

**Names for (ISBT\_102) GATA1 Alleles**

General description: This gene encodes a hematopoietic-specific transcription factor that induces high-level expression of adult beta-globin and other erythroid genes. The zinc-finger protein binds to the DNA sequence CCACACCCT found in the beta hemoglobin promoter. Heterozygous loss-of-function mutations in this gene result in the dominant In(Lu) blood phenotype. *GATA1* does not represent a blood group system, but a mutation in this gene is responsible for an X-linked form of Lutheran-mod phenotype, XS2.

General Description (from GeneCards) *GATA1* (GATA Binding Protein 1) is a Protein Coding gene. Diseases associated with *GATA1* include Thrombocytopenia With Beta-Thalassemia, X-Linked and Thrombocytopenia, X-Linked, With Or Without Dyserythropoietic Anemia. Among its related pathways are C-MYB transcription factor network and DREAM Repression and Dynorphin Expression. Gene Ontology (GO) annotations related to this gene include DNA-binding transcription factor activity and chromatin binding. An important paralog of this gene is *GATA2*.

Gene name: *GATA1*

LRG Ref LRG\_559

Number of exons: 6  
 Initiation codon: within exon 2  
 Stop codon: within exon 6

Entrez Gene ID: 2623

LRG sequence: NG\_008846.2 (genomic)  
 NM\_002049.3 (transcript)

Antigens on reference allele n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
Common	<i>GATA1*01</i>						
XS2 Lu-mod	<i>GATA1*BGM01</i>	c.1240T>C	6	p.Ter414Argext*41	PMID: 23278136	none	rs587776456

## References

- PMID 23278136 Singleton BK, Roxby DJ, Stirling JW, Spring FA, Wilson C, Poole J and DJ Anstee. A novel GATA1 mutation (Stop414Arg) in a family with the rare X-linked blood group Lu(a-b-) phenotype and mild macrothrombocytic thrombocytopenia. *Br J Haematol.* 2013. 161(1):139-142.

<b>Track of changes</b>		<b>from version</b>		<b>to version</b>
<b>1</b>	<b>Version</b>			<b>v1.0 30-JUN-2021</b>
<b>2</b>	Author	created:	n.a.	Margaret Keller
<b>3</b>	Review	reviewed:	n.a.	unreviewed
<b>4</b>	General		n.a.	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
<b>5</b>	<b>End Version</b>			<b>v1.0 30-JUN-2021</b>