CASE REPORT – VESICAL PLEXIFORM NEUROFIBROMA IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1

Ana Carolina Bonini Domingos, Barbara Nasr Pinto, Mireille Caroline Silva de Miranda Gomes, Anna Claudia Evangelista dos Santos
Instituto Nacional do Cancer (INCA), Rio de Janeiro - RJ, Brasil
Ana Carolina Bonini <anacarolinabonini@gmail.com>

INTRODUCTION

Neurofibromatosis type 1 (NF1) is an autossomic dominant condition with 100% penetrance and variable phenotipic expression. The prevalence of NF1 is 1:2000 among alive born and its proportion between male:female is 3:1. The signals of the disease are cafe-au-lait spots in the skin, Lisch nodules, bone malformations, neurofibromas and plexiform neurofibromas. Complications like visceral neurofibroma are present in only 1% of all the NF1 cases being more frequent in the GI tract. Considering that the vesical plexiform neurofibromas are extremely rare, we will describe a case of vesical plexiform neurofibroma in a child with NF1.

CASE REPORT

That's a case of a 4yo male patient who firstly presented an important deviation at lumbosacral region, urinary urgency and anal sphincter dysfunction.

The MRI from abdomen and pelvis with contrast revealed dural ectasia and important thickening of the bladder wall with an amorphous image interposed between bladder and rectum.

The CT showed voluminous expansive formation in the pelvis, without a well-defined cleavage plane between rectum and bladder, measuring 10.5 x 5.4 x 4.8 cm; poorly visualized bladder, inferiorly pushed by the lesion.

The abdominal and pelvic US showed distension of the pelvis and ureters, suggesting bilateral hydronephrosis, and presence of infiltrative-looking hyperechoic tissue near the right and posterior wall of the bladder.

Anatomopathological assessment of abdominal fluid, urine and tumor fragments were performed, and showed no malignant cells and confirmed the diagnosis of plexiform neurofibroma.

Physical examination showed walking abnormalities, tumors in the right side of lumbosacral region, cafe-au-lait spots of variable sizes throughout the body, hypertrichosis in the sacral region, mass in the scrotal sac on the right and palpable bladder. Based on the signs and clinical findings the NF1 diagnosis was confirmed. The ophthalmologic evaluation did not demonstrate Lisch's nodules and the bone inventory haven't evidentiated any changes.

The boy was submitted to surgery for the removal of the tumor, but no quimotherapy or radiotherapy was taken.

DISCUSSION AND CONCLUSION

Plexiform neurofibromas are benign tumors of the nerve sheath that involve fascicles and nerve branches; it usually affects upper trigeminal or cervical nerves and is histologically characterized by elongated and diffuse fibroids.

The main sites affected by PNF are head and neck (38%), extremities (22%) and trunk (17%). The involvement of the pelvic region and the genitourinary tract is uncommon and when present, the bladder is the most frequently affected organ followed by the upper urinary tract and genital tract. To date, less than 70 bladder NFP cases have been described in patients with NF1 being 25 cases in children.

The manifestations of the urinary tract neurofibromas are variable, the lower urinary tract symptoms (LUTS) are the most common, being followed by asymptomatic hematuria and urinary tract infections (UTI), as well as asymptomatic peripheral mass. Other manifestations involve urinary retention, frequency and urgency. The majority of patients present hydronephrosis, decreased bladder capacity and thickening of the bladder wall. In long-term disease, urinary complications such as atonia and neurogenic bladder may occur.

Due to the high growth rates of tumors and their origin in the nerve plexuses of the pelvis, bladder and/or prostate, a continuous network is formed that allows the tumor to spread and reach other organs. In our case, the patient already had high urinary tract changes and signs of disseminated disease.

NF1 diagnoses is based on clinical aspects (Table 1) and it´s associated with significant morbidity and mortality in children and adults and tends to present higher mortality rates among patients with associated PNF. In addition, patients with NF1 and PNF have higher frequencies of other tumors related to NF1.

Thus, because NF1 is a very frequent genetic disease, it is necessary to highlight the need of a correct diagnosis, training general practitioners and pediatricians skills to identify the signs and symptoms of the disease. Patients with this genetic disease should be continuously and carefully monitored in order to have diagnosed and treated the conditions that may lead to irreversible damage, such as PNF and malignant tumors.

Table 1: Diagnostic criteria for Neurofibromatosis Type 1.

Diagnostic Criteria for Neurofibromatosis Type 1

Presence of two or more of the following features is considered diagnostic

- Six café au lait spots >5 mm in diameter in prepubertal children or >15 mm in postpubertal children
- Two or more neurofibromas or one plexiform neurofibroma
- Freckling in the axillary or inguinal region
- Optic glioma
- Two or more iris hamartomas (Lisch nodules)
- A distinctive osseous lesion (sphenoid dysplasia or thinning of long bones)
- A first-degree relative with neurofibromatosis type 1

REFERENCES

- Rubenstein AE, Korf BR. Neurofibromatosis. A handbook for patients, families, and health care professionals. 1st ed. New York: Thieme; 1990.
- Korf BR. Plexiform neurofibromas. Am J Med Genet 1999; 89: 31-37.
- Pivnick EK, Riccardi VM. The Neurofibromatoses. In : Freedberg IM, Eisen AZ,Wolff K, Austen KF, Goldsmith LA, Kartz SI, Fitzpatrick TB. Fitzpatrick's Dermatology in General Medicine. New York: Mc graw Hill ; 1999; 2152-2158.
- Listernick R, Charrow J. The Neurofibromatoses. Wolff K, Goldsmith L, Katz S, Gilchrest B, Paller A, Leffell D. Editors: Fitzpatrick's Dermatology in General Medicine, 7th ed New York: Mc graw Hill ;2008. p.1331-1339
- Dombi E, Solomon J, Gillespie AJ, Fox E, Balis FM, Patronas N, et al. NF1 plexiform neurofibroma growth rate by volumetric MRI: relationship to age and body weight. Neurology 2007;68:643-7
- Darrigo LG Jr., Geller M, Bonalumi Filho A, Azulay DR. Prevalence of plexiform neurofibroma in children and adolescents with type I neurofibromatosis. J Pediatr (Rio J) 2007;83:571-3.
- Winfield HN, Catalona WJ. An isolated plexiform neurofibroma of the bladder. J Urol 1985; 134: 542-543
- Cheng L, Scheithauer BW, Leibovich BC, Ramnani DM, Cheville JC, Bostwick DG. Neurofibroma of the urinary bladder. Cancer 1999;86:505e13.
- Scheithauer BW, Santi M, Richter ER, Belman B, Rushing EJ. Diffuse ganglioneuromatosis and plexiform neurofibroma of the urinary bladder: report of a pediatric example and literature review. Hum Pathol. 2008; 39: 1708-1712.

Financial Support: CNPq CAPES

Projeto Gráfico: Setor de Edição e Informação Técnico-Científica / INCA