# Specific localization of missense mutations in the vhl gene in clear cell renal cell carcinoma

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**Abstract**  
Missense mutations in the VHL gene during sporadic clear cell renal cell carcinoma were studied to evaluate their localization in relation to functionally important motifs of the VHL protein. Somatic mutations were identified in 124 of 307 samples. All missense mutations in the α-domain were localized in the binding site for elongin C. Substitutions in the β-domain (77%) were found in the HIF-binding site. Five missense mutations were absent in these sites, which illustrates their role in VHL protein formation or suppressor function of other protein cofactors. Mutation c.392A→T (p.N131I) was identified for the first time. Our results hold much promise to estimate the boundaries of functionally important sites in the VHL suppressor gene and contribute to the interpretation of a pathogenic role of mutations in direct DNA diagnostics.

KEYWORDS:  
VHL gene; clear cell renal cell carcinoma; somatic mutation

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