

GATA1 (GATA binding protein 1 (globin transcription factor1))

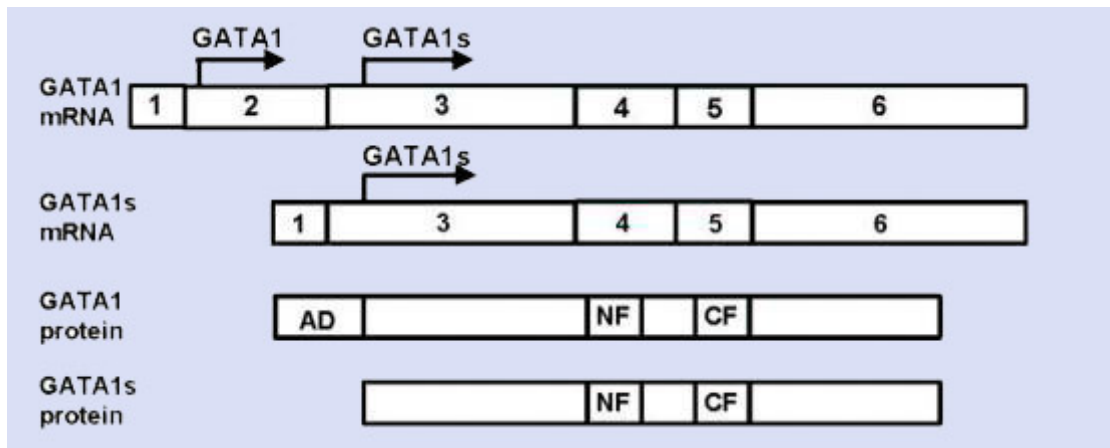
Identity

Other names **ERYF1**
GF1
NFE1
 Hugo [GATA1](#)
 Location Xp11.23

DNA/RNA

Description Genomic sequence 7,757 bases, mRNA six exons (five coding) 1497bp.
 Plus strand

Protein



Alternative models for generation of GATA1 isoforms. The full GATA1 protein can only be translated from the full GATA1 mRNA, whereas the GATA1s protein can be translated either from the full gata-1-mRNA or from the shorter splice variant in which exon 2 is skipped.

Description The full length GATA1 protein is 413 amino acids; 42.7 KDa. However two major protein isoforms are formed by alternative splicing of the mRNA and alternative translation initiation sites as shown in the figure. The shorter GATA1 protein (GATA1s) lacks the first 83 aa. ("The N-terminal activation domain AD"). GATA1s is less active in activation of megakaryocytic promoters. Both proteins contain two Zinc finger domains mediating protein interactions and DNA binding.

Expression Bone-Marrow - Erythroid, Megakaryocytic, Mast and Eosonophilic precursors.

Localisation Nuclear

Function Transcription Factor, essential for erythroid and megakaryocytic development

Homology A member of the family of GATA proteins.

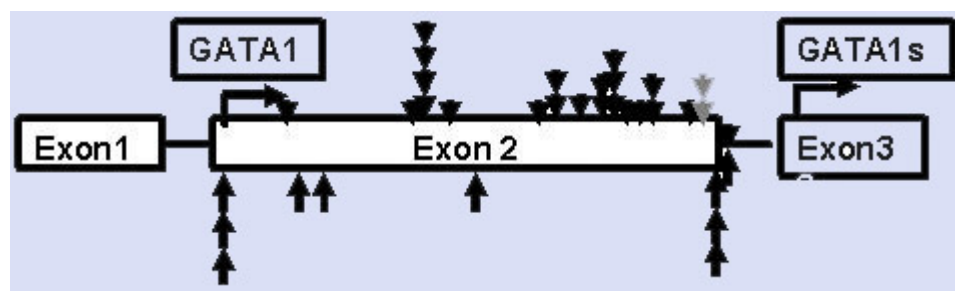
Mutations

Germinal Implicated in germline mutations in the N- Zinc finger domain that mediates the interaction with FOG1 and/or binding to DNA, cause X-linked Dyserythropoietic anemia with thrombocytopenia. The syndrome can be also be manifested only by X-linked macrothrombocytopenia

