

## Names for GE (ISBT 020) Blood Group Alleles

### Intro

**General description**      The Gerbich blood group system consists of 13 antigens carried on a single pass type I membrane glycoprotein with called glycophorin C (GPC) and/or glycophorin D (GPD). GPC consists of 128 amino acids and GPD, the shorter isoform, has 107 amino acids. The glycoproteins are encoded by *GYPC*, or *GE* if analysis is to predict a blood group antigen.

**Gene name**                    *GYPC*  
**Number of exons**            4  
**initiation codon:**        Within exon 1 for GPC and within exon 2 for GPD  
**Stop codon:**                Within exon 4  
**Entrez Gene ID:**         2995  
**LRG\_number:**              LRG\_813  
**LRG sequence**             NG\_007479.1 (genomic)  
                                      NM\_002101.4 (transcript)  
                                      NP\_002092.1 (protein)  
**Reference allele**            *GE\*01* (shaded)  
                                      Acceptable: Ge if inferred by haemagglutination  
**Reference allele**            GE2, GE3, GE4, GEPL, GEAT, GETI, GECT, GEAR  
***GE\*01* encodes:**  
**Antithetical antigens:**    n.a.

Phenotype	Allele name	Nucleotide change †	Exon Intron	Predicted amino acid change	Reference (PMID)	Accession number	rs number
GE:2,3,4	<b>GE*01</b>						
Ge:-2,3,4 or Yus type	<b>GE*01.-02.01</b>	NG_007479.1: g.39158_42766del c.60_116del	2 - 3	p.(Ala23_Met41del) §	PMID: 1991173	LN901212	n.a.
	<b>GE*01.-02.02</b>	NG_007479.1: g.37523_41132del c.50- 1625_107-1625del	i1 - i2	p.(Glu17_Ala35del)	PMID: 28272739	LN901213	n.a.
	<b>GE*01.-02.03</b>	NG_007479.1: g.38410_42021del c.50- 738_107-736del	i1 -i2	p.(Glu17_Ala35del)	PMID: 28272739	LN901214	n.a.
	<b>GE*01.-02.04</b>	NG_007479.1: g.37342_40948del c.50- 1806_106+1744del	i1 - i2	p.(Glu17_Ala35del)	PMID: 28272739	LN901215	n.a.
GE:-2,-3,4 or Gerbich type	<b>GE*01.-03.01</b>	NG_007479.1: g.40492_44103del c.106+1288_191-736del	i2- i3	p.(Glu36_Ala63del)	PMID: 1991173 PMID: 18407531	EF434170	n.a.
	<b>GE*01.-03.02</b>	NG_007479.1: g.40117_43777del c.106+913_190+937del	i2- i3	p.(Glu36_Ala63del)	PMID: 28272739	LN901216	n.a.
	<b>GE*01.-03.03</b>	NG_007479.1: g.39347_42996del c.106+143_190+156del	i2- i3	p.(Glu36_Ala63del)	PMID: 28272739	LN901217	n.a.
GE:5 or Wb+	<b>GE*01.05</b>	c.23A>G	1	p.Asn8Ser in GPC	(1), Abstract		rs121912760
GE:6 or Ls(a+)	<b>GE*01.06.01</b>	Duplicated Exon 3	3	in frame duplication	PMID: 7526492		n.a.
GE:6 or Ls(a+)	<b>GE*01.06.02</b>	Tripllicated Exon 3	3	in frame triplication	(2), Abstract		n.a.
GE:7 or An(a+)	<b>GE*01.07</b>	c.67G>T	2	p.Ala23Ser in GPC p.Ala2Ser in GPD ‡	PMID: 8219208		rs774359594
GE:8 or Dh(a+)	<b>GE*01.08</b>	c.40C>T	1	p.Leu14Phe in GPC	PMID: 1413665		rs121912761

