Rare Blood, Rarer Genes: Mumbai's First Réunion Phenotype Blood Donor with a Novel FUT2 Mutation.

Elvis A¹, Rati D², Monali L², Harita M², Sangeeta S¹, Amruta I¹, Swati K², Lincy J³

¹S.L. Raheja Hospital -A Fortis Associate Mahim, Mumbai - 400016

²ICMR-National Institute of Immunohaematology, Parel, Mumbai 40012

³Dr L. H. Hiranandani Hospital, Powai, Mumbai, 400076

Background & Objectives: The Réunion phenotype, an H-deficient blood group variant, arises due to mutations in the FUT1 and FUT2 genes, leading to <u>partial absence of H antigen</u> on RBCs and non-secretor status. This case highlights a 37-year-old male donor with a blood grouping discrepancy later identified as A_h (Réunion phenotype), stressing the role of molecular diagnostics in differentiating it from the Para-Bombay phenotype.

	H-deficient non- secretor (Bombay)	H-deficient secretor (Para Bombay)	H-partially deficient Non- Secretor (Réunion)
Secretory status	Non- secretor	Secretor (H,A,B)	Non-secretor
Adsorption and elution	Neg (H, A, B)	H (neg/ +/wk+), A (neg/ +/wk+), B (neg/ +/wk+).	H (wk+)
Antibodies	Anti H	Anti IH	Anti H
Notations	O _h Indian	O _h ⁰ -secretor O _h ^A -secretor O _h ^B -secretor	A _h , B _h , AB _h

A antig B antig	RI	ED CEL	LS TESTS	t	Discordant Red cell	and serum Gro							
A antig B antig		ED CEL	LSTESTS				ouping (Type	II / Ty	oe IV)				
A antig B antig	Cell		12010	•		SERUM TESTS	1				-	SALIVA TESTS	
A antig B antig		Anti	Anti B		Reverse t	yping		IAT		Negative		Secretory Study	1
B antig	yping	Anti	Neg	Temperature of	Acel	B cell	O cell/ S	creening	cells	No		Secretory study	J
		neg neg	+ neg	testing 4°c	IgM type Anti A	IgM type anti B	? cold agglu	tinin/Ar	ti IH	antibody detected In serum			_
				22°C/RT	IgM type Anti A	IgM type anti B	? cold agglu	tinin/Ar	ti IH			Non secretor	Secretor
Ant	ti H with (Co tisera (Anti a Bombay p	H lectin) / Serum	37°C/ AHG	IgG type Anti A with wide thermal amplitude	IgG type Anti B with wide thermal amplitude	IgG type and High freque Anti IH with amplitude)	ncy Anti	body/				
	_	_		-				+		1			
+		Ne	gative					Anti	Anti H	Hm		1	
1 1	Adsorptio	on and	lution				Cord cells	neg	+			i i	
1 1	**wk*	/ \.					Bombay RBC	neg	neg	4		1	
i ian	DAT)	i i	Vegative			1	Adult O RBC	+	+			1	
1	H antigen		w !					-		O _h Phe	notype	→ Bombay O ₅	O _b o secret
1 -									i				
1	A antigen	wk+	V							- An Phen	otype	-	O _h ^A secreto
(+)	B antigen		,							B _h Phen	otype		Oh B secreto
Hm	1						1	i					I
				ulex europaeus lectin cells (A ided as polyclonal in nature		tibodies from sera of B	iembay	+	+	H-partially deficient Non-seco		H deficient Non secretor (Bombay)	H deficient Secretor (Para Bomi

	Anti A	Anti B	Anti AB	Anti-D	A cell	B cell	O cell
Tube Method	4+	0+	4+	4+	2+	4+	4+
CAT Method	4+	0+	4+	4+	2+	4+	2+

SEROLOGICAL TEST RESULTS

TEST	METHOD	RESULT
Anti-H Lectin	Tube	Negative
Anti-A1 Lectin	Tube	Negative
DAT	CAT	Negative
IAT	CAT	Positive
Antibody Screening	NEO 2.0	Negative
IAT with Bombay Positive Cells	Tube	Negative
Secretor Studies	Tube	Non-secretor for A, B, H.
Impression		A _h Réunