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Serology and molecular genetic analysis of weakened expression of ABO blood group antigens in Chinese pediatric population

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Background - The ABO blood group system is vital for red blood cell antigens. Weakened expression of ABO antigens often leads to misidentification of blood types, challenging transfusion safety.

Materials and methods - Blood samples from 44 cases underwent serological analysis using microcolumn agglutination and tube tests. To investigate genetic variations, we sequenced the promoter region and exons 1-7 of the *ABO* gene. We also examined the relationship between ABO haplotypes and the presence of rare alleles. We used PolyPhen-2 to predict the structure and function of glycosyltransferase B (GTB) and classify missense variants. We also conducted pedigree analysis on 4 selected cases to further understand the inheritance patterns.

Results - We detected 18 different ABO subgroup alleles in 44 cases of weakened ABO antigen expression. We found 7 subtype A (predominantly A2), 7 subtype B (which included Bw and B3), one cisAB, and 3 group B(A). The complete phenotypes we observed included A2, Bw, B3, A2B, ABw, AB3, cisAB, and B(A). Notably, we discovered a rare B allele characterized by the c.538C>T variant, which alters residue 180 to p.Arg180Cys. This variant received a PolyPhen-2 score of 1.00, suggesting a high likelihood of damaging effects. Furthermore, pedigree analysis supported an autosomal inheritance pattern for this allele.

Discussion - The weakened expression of ABO antigens in young children may result from their immature immune systems or subtyping. ABO genotyping is essential to accurately identify blood groups and ensure transfusion safety.

Keywords: *ABO system, genotyping, subtyping, gene variant, pediatric.*

INTRODUCTION

The ABO blood group system, identified by Karl Landsteiner in 1901, plays a crucial role in ensuring the safety of blood transfusions[1,2]. Determining ABO blood types poses greater challenges in infants than in adults because these young patients are still developing antigens and antibodies[3]. Moreover, our findings in patients who are no longer infants are explained by the presence of variant alleles.

Since the cloning of cDNA corresponds to the mRNA located at the ABO locus, a better understanding of the molecular genetic foundation of the ABO system has emerged.

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The molecular genetic framework of the ABO system has been recognized since the identification of cDNA associated with the mRNA at the ABO locus. This gene comprises a coding region of 1065 base pairs, which includes seven exons and spans more than 19.5 kb in total[4]. Exons 6 and 7 encode the catalytic domain of ABO glycosyltransferases (GTs), collectively making up 77% of the entire enzyme structure. Changes in these sequences can affect the expression and activity of glycosyltransferase, potentially altering ABO blood group classifications[5]. In addition to exons, the roles of intron 1 enhancer and both the 5' and 3' non-coding regions are crucial in influencing the transcription process[3,6]. Weak ABO subgroups frequently arise from the inheritance of uncommon alleles at the ABO locus, which can result from missense variants, insertions, or deletions[7-9]. Nevertheless, the underlying mechanisms of certain rare ABO blood group subgroups are still not completely understood. Consequently, examining variations in the *ABO* gene sequence is vital for accurately reporting ABO phenotypes and providing significant guidance for blood transfusions in pediatric cases.

In this research, serological assessments and DNA sequencing were conducted on 44 infants and young children who had reduced expression of ABO blood group antigens, leading to the identification of 18 subtypes of ABO. Notably, based on ABO subtyping, among the 26 samples from infants aged <4 months, 4 demonstrated weakened antigen: *ABO*A2.05/B.01, ABO*A2.05/O.01.01, ABO*A1.02/Bw.07, and ABO*A1.02/ B311.*

MATERIALS AND METHODS

Blood samples

Blood samples from 44 children admitted to Jinan Children's Hospital were collected from August 20, 2020, to February 14, 2024, revealing reduced levels of ABO blood group antigens. Of these 44 pediatric patients with serological ABO discrepancies, 12 were ≤6 months old and 32 were >6 months old. Among the 26 patients without identified variant alleles, 5 were ≤6 months old, with 21 >6 months old.

In total, blood was collected from 9 relatives belonging to 4 different families. All procedures that included human participants adhered to the ethical guidelines established by relevant institutional and national research committees. Each participant provided informed consent to take part in the study.

Serological testing

The phenotypes of the ABO blood groups in the samples were evaluated using a microcolumn agglutination assay (supplied by Shanghai Runpu Biological, Shanghai, China), as well as a saline test tube technique employing monoclonal anti-A and anti-B (Shanghai Hemo-Pharmaceutical Biological, Shanghai, China), anti-H (Sanquin, Amsterdam, the Netherlands), and an ABO reverse typing kit (Shanghai Hemo-Pharmaceutical Biological).

Polymerase chain reaction

Genomic DNA extraction was performed, and ABO genotyping was carried out using a fluorescence polymerase chain reaction (PCR) kit provided by Jiangsu Zhongjiwat Biotechnology Medicine Co. Ltd (Jiangsu, China), according to the manufacturer's guidelines. The preliminary sequencing analysis focused on exons 6-7 and the adjacent areas, which encompass the prevalent variants associated with variations in ABO subgroups.

DNA sequencing for exons and promoter of the *ABO* gene

Following the purification of PCR products, we conducted a sequencing analysis targeting the entire coding sequences (CDS) for exons 1-7, along with the promoter

Table I - *Primers used for amplification of ABO gene fragments*

Primer	Primer sequence(5'→3')	Amplification region
PrE1-F	GGCGCGTCTCCTAG	Promoter, exon 1
PrE1-R	AGGGACCCCGCTCTCAC	
E2-F	CTGAGTGGAAGGAGGTCAAT	Exon 2
E2-R	AATGGGAGTGAGATGAGCC	
E3-F	CCAGAACCAAGAGTGAAGTC	Exon 3
E3-R	AAAGGGAGAGTTGAGGATGG	
E4-F	AGACAGAAGCCAGATCACCT	Exon 4
E4-R	CACACACACACACAAAAGCC	
E5-F	TGTCACTCTCCCACTTCTCA	Exon 5
E5-R	GTACCTATCAGGCCTTTGCA	
E6-F	GCTGCATGAATGACCTTTCCC	Exon 6
E6-R	GATTTGCCCGGTTGGAGTCG	
E7-F	GCCTAGGCTTCAGTTACTCACAACAG	Exon 7
E7-R	TCGCTGGGAAGAGGATGAAG	

region of the *ABO* gene (**Table I**). By using bidirectional Sanger sequencing, we aimed to analyze the entire transcript and detect rare alleles via haplotype analysis. To computationally predict the structural integrity and catalytic functionality of glycosyltransferase B (GTB) and assess the impacts of new variants, we used PolyPhen-2 (*http://genetics.bwh.harvard.edu/pph2/*), an established algorithm designed to evaluate protein damage induced by variants. The PolyPhen-2 score classifies the likelihood of the substitution causing damage.

Pedigree analysis

Our objective was to explore the correlations between genotype and phenotype that lead to reduced antigen expression through familial segregation analysis of 4 pediatric cases. We performed *ABO* gene profiling of the parents using bidirectional Sanger sequencing, concentrating on the complete coding region (exons 1-7) as well as the regulatory sequences of the promoter. This enabled us to systematically examine inheritance patterns and confirm the genetic variants.

RESULTS

Identification of 18 ABO subgroups

A total of 44 blood samples exhibiting weakened ABO antigen strength were discovered. Serological analysis showed that these comprised 18 distinct ABO subgroups.

PCR sequencing results

The sequencing of genes from 44 samples revealed 7 subtype A, 7 subtype B, one cisAB, and 3 group B (A). Subtype A, which is predominantly made up of A2 variants, consists of 5 samples of *ABO*A2.01/B.01*, one sample of *ABO*A2.05/B.01*, and one sample of *ABO*A2.05/O.01.01*. In contrast, subtype B exhibited greater heterogeneity, encompassing two instances of *ABO*A1.02/Bw.07*, along with separate instances of *ABO*A1.02/B311*, *ABO*B311/O.01.02*, *ABO*B311/O.01.01*, *ABO*Bw03/O.01.01*, and a combined case of *ABO*A1.02/B.01* featuring a c.538C>T substitution. Detailed genotypes can be found in **Table II**.

