SMARCB1/INI1 inactivation in renal medullary carcinoma

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Résumé en anglais	**Aims:** Renal medullary carcinoma (RMC), a rare and highly aggressive tumour which occurs in patients with sickle-cell disease, shares many clinicopathological features with collecting duct carcinoma (CDC). The molecular mechanisms underlying RMC and CDC are mainly unknown, and there is ongoing debate about their status as distinct entities. Loss of expression of SMARCB1/INI1, a chromatin remodelling regulator and repressor of cyclin D1 transcription, has been reported recently in RMC. The aim of our study was to investigate if such loss of expression is specific for RMC. SMARCB1/INI1 genetic alterations and cyclin D1 expression were also studied. **Methods and results:** Using immunochemistry, neoplastic cells showed complete loss of SMARCB1/INI1 expression in all six cases of RMC but in only one of 22 cases of CDC. In two RMC cases investigated, comparative genomic hybridization demonstrated complete loss of one SMARCB1/INI1 allele, with no other genomic imbalances, and no mutations were found on the remaining allele. Cyclin D1 was expressed in all RMCs, suggesting that SMARCB1/INI1 inactivation may result in increased cyclin D1 transcription. **Conclusions:** The specific SMARCB1/INI1 inactivation observed in RMCs suggests that RMC and CDC are different entities.
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