Adenocarcinoma of exstrophy bladder in a child with gender dysphoria: A rare entity with possible management

D. Bhadoo, M. Bajpai, S. S. Panda, N. Sharma, M. K. Vijay

Department of Paediatric Surgery, Pathology, All India Institute of Medical Sciences, New Delhi-110029, India

Abstract. Bladder exstrophy is a rare congenital defect and is associated with an increased incidence of bladder carcinoma. Majority of these malignancies occur during fourth and fifth decade of life. We report a case of adenocarcinoma of bladder in a 17-year-old female child with unoperated exstrophy bladder with gender dysphoria. This case represents the youngest patient with adenocarcinoma of an unreconstructed exstrophy of bladder.

Key words: Adenocarcinoma, Bladder Exstrophy, Gender Dysphoria

Introduction

Bladder exstrophy is a rare congenital defect with incidence varying between 1 in 10000 to 1 in 50000 live births. It is associated with an increased incidence of bladder carcinoma majority of which occur during 4th and 5th decade of life. We report a rare case of primary adenocarcinoma of the bladder in a 17-year-old female child with unreconstructed exstrophy of bladder.

Case Report

A 17-year-old female child presented with continuous dribbling of urine from lower abdominal mass since birth. She was reared as a male and had gender dysphoria. She also complained of low mood and decreased social interaction. Examination revealed classical exstrophy-epispadias complex with polypoidal bladder mucosa. A normal vaginal orifice was seen along with an anteriorly placed anal opening. Rest of the systemic examination was within normal limits. Investigatory work up revealed a normal female (46, XX) karyotype. Her renal function tests were normal. Hormone analysis showed normal serum T4, TSH, LH, FSH and testosterone. Ultrasonography showed bilateral normal kidneys. Contrast enhanced CT scan of the abdomen showed classical skeletal deformities of exstrophy-epispadias complex with visualization of uterus and bilateral ovaries. It also showed a right ovarian dermoid (Fig. 1). Primary bladder closure was planned. Intraoperative frozen section was sent in view of extensive squamous metaplasia of bladder mucosa which revealed an adenocarcinoma.

Cystectomy with retroperitoneal lymph node dissection, right ovarian dermoid excision plus neobladder reconstruction using gastric patch with appendicovesicostomy and abdominal wall closure was done. Histopathological analysis of the cystectomy specimen was suggestive of adenocarcinoma of bladder exstrophy (Fig. 2). Metastatic work up (chest X-ray, liver function test, contrast enhanced CT of abdomen and pelvis) was negative. Follow up contrast enhanced CT scan of abdomen and pelvis did not show any evidence of residual/recurrent disease. She is disease free at her last follow up 9 months post surgery. She is also under treatment by paediatric psychologist for her gender dysphoria.

Discussion

Bladder exstrophy is a rare midline congenital defect caused by abnormal development of the cloacal membrane. The incidence varies between 1 in 10000 to 1 in 50000 live births. Malignant potential of the extrophied bladder mucosa is well known; 95% are adeno-carcinoma and 3 to 5% are squamous cell carcinomas. It is associated with an increased incidence of bladder cancer with incidence of adenocarcinoma reaching 250 to 800 times the normal population by the age of 40 years. Only about 115 cases of cancer occurring in unreconstructed exstrophy of bladders have been reported. We report a case of adenocarcinoma in a 17-year-old child with unoperated exstrophy bladder with gender dysphoria. This case, to our best knowledge,
represents the youngest reported bladder carcinoma in an unoperated exstrophy of bladder.

We usually do not send frozen sections of bladder exstrophy before or during reconstruction in newborns, infants and children with normal looking bladder mucosa with very less polypoidal and metaplastic changes. As this case was a 17-years female with polypoidal and metaplastic changes of exstrophy bladder and friable bladder mucosa intraoperatively, we decided to send frozen section of bladder wall before reconstruction which came out to be adenocarcinoma. So we strongly recommend frozen section of bladder in older patients with polypoidal and metaplastic changes before reconstructive surgery.

Patients of adenocarcinoma of exstrophy bladder after surgery should be followed up regularly. Metastatic work up includes CECT abdomen and chest, chest X-ray, liver function test. These investigations are also advised during follow up. In primary adenocarcinoma of bladder, surgery is the treatment. Chemotherapy has very less role. These patients should be kept on regular follow up both for outcome of the surgical procedure and metastasis.

**Conclusion**

Adenocarcinoma of unreconstructed bladder exstrophy in childhood is very rare but one should be very vigilant during preoperative work up of these patients. We strongly recommend frozen section of bladder in older patients with polypoidal and metaplastic changes before reconstructive surgery.

Fig. 1. Preoperative CECT pelvis showing pubic diastasis (asterix), bladder plate (white thin arrow), right ovarian dermoid (white thick arrow), vagina (black thick arrow), rectum (black arrow head)

Fig. 2. Photomicrographs (A and B) shows infiltrating glands lined with cells showing nuclear stratification and nuclear atypia along with presence of an occasional mitotic figure at places

**References**