



Alelos del sistema ABO (ISBT 001)

El sistema ABO se descubrió como en 1900 y se considera el primer sistema clínicamente más importante.

El gen ABO y sus 7 exones codificadores dan lugar a una de dos glicosiltransferasas principalmente diferentes.

La α glicosiltransferasa (GTA) cataliza la adición de un sustrato donante, UDP-N-acetilgalactosamina, a un sustrato aceptor conocido como el antígeno H.

La β glicosiltransferasa (GTB) difiere en solo cuatro sustituciones de aminoácidos de GTA y realiza la misma reacción enzimática, pero utiliza UDP-galactosa como sustrato donante.

De esta manera, el polimorfismo genético da lugar a dos antígenos relacionados en este sistema. Por lo tanto, cualquier polimorfismo o mutación que cambie la actividad o la especificidad de la enzima codificada puede alterar el fenotipo ABO.

Las alteraciones que eliminan por completo la actividad enzimática dan lugar al fenotipo del grupo sanguíneo O, en el que el antígeno H permanece sin convertir y no se puede detectar ningún antígeno A o B.

Si la alteración genética disminuye la actividad de la enzima, o altera su ubicación subcelular y, por lo tanto, disminuye la conversión de H a A o B, puede producirse un fenotipo de subgrupo A o B débil.

Además, ciertos polimorfismos dan como resultado enzimas promiscuas que pueden sintetizar los antígenos A y B, dando como resultado los denominados fenotipos cisAB o B (A).

El fenotipo A se divide en A1 y A2. El primero es más frecuente en todas las poblaciones y tiene aproximadamente 5 veces más epítomos A por glóbulo rojo. El GTA1 también es mejor que el GTA2 para sintetizar ciertas formas de A, por ejemplo. Un tipo 3 y 4.

Además de los antígenos A y B, se incluyen otros dos antígenos en el sistema ABO, a saber, A₁, B y A₁.

El primero es un epítipo conjunto en el antígeno A o B y, por lo tanto, está presente en los fenotipos A, B y AB. La naturaleza bioquímica exacta del antígeno A₁ ha sido más controvertida, pero se ha propuesto que represente A tipo 4.

Nombre del Gen: *ABO*

Número de exones: 7



Bases moleculares asociadas a fenotipos débiles A1 y A2

Fenotipo	Nombre de alelo	Cambio de nucleótido	Exón	Cambio de aminoácido
A ₁	ABO*A1.01			
A ₁	ABO*A1.02	c.467C>T	7	p.Pro156Leu
A ₂	ABO*A2.01	c.467C>T; c.1061delC	7	p.Pro156Leu; p.Pro354Argfs*23
A ₂	ABO*A2.02	c.1054C>T	7	p.Arg352Trp
A ₂	ABO*A2.03	c.1054C>G	7	p.Arg352Gly
A ₂	ABO*A2.04	c.297A>G; c.526C>G; c.657C>T; c.703G>A; c.771C>T; c.829G>A	6 7	p.Arg176Gly; p.Gly235Ser; p.Val277Met
A ₂	ABO*A2.05	c.467C>T; c.1009A>G	7	p.Pro156Leu; p.Arg337Gly
A ₂	ABO*A2.06	c.1061delC	7	p.Pro354Argfs*23
A ₂	ABO*A2.07	c.539G>C	7	p.Arg180Pro
A ₂	ABO*A2.08	c.467C>T; c.539G>C	7	p.Pro156Leu; p.Arg180Pro
A ₂	ABO*A2.09	c.467C>T; c.527G>A; c.1061delC	7	p.Pro156Leu; p.Arg176His; p.Pro354Argfs*23
A ₂	ABO*A2.10	c.268T>C; c.467C>T	6 7	p.Trp90Arg; p.Pro156Leu
A ₂	ABO*A2.11	c.266C>T; c.467C>T	6 7	p.Pro89Leu; p.Pro156Leu
A ₂	ABO*A2.12	c.190G>A; c.527G>A; c.1061delC	4 7	p.Val64Ile; p.Arg176His; p.Pro354Argfs*23
A ₂	ABO*A2.13	c.467C>T; c.742C>T	7	p.Pro156Leu; p.Arg248Cys
A ₂	ABO*A2.16	c.106G>T; c.188G>A; c.189C>T; c.467C>T; c.1061delC	3 4 7	p.Val36Phe; p.Arg63His; p.Pro156Leu; p.Pro354Argfs*23



