

KIT (updated: old version not available)

Identity

Other names

SCFR (Stem Cell Factor Receptor)

CD117

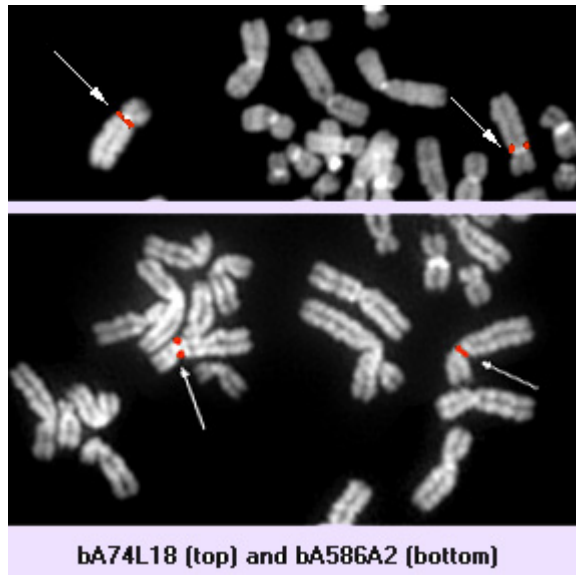
Hugo

KIT

Location

4q12

centromere-PDGFR α -KIT-KDR-telomere



[KIT](#) (4q12) - Courtesy Mariano Rocchi, [Resources for Molecular Cytogenetics](#).
Laboratories willing to validate the probes are welcome : contact rocchi@biologia.uniba.it

DNA/RNA

Description spans 89 kb; 21 exons; size of intron 1 : 37,4 kb

Transcription 5,23 kb mRNA; alternative splicing of exon 9 gives rise to two isoforms, KitA and Kit, that differ by the presence or absence of four aminoacids.

Protein

Description 976 aa: 145 kDa: type III receptor tyrosine kinase: contains an

extracellular domains with 5 Ig-like loops, a highly hydrophobic transmembrane domain (23 aa), and an intracellular domain with tyrosine kinase activity split by a kinase insert (KI) in an ATP-binding region and in the phosphotransferase domain

Expression	hematopoietic stem cells, mast cells, melanocytes, germ-cell lineages and ICCs (Interstitial cells of Cajal).
Localisation	plasma membrane
Function	SCF/ MGF receptor with tyrosine kinase activity; binding of ligand (SCF) induces receptor dimerization, autophosphorylation and signal transduction via molecules containing SH2- domains.
Homology	with CSF-1R, PDGFRb , PDGFRa, and FLT3 .

Mutations

Note	see diagrams: Loss-of-function mutations , and Gain-of-function mutations
Germinal	in piebaldism, and in familial gastrointestinal stromal tumours (see below).
Somatic	in aggressive mastocytosis, mast cell leukemia, ANLL with/without mast cell involvement, myeloproliferative disorders, colon carcinoma and gastrointestinal stromal tumours and germ cell tumors (GCCs)

Implicated in

Entity	Piebaldism .
Disease	autosomal dominant disorder of pigmentation; loss of function abnormalities of the c-kit gene have been demonstrated in 59% of the typical patients.
Entity	Familial gastrointestinal stromal tumours and sporadic gastrointestinal stromal tumours (GISTs).
Disease	GISTs are the most common mesenchymal tumors in the human digestive tract; they originate from kit-expressing cells (ICCs), and often have activating c-kit mutations clustered in the juxtamembrane domain.
Entity	systemic mast cell disease (SMCD)
Disease	mast cell hyperplasia in the bone marrow, liver, spleen, lymph nodes, gastrointestinal tract and skin; gain of function mutations are detected in most patients
Prognosis	depending on the four clinical entities recognized: indolent form, form associated with hematologic disorder, aggressive SMCD and mast cell leukemia; leukemic transformation with mast cell involvement is characterized by rapid progression of disease with a survival time less than 1 year
Oncogenesis	clinical features of malignant hematopoietic cell growth are influenced by the time, the location of c-kit mutative events, and the number of

associated lesions.

Entity	Core binding factor leukemias (ANLL-M2 with t(8;21) (link), (ANLL-M4Eo with inv(16))
Disease	characterized by disruption and loss of CBFa2/AML1 - CBFb/PEBP2b function. Myelomonoblastic leukemia cells are marked by combined positivity for the stem cell antigens CD34, CD117 and high frequency of c-kit mutations (see Figure on CBF leukemia and KIT mutations)

To be noted

loss of expression of c-KIT appears to be associated with progression of some tumors (melanoma) and autocrine/paracrine stimulation of the c-kit/SCF system may participate in human solid tumors such as [lung](#), [breast](#), [testicular](#) and gynecological malignancies.

