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Gene Section
Mini Review

SNAI2 (SNAIL homolog 2)

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Identity

Other names: SLUG; SLUGH; Neural Crest Transcription Factor SLUG

HGNC (Hugo): SNAI2

Location: 8q11.21

DNA/RNA

Description

SNAI2 is a neurogenic transcription factor belonging to the SNAIL family implicated in the epithelial-mesenchymal transition and cell survival, in important morphogenetic processes during embryo development and in tumor metastasis. The gene has 3 exons (243bp, 546bp, 1299bp).

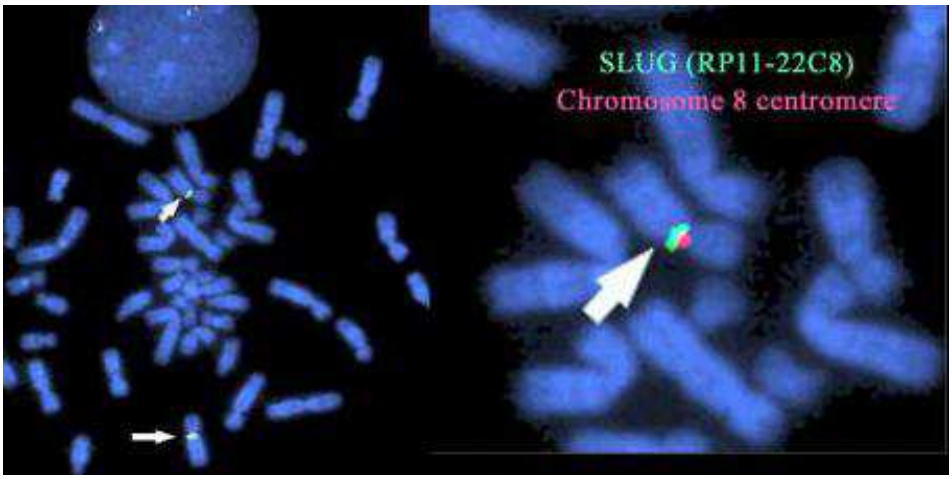
Transcription

Transcript length: 2.2Kb.

Protein

Description

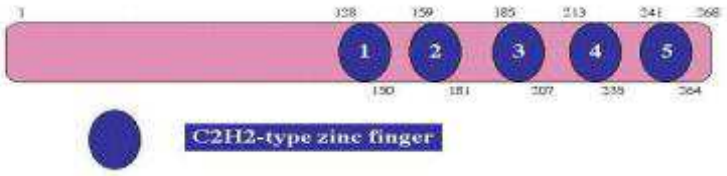
SNAI2 is a zinc-finger transcription factor. Translation length: 268 residues (79bp 1st exon, 546bp 2nd exon, 181bp 3rd exon).
Zinc-finger information: Type: C2H2; Number of domains: 5.
1st(from residue 128 to 150)
2nd(from residue 159 to 181)
3rd(from residue 185 to 207)
4th(from residue 213 to 235)
5th(from residue 241 to 264)



Sequence :

MPRSFLVKKHFNASKKPNYSELDTHTVIISPYLYESYSMPVIPQPEILSSGAYSPITVWTTAAPFHAQL
PNGLSPLSGYSSSLGRVSPPPPSDTSSKDHSGSESPISDEEERLQSKLSDPHAIEAEKFQCNLCNKTYST
FSGLAKHKQLHCDAQSRKSFSCKYCDKEYVSLGALKMHIRTHTLPCVCKICGKAFSRPWLLQGHIRT
HTGEKPFSCPHCNRAFADRSNLRAHLQTHSDVKKYQCKNCSKTFSRMSLLHKHEESGCCVAH

Domains :



Expression

Placenta, adult heart, pancreas, liver, kidney and skeletal muscle.

Localisation

Nuclear (probable).

Function

Transcriptional repressor implicated in the epithelial-mesenchymal transition and cell survival.

Homology

The human SLUG protein is 95, 93, and 88% homologous to mouse, chicken, and Xenopus slug, respectively, but it shows only 47% homology to mouse Snail. The zinc finger region is 100% identical between human and mouse Slug.

Implicated in

Note

SNAI2 is a vertebrate gene encoding a zinc finger protein of the Snail family implicated in the epithelial-mesenchymal transition and cell survival. It was identified in the neural crest and in mesodermal cells emigrating from the primitive streak in chick embryos. It is involved in chick limb development and has conserved and divergent roles in the chick and mouse embryo. Human SNAI2 maps to the long arm of chromosome 8 (8q11.21), contains 3 exons and codes for a protein of 268bp (29KDa) with 5 zinc finger regions. This gene has been identified as downstream target of E2A-HLF oncoprotein and its expression is strongly correlated with loss of E-cadherin. SNAI2 contributes to the function of the stem cell factor c-kit signaling pathway and mediates the radioresistance biological function of the SCF/kit. The alterations of this gene have been associated to different human syndromes.

Disease

Missense mutation has been identified in patients with neural tube defects. Deletions of the SNAI2 gene results in human piebaldism, a cancer prone disease, and has been detected in patients with Waardenburg disease. Duplication of SNAI2 gene is implicated in a rare congenital heart disease (data submitted).

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