fig. 1 and 2). Laboratory test showed no abnormalities. The patient was admitted to our Department, in order to remove both visceral tumors. Through total midline incision, extensive mobilization of the inferior vena cava (Fig. 3), right renal vessels and right kidney-adrenal were performed with careful division of three posterior lumbar veins and of right adrenal vein, in order to achieve a complete resection of the retrocaval mass. Excision of the left iliac tumor required careful dissection of left ureter and both internal and external iliac vessels. The retrocaval and iliac masses measured respectively about 5.0 and 4.5 cm in diameter (Fig. 4), and showed, irregular shape, greyish colour and elastic consistence.

Postoperative course was uneventful.

At histology, both tumors showed nerve-like fascicular architecture, cells with sinusoidal nucleus leaking chromatin and eosinophilic cytoplasm with indistinct margins. These fascicles were in myxoid or fibrous matrix. Immunohistochemistry revealed positive reaction to anti-neurofilament antibodies.

Discussion

Abdominal manifestations of NF1 include five kinds of tumors: neurogenic tumors (neurofibromas, malignant peripheral nerve sheath tumors [MPNSTs], and ganglioneuromas); neuroendocrine tumors (pheochromocytomas and carcinoids); non-neurogenic gastrointestinal stromal tumors (GISTs); embryonal tumors; and miscellaneous (1, 8). Non-neoplastic associations may also include cardiovascular disorders such as hypertension and a high frequency of congenital heart and vascular diseases (valvular pulmonary stenosis, arterial aneurysms) (9).

Tumors of neuroendocrine origin are gastrointestinal neuroendocrine tumors (carcinoids) and mainly pheochromocytomas. The association between pheochromocytoma and NF1 is well defined and accepted (10). Furthermore, patients with NF1 present an increased risk
to develop GISTs, mesenchymal neoplasms of the gastrointestinal tract arising from the interstitial cells of Cajal of the myenteric plexus (11, 12). In patients with NF1, the incidence of GISTs is increased up to 25 per cent (13); moreover, NF1-associated GISTs present peculiar characteristics compared with sporadic GIST. They usually arise from the small bowel rather than the stomach, are often multiple, present early (median age 50 years), and have a less aggressive behaviour (14-16). The miscellaneous group of tumors related to NF1 are adenocarcinomas, involving the whole gastrointestinal tract (17).

Neurogenic tumors include neurofibromas, ganglioneuromas and MPNSTs. MPNSTs are the most common malignancy in patients with NF1. These tumors can arise from pre-existent plexiform neurofibromas (10%). Ganglioneuromas are benign tumors that originate from sympathetic ganglia and are frequently localized in the paravertebral plexus, in the adrenal glands and rarely, as polyps, in the gastrointestinal tract. Neurofibromas are the most common abdominal and gastrointestinal tumors in NF1 patients. They are composed of Schwann cells, fibroblasts and myxoid matrix and take origin from a peripheral nerve or a plexus (plexiform neurofibromas) (18). The former are well-defined lesions confined to the nerve, the latter are complex and disordered masses involving the entire plexus or multiple fascicles of a large-sized nerve with totally modified architecture (18). Moreover plexiform neurofibromas are exclusive of NF1 and may develop into MPNST.

Neurofibromas are often asymptomatic (65%) but when growing up they may cause symptoms and signs due to compression, displacement or obstruction of surrounding structures, such as small or large bowel. They may present with chronic or acute pain, or gastrointestinal bleeding when the mucosa or submucosa are involved (19, 20). Compression of large abdominal vessels (vena cava, aorta, etc.) seems to be very rare.

Imaging findings depend on tumor localization. On CT scan, neurofibromas appear as smooth, round, or tubular masses homogenously hypoattenuating (21), whe-
In patients with von Recklinghausen disease, retrocaval masses are often detected during surgery for associated tumors. Extraperitoneal neurofibromas are often single and can be localized everywhere in the retroperitoneal space, mainly in the paraspinal position.

Surgical removal, when technically feasible, prevents local infiltration and malignant transformation. However, complete removal is often not possible and involves large vessels or other vital structures. Surgery becomes very demanding and requires technical expertise. In case removal of the retrocaval mass required extensive and careful isolation of the inferior vena cava and right renal vessels and division of posterior lumbar veins.

Early diagnosis of these abdominal manifestations is very important because of the risk of malignancy, organic complications, or hemorrhagic-obstructive complications, such as in the case of tumors of the gastrointestinal tract.

Surgical removal, when technically feasible, prevents local infiltration and malignant transformation. However, complete removal is not always possible and often involves large vessels or other “vital” structures. Surgery becomes very demanding and requires technical expertise. In case removal of the retrocaval mass required extensive and careful isolation of the inferior vena cava and right renal vessels and division of posterior lumbar veins.

Early diagnosis of these abdominal manifestations is very important because of the risk of malignancy, organic complications, or hemorrhagic-obstructive complications, such as in the case of tumors of the gastrointestinal tract.

References


