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Gene Section
Short Communication

ARID5B (AT rich interactive domain 5B (MRF1-like))

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Abstract

Review on ARID5B, with data on DNA/RNA, on the protein encoded and where the gene is implicated.

Identity

Other names: DESRT, MRF-2, MRF2

HGNC (Hugo): ARID5B

Location: 10q21.2

Note

ARID5B (AT rich interactive domain 5B (MRF1-like)) encodes a possible transcription factor with chromatin remodeling activities. It may be involved in hematopoietic cell development and differentiation (Novershtern et al., 2011). Single nucleotide polymorphisms (SNPs) within this gene are associated with susceptibility to childhood acute lymphoblastic leukemia (Papaemmanuil et al., 2009; Treviño et al., 2009), rheumatoid arthritis (Okada et al., 2012), and systemic lupus erythematosus (Yang et al., 2013).

DNA/RNA

Description

The ARID5B gene is composed of 10 exons spanning ~200 kb on chr 10q21.2.

Two alternative splicing variants have been identified.

Both transcripts share exons 5 to 10 but the short isoform (7032 bp) lacks a portion of the 5' coding region of the long transcript (7948 bp).

Protein

Description

ARID5B belongs to a family of DNA binding proteins characterized by a conserved AT-rich interaction domain (ARID).

The ARID domain, a helix-turn-helix motif, recognizes a core sequence AAT(C/T). Alternatively-spliced transcription variants encoding two isoforms have been identified, and the short isoform has a truncated N-terminal compared to the long isoform.

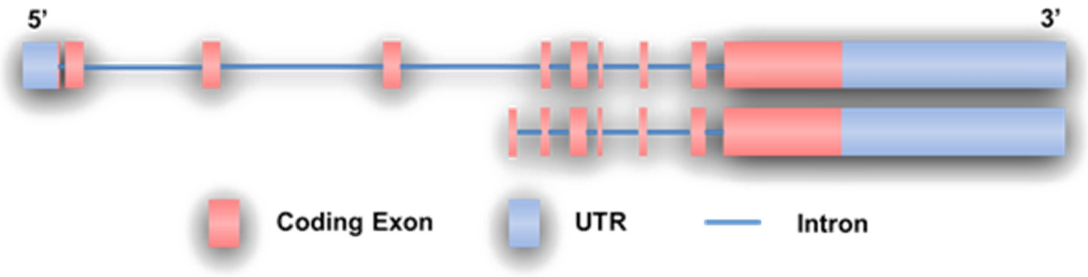

Illustration of transcriptions of human ARID5B gene.

Expression

ARID5B is expressed is in a variety of tissues and cell types, particularly in uterus, smooth muscle, placenta, and CD19+ B cells.

Localisation

ARID5B contains nuclei location signal (NLS) and may primarily function as a transcription regulator within the nucleus.

Function

ARID5B was identified as one of the intermediate early effector genes of the platelet-derived growth factor (PDGF). It forms a histone H3K9Me2 demethylase complex with PHD finger protein 2 and regulates the transcription of target genes involved in adipogenesis and liver development. ARID5B also plays a role in hematopoietic cell development and differentiation, plausibly as a transcription factor.

Implicated in

Acute lymphoblastic leukemia (ALL)

Note

Single nucleotide polymorphisms (SNPs) in this gene (e.g., rs10821936) are strongly associated with susceptibility to childhood ALL, particularly hyperdiploid B-lineage ALL. Also, ARID5B SNPs were also related to ALL relapse, with the disease susceptibility alleles always linked to poorer treatment outcome. However, the molecular mechanisms of how ARID5B contributes to leukemogenesis and relapse remain unclear.

Endometrial carcinoma

Note

Recurrent somatic mutations in ARID5B have also been described in endometrial carcinoma and is particularly enriched in the MSI subtype (23,1%).

Rheumatoid arthritis (RA) and systemic lupus erythematosus (SLE)

Note

SNPs in this gene (e.g., rs10821944 in RA and rs4948496 in SLE) are associated with RA and SLE, both of which are autoimmune diseases but with distinct clinical characteristics.

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