DIRC3 (disrupted in renal carcinoma 3)

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**Identity**

HGNC (Hugo): DIRC3
Location: 2q35

**DNA/RNA**

**Description**

The gene spans 3071 bp and contains 12 exons. The last exon contains a consensus polyadenylation site sequence (AGTAA) at 20 nt upstream up the poly(a) addition site. DIRC3 expression could be detected in the placenta, but low expression was found in most tissues and the gene may act as a non-coding RNA.

**Implicated in**

* t(2;3)(q35;q21) and hereditary renal cell cancer

**Disease**

Familial renal cell cancer.

**Cytogenetics**

Disruption of the gene because of the t(2;3) translocation.

**Abnormal protein**

DIRC3-HSPBAP1 is formed by replacing the first coding exon of HSPBAP1 by the first two exons of DIRC3. The fusion transcript most likely encodes a truncated HSPBAP1 protein starting from an internal initiation side embedded in a strong Kozak consensus sequence.

A schematic overview of the breakpoint regions on chromosome 2 and 3 in a family with a t(2;3) translocation with the breakpoint genes DIRC2 and DIRC3, the fusion protein DIRC3-HSPBAP1 and their neighbouring genes. (TNS: tensin; SEMA5B: sema domain 5B).
References


This article should be referenced as such: