Abstract

Objective: The prevalence of the disease is 1/3000 newborns; it is more frequent in men than in women with a 3:1 ratio in all races. Genitourinary tract neurofibromas usually arise from the pelvic and bladder nerves, and the prostatic plexus. Bladder is the most frequently affected organ of the urinary tract; bladder neurofibromatosis may present as a diffuse infiltrative process or an isolated neurofibroma. Bladder neurofibromas arise from nervous ganglia of the bladder wall and stain positive for protein S-100 and type IV collagen with immunohistochemical techniques. Methods: We performed a bibliographic review about urinary tract neurofibromas, and specifically of bladder neurofibroma. We report the case of a 45-year-old female consulting for voiding symptoms and recurrent urinary tract infections. Imaging tests showed a mass in the left lateral wall of the bladder and diffuse thickening of the bladder wall. The thickening of the bladder wall is the most characteristic finding in imaging tests, which may also be present in other diseases such as inflammatory pseudotumor and leiomyoma, so that final diagnosis should be achieved by pathologic study. Results: The case is relevant for the absence of previous diagnosis of neurofibromatosis, being bladder involvement its clinical debut. This is why it was difficult to suspect the final diagnosis: the absence of other characteristic clinical manifestations of the disease. Transurethral resection of the tumor was performed and pathologic and immunohistochemical studies offered the final diagnosis. The patient was followed in the urology clinic and also sent to the internal medicine department to rule out other organs involvement of the disease. Conclusions: Neurofibromatosis is a rare systemic disease, and urinary tract involvement is rarer. Bladder is the most frequently involved organ in the urinary tract, generally as a diffuse infiltration or more rarely a solitary tumor. The final diagnosis is pathological and immunohistochemical. Treatment is usually conservative. The patient should be worked up to rule out other manifestations of the disease, and followed to evaluate the development of new lesions.

Keywords

Neurofibromatosis, Bladder, Immunohistochemical study.