Table II - *The phenotypes of 18 ABO subgroups*

Sample No.	Age	Agglutination with RBCs plasma			Agglutination with		Phenotype	Genotype
		Anti-A	Anti-B	Anti-H	A1 cell	B cell		
1	5y	4+	2+	4+	–	–	AB_{weak}	*ABO*A1.02/BW.07*
2	18d	4+	2+	3+	–	–	AB_{weak}	*ABO*A1.02/BW.07*
3	3y	2+	4+	2+	–	–	$A_{weak}B$	*ABO*A2.01/B.01*
4	2d	MF	4+	NT	–	–	$A_{weak}B$	*ABO*A2.05/B.01*
5	10m	2+	4+	3+	–	–	$A_{weak}B$	*ABO*B(A)*
6	5y	4+	MF	3+	–	±	AB_{weak}	*ABO*A1.02/B311*
7	7y	-	2+	4+	4+	–	B_{weak}	*ABO*BW.03/O.01.01*
8	1m	MF	-	2+	–	–	A_{weak}	*ABO*A2.05/O.01.01*
9	4y	4+	3+	3+	–	2+	$A_{weak}B$	*ABO*cisAB.01/O.01.01*
10	7y	-	3+	4+	–	–	B_{weak}	*ABO*B311/O.01.01*
11	5y	3+	4+	2+	–	–	$A_{weak}B$	*ABO*A2.01/B.01*
12	6y	3+	4+	NT	–	–	$A_{weak}B$	*ABO*A2.01/B.01*
13	7y	3+	3+	±	–	–	$A_{weak}B_{weak}$	*ABO*A2.01/B.01*
14	8y	2+	4+	2+	3+	–	$A_{weak}B$	*ABO*B(A)02/O.01.01*
15	7y	3+	4+	+	–	–	$A_{weak}B$	*ABO*A2.01/B.01*
16	6y	2+	4+	3+	4+	–	$A_{weak}B$	*ABO*B(A)02/O.01.01*
17	3d	4+	2+	4+	–	–	AB_{weak}	*ABO*A1.02/B311*
18	4y	4+	1+	-	–	–	AB_{weak}	*ABO*A1.02/B.01*(with c.538 C>T)

d: days; m: months; MF: mixed-field agglutination; NT: not tested RBC: red blood cells; y: years.

Table III - *The variants in the 5'UTR region, exons 6, 7 of the 14 ABO alleles*

ABO allele	5'UTR region -35-18	Exon 6		Exon 7														
		261	297	467	526	646	657	681	703	721	771	796	803	829	930	1009	1055	1061
A101*	GGCGGAAGGCGGAGGCCG	G	A	C	C	T	C	G	G	C	C	C	G	G	G	A	G	C
A102				T														
A106			G	T														
A201				T														del
A205				T												G		
B101			G		G		T		A			A	C		A			
Bw03			G		G		T		A	T		A	C		A			
Bw07			G		G		T		A			A	C		A		A	
B311	del		G		G		T		A			A	C		A			
B(A)new			G		G		T		A			A	C		A			
B(A)02			G		G		T		A			A	C		A			
cisAB01				T									C					
O01		del																
O02		del	G			A		A			T			A				

*The A101 sequence was used as a genetic reference in this analysis.

Analysis of gene variant sites

Table III shows the primary variant locations. These locations are situated within the promoter region and in exons 6 and 7 of the 14 ABO alleles identified in the subtyped samples.

The sequence of *ABO*B311*

According to the results of the nucleotide sequence, three genotypes, *ABO*A1.02/B311*, *ABO*B311/O.01.02*, and *ABO*B311/O.01.01*, were found. Furthermore, *ABO*B311* was identified using the ABO haplotype analysis. Compared with the *ABO*B.01* allele, *ABO*B311* had the deletion from positions -35 to -18 in their promoter region, along with nucleotides consistent with the common B allele in the coding region (**Figure 1**).

The rare allele *ABO*B.01+c.538C>T*

Analysis of the sequence identified an uncommon *ABO*B.01* allele, characterized by a change at nucleotide position c.538 in exon 7, where T replaces C. This rare variant results in the alteration of arginine to cysteine at position 180 (p.Arg180Cys). According to the PolyPhen-2

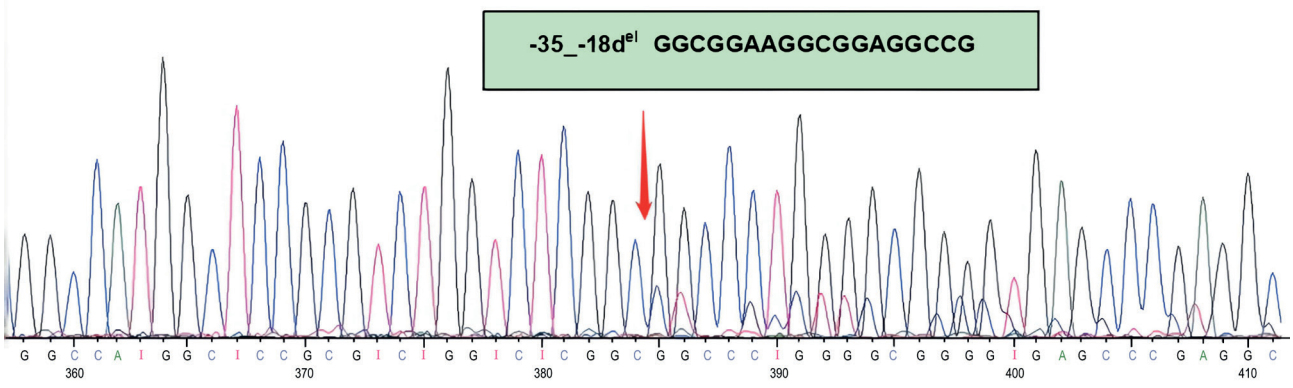

Figure 1 - The haplotype analysis result of *ABO*B311*, the deletion from positions -35 to -18 in the promoter region

software, the amino acid change received a score of 1.00, indicating that it is confidently predicted to be damaging (**Figure 2**).

Pedigree analysis

Analysis of the family indicated that the proband inherited weak antigen alleles from both parents, suggesting a potential lack of association with the disease. The pedigree analysis of the proband along with his family is shown in **Figure 3**.

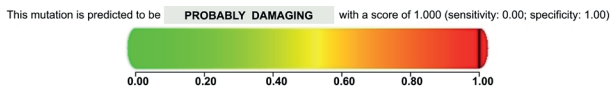

Figure 2 - Prediction results by PolyPhen-2 software
The score is the probability of the substitution being damaging

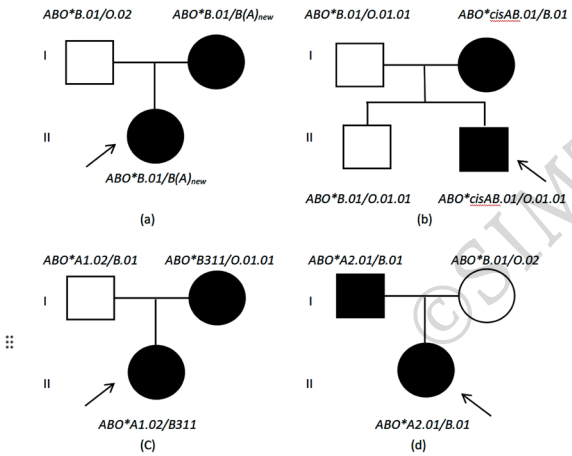

Figure 3 - Pedigree analysis
Roman numerals represent the generations, while arrows indicate the proband "□" represents normal males, "■" represents males with weakened ABO antigens, "○" represents normal females, and "●" represents females with weakened ABO antigens. (**a**) Family lineage analysis of patient B(A)$_{new}$, the proband and her mother are *ABO*B.01/ ABO*B(A)$_{new}$*, her father is *ABO*B.01/O.02*, and no blood sample was collected from her sister. (**b**) Analysis of the family line of patient cisAB01, the genotype of the proband is *ABO*cisAB.01/O01.01*, the genotype of his mother is *ABO*cisAB.01/B.01*, and the genotypes of his father and brother are *ABO*B.01/O01.01*.(**c**) Regarding the lineage of patient B311, the proband is *ABO*A1.02/B311*, with her mother showing a genotype of *ABO*B311/O.01.01*, and her father exhibiting the *ABO*A1.02/ABO*B.01*. (**d**) In the familial lineage for A201, the proband is *ABO*A2.01/B.01*, her mother is *ABO*B.01/ABO*O.02*, and her father is *ABO*A2.01/ABO*O.02*.

DISCUSSION

In this study, we conducted a molecular genetic analysis involving 44 pediatric cases. It is well known that the expression levels of ABO antigens in infants are much less pronounced. Variant alleles, including a newly identified variant site (c.538C>T, p.Arg180Cys) were found in 18 patients across different age groups, underscoring that genetic variants are a key cause of weakened expression beyond infancy. The findings highlight the essential importance of serology and molecular genetics in understanding the mechanisms behind the weakened expression of ABO antigens. This study offers a significant theoretical foundation and practical suggestions aimed at enhancing the safety of blood transfusions in pediatric patients.

The weakened expression of ABO antigens in infants is frequently linked to the underdevelopment of their immune systems, as well as the growth stage of the glycosyltransferase enzymes involved in synthesizing these antigens[10]. This research examined 44 instances with lowered levels of ABO antigens and discovered various ABO subtypes: A2, Bw, B3, A2B, ABw, AB3, cisAB, and B(A). We found that only 4 of the 12 infants aged ≤6 months carried variant alleles. Specifically, the *ABO*A2.05* allele was found in 2 infants, but its overall frequency in our cohort was limited. In the majority of cases (14 of 18), and in particular in those infants aged >6 months, weakened expression is directly linked to the variant alleles. These findings underscore the genetic intricacies associated with ABO antigen expression and reinforce the importance of performing molecular genotyping in addition to the serological testing.

The ABO blood group system is dictated by the *ABO* gene, responsible for encoding enzymes that facilitate the synthesis of A and B antigens. Changes in the *ABO* gene, especially within exons 6 and 7, are understood to considerably influence the activity of glycosyltransferases, enzymes essential for producing A and B antigens on red blood cells. Such variants can weaken or modify antigen expression, resulting in the emergence of various ABO blood group subtypes. In their research, Cai *et al.* found 29 novel alleles of the ABO subgroups, many identified as point variants within exons 6 and 7[8]. Additional investigations have shown that particular variants in these exons can give rise to new ABO alleles that exhibit distinct phenotypic

characteristics[11,12]. Our study, involving sequencing the *ABO* gene, uncovered multiple variants linked to different ABO subtypes, including a rare variant (c.538C>T) that leads to the p.Arg180Cys change. The PolyPhen-2 analysis suggested that this variant is likely to be damaging, implying it may significantly impact the functionality of glycosyltransferases. The p.Arg180Cys substitution occurs in a crucial area of the glycosyltransferase enzyme, potentially disrupting its structural integrity and functional capacity[13]. Comparable variants documented in other research highlight the significant role of genetic variants in the evolution of ABO subtypes[14,15].

Identifying the cisAB and B(A) phenotypes effectively demonstrates the considerable genetic diversity present within the ABO blood group system[16]. The uncommon cisAB phenotype is characterized by the presence of both A and B antigens on a single allele, often arising from intricate genetic rearrangements. In the current research, a single instance of the cisAB phenotype was recognized, and pedigree analysis validated its inheritance from the mother. The cisAB blood group exhibits unique genetic characteristics; one haplotype possesses the *AB* gene, while another may contain A, B, or O, leading to a distinctive pattern of serological reactions. Those who are heterozygous for the O haplotype display an A_2B_3/A_2B_x phenotype, accompanied by increased anti-H agglutination. In contrast, individuals heterozygous for the conventional A/B allele exhibit an ABx/AxB phenotype, which displays an antigenic dosage effect along with a weakened anti-H response[17,18]. In this study, the child's serology (*ABO*cisAB.01/O.01.01*) revealed a normal A antigen level (4+) and a reduced B antigen level (3+), alongside anti-B antibodies. Meanwhile, the mother's serology (*ABO*cisAB.01*/B.*01*) indicated a decreased A antigen level (2+), a standard B antigen level (4+), and the absence of anti-B antibodies. Genetic analysis revealed that the children possessed the variants 467C>T, 803G>C, and 261delG. This implies that variations in phenotype among individuals sharing the same cisAB genotype might be attributed to modifier genes or epigenetic influences. Specific recombinations of cisAB with traditional *ABO* genes can produce phenotypes that are serologically indistinguishable yet genetically distinct. This underscores the limitations inherent in conventional serological testing. Similarly, there were 3 instances of the B(A) phenotype, characterized by subtle A antigens present on a B allele. These results underscore the importance of molecular genotyping in ensuring accurate ABO phenotyping, particularly in cases of unusual or weakened antigen expression. There are currently 7 B(A) alleles, each carrying variants at the 796C>A locus[19]. No variants at this locus were found in the gene sequences of the participants in this study and their mothers, indicating that both groups might carry a novel allele associated with the B(A) subtype. In addition to exons 6 and 7, nucleotide variations in the intronic and non-coding DNA region could also affect the transcription activity of the element, leading to the reduced ABO antigen expression in erythroid cells[20,21]. In this study, our data showed the promoter mutation spanning positions -35 to -18 was consistent with a previous study by Cai *et al.*[8]. Their study showed that the deletion mutation affected the promoter activity of the *ABO* gene and might influence the ability of RNA polymerase II or other transcription factors to bind to a promoter[8]. However, given that both a 5-year-old (with M expression) and a 3-day-old infant (with weakened B) had the same *ABO*A1.02/B311* genotype, other regulatory or developmental factors may also play a role. This functional uncertainty has now been noted.

This research demonstrated that the proband exhibiting the *ABO*A1.02/B311* genotype showed mixed-field agglutination (MF) when tested with anti-B antibodies, while displaying complete agglutination with anti-A antibodies. The erythrocytes derived from the proband's mother, who has the *ABO*B311/O.01.01* genotype, showed complete agglutination with anti-B antibodies as well. This observation is at odds with the expected serological characteristics associated with the B3 subgroup, which previous studies have described as displaying mixed-field agglutination in reactions with anti-B antibodies. Research suggests that ABO antigen levels present in cord blood erythrocytes are lower than those in adults[22]. This difference is attributed to reduced levels of A and B antigens, resulting in the number of ABO antigens being only 25-50% of those typically found in adults[23]. Furthermore, ABO(H) antigens have not yet fully matured[3] and are only expected to reach adult levels by the age of 2 to 3 years[24]. Daniels indicated that two glycosyltransferases vie for co-receptor materials due to competitive alleles[3]. This contest results in the A antigen

being less pronounced on A2B cells than on A2 cells, and likewise on A1B cells when compared to A1 cells. Consequently, this leads to a notably weakened reaction with human serum, and the incorporation of monoclonal antibodies in our investigation may have additionally influenced the normal serological outcomes for the mother.

Inadequate expression of antigens can lead to the incorrect classification of ABO blood types, which may cause significant clinical issues, such as hemolytic transfusion reactions. This is of critical significance during blood transfusions and hematopoietic stem cell transplants, where accurate blood group typing is essential to avert severe adverse reactions. Research has indicated that acute hemolytic reactions from transfusions occur due to the administration of ABO-incompatible platelets, underscoring the need to use ABO-compatible transfusions, especially in pediatric patients[25]. This research illustrates the essential function of ABO genotyping in transfusion medicine for children, as it guarantees precise blood type identification and improves safety during transfusions.

The analysis of the family trees from 4 selected cases indicates that the weakened expression of ABO antigens was transmitted from the parents, aligning with the autosomal dominant inheritance pattern of ABO blood groups. In the first case, the mother carries the B(A) phenotype, whereas in the second case, the father transmitted the cisAB phenotype. These results underscore the importance of familial studies in understanding the genetics of ABO subtypes and their transmission. The inheritance patterns of ABO subtypes have significant consequences for genetic counseling. It is crucial for parents with ABO blood group variants to be aware that their offspring may inherit these particular traits, potentially heightening the risks related to blood transfusions and organ transplantation. Molecular genotyping serves as a valuable resource for genetic counseling, enhancing the care of pediatric patients who exhibit reduced levels of ABO antigens.

This study provides significant insights into the molecular genetic factors underlying reduced expression of ABO antigens. However, it is essential to acknowledge several limitations. Firstly, due to the relatively small sample size, additional research involving larger cohorts is needed to validate the findings of this study. Secondly, while bioinformatics analyses have indicated the potential functional implications of the rare variant (c.538C>T), experimental validation is required to ascertain its influence on glycosyltransferase function. Lastly, this investigation concentrated on a pediatric population in China, meaning that the conclusions may not be applicable to wider demographic groups. Future studies should aim to evaluate the prevalence of ABO subtypes among diverse populations and to investigate the impact of specific variants. Moreover, it is vital to develop standardized protocols for ABO genotyping in children to improve transfusion safety and overall clinical outcomes.

CONCLUSIONS

This research illustrates that certain genetic variants are linked to weakened ABO antigen expression in infants and young children. Among these is a rare variant (c.538C>T) leading to the alteration p.Arg180Cys. The investigation revealed that 4 infants aged <4 months had decreased expression of ABO subgroup-related antigens. Specific combinations of genotypes (*ABO*A1.02/Bw07*, *ABO*A2.05/B.01*, *ABO*A2.05/O.01.01* and *ABO*A1.02/B311*) demonstrated a reduced expression pattern typical of ABO blood group variants. These results underscore the importance of molecular genotyping in accurately identifying ABO blood groups, which is essential for ensuring safe transfusion practices in pediatric patients. Further studies are necessary to clarify how particular variants influence functionality and to develop standardized protocols for ABO genotyping in clinical settings.

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Authorship contributions

QR conceived the study, curated the data and visualization, and wrote the original draft. JL provided samples. CL

curated the data. XL supervised the study. XM conceived and supervised the study. All Authors reviewed and edited the manuscript for publication.

The Authors declare no conflicts of interest.

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