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A DIFFUSE FORM OF NEUROFIBROMA OF BLADDER IN A CHILD WITH VON RECKLINGHAUSEN DISEASE*

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Neurofibromatosis in infants is uncommon and involvement of the bladder is rare. The reported bladder lesions are rare, and in such patients include neurofibromas, neurofibrosarcomas and rhabdomyosarcomas. Neurofibromas can be of different histologic types. The histologic type of the about 20 reported bladder neurofibromas in children is not clarified or is stated to be of plexiform type. We describe herein an unusual case of a neurofibroma of bladder in a child with von Recklinghausen disease. The therapeutic management and the possible prognostic implication of the type of the bladder neurofibroma and bladder lesions other than neurofibroma are discussed. *Key words: diffuse neurofibroma, bladder, neurofibromatosis.*

Neurofibromatosis is a rare hamartomatous growth of neural crest derivation that indirectly affects supporting mesenchymal elements. The minimal criteria for the clinical diagnosis of neurofibromatosis are: 1. The presence of neurofibromas (NFs), or multiple cafe-au-lait spots with positive family story, or both; 2. At least six cafe-au-lait spots over 1.5 cm in diameter; 3. Multiple NFs[1, 2]. The involvement of the genitourinary tract or of the urinary bladder is rare. In fact, upon search of literature we have encountered about 60 cases of NFs of urinary bladder in patients with von Recklinghausen (vRH) disease. Only one-third of these cases are reported to occur in children[3-8]. The morphologic forms of these cases are either stated to be of plexiform type or are not clarified at all. We report a rare case of a diffuse form of NF involving the urinary bladder of a 17-month-old male infant with clinically diagnosed vRH disease.

Case Report

A 17-month-old white male infant was evaluated for motor retardation. His medical history included frequent upper respiratory and urinary tract infections.

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On physical examination, his height was at the tenth percentile (77 cm) and weight was at the fifth percentile (10.6 kg). Approximately 10 cafe-au-lait spots were present on his trunk, neck and extremities.

Laboratory studies revealed normal complete blood counts and serum chemistry. Urinalysis demonstrated positive protein,[8-10] neutrophils and few red blood cells per high power field. Urine culture was positive for Pseudomonas aeruginosa.

Further investigation of the patient included abdominal ultrasonography, intravenous urography (IVU), voiding cystourethrography (VCUG), abdominal computed tomography (CT) and TC-99m DMSA that demonstrated left-sided hydronephrosis and hydroureter, irregular thickening of the urinary bladder wall and a retrovesical mass compressing the urinary bladder posteriorly.

Exploratory laparatomy revealed multiple tumor masses involving the wall of urinary bladder which were more prominent in the posterior and superior walls. A partial cystectomy of the superior-posterior wall of the urinary bladder and a bilateral ureterocystostomy were performed. The pathologic examination showed the entire material to be composed of a diffuse form of NF. He was diagnosed as having vRH disease.

One month postoperatively, abdominal ultrasonography showed bilateral hydroureter and hydronephrosis. A cystoscopy at this time revealed complete obstruction of the orifices of both ureters. A bilateral percutaneous nephrostomy was performed. However, right-sided nephrostomy failed to function and approximately one month after this procedure, his serum BUN raised to 22 mg/dl, creatinine was 7 mg/dl and glomerular filtration rate was 46 ml/1.73/m^2. The blood electrolytes were within normal ranges. Abdominal computed tomography (CT) scan showed multiple tumoral masses involving the entire urinary bladder wall. A total cystectomy with ileocecal conduit was performed. The patient's postoperative course was uneventful.