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

A ₂	ABO*A2.17	c.407C>T; c.467C>T	7	p.Thr136Met; p.Pro156Leu
A ₂	ABO*A2.18	c.467C>T; c.722G>A	7	p.Pro156Leu; p.Arg241Gln
A ₂	ABO*A2.19	c.467C>T; c.778G>A	7	p.Pro156Leu; p.Glu260Lys

A ₂	ABO*A2.20	c.467C>T; c.829G>A	7	p.Pro156Leu; p.Val277Met
A ₃	ABO*A3.01	c.871G>A	7	p.Asp291Asn
A ₃	ABO*A3.02	c.829G>A; c.1061delC	7	p.Val277Met; p.Pro354Argfs*23
A ₃	ABO*A3.03	c.838C>T	7	p.Leu280Phe
A ₃	ABO*A3.04	c.467C>T; c.539G>A; c.1061delC	7	p.Arg180His; p.Pro156Leu; p.Pro354Argfs*23
A ₃	ABO*A3.05	c.820G>A	7	p.Val274Met
A ₃	ABO*A3.06	c.467C>T; c.820G>A	7	p.Pro156Leu; p.Val274Met
A ₃	ABO*A3.07	c.467C>T; c.745C>T	7	p.Pro156Leu; p.Arg249Trp
A _{weak}	ABO*AW.01	c.407C>T; c.467C>T; c.1061delC	7	p.Thr136Met; p.Pro156Leu; p.Pro354Argfs*23
A _{weak}	ABO*AW.02	c.350G>C; c.467C>T; c.1061delC	6 7	p.Gly117Ala; p.Pro156Leu; p.Pro354Argfs*23
A _{weak}	ABO*AW.03	c.203G>C; c.467C>T; c.1061delC	4 7	p.Arg68Thr; p.Pro156Leu; p.Pro354Argfs*23
A _{weak}	ABO*AW.04	c.721C>T	7	p.Arg241Trp
A _{weak}	ABO*AW.05	c.965A>G	7	p.Glu322Gly
A _{weak}	ABO*AW.06	c.502C>G	7	p.Arg168Gly
A _{weak}	ABO*AW.07	c.467C>T; c.592C>T; c.1061delC	7	p.Pro156Leu; p.Arg198Trp; p.Pro354Argfs*23
A _{weak}	ABO*AW.08	c.220C>T; c.297A>G; c.488C>T; c.526C>G; c.802G>A	5 6 7	p.Pro74Ser; p.Thr163Met; p.Arg176Gly; p.Gly268Arg



Aweak	ABO*AW.09	c.46G>A; c.106G>T; c.188G>A; c.220C>T; c.467C>T; c.1061delC	2 3 4 5 7	p.Ala16Thr; p.Val36Phe; p.Arg63His; p.Pro74Ser; p.Pro156Leu; p.Pro354Argfs*23
Aweak	ABO*AW.10	c.784G>A	7	p.Asp262Asn
Aweak	ABO*AW.11	c.523G>A; c.721C>T	7	p.Val175Met; p.Arg241Trp

Aweak	ABO*AW.12	c.467C>T; c.556A>G	7	p.Pro156Leu; p.Met186Val
Aweak	ABO*AW.13	c.2T>C	1	p.Ala2_Met20del
Aweak	ABO*AW.14	c.467C>T; c.699C>A	7	p.Pro156Leu; p.His233Gln
Aweak	ABO*AW.15	c.374+4A>T	Intron 6	Altered splicing
Aweak	ABO*AW.16	c.1A>G; c.467C>T; c.1061delC	1 7	p.Ala2_Met20del; p.Pro156Leu; p.Pro354Argfs*23
Aweak	ABO*AW.17	c.236C>T; c.467C>T; c.1061delC	5 7	p.Pro79Leu; p.Pro156Leu; p.Pro354Argfs*23
Aweak	ABO*AW.18	c.347T>C; c.467C>T; c.1061delC	6 7	p.Ile116Thr; p.Pro156Leu; p.Pro354Argfs*23
Aweak	ABO*AW.19	c.434A>G; c.467C>T; c.1061delC	7	p.His145Arg; p.Pro156Leu; p.Pro354Argfs*23
Aweak	ABO*AW.20	c.467C>T; c.607G>A; c.1061delC	7	p.Pro156Leu; p.Glu203Lys; p.Pro354Argfs*23
Aweak	ABO*AW.21	c.467C>T; c.607G>C; c.1061delC	7	p.Pro156Leu; p.Glu203Gln; p.Pro354Argfs*23
Aweak	ABO*AW.22	c.467C>T; c.634G>A; c.1061delC	7	p.Pro156Leu; Val212Met; p.Pro354Argfs*23
Aweak	ABO*AW.23	c.467C>T; c.722G>A; c.1061delC	7	p.Pro156Leu; p.Arg241Gln; p.Pro354Argfs*23
Aweak	ABO*AW.24	c.467C>T; c.742C>T; c.1061delC	7	p.Pro156Leu; p.Arg248Cys; p.Pro354Argfs*23



