
Platinum-based chemotherapy is the treatment of choice for metastatic urothelial carcinoma, which is limited by primary and secondary resistance of the tumour. The Cu(2+) transporting beta polypeptide ATPase (ATP7B) is believed to play a role in this resistance. The aim of this study was to screen the ATP7B gene for mutations and loss of heterozygosity in bladder cancer and to evaluate their impact on chemotherapy resistance. DNA extracted from 17 patients with metastatic bladder cancer was analyzed by DNA sequencing, and microsatellite analysis. We found 12 non-synonymous mutations and 20 synonymous mutations out of which 11 and 15, respectively, have not been previously described. Results were correlated with response to platinum-based chemotherapy: 65% of patients exhibited LOH of the ATP7B locus on chromosome 13q14.3, with a tendency to have a better response to chemotherapy. Although resistance is complex, LOH at the ATP7B locus might be useful in predicting chemotherapy response and needs further evaluation.