A Rare Case of BRCA2 Squamous Cell Carcinoma
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Introduction

BRCA2 mutations in men have been known to increase the risk of breast and prostatic adenocarcinoma but this case presents the clinical course of a patient with a BRCA2 mutation who presented with squamous cell carcinoma (SCC) of the prostate.

Staging and treatment differs between adenocarcinoma and SCC of the prostate. The prostate is composed of columnar glandular epithelium only and Gleason score is used to stage prostatic adenocarcinoma so the presence of SCC in the prostate posed a unique challenge in the management of this patient.

Case Report

A 56-year-old previously healthy Caucasian male was referred from urology for a 6 week history of having a weak urinary stream which progressed to the point of no urination but denied hematuria, dysuria or nocturia. He was seen by urology due to no urine output and was subsequently catheterized. Past medical history includes hypospadias repair as a child.

Family history includes father deceased from gastric cancer, younger brother with liposarcoma and maternal grandmother with breast cancer.

Trans rectal ultrasound showed nodularity and fine needle aspiration biopsy confirmed SCC in 9 out of 12 biopsies with urothelial differentiation (Fig 1, Table 1).

PET/CT with contrast of the chest, abdomen and pelvis was performed for staging purposes and showed a prostatic mass extending toward the bladder and the presence of multiple peripheral lymph nodes. Biopsy confirmed metastatic SCC. Genetic testing confirmed presence of BRCA 2 mutation.

Treatment & Management

As there is no squamous epithelium in the prostate, the origin was determined to be the prostatic urethra with extension into the prostate (Figure 2).

The patient was started on dose dense Methotrexate, Vinblastine, Adriamycin, Cisplatin (MVAC) with remission of disease. Maintenance immunotherapy using Pembrolizumab (Keytruda) was then used with continuation of chemotherapy (Figure 3) but led to unrelated SCC of the scalp and subsequent treatment with Mohs procedure.

The patient had recurrence of disease 20 months later and treatment with Poly ADP Ribose Polymerase (PARP) inhibitors was recommended given underlying mutation and unique presentation.

Pt. continues to be treated with Niraparib 27 months from diagnosis. He had restoration of normal sexual activity but continued inability to urinate and suprapubic catheter is still in place. Patient is doing well 27 months from diagnosis.

Discussion

Management of metastatic prostatic adenocarcinoma involves Gleason score staging and treatment with LHRH agonist therapy in combination with oral hormonal modulation therapy.

BRCA 2 mutations have been associated with worse prognosis due to greater lymph node involvement and distant metastasis compared to non BRCA 2 mutations. However, these mutations are known to respond well to chemotherapy, although most trials have studied adenocarcinomas.

The patient’s previous history of hypospadias increases the risk of testicular cancer but no evidence exists for an increased risk of urethral/prostatic SCC. Only one case has been reported in the distal urethra in a patient with hypospadias.

Conclusion

This case was rare due to the unusual location and presentation of a BRCA2 cancer along with the delay in symptoms the patient experienced. At presentation, the patient had stage IV metastatic SCC. The prognosis for metastatic SCC is dismal with a median survival of 14 months and a 3 year survival of 9%.

References

2. Klein-Jones, L., Leach, M.O., Reilly, J.A., et al. BRCA2 is a moderate penetrance gene contributing to prostate cancer susceptibility: evidence for genetic testing in prostate cancer patients. JNCI: Journal of the National Cancer Institute. 2011; 103(12); 1243-1254