Aweak	ABO*AW.25	c.467C>T; c.829G>A; c.1061delC	7	p.Pro156Leu; p.Val277Met; p.Pro354Argfs*23
Aweak	ABO*AW.26	c.467C>T; c.527G>A; c.1061delC	7	p.Pro156Leu; p.Arg176His; p.Pro354Argfs*23
Aweak	ABO*AW.27	c.527G>A; c.1061delC	7	p.Arg176His; p.Pro354Argfs*23
Aweak	ABO*AW.28	c.98+2T>C	Intron 1	Altered splicing
Aweak	ABO*AW.29	c.311T>A	6	p.Ile104Asn
Ax/Aweak	ABO*AW.30.01	c.646T>A	7	p.Phe216Ile
Ax/Aweak	ABO*AW.30.02	c.646T>A; c.681G>A	7	p.Phe216Ile

Ax/Aweak	ABO*AW.31.01	c.297A>G; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Phe216Ile; p.Val277Met
Ax/Aweak	ABO*AW.31.0205	c.646T>A; c.681G>A; c.771C>T; c.829G>A	7	p.Phe216Ile; p.Val277Met
Ax/Aweak	ABO*AW.32	c.996G>A	7	p.Trp332Ter
Ax/Aweak	ABO*AW.33	c.467C>T; c.543G>T	7	p.Pro156Leu; p.Trp181Cys
Ax/Aweak	ABO*AW.34	c.467C>T; c.829G>A; c.1009A>G	7	p.Pro156Leu; p.Val277Met; p.Arg337Gly
Ax/Aweak	ABO*AW.35	c.467C>T; c.860C>T	7	p.Pro156Leu; p.Ala287Val
Ax/Aweak	ABO*AW.36	c.607G>A	7	p.Glu203Lys
Ax/Aweak	ABO*AW.37	c.940A>G	7	p.Lys314Glu
Ax/Aweak	ABO*AW.38	c.426G>C	7	p.Met142Ile
Ax/Aweak	ABO*AW.39	c.385T>C	7	p.Phe129Leu
Ax/Aweak	ABO*AW.40	c.499G>T	7	p.Gly167Cys
Ax/Aweak	ABO*AW.41	c.370A>G	6	p.Lys124Glu
Ax/Aweak	ABO*AW.42	c.467C>T; c.905A>G	7	p.Pro156Leu; p.Asp302Gly
Ax/Aweak	ABO*AW.43	c.467C>T; c.721C>T	7	p.Pro156Leu; p.Arg241Trp
Afinn/Aweak	ABO*AW.44	c.374+4A>G	Intron 6	Altered splicing



Abantu/Aweak	ABO*AW.45	c.203+1delG; c.467C>T; c.1061delC	Intron 4 7	Altered splicing
Am	ABO*AM.01	c.467C>T; c.761C>T	7	p.Pro156Leu; p.Ala254Val
Am	ABO*AM.02	c.664G>A	7	p.Val222Met
Ael	ABO*AEL.01	c.804dupG	7	p.Phe269Valfs*124
Ael	ABO*AEL.02	c.467C>T; c.646T>A; c.681G>A	7	p.Pro156Leu; p.Phe216Ile
Ael	ABO*AEL.03	c.804delG	7	p.Phe269Serfs*20
Ael	ABO*AEL.04	c.374+5G>A	Intron 6	Altered splicing
Ael	ABO*AEL.05	c.467C>T; c.767T>C	7	p.Pro156Leu; p.Ile256Thr
Ael	ABO*AEL.06	c.425T>C; c.467C>T	7	p.Met142Thr; p.Pro156Leu
Ael	ABO*AEL.07	c.467C>T; 681G>A; 771C>T; c.829G>A	7	p.Pro156Leu; p.Val277Met
Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
Ael	ABO*AEL.08	c.467C>T; c.804dupG	7	p.Pro156Leu; p.Phe269Valfs*124

