

Gene Section

Short Communication

HSPBAP1 (HSPB (heat shock 27kDa) associated protein 1)

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Identity

Other names: PASS1; FLJ22623; 3_123833568

HGNC (Hugo): HSPBAP1

Location: 3q21.1

DNA/RNA

Description

The HSPBAP1 gene covers 54.67 kb. The gene contains 13 confirmed introns, 12 of which are alternative. In the t(2;3)(q35;q21) translocation, HSPBAP1 was found to be fused to DIRC3.

Protein

Description

488 amino acids.

Localisation

Plasma membrane.

Function

The protein encodes a set of transcription factor jumonji, jmjC family members. The Transcription factor jumonji, jmjC motif, is found in 2 isoforms from this gene. Jumonji protein is required for neural tube formation in mice. There is evidence of domain swapping within the jumonji family of transcription factors. This domain is often associated with jmjN.

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.

Hybrid/Mutated gene

DIRC3/HSPBAP1.

References

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