The first partial cystectomy material consisted firm, grayish-white tissue, which appeared to be covered by mucosal tissue on one side, measuring 7 x 5 x 3 cm. The second cystectomy material measured 9 x 7 x 6 cm. The full thickness of almost the entire bladder wall was involved by grayish-white, firm tumor tissue which was bulging into the lumen as multiple, polypoid masses, ranging from 0.5 to 3.5 cm in greatest dimension (Fig. 1). The orifices of both ureters could not be probed. Light microscopic examination of both specimens revealed a diffuse form of neurofibroma, involving the entire thickness of the bladder wall except for the mucosa. The overlaying mucosa was attenuated. The tumor was composed of short, fusiform or round cells which were dispersed in a uniform matrix of fine fibrillary collagen. The tumor contained numerous, large clusters of Wagner-Meissner bodies (Fig. 2). In spite of extensive search, no cytologic atypia or mitotic activity was found.

Fig. 1: The cystectomy material showing multiple masses protruding into the lumen.

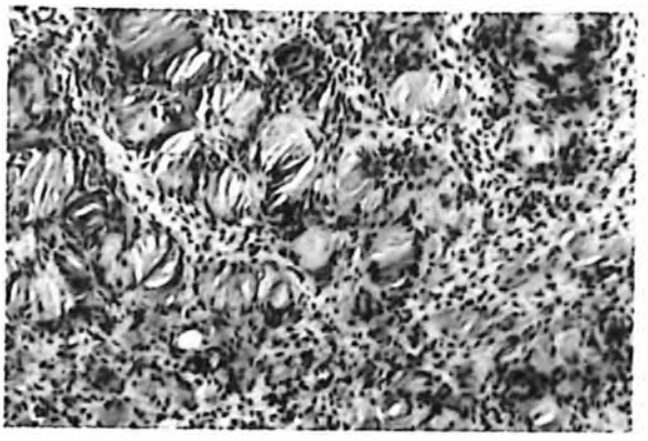

Fig. 2: An area showing diffuse sheets of Wagner-Meissner bodies (HE, 100X).

Discussion

Diffuse NF is a uncommon form of NF that occurs principally in children and young adults and is most common in the skin and subcutaneous tissues of the head and neck region. It is reported to occur in 10 percent of all neurofibromatosis cases. However, the literature on this form is scarce. The tumor has an ill-defined border with an infiltrative growth pattern. The histologic features of diffuse NFs are somewhat different from the plexiform or nonplexiform types of NFs. The tumor cells are short fusiform or round as opposed to the more elongated, wavy cells of the other forms. They are dispersed in a uniform matrix of fine fibrillary collagen. The characteristic feature of this lesion is the presence of tactoid (Wagner-Meissner) bodies[1].

Even though NFs can occur in any site in patients with vRH disease, visceral involvement is not frequent and when it does occur, the gastrointestinal tract is most often affected. Urinary tract involvement is rare and is reported to be of plexiform type in those cases in which the histologic type is clarified.

Although the treatment of bladder NFs is controversial, a conservative treatment with partial cystectomy is recommended for those localized bladder lesions. But some patients with concurrent ureteral, prostatic and urethral involvement require radical prostatectomy with urinary diversion[8]. However, the histologic type of NF may also be an important factor in deciding the extent and, therefore, the cohice of treatment. Even though the tumoral masses of the diffuse form of NF may appear as localized lesions, as was initially seen in our case, they are indeed ill-defined, infiltrative growths. Therefore, it is possible that the residual infiltrative lesion in the remaining bladder wall was responsible for the rapid progression of the tumor after the partial cystectomy in our case. This progressive feature of the illness was the cause of the obstruction of the neo-ureteral orifices.

Upon search of the literature, we found two cases of neurofibrosarcoma in the bladder and nine cases of rhabdomyosarcoma in the genitourinary tract of young children with neurofibromatosis[2, 9, 10]. The site of involvement was cited as the urinary bladder in four of nine cases and was not clarified in the remaining five cases. Since the total number of reported cases of bladder neurofibromas in children with neurofibromatosis does not exceed 22, it appears that sarcomas made up to second most common bladder tumor in those children. Because of the significant difference between the therapeutic management of those patients with bladder neurofibroma and sarcoma and the possible prognostic implication of the type of neurofibroma, it becomes important to obtain tissue confirmation of histologic findings before persuing or withholding any treatment.

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