Bases moleculares para las variantes B

Fenotipo	Nombre del Alelo	Cambio de nucleóido	Exón	Aminoácido cambiado
B	ABO*B.01	c.297A>G; c.526C>G; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
B	ABO*B.02	c.892G>T	7	p.Ala298Ser
B	ABO*B.03	c.559C>T	7	p.Arg187Cys
B ₃	ABO*B3.01	c.1054C>T	7	p.Arg352Trp
B ₃	ABO*B3.02	c.646T>A	7	p.Phe216Ile



B ₃	<i>ABO*B3.03</i>	c.155+5G>A	Intron 3	Altered splicing
B ₃	<i>ABO*B3.04</i>	c.247G>T	6	p.Asp83Tyr
B ₃	<i>ABO*B3.05</i>	c.425T>C	7	p.Met142Thr
B ₃	<i>ABO*B3.06</i>	c.547G>A	7	p.Asp183Asn
B ₃	<i>ABO*B3.07</i>	c.410C>T	7	p.Ala137Val
B ₃	<i>ABO*B3.08</i>	c.938A>C	7	p.His313Pro
B _x /B _{weak}	<i>ABO*BW.01</i>	c.871G>A	7	p.Asp291Asn
B _{weak}	<i>ABO*BW.02</i>	c.873C>G	7	p.Asp291Glu
B _{weak}	<i>ABO*BW.03</i>	c.721C>T	7	p.Arg241Trp
B _{weak}	<i>ABO*BW.04</i>	c.548A>G	7	p.Asp183Gly
B _{weak}	<i>ABO*BW.05</i>	c.539G>A	7	p.Arg180His
B _{weak}	<i>ABO*BW.06</i>	c.1036A>G	7	p.Lys346Glu
B _{weak}	<i>ABO*BW.07</i>	c.1055G>A	7	p.Arg352Gln
B _{weak}	<i>ABO*BW.08</i>	c.863T>G	7	p.Met288Arg
B _{weak}	<i>ABO*BW.09</i>	c.1037A>T	7	p.Lys346Met
B _{weak}	<i>ABO*BW.10</i>	c.556A>G	7	p.Met186Val
B _{weak}	<i>ABO*BW.11</i>	c.695T>C	7	p.Leu232Pro
B _{weak}	<i>ABO*BW.12</i>	c.278C>T	6	p.Pro93Leu
B _{weak}	<i>ABO*BW.14</i>	c.523G>A	7	p.Val175Met
B _{weak}	<i>ABO*BW.15</i>	c.565A>G	7	p.Met189Val
B _{weak}	<i>ABO*BW.16</i>	c.575T>C	7	p.Ile192Thr
B _{weak}	<i>ABO*BW.17</i>	c.784G>A	7	p.Asp262Asn
B _{weak}	<i>ABO*BW.18</i>	c.802G>A	7	p.Gly268Thr
B _{weak}	<i>ABO*BW.19</i>	c.646T>A; c.681G>A	7	p.Phe216Ile
B _{weak}	<i>ABO*BW.20</i>	c.815_816insG	7	p.Ser273Valfs*?
B _{weak}	<i>ABO*BW.21</i>	c.688G>C	7	p.Gly230Arg
B _{weak}	<i>ABO*BW.22</i>	c.503G>T	7	p.Arg168Leu



