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Investigating upper urinary tract urothelial carcinomas

Sami Hayek
Cambridge University, UK

Objectives: Evidence of the accuracy of predictive tests in confirming the presence and grade of upper urinary tract urothelial carcinomas (UUTUC) is limited. We present the largest series evaluating the diagnostic value of pre- and intra-operative parameters in the detection of UUTUC.

Materials & Methods: We retrospectively analyzed records of patients who underwent diagnostic ureteroscopy between 2005 and 2014 for suspected UUTUC. Pre-operative workup included voided urine cytology and CT imaging. Intra-operative assessments involved ureteroscopy to directly visualize suspicious lesions, and where possible selective cytology and biopsy. Primary outcomes were the visualization of UUTUC and histopathological confirmation of tumor.

Results: Hundred out of 160 (63 %) patients presenting with suspected upper tract malignancy had UUTUC. Voided and selective urine cytology and CT individually predicted UUTUC with a sensitivity/specificity of 63/67, 76/73, and 95/26 %, respectively. Forty out of 48 (83 %) patients who had abnormal CT and abnormal voided urine cytology had UUTUC, while 100 % of those with normal CT and normal voided cytology (investigated for ongoing symptoms) were normal. Comparing endoscopic biopsy to nephroureterectomy specimen grade, 19 (46%), 18 (44%), and 4 (10%) were identical, upgraded, and downgraded, respectively.

Conclusion: Pre-operative investigations can predict UUTUCs. When these investigations were normal, the risk of UUTUC is negligible. In selective patients with abnormal investigations, ureteroscopy should be performed to confirm and predict the grade of UUTUC, in order to guide future management. Selective cytology is unlikely to significantly contribute to the diagnostic workup of UUTUC

hayek@addenbrookes.nhs.uk

Identification of disease-related genes is a critical step in understanding the molecular basis of disease and developing targeted therapies

Jameela Abdulaziz Kari
King Abdul Aziz University, Saudi Arabia

The genetic study of diseases occurring in the offspring of consanguineous unions is a powerful way to discover new disease genes. Pediatric nephrology provides an excellent example because approximately 70% of cases of kidney disease in childhood are congenital with a likely genetic basis. This percentage is likely to be even higher in countries with a high consanguinity rate, such as the Kingdom of Saudi Arabia. However, there are a number of cMadisonenges, such as cultural, legal, and religious restrictions, that should be appreciated before carrying out genetic research in a tradition-bound country. In this presentation, we discuss the background, opportunities, and cMadisonenges involved with this unique opportunity to conduct studies of such genetic disorders. Keys to success include collaboration and an understanding of local traditions and laws.

jkari@doctors.org.uk