KIDNEY CANCER

BCOR mutations that might help in diagnosis of CCSK

A new paper published in *Nature Genetics* reports the identification of characteristic internal tandem duplications that occur in the *BCOR* gene in clear cell sarcoma of the kidney (CCSK) but not in other renal tumour types.

CCSK, which is responsible for 5% of renal neoplasms in children, is commonly misdiagnosed. “Diagnosis with CCSK ... is difficult owing to its rarity, morphological similarity to other renal tumours and lack of immunohistochemical markers”, commented Ueno-Yokohata *et al.*, the authors of the latest paper.

BCOR has been shown by expression profiling to be highly expressed in CCSK. Using reverse transcriptase PCR and sequencing analysis, Ueno-Yokohata and colleagues identified internal tandem duplications in the 3' part of the exon 16 coding sequence in all 20 of the CCSK tumour samples that they tested.

In addition, the researchers found that none of the 193 non-CCSK paediatric renal tumours that they analysed showed these sequence abnormalities, not even tumours with histologies closest to that of CCSK (congenital mesoblastic nephroma or rhabdoid tumour of the kidney).

“The partial duplication of *BCOR* is markedly specific to CCSK and should assist in the differential diagnosis of pediatric renal tumours,” say the authors.

A single *BCOR* allele is thought to be abnormal in CCSK tumours, which the authors say indicates that *BCOR* duplication might have a key role in CCSK tumorigenesis and could offer a potential therapeutic target.

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Original article Ueno-Yokohata, H. *et al.* Consistent in-frame internal tandem duplications of *BCOR* characterize clear cell sarcoma of the kidney. *Nat. Genet.* doi:10.1038/ng.3338