Bweak	<i>ABO*BW.23</i>	c.743G>C	7	p.Arg248Pro
Bweak	<i>ABO*BW.24</i>	c.558G>T	7	p.Met186Ile
Bweak	<i>ABO*BW.25</i>	c.103G>A; c.619C>G	3 7	p.Gly35Arg; p.Leu207Val
Bweak	<i>ABO*BW.26</i>	c.53G>T	2	p.Arg18Leu
Bweak	<i>ABO*BW.27</i>	c.905A>G	7	p.Asp302Gly
Bweak	<i>ABO*BW.28</i>	c.541T>C	7	p.Trp181Arg
Bweak	<i>ABO*BW.29</i>	c.588C>G	7	p.Cys196Trp
Bweak	<i>ABO*BW.30</i>	c.976G>T	7	p.Asp326Tyr
Bweak	<i>ABO*BW.31</i>	c.900G>C	7	p.Trp300Cys
Bweak	<i>ABO*BW.32</i>	c.808T>A	7	p.Phe270Ile
Bweak	<i>ABO*BW.33</i>	c.550G>A	7	p.Val184Met
Bweak	<i>ABO*BW.34</i>	c.889G>A	7	p.Glu297Lys
Bel	<i>ABO*BEL.01</i>	c.641T>G	7	p.Met214Arg
Bel	<i>ABO*BEL.02</i>	c.669G>T	7	p.Glu223Asp
Bel	<i>ABO*BEL.03</i>	c.502C>T	7	p.Arg168Trp
Bel	<i>ABO*BEL.05</i>	c.952G>A	7	p.Val318Met
Bel	<i>ABO*BEL.04</i>	c.467C>T; c.646T>A; c.681G>A; c.771C>T; c.796C>A; c.803G>C; c.829G>A	7	p.Pro156Leu; p.Phe216Ile; p.Leu266Met; p.Gly268Ala; p.Val277Met

Bases moleculares asociados a formas cisAB y fenotipos B(A)

Fenotipo	Nombre de alelo	Cambio de nucleótido	Exón	Cambio de aminoácido
cisAB	<i>ABO*cisAB.01</i>	c.467C>T; c.803G>C	7	p.Pro156Leu; p.Gly268Ala
cisAB	<i>ABO*cisAB.02</i>	c.526C>G; c.657C>T; c.703G>A; c.803G>C	7	p.Arg176Gly; p.Gly235Ser; p.Gly268Ala



cisAB	<i>ABO*cisAB.03</i>	c.297A>G; c.526C>G; c.657C>T; c.700C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Pro234Ser; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
cisAB	<i>ABO*cisAB.04</i>	c.467C>T; c.796C>A	7	p.Pro156Leu; p.Leu266Met
cisAB	<i>ABO*cisAB.05</i>	c.297A>G; c.526C>G; c.657C>T; c.703G>A; c.796C>A; c.930G>A	6 7	p.Arg176Gly; p.Gly235Ser; p.Leu266Met
cisAB	<i>ABO*cisAB.06</i>	c.297A>G; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Gly235Ser; p.Leu266Met; p.Gly268Ala
B(A)	<i>ABO*BA.01</i>	c.297A>G; c.526C>G; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Leu266Met; p.Gly268Ala
B(A)	<i>ABO*BA.02</i>	c.297A>G; c.526C>G; c.657C>T; c.700C>G; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Pro234Ala; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
B(A)	<i>ABO*BA.03</i>	c.297A>G; c.526C>G; c.657C>T; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Leu266Met; p.Gly268Ala
B(A)	<i>ABO*BA.04</i>	c.297A>G; c.526C>G; c.640A>G; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Met214Val; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
B(A)	<i>ABO*BA.05</i>	c.297A>G; c.526C>G; c.641T>C; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; p.Met214Thr; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
B(A)	<i>ABO*BA.06</i>	c.297A>G; c.526C>G; c.657C>T; c.703G>A; c.796C>A; c.930G>A	6 7	p.Arg176Gly; p.Gly235Ser; p.Leu266Met



Bases moleculares del fenotipo O (null)

Fenotipo	Nombre del alelo	Nombre alternativo	Nucleótido cambiado	Exón	Cambio de aminoácido
O	<i>ABO*O.01.01</i>	O01 (O^1)	c.261delG	6	p.Thr88Profs*31
O	<i>ABO*O.01.02</i>	O02 (O^{1v})	c.106G>T; c.188G>A; c.189C>T; c.220C>T; c.261delG; c.297A>G; c.646T>A; c.681G>A; c.771C>T; c.829G>A	3 4 5 6 7	p.Val36Phe; p.Arg63His; p.Pro74Ser; p.Thr88Profs*31
O	<i>ABO*O.01.04</i>	O04	c.261delG; c.579T>C	6	p.Thr88Profs*31
O	<i>ABO*O.01.05</i>	O05	c.261delG; c.297A>G	6	p.Thr88Profs*31
O	<i>ABO*O.01.06</i>	O06, O30	c.261delG; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	<i>ABO*O.01.07</i>	O07	c.261delG; c.297A>G; c.646T>A; c.681G>A; c.721C>T; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	<i>ABO*O.01.09</i>	O09	c.261delG; c.318C>T; c.467C>T	6 7	p.Thr88Profs*31
O	<i>ABO*O.01.10</i>	O10	c.261delG; 657C>T	6 7	p.Thr88Profs*31
O	<i>ABO*O.01.11</i>	O11	c.261delG; c.297A>G; c.542G>A; c.646T>A;	6 7	p.Thr88Profs*31



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

			c.681G>A; c.771C>T; c.829G>A		
O	ABO*O.01.12	O12	c.261delG; c.297A>G; c.595C>T; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.13	O13	c.261delG; c.297A>G; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.22	O22	c.261delG c.467C>T; c.1061delC	6 7	p.Thr88Profs*31

O	ABO*O.01.23	O23	c.261delG; c.297A>G; c.646T>A; c.771C>T; c.829G>A; 1054C>T	6 7	p.Thr88Profs*31
O	ABO*O.01.24	O24	c.106G>T; c.188G>A; c.189C>T; c.261delG; c.297A>G; c.526C>G; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	3 4 6 7	p.Val36Phe; p.Arg63His; p.Thr88Profs*31
O	ABO*O.01.25	O25	c.261delG; c.454T>C	6 7	p.Thr88Profs*31
O	ABO*O.01.26	O26	c.261delG; c.768C>A	6 7	p.Thr88Profs*31
O	ABO*O.01.27	O27	c.261delG; c.318C>T; c.467C>T; c.729C>T	6 7	p.Thr88Profs*31



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

O	ABO*O.01.28	O28	c.261delG; c.926A>G	6 7	p.Thr88Profs*31
O	ABO*O.01.29	O29	c.261delG; c.318C>T	6	p.Thr88Profs*31
O	ABO*O.01.31	O31	c.261delG; c.297A>G; c.529G>A; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.32	O32	c.261delG; c.297A>G; c.538C>T; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.33	O33	c.261delG; c.297A>G; c.498C>T; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.34	O34	c.261delG; c.297A>G; c.351G>A; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.35	O35	c.106G>T; c.188G>A; c.189C>T; c.220C>T; c.261delG; c.297A>G; c.681G>A; c.771C>T; c.829G>A	3 4 5 6 7	p.Val36Phe; p.Arg63His; p.Pro74Ser; p.Thr88Profs*31



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

O	ABO*O.01.36	O36	c.106G>T; c.188G>A; c.189C>T; c.220C>T; c.261delG; c.297A>G; c.646T>A; c.681G>A; c.829G>A	3 4 5 6 7	p.Val36Phe; p.Arg63His; p.Pro74Ser; p.Thr88Profs*31
O	ABO*O.01.39	O39	c.220C>T; c.261delG; c.297A>G; c.681G>A; c.771C>T; c.829G>A	5 6 7	p.Pro74Ser; p.Thr88Profs*31
O	ABO*O.01.40	O40	c.106G>T; c.188G>A; c.189C>T; c.261delG; c.297A>G; c.646T>A; c.681G>A; c.829G>A	3 4 6 7	p.Val36Phe; p.Arg63His; p.Thr88Profs*31
O	ABO*O.01.41	O41	c.261delG; c.297A>G; c.526C>G; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.44	O44	c.261delG; c.297A>G; c.646T>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.45	O45	c.261delG; c.646T>A; c.771C>T	6 7	p.Thr88Profs*31
O	ABO*O.01.46	O46	c.261delG; c.646T>A; c.771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.56	O56; O70	c.261delG; c.496delA	6 7	p.Thr88Profs*31



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

O	ABO*O.01.57	O57	c.261delG; c.802G>A	6 7	p.Thr88Profs*31
---	-------------	-----	------------------------	--------	-----------------

O	ABO*O.01.58	O58	c.261delG; c.297A>G; c.646T>A; c.681G>A; c.687C>T; 771C>T; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.61	O61	c.261delG; c.743G>C	6 7	p.Thr88Profs*31
O	ABO*O.01.67	O67	c.103G>A; c.106G>T; c.188G>A; c.189C>T; c.220C>T; c.261delG; c.297A>G; c.646T>A; c.681G>A; c.771C>T; c.829G>A	3 4 5 6 7	p.Gly35Arg; p.Val36Phe; p.Arg63His; p.Pro74Ser; p.Thr88Profs*31
O	ABO*O.01.68	O68	c.106G>T; c.188G>A; c.189C>T; c.261delG; c.297A>G; c.646T>A; c.681G>A; c.771C>T; c.829G>A	3 4 6 7	p.Val36Phe; p.Arg63His; p.Thr88Profs*31
O	ABO*O.01.71	O71	c.261delG; c.829G>A	6 7	p.Thr88Profs*31
O	ABO*O.01.75	O75	c.106G>T; c.188G>A; c.189C>T; c.220C>T; c.261delG; c.297A>G; c.542G>A; c.646T>A; c.681G>A; c.771C>T; c.829G>A	3 4 5 6 7	p.Val36Phe; p.Arg63His; p.Pro74Ser; p.Thr88Profs*31