Somatic see below

Implicated in

Entity Acquired somatic mutations in GATA1 occur in virtually all children with Down Syndrome (DS) and congenital transient myeloproliferative syndrome (TMD) or [acute megakaryocytic leukemia](#) (AMKL, M7-ANLL). The mutations have also been detected in umbilical cord blood of DS patients and in fetal liver of aborted DS embryos. The mutations occur in-utero probably during fetal liver hematopoiesis. They consist of insertions, deletions and base substitution in exon 2 and vicinity and all result in elimination of the full length GATA1 protein with preservation of the GATA1s isoform. The presence of GATA1s in the absence of full length GATA1 blocks megakaryocytic differentiation and promote proliferation of megakaryoblasts. The genes on chromosome 21 that promote this abnormality are unknown. GATA1 mutations are almost always associated with the M7 leukemia in DS although they were also described in a pair of twins with [acquired trisomy 21](#) and in one adult non DS patient with M7. The down-regulations of genes regulated by GATA1 may explain the exquisite sensitivity of DS leukemic blasts to ACA-C.



Legend: Example to the distribution of the mutations in children with M7 and DS described in Rainis et al.

External links

Nomenclature

[Hugo](#)

[GATA1](#)

[GDB](#) [GATA1](#)
[Entrez Gene](#) [GATA1 2623](#) GATA binding protein 1 (globin transcription factor 1)

Cards

[Atlas](#) [GATA1ID40689chXp11](#)
[GeneCards](#) [GATA1](#)
[Ensembl](#) [GATA1](#)
[CancerGene](#) [GATA1](#)
[Genatlas](#) [GATA1](#)
[GeneLynx](#) [GATA1](#)
[eGenome](#) [GATA1](#)
[euGene](#) [2623](#)

Genomic and cartography

[GoldenPath](#) [GATA1 - Xp11.23 chrX:48401210-48408964 + Xp11.23](#) (hg17-May_2004)
[Ensembl](#) [GATA1 - Xp11.23 \[CytoView\]](#)
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]
[OMIM](#) [Disease map \[OMIM\]](#)
[HomoloGene](#) [GATA1](#)

Gene and transcription

[Genbank](#) [AI057349](#) [SRS] [AI057349](#) [ENTREZ]
[Genbank](#) [BC009797](#) [SRS] [BC009797](#) [ENTREZ]
[Genbank](#) [M30601](#) [SRS] [M30601](#) [ENTREZ]
[Genbank](#) [X17254](#) [SRS] [X17254](#) [ENTREZ]
[RefSeq](#) [NM_002049](#) [SRS] [NM_002049](#) [ENTREZ]
[RefSeq](#) [NT_086942](#) [SRS] [NT_086942](#) [ENTREZ]
[AceView](#) [GATA1](#) AceView - NCBI
[TRASER](#) [GATA1](#) Traser - Stanford
[Unigene](#) [Hs.765](#) [SRS] [Hs.765](#) [NCBI] [HS765](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [P15976](#) [SRS] [P15976](#) [EXPASY] [P15976](#) [INTERPRO]
[Prosite](#) [PS00344 GATA_ZN_FINGER_1](#) [SRS] [PS00344 GATA_ZN_FINGER_1](#) [Expasy]
[Prosite](#) [PS50114 GATA_ZN_FINGER_2](#) [SRS] [PS50114 GATA_ZN_FINGER_2](#) [Expasy]
[Interpro](#) [IPR000679 Znf_GATA](#) [SRS] [IPR000679 Znf_GATA](#) [EBI]
[CluSTr](#) [P15976](#)
[Pfam](#) [PF00320 GATA](#) [SRS] [PF00320 GATA](#) [Sanger] [pfam00320](#) [NCBI-CDD]
[Blocks](#) [P15976](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [305371](#) [[map](#)]
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[SNP](#) [NM_002049](#) [SNP-NCI]
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General knowledge

[Family Browser](#) [GATA1](#) [UCSC Family Browser]
[SOURCE](#) [NM_002049](#)
[SMD](#) [Hs.765](#)
[SAGE](#) [Hs.765](#)
[Amigo](#) [process|development](#)
[Amigo](#) [component|nucleus](#)
[Amigo](#) [component|nucleus](#)
[Amigo](#) [process|regulation of transcription from Pol II promoter](#)
[Amigo](#) [process|regulation of transcription, DNA-dependent](#)
[Amigo](#) [function|transcription factor activity](#)
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[BIOCARTA](#) [Hemoglobin's Chaperone](#)
[PubGene](#) [GATA1](#)

Other databases

Probes

[Probe](#) [GATA1 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [37 Pubmed reference\(s\) in LocusLink](#)

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Look AT

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Medline [15630411](#)

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Contributor(s)

Written 02-2005 Shai Izraeli

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URL :

<http://www.infobiogen.fr/services/chromcancer/Genes/GATA1ID40689chXp11.html>

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