CONSERVATIVE TREATMENT OF HUGE BLADDER NEUROFIBROMA: A CASE REPORT

A. E. J. Mohd[1], F. A. F. Omar[1], L. K. S. Christopher[1], A. M. G. Khairul[1], S. K. Lai[2]

[1]Department of Urology, Faculty of Medicine, Universiti Putra Malaysia, Seri Kembangan, Selangor, Malaysia
[2]Department of Pathology and Microbiology, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia

Correspondence author: A. E. J. Mohd, Department of Urology, Faculty of Medicine, Universiti Putra Malaysia, Seri Kembangan, Selangor, Malaysia. Telephone: +60195525878
Email: azrul.eimirul@upm.edu.my

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ABSTRACT:
Neurofibroma with involvement of the urinary bladder is a rare condition, that has been reported in less than 80 cases reported worldwide. We present a 22-year-old lady with a known history of childhood neurofibroma type 1. Incidental findings of a huge pelvic mass during laparoscopic surgery for initially thought of a gynae pathology. Further investigation after that with Computed Tomography (CT) scan displayed a bladder mass occupying the abdomen cavity with regards to neurofibroma of the bladder. A core Biopsy of the mass was done and confirmed the pathology. Clinically patient was asymptomatic with the mass. The patient refused surgical intervention. Follow-up for 2 years with serial imaging showed no significant progression of the disease. It is important to determine any Sarcomatoid/malignant changes to decide on further management of bladder neurofibroma.

Keywords: bladder , neurofibroma , neurofibromatosis , NF1 , plexiform neurofibroma

INTRODUCTION:

Neurofibroma of bladder are approximately less than one-third of these cases are in the pediatric population. In the bladder, neurofibromas arise in the nervous ganglia of the bladder wall. Most Neurofibroma of bladder are identified at young age and no exception in our case. Majority of those patient that are symptomatic presented with pain, haematuria, lower urinary tract symptoms or obstruction. It was different in this case where it was incidentally found during gynaecology surgery and patient was asymptomatic. The way mass appeared in CT are worrying of malignancy or possibility presence of sarcomatoid changes. So far to date, surgical removal of bladder mass is the only way to successfully eliminate the risk of possible malignancy changes in the future. Inadequate evidence of other treatment modalities such as radiotherapy and chemotherapy able to salvage of this condition have been reported. However, managing by conservative treatment is considered still good choice as risk of malignant changes are varied from 5-20% from collection of reported case so far. We present a massive plexiform Neurofibroma of bladder, managed by conservative management for up to 2 years following patient decision without evidence of significant changes.

CASE PRESENTATION:

A 22-year-old lady with Neurofibroma type 1 since childhood was under gynecology follow up

for irregular menses. Ultrasound abdominal assessment showed possible right ovarian cyst, 10 cm in the largest diameter, suspected of an ovarian mass. She was scheduled for laparoscopic right oophorectomy by gynecology team. Intraoperatively a huge pelvic mass possibly arising from the bladder was found, and it was attached to the anterior abdominal wall. However reproductive organs were normal in appearance. Urologist was called-in the operating theatre for opinion. The procedure was abandoned in view of uncertainty of the diagnosis and possible dealing with malignant tissue without proper investigation. CT abdomen pelvic was arrange post procedure. Further history, patient had normal urination without any history of lower urinary tract symptoms, haematuria, or history of recurrent urinary tract infection.

Flexible cystoscopy revealed bladder irregular mucosal surface suggesting of bladder neurofibroma with no intravesical mass. Multiple flat sessile lesions generalized scattered over bladder. Biopsy was performed by an interventional radiologist. Core tissue bladder mass was successfully obtained and sent for histopathology. Plexiform bladder neurofibroma was confirmed without malignant or sarcoma features seen. Findings were discussed with patient and offered for surgery partial cystectomy. However, patient was not keen for surgery. Serial imaging follow-up with CT and ultrasound was done for 2 years that showed no signs of progression, suggesting a stable disease. There is no hydronephrosis on both kidneys with renal function urine examination showed normal parameters.

No genetic test was done for the patient. On examination pelvic mass was palpable, however no stigmata of neurofibroma were found on examination externally.