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

O	ABO*O.01.76	O76	c.261delG; c.579T>C; c.1046_1048delAGG	6 7	p.Thr88Profs*31
O	ABO*O.01.82		c.190G>A; c.261delG	4 6	p.Val64Ile; p.Thr88Profs*31
O	ABO*O.01.83		c.106G>T; c.188G>A; c.261delG; c.297A>G	3 4 6	p.Val36Phe; p.Arg63His; p.Thr88Profs*31
O	ABO*O.02.01	O03 (O ²)	c.53G>T; c.220C>T; c.297A>G; c.526C>G; c.802G>A	2 5 6 7	p.Arg18Leu; p.Pro74Ser; p.Arg176Gly; p.Gly268Arg

O	ABO*O.02.02	O48 (O ² -2)	c.53G>T; c.220C>T; c.297A>G; c.526C>G; c.649C>T; c.689G>A; c.802G>A	2 5 6 7	p.Arg18Leu; p.Pro74Ser; p.Arg176Gly; p.Arg217Cys; p.Gly230Asp; p.Gly268Arg
O	ABO*O.02.03	O49 (O ² -2)	c.53G>T; c.220C>T; c.297A>G; c.526C>G; c.689G>A; c.802G>A	2 5 6 7	p.Arg18Leu; p.Pro74Ser; p.Arg176Gly; p.Gly230Asp; p.Gly268Arg
O	ABO*O.02.04	O50 (O ² -4)	c.53G>T; c.220C>T; c.297A>G; c.488C>T; c.526C>G; c.802G>A	2 5 6 7	p.Arg18Leu; p.Pro74Ser; p.Thr163Met; p.Arg176Gly; p.Gly268Arg
O	ABO*O.03	O08 (O ³)	c.467C>T; c.804dupG; c.1061delC	7	p.Pro156Leu; p.Phe269Valfs*87
O	ABO*O.04.01	O41 (O ⁴)	c.87_88insG	2	p.Val30Glyfs*27
O	ABO*O.04.02	O21 (O ⁴)	c.87_88insG; c.261delG; c.467C>T	2 6 7	p.Val30Glyfs*27
O	ABO*O.05	O52 (O ⁵)	c.322C>T	6	p.Gln108Ter
O	ABO*O.06	O53 (O ⁶)	c.542G>A	7	p.Trp181Ter



Prof. Marcelo Pedro Russo
Medicina
Hemoterapia e Imunohematologia

O	ABO*O.07	O14 (O301)	c.467C>T; c.893C>T	7	p.Pro156Leu; p.Ala298Val
O	ABO*O.08	O15 (O302)	c.927C>A	7	p.Tyr309Ter
O	ABO*O.09.01	O19 (R102)	c.646T>A; c.681G>A; c.771C>T; c.829G>A	7	p.Phe216Ile; p.Val277Met
O	ABO*O.09.02	O20 (R103)	c.297A>G; c.646T>A; c.681G>A; c.771C>T; c.829G>A	6 7	p.Phe216Ile; p.Val277Met
O	ABO*O.10	O72	c.66_67insG	2	p.Phe23Valfs*34
O	ABO*O.11	O74	c.297A>G; c.505_507delCAG; c.526C>G; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Gln169del; p.Arg176Gly; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
O	ABO*O.12	O77	c.297A>G; c.526C>G; c.563G>A; c.657C>T; c.703G>A; c.796C>A; c.803G>C; c.930G>A	6 7	p.Arg176Gly; Arg188His; p.Gly235Ser; p.Leu266Met; p.Gly268Ala
O	ABO*O.13	O78	c.452T>G	7	p.Val151Gly
O	ABO*O.14	O79	c.635T>A	7	p.Val212Glu
O	ABO*O.15	O81	c.793T>C	7	p.Tyr265His
O	ABO*O.16		c.106G>T; c.188G>A; Deletion of exons 5-7	3 4 5-7	p.?