

## Names for Er (ISBT 044) Blood Group Alleles

### Intro

General description: The Er blood group system consists of five antigens carried on Piezo Type Mechanosensitive Ion Channel Component 1 (Piezo1). The protein comprises 2521 amino acid residues, 36 transmembrane domains, and one large extracellular domain (amino acids 2198 to 2431). Piezo1 is an important non-selective cation channel in a variety of tissues. The protein is encoded by a highly polymorphic single gene *PIEZO1*, associated with pathogenic mutations (e.g. dehydrated hereditary stomatocytosis, generalized lymphatic dysplasia, hemolytic anemia).

Gene name: *PIEZO1*

Number of exons: 51

Initiation codon: Within exon 1

Stop codon: Within exon 51

Entrez Gene ID: 9780

LRG: LRG\_1137

LRG sequence: NG\_042229.1 (genomic)

NM\_001142864.4 (transcript)

NP\_001136336.2 (protein)

Reference allele: *ER\*01* (shaded)

Acceptable: *ER\*A*, if inferred by haemagglutination

Reference allele *ER\*01*  
encodes: ER1, ER3, ER4, ER5

Antithetical antigens: [ER1 ER2]

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	Reference (PMID)	Accession number	rs number
Er(a+b-), Er3+, ERSA+, ERAMA+ ER:1,-2,3,4,5	<i>ER*01 or ER*A</i>					NM_001142864.4	
Er(a-b+) ER:-1,2	<i>ER*02 or ER*B</i>	c.7180G>A	50	p.Gly2394Ser	PMID: 36122374	OM914592.1	rs201950081
Er(a-b-), Er3-, ERSA+, ERAMA+ ER:-1,-2,-3,4,5	<i>ER*01.-03</i>	c.7174G>A	50	p.Glu2392Lys	PMID: 36122374	OM914597.1	rs528448732
ERSA- ER:-4	<i>ER*01.-04.01</i>	c.7219G>C	50	p.Glu2407Gln	PMID: 36122374	OM914593.1	rs200291894
ERSA- ER:-4	<i>ER*01.-04.02</i>	c.7219G>A	50	p.Glu2407Lys	PMID: 36122374	OM914594.1	rs200291894
ERAMA- ER:-5	<i>ER*01.-05</i>	c.6734G>A	46	p.Arg2245Gln	PMID: 36122374	OM914595.1	rs2290901
Null alleles							
Er(a-b-), Er3-, ERSA-, ERAMA- ER:-1,-2,-3,-4,-5	<i>ER*01N.01</i>	c.5289C>G	38	p.Tyr1763Ter	PMID: 36122374	OM914596.1	rs72811487

## References

- PMID 36122374 Karamatic Crew V, Tilley LA, Satchwell TJ, AlSubhi SA, Jones B, Spring FA, Walser PJ, Martins Freire C, Murciano N, Rotordam MG, Woestmann SJ, Hamed M, Alradwan R, AlKhrousey M, Skidmore I, Lewis S, Hussain S, Jackson J, Latham T, Kilby MD, Lester WA, Becker N, Rapedius M, Toye AM, Thornton NM. Missense mutations in *PIEZO1*, encoding the Piezo1 mechanosensor protein, define the Er red blood cell antigens. *Blood* 2022; blood.2022016504. doi: 10.1182/blood.2022016504.

## Track of changes

<b>1</b>	<b>Version</b>		<b>v1.0 31-DEC-2022</b>
<b>2</b>	Author	created	Vanja Crew, December 2022
<b>3</b>	Reviewer	reviewed	Nicole Thornton, December 2022
<b>4</b>	General	Document created	Spread-sheets "Intro", "Allele Table", "References" and "Versioning" created
<b>5</b>	<b>End Version</b>		<b>v1.0 31-DEC-2022</b>