External links

Nomenclature

Hugo	KIT
GDB	KIT
Entrez_Gene	KIT 3815 v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog

Cards

Atlas	KITID127
GeneCards	KIT
Ensembl	KIT
Genatlas	KIT
GeneLynx	KIT
eGenome	KIT
euGene	3815

Genomic and cartography

GoldenPath	KIT - 4q12 chr4:55218918-55301612 + 4q12 (hg18-Mar_2006)
Ensembl	KIT - 4q12 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	KIT

Gene and transcription

Genbank	AJ438313 [ENTREZ]
Genbank	BC071593 [ENTREZ]
Genbank	X06182 [ENTREZ]
RefSeq	NM_000222 [SRS] NM_000222 [ENTREZ]
AceView	KIT AceView - NCBI
TRASER	KIT Traser - Stanford
Unigene	Hs.479754 [SRS] Hs.479754 [NCBI] HS479754 [spliceNest]

Protein : pattern, domain, 3D structure

SwissProt	P10721 [SRS] P10721 [EXPASY] P10721 [INTERPRO]
Prosite	PS50835 IG LIKE [SRS] PS50835 IG LIKE [Expasy]
Prosite	PS00107 PROTEIN KINASE ATP [SRS] PS00107 PROTEIN KINASE ATP [Expasy]
Prosite	PS50011 PROTEIN KINASE DOM [SRS] PS50011 PROTEIN KINASE DOM [Expasy]
Prosite	PS00109 PROTEIN KINASE TYR [SRS] PS00109 PROTEIN KINASE TYR [Expasy]
Prosite	PS00240 RECEPTOR TYR KIN III [SRS] PS00240 RECEPTOR TYR KIN III [Expasy]
Interpro	IPR007110 Iq-like [SRS] IPR007110 Iq-like [EBI]
Interpro	IPR000719 Prot kinase [SRS] IPR000719 Prot kinase [EBI]
Interpro	IPR001824 RecepttyrkinsIII [SRS] IPR001824 RecepttyrkinsIII [EBI]
Interpro	IPR008266 Tyr_pkinase_AS [SRS] IPR008266 Tyr_pkinase_AS [EBI]
Interpro	IPR009134 VEGFR [SRS] IPR009134 VEGFR [EBI]
CluSTr	P10721
Pfam	PF00047 ig [SRS] PF00047 ig [Sanger] pfam00047 [NCBI-CDD]
Smart	SM00409 IG [EMBL]
Smart	SM00408 IGc2 [EMBL]
Smart	SM00219 TyrKc [EMBL]
Prodom	PD000001 Prot kinase [INRA-Toulouse]
Prodom	P10721 KIT HUMAN [Domain structure] P10721 KIT HUMAN [sequences sharing at least 1 domain]
Blocks	P10721
PDB	1PKG [SRS] 1PKG [PdbSum], 1PKG [IMB]
PDB	1T45 [SRS] 1T45 [PdbSum], 1T45 [IMB]
PDB	1T46 [SRS] 1T46 [PdbSum], 1T46 [IMB]

Protein Interaction databases

DIP	P10721
IntAct	P10721

Polymorphism : SNP, mutations, diseases

[OMIM](#) [164920;273300;601626;606764](#) [[map](#)]

[GENECLINICS](#) [164920;273300;601626;606764](#)

[SNP](#) [KIT](#) [dbSNP-NCBI]

[SNP](#) [NM_000222](#) [SNP-NCI]

[SNP](#) [KIT](#) [GeneSNPs - Utah] [KIT](#) [HGBASE - SRS] [KIT](#) [SNP - HAPMAP]

General knowledge

[Family Browser](#) [KIT](#) [UCSC Family Browser]

[SOURCE](#) [NM_000222](#)

[SMD](#) [Hs.479754](#)

[SAGE](#) [Hs.479754](#)

[Enzyme](#) [2.7.1.112](#) [Enzyme-SRS] [2.7.1.112](#) [Brenda-SRS] [2.7.1.112](#) [KEGG] [2.7.1.112](#) [WIT]

[Amigo](#) [nucleotide binding](#)

[Amigo](#) [receptor signaling protein tyrosine kinase activity](#)

[Amigo](#) [receptor activity](#)

[Amigo](#) [vascular endothelial growth factor receptor activity](#)

[Amigo](#) [ATP binding](#)

[Amigo](#) [protein amino acid phosphorylation](#)

[Amigo](#) [protein amino acid dephosphorylation](#)

[Amigo](#) [signal transduction](#)

[Amigo](#) [transmembrane receptor protein tyrosine kinase signaling pathway](#)

[Amigo](#) [membrane](#)

[Amigo](#) [integral to membrane](#)

[Amigo](#) [transferase activity](#)

[BIOCARTA](#) [Regulation of BAD phosphorylation](#) [[Genes](#)]

[BIOCARTA](#) [Melanocyte Development and Pigmentation](#) [[Genes](#)]

[PubGene](#) [KIT](#)

Other databases

[Other database](#) [Somatic mutation \(COSMIC-CGP-Sanger\)](#)

Probes

[Probe](#) [Cancer Cytogenetics \(Bari\)](#)

[Probe](#) [KIT Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [159 Pubmed reference\(s\) in LocusLink](#)

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URL : <http://AtlasGeneticsOncology.org/Genes/KITID127.html>

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