DIRC2 (disrupted in renal carcinoma 2)

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Identity
HGNC (Hugo): DIRC2
Location: 3q21

DNA/RNA
Description
The gene spans 73 kb, 9 exons. The first exon and 5-prime UTR contain a CpG island. The gene contains 12 transmembrane segments. It contains a conserved motif, shared with the major facilitator superfamily of transporters, between membrane-spanning domains 2 and 3, and a proline-rich region between membrane-spanning domains 6 and 7. It also contains a putative N-glycosylation site and several putative phosphorylation sites.

Protein
Description
478 amino acids.

Expression
Expression in pancreas, kidney (proximal tubular cells of the kidney), skeletal muscle, liver, lung, placenta, brain and heart.

Function
See below, may be a transporter.

Homology
Computer predictions of the putative DIRC2 protein showed significant homology to different members of the major facilitator superfamily of transporters. DIRC2 shares 43% similarity with the human homolog of feline leukemia virus type C receptor (FLVXR), which has been classified as a major facilitator superfamily transporter, and over 85% homology with Dirc2 from monkey, pig, dog, and mouse.

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.

Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.
References


This article should be referenced as such: