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REPORT OF NEW ALLELES OR ANTIGENS

TRANSFUSION

Identification of a stably inherited novel A subtype with c.625T>G mutation

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1 | INTRODUCTION

Although 43 human erythrocytic BG (blood group) systems had been identified up to date,[1] the ABO BG system is still recognized as one of the most important roles in clinic[2] due to its close relationship to blood transfusion as well as transplantation. Especially with the continuous improvement of detection methods,[3] numerous subtypes have been identified. Here, we described a novel A subtype caused by c.625T>G mutation on Exon 7, which has an Ael serological characteristics, and this mutation can stably be inherited to offspring.

2 | BRIEF METHODS

An incongruent phenomenon of forward and reverse blood typing by using microcolumn gel method (Ortho

TABLE 1 Results of serological detections

	ABO BG initial screening results								Absorption-elution test		
	Anti-A	Anti-B	Anti-H	Anti-A1	Anti-AB	A1 RBC	B RBC	O RBC	A1 RBC	B RBC	O RBC
Member 1	0	0	3+	0	0	0	2+	0	+	0	0
Member 2	0	0	3+	0	0	0	4+	0	+	0	0
Member 3	0	0	3+	0	0	0	4+	0	+	0	0
Member 4	0	0	3+	0	0	+	4+	0	+	0	0
Member 5	0	0	3+	0	0	+	4+	0	+	0	0
Member 6	0	0	3+	0	0	0	4+	0	+	0	0

Abbreviations: BG, blood group; Cys, Cystine; Gly, Glycine; RBC, red blood cell.

Clinical Diagnostics) in a male patient diagnosed with bronchiectasis was found. Classical serological detections (hemagglutination tube method) were executed for ABO forward and reverse blood typing (Shanghai Hemopharmaceutical Biological Company) to the patient and some members of this family. Reverse blood typing enhanced test (200 μl patient's plasma reacted with 50 μl standard RBCs at 4°C for 30 min, followed by immediate centrifugation at 1000 *g*/min for 15 s, and then observe the agglutination intensity), H antigen identification, absorption-elution test, and salivary substance test were also performed according to standardized operating procedure. Genomic DNA was extracted and *ABO* genotyping was performed by using *ABO* genotyping kit (Jiangsu Zhongji Wantai Biomedicine Corporation) according to the manufacturers' instructions. Exons 1–7 of *ABO* gene were sequenced by using the specific amplification primers.

3 | RESULTS AND DISCUSSION

Twenty family members were recruited with informed consent, and 19 members' specimens were detected. We identified 6 of the 20 family members as Anovel and named them members 1 to 6 (in which member 2 is the proband). The results of serological detections are shown in Table 1. The c.625T>G site mutation occurred on Exon 7 of *A* gene, encodes p.Cys209Gly of A transferase, leading to the weakening of A antigen, which can only be detected by the most sensitive absorption-elution test. The relevant data had been deposited in GenBank with the accession number MT434876. According to the exon sequencing of *ABO* gene (with *ABO*A1.01* as the reference sequence), the genotype of members 1 and 4 was considered to be *Anovel/O.01.02*, and the genotype of members 2, 3, 5, and 6 was considered to be *Anovel/O.01.01*. In addition, pedigree investigation showed that c.625T>G occurred on seven members of three generations and suggested that c.625T>G can be stably inherited.

In conclusion, we identified a novel mutation c.625T>G on Exon 7 of *ABO* gene and indicated that this site mutation can stably entail offspring that arise serological characteristics of Ael subtype.

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CONFLICT OF INTEREST

The authors have disclosed no conflicts of interest.

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