

Gene Section

Mini Review

GATA2 (GATA binding protein 2)

Franck Viguié

Laboratoire de Cytogénétique - Service d'Hématologie Biologique, Hopital Hotel-Dieu - 75181 Paris Cedex 04, France (FV)

Published in Atlas Database: January 2009

Online updated version : <http://AtlasGeneticsOncology.org/Genes/GATA2ID44160ch3q21.html>

DOI: 10.4267/2042/44636

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2009 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Identity

Other names: MGC2306; NFE1B

HGNC (Hugo): GATA2

Location: 3q21.3

DNA/RNA

Description

Genomic DNA 13,759bp, 6 exons (one non coding).

Transcription

mRNA 3383bp, transcription is oriented from telomere to centromere. It contains 2 alternative first exons, one distal (IS) specifically transcribed in hematopoietic and neural cells, and a proximal one (IG) transcribed in other cell types.

Protein

Description

Contains 2 zinc finger domains, ZF1 (aa294 to 344) and ZF2 (aa349 to 398).

Expression

Strictly regulated and tissue specific. Gene activity depends on several trans regulators and cis-acting regulatory elements located in the vicinity of the gene.

Localisation

Nuclear.

Function

Binds to the consensus sequence 5'-(A/T)GATA(A/G)-3'. Transcriptional activator

which is expressed very early in hematopoiesis and plays a role in development and regulation of every early pluripotent hematopoietic precursor, but also of non hematopoietic embryonic stem cells. Early stages of erythroid differentiation depends of GATA2, but during maturation GATA2 expression decreases progressively at the benefit of GATA1.

Homology

Member of the GATA family which contains 6 known members; only GATA1, GATA2 and GATA3 are involved in hematopoiesis.

Mutations

Germinal

No known mutation.

Somatic

In CML acute myeloid transformation (see below).

Implicated in

Acute promyelocytic leukaemia

Disease

GATA2 may be involved in APL leukemogenesis by physical interaction with the PML component of PML-RARa fusion or with the variant PLZF-RARa fusion, generated respectively by t(15;17) or t(11;17) translocation.

Myelodysplastic syndrome

Disease

GATA2 is expressed in MDS, but not in normal controls; the frequency of expression increases with the severity of dysplasia (100% in RAEB/RAEB-T).

Myeloid transformation of chronic myeloid leukemia CML

Disease

Out of 85 unselected cases of CML blast transformation, 9 showed a GATA2 mutation: 8 with a T->G substitution at aa359 in ZF2 (L359V) and 1 with a 6 aa deletion (aa 341 to 346) in ZF1. All 9 transformations were myeloid, with a myeloblastic or monoblastic morphology. L359V leads to a gain of function of GATA2 protein.

Aplastic anemia

Disease

Hypothetical. In knockout mice, GATA2 haploinsufficiency leads to a decrease of hematopoietic stem cells number and efficiency. In human, GATA2 mRNA expression is largely reduced in patients with AA.

References

Tsuzuki S, Towatari M, Saito H, Enver T. Potentiation of GATA-2 activity through interactions with the promyelocytic leukemia protein (PML) and the t(15;17)-generated PML-

retinoic acid receptor alpha oncoprotein. *Mol Cell Biol.* 2000 Sep;20(17):6276-86

Fujimaki S, Harigae H, Sugawara T, Takasawa N, Sasaki T, Kaku M. Decreased expression of transcription factor GATA-2 in haematopoietic stem cells in patients with aplastic anaemia. *Br J Haematol.* 2001 Apr;113(1):52-7

Fadilah SA, Cheong SK, Roslan H, Rozie-Hanisa M, Yen GK. GATA-1 and GATA-2 gene expression is related to the severity of dysplasia in myelodysplastic syndrome. *Leukemia.* 2002 Aug;16(8):1563-5

Harigae H. GATA transcription factors and hematological diseases. *Tohoku J Exp Med.* 2006 Sep;210(1):1-9

Wu X, Li Y, Zhu K, Wang Z, Chen S, Yang L. GATA-1, -2 and -3 genes expression in bone marrow microenvironment with chronic aplastic anemia. *Hematology.* 2007 Aug;12(4):331-5

Zhang SJ, Ma LY, Huang QH, Li G, Gu BW, Gao XD, Shi JY, Wang YY, Gao L, Cai X, Ren RB, Zhu J, Chen Z, Chen SJ. Gain-of-function mutation of GATA-2 in acute myeloid transformation of chronic myeloid leukemia. *Proc Natl Acad Sci U S A.* 2008 Feb 12;105(6):2076-81

This article should be referenced as such:

Viguié F. GATA2 (GATA binding protein 2). *Atlas Genet Cytogenet Oncol Haematol.* 2009; 13(12):939-940.
