

Gene Section

Short Communication

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.

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