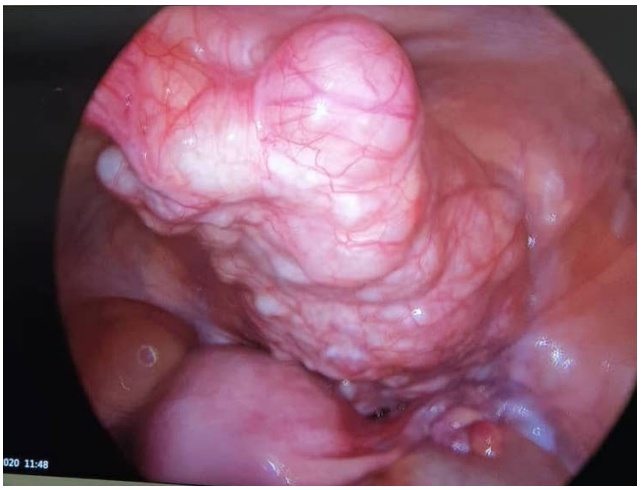

Figure 1: An image taken during laparoscopic surgery showed huge pelvic mass occupying anterior part of pelvic extend superiorly near anterior abdominal wall. Reproductive organ visualised in this image showed normal findings.

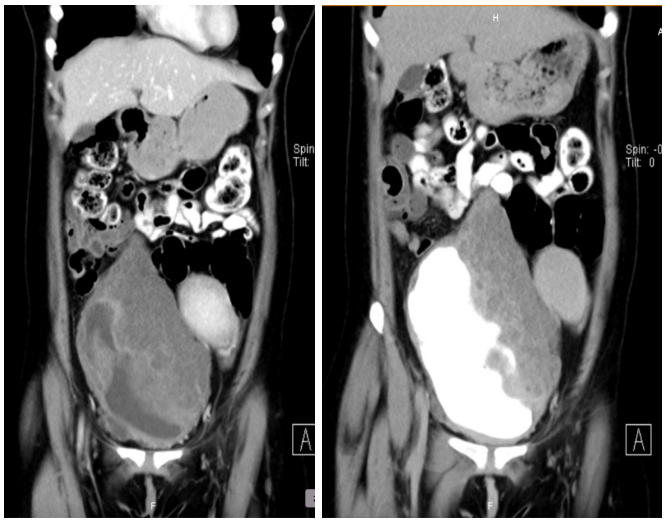

Figure 2: Delayed phase coronal view of CT scan showed huge bladder mass occupying abdomen with Irregular bladder mucosa of bladder.

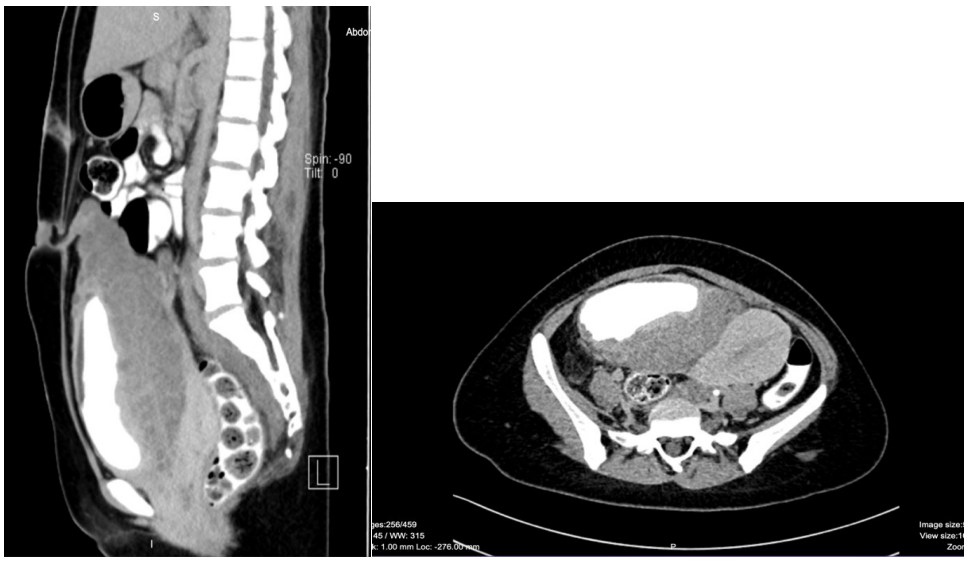

Figure 3: Sagittal and axial view of CT kidney in delayed phase. There is multiple nodule similar density noted in the umbilical region, anterior to bladder and subcutaneous tissue suggestive of neurofibroma.

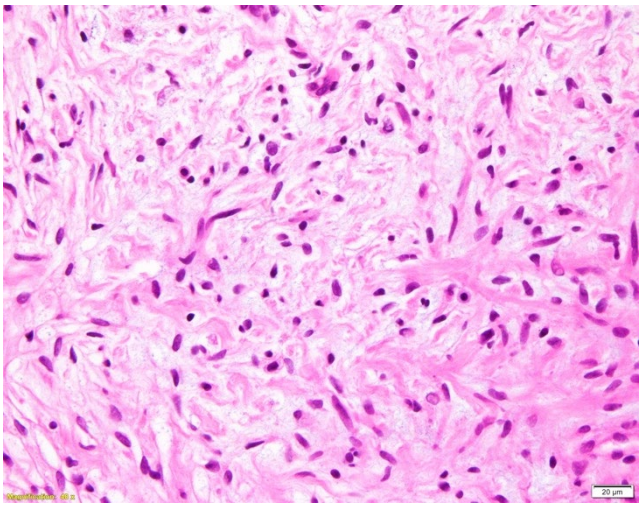

Figure 4: Figure 4 shows histological image of bladder neurofibroma, sample taken using CT guided biopsy at the core of the mass. There are spindle cells proliferation in variable cellularity forming nodular pattern in areas. The spindle cells show mild pleomorphism with wavy nuclei and inconspicuous nucleoli. Mitosis is not observed. No necrosis is noted. Mast cells are present. Immunohistochemical study showed the spindle cells are focally positive for S100. No sarcomatoid changes were seen.

DISCUSSION:

Neurofibromatosis type 1 (NF1) is an autosomal dominant transmitted disease with various clinical manifestations. Neurofibromatosis (NF1) affecting approximately 1 in 3000 individuals. It can be presented with alterations of skin pigmentation, iris Lisch nodules, and multiple benign neurofibromas usually constitute the clinical picture. The genitourinary tract is rarely involved in NF1, and less than 80 cases were reported in the literature to date. [2]

Bladder neurofibroma typically occur in young patients with neurofibromatosis type 1. Neurofibroma of the bladder presents early in life and occurs more often in the setting of generalized disease than as isolated visceral neurofibromatosis. The mean age at diagnosis is reportedly 17 years (range, 1 month to 54 years), and the male-to-female ratio is 2.3:1.

Clinical presentations can be voiding/storage symptoms, flank pain with urinary incontinence. The diagnosis is confirmed by histopathological and immunohistochemical examination. Histologically, the tumors are usually of the plexiform and diffuse type. Histopathologically, they stain positive for protein S100 with immunohistochemical techniques, and this is the pathognomonic pathology finding of schwannomas.

In our case we present a young lady with a known case childhood NF type 1 presented with irregular menses without any urinary symptoms. For bladder neurofibroma possibility of malignant transformation or sarcomatoid transformation is a must to look for. From the literature, it was found out about 5-10% tumors underwent malignant transformation during follow-up; and 12-29% for non-genitourinary involvement. [4] None of these occurred in children. The risk of malignant degeneration changes increases with advancing age [3] and after an operation for benign neurofibromas. [4]

Looking at the CT image and intraoperative laparoscopic findings, the bladder mass was huge with the largest diameter of 15 cm occupying the anterior abdominal wall and pelvic space displacing bowel superiorly and reproductive organ posteriorly. Possible malignant changes need to rule out so possible surgical intervention can be offered. Multidisciplinary discussion with oncology, intervention radiologist (IR) and urology team was conducted. Core biopsy was obtained with help of IR to give more ideas of diagnosis.

The histologic appearance of neurofibroma may simulate a differential diagnosis of low grade malignant peripheral nerve sheath tumour, leiomyoma, low grade leiomyosarcoma and rhabdomyosarcoma. In this case the was absent of mitotic figure, necrotic cell and spindle cell proliferation in nodular pattern suggesting benign findings. Immunohistochemical study shows the spindle cells are focally positive for S100 with presence of Mast cells. This distinctive clinical, histologic and immunohistochemical findings proved a definitive diagnosis. [3]

Treatment of these tumors has included cystectomy, transurethral resection, observation, radiotherapy, chemotherapy, and urinary diversion. Some of patients who presents with symptoms of bladder overactivity were treated with botox intravesical injection after fail treatment of oral anticholinergic.[6]

For malignant transformation or malignant peripheral nerve sheath tumor (MPNST), surgery is the mainstay of treatment. However, oncology treatment such as chemotherapy and radiotherapy are also offered even the outcome is still uncertain. From a meta-analysis that was done by Cai et al that study all reported case of MPNST on different extremities was studied and conclude the prognosis are poor with high chance of local recurrence. Worse prognosis factors mainly associated with NF 1 mutation, large size, deep to fascia, high grade, metastases, and location (trunk and head and neck). [7]

Significant morbidity may be associated with neurofibroma of the bladder. Involvement of the bladder is often extensive, necessitating cystectomy in approximately one-third of cases. For surgical treatment option that available are mainly pelvic exenteration with urinary diversion with formation of ileal conduit.

For conservative treatment, a study with follow up patient for over a mean follow-up of 9.6 years with none of the patients experienced malignant transformation.[1] Our patient was following up for 2 years with regular ultrasound kidney, ureter, bladder surveillance so far did not show any change with symptoms and radiologically. Was offered for surgical treatment in keeping worried of malignant transformation, refused of it in keeping mind of morbidity of the post operation.

No proper suggested tools for follow up assessment of mass has been suggested. From radiological aspects of neurofibromas are characteristic; especially on MRI and they can frequently evoke the diagnosis, which is confirmed by biopsy. We believe MRI could be an excellent tool for non-invasive follow-up [8] as it gives more detail in possible sarcomatoid changes of mass during follow up. No specific guideline in follow up neurofibroma of bladder, in our case we used regular ultrasound in 6 month and yearly CT to look for any significant change in size and symptoms.

CONCLUSION:

Presentation of bladder neurofibroma are rare. In this case with huge bladder mass with mass effect in abdomen are alarming for malignant changes, however no evidence of it on histopathology examination. Surgery treatment was offered however patient preferred to be regular imaging surveillance. Follow up for 2 years showed no significant changes clinically and radiologically.

ACKNOWLEDGEMENT:

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STATEMENT OF ETHICS:

Ethical approval was not required as this is a case report. Informed written consent to participate was provided by all participants.

CONFLICTS OF INTEREST:

The authors declare they have no competing interests.

FUNDING:

Not applicable.

DATA AVAILABILITY STATEMENTS:

The detailed that support the findings of this case report are available from the corresponding

author, [A.E.J.Mohd], upon reasonable request. No additional data than the one presented in this article was used.

REFERENCES:

1. Cheng L, Scheithauer BW, Leibovich BC, Ramnani DM, Cheville JC, Bostwick DG. Neurofibroma of the urinary bladder. Cancer. 1999 Aug 1;86(3):505-13.
2. Üre I, Gürocak S, Gönül II, Sözen S, Deniz N. Neurofibromatosis type 1 with bladder involvement. Case Reports in Urology. 2013 Jul 28;2013.
3. Umakanthan S, Naik R, Bukelo MM, Rai S, Prabhu L. Primary bladder neurofibroma: a rare case with clinical implications and diagnostic challenges. Journal of Clinical and Diagnostic Research: J Clin Diagn Res. 2015 Sep;9(9): ED05.
4. Baugh B, Stencel M, Patel A, Hale N. An isolated case of an incidentally discovered neurofibroma of the urinary bladder. Urol. Case Rep. 2020 Sep 1; 32:101215.
5. Zugail AS, Benadiba S, Ferlicot S, Irani J. Oddities sporadic neurofibroma of the urinary bladder. A case report. Urol. Case Rep. 2017 Sep 1;14:42-4.
6. Cai Z, Tang X, Liang H, Yang R, Yan T, Guo W. Prognosis and risk factors for malignant peripheral nerve sheath tumor: a systematic review and meta-analysis. World Journal of Surgical Oncology. 2020 Dec;18(1):1-2.
7. Dogan GM, Siğirci A, Karaca L. Neurofibromas of the bladder in a child with neurofibromatosis type 1. International braz j urol. 2018 Nov; 44:1256-7.