# Names for GE (ISBT 020) Blood Group Alleles

## Intro

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| General description | The Gerbich blood group system consists of 11 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)<br>NM_002101.4 (transccript)<br>NP_002092.1 (protein) |
| Reference allele | *GE*01* (shaded)<br>Acceptable: Ge if inferred by haemagglutination |
| Reference allele *GE*01* encodes: | GE2, GE3, GE4, GEPL, GEAT, GETI |
| Antithetical antigens: | n.a. |

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

| Phenotype | Allele name | Nucleotide change † | Exon | Predicted amino acid change | Reference (PMID) | Accession number | rs number |
|---|---|---|---|---|---|---|---|
| GE:9 or GEIS+ | *GE*01.09* | c.95C>A | 2 | p.Thr32Asn in GPC<br>p.Thr11Asn in GPD | [9] | | n.a. |
| GE:–10 or GEPL– | *GE*01.–10* | c.134C>T | 3 | p.Pro45Leu in GPC<br>p.Pro24Leu in GPD | [10] | | rs139780142 |
| GE :–11 or GEAT– | *GE*01.–11* | c.56A>T | 2 | p.Asp19Val in GPC | [10] | | rs749522569 |
| GE:–12 or GETI– | *GE*01.–12* | c.80C>T | 2 | p.Thr27Ile in GPC<br>p.Thr6Ile in GPD | [10] | LT605061 | rs776682317 |
| Ge:–2,–3,–4,<br>or Leach type (PL) | *GE*01N.01* | del Exons 3 & 4 | intron 2 - intron 4 | in frame deletion | n.a. | | n.a. |
| Ge:–2,–3,–4,<br>or Leach type (LN) | *GE*01N.02* | c.131G>T<br>c.134delC | 3 | p.Trp44Leu;<br>p.Pro45Argfs*12 | [11]; PMID: 2818576;<br>[12]; PMID: 1884026 | | n.a.<br>rs139780142 |

† Nucleotide changes are based on the *GYPC* transcript

‡ An$^{a}$ is only expressed by GPD

§ Because of the similarity of the beginnin of exon 2 and exon 3 this deletion has the same effect as p.(Glu17_Ala35del)

## References


| | | |
|---|---|---|
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| 3 | PMID: 18407531 | Scott B, Easteal S A single-step assay for the Gerbich-negative allele of glycophorin C Blood Cells. Molecules & Diseases 2008; 41, 1-4 |
| 4 | abstract | Telen MJ, Le Van Kim C, Guizzo ML et al. Erythrocyte Webb-type glycophorin C variant lacks N-glycosylation due to an asparagine to serine substitution. Am J Hematol 1999; 37(1), 51-2 |
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| 6 | congress communi-cation | Uchikawa M Rare blood group variants in Japanese. 10th Regional Congr Int Soc Blood Transfus Western Pacific Region 1999; 198-201 |
| 7 | PMID: 8219208 | Daniels G, King MJ, Avent ND et al. A point mutation in the GYPC gene results in the expression of the blood group Ana antigen on glycophorin D but not on glycophorin C: further evidence that glycophorin D is a product of the GYPC gene. Blood 1993; 82(10) 3198-3203 |
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| 9 | abstract | Yabe R, Uchikawa M, Tuneyama H et al. IS: a new Gerbich blood group antigen located on the GPC and GPD. Vox Sang 2004; 87(S3) P 17.52, 79 |
| 10 | abstract | 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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| 12 | 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 |

| **Track of changes** | | **from v3.0 160622** | **to v4.0 15th January 2020** |
|---|---|---|---|
| | created | Peter Ligthart | Peter Ligthart, December 2019 |
| | checked | n.a. | Christoph Gassner, January 2020 |
| General | | n.a. | First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created. |
| Intro | LRG ID line added: | n.a. | LRG_813 |
| 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. |
| Allele table | n.a. | subdivided *GE*01*-02* into 4 groups | new alleles |
| | n.a. | subdivided *GE*01*-03* into 3 groups | new alleles |
| References | | reference [3]; PMID: 18407531 | original publication of allel |
| Allele table & References | | re-numberd the references | top to bottom |

| **End of changes** | **from v3.0 160622** | **to v4.0 15th January 2020** |
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