Phenotype	Allele name	Nucleotide change †	Exon Intron	Predicted amino acid change	Reference (PMID)	Accession number	rs number
GE:9 or GEIS+	<b>GE*01.09</b>	c.95C>A	2	p.Thr32Asn in GPC p.Thr11Asn in GPD	(3), Abstract		n.a.
GE:-10 or GEPL-	<b>GE*01.-10</b>	c.134C>T	3	p.Pro45Leu in GPC p.Pro24Leu in GPD	(4), Abstract		rs139780142
GE :-11 or GEAT-	<b>GE*01.-11</b>	c.56A>T	2	p.Asp19Val in GPC	(4), Abstract		rs749522569
GE:-12 or GETI-	<b>GE*01.-12</b>	c.80C>T	2	p.Thr27Ile in GPC p.Thr6Ile in GPD	(4), Abstract	LT605061	rs776682317
GE:-13 or GECT-	<b>GE*01.-13</b>	c.59C>T	2	p.Pro20Leu in GPC	(5), Abstract		rs143216051
GE:-14 or GEAR-	<b>GE*01.-14</b>	c.118G>A c.333A>C (non-coding)	3 4	p.Gly40Arg in GPC p.Gly19Arg in GPD	(6), Abstract		rs772372126 rs1050967
Ge:-2,-3,-4, or Leach type (PL)	<b>GE*01N.01</b>	del Exons 3 & 4	i2 - i4	in frame deletion	n.a.		n.a.
Ge:-2,-3,-4, or Leach type (LN)	<b>GE*01N.02</b>	c.131G>T c.134delC	3	p.Trp44Leu p.Pro45Argfs*12	PMID: 2818576 PMID: 1884026		n.a. rs139780142

† Nucleotide changes are based on the *GYPC* transcript

‡ An<sup>a</sup> is only expressed by GPD

§ Because of the similarity of the beginning of exon 2 and exon 3 this deletion has the same effect as p.(Glu17\_Ala35del)

## References

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- Abstract (4) Poole J, Tilly, Hudler P et al. Novel mutations in GYPC giving rise to lack of ge epitopes and anti Ge production. *Vox Sang* 2008; 95(S1), P-324, 181
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- PMID 1884026 Telen MJ, Le van Kim C, Chung A et al. Molecular basis for elliptocytosis associated with glycophorin C and D deficiency in the Leach phenotype. *Blood* 1991; 78(6) 1603-1606
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<b>Track of changes</b>		<b>from</b>	<b>to</b>
<b>1</b>	<b>Version</b>	<b>v4.0 15th January 2020</b>	<b>v4.1 30-NOV-2021</b>
<b>2</b>	Author	created	Peter Ligthart, December 2019
<b>3</b>	Reviewer	checked	Christoph Gassner, January 2020
<b>4</b>	Versioning	updated	updated to newest project-2-format
<b>5</b>	Intro	changed	number of antigens updated to 13
<b>6</b>	Intro	Antigens added	added GECT and GEAR to the antigens encoded by <i>GE*01</i>
<b>7</b>	Allele table	Allele added	added the new alleles <i>GE*1.-13</i> and <i>GE*1.-14</i>
<b>8</b>	References	changed	re-numbered the references
<b>9</b>	References	added	added the references for the two new alleles
<b>10</b>	<b>End Version</b>	<b>v4.0 15th January 2020</b>	<b>v4.1 30-NOV-2021</b>

<b>Track of changes</b>		<b>from</b>	<b>to</b>
<b>1</b>	<b>Version</b>	<b>v3.0 160622</b>	<b>v4.0 15th January 2020</b>
2	Author	created	Peter Ligthart
3	Reviewer	checked	n.a. Christoph Gassner, January 2020
4	General	n.a.	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	LRG ID line added:	n.a. LRG_813
6	Allele Table	Table column and header additions	n.a. Table columns "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.
7	Allele table	n.a.	subdivided <i>GE*01*-02</i> into 4 groups new alleles
8		n.a.	subdivided <i>GE*01*-03</i> into 3 groups new alleles
9	References	reference [3]; PMID: 18407531	original publication of allele
10	Allele table & References	re-numbered the references	top to bottom
<b>11</b>	<b>End Version</b>	<b>v3.0 160622</b>	<b>v4.0 15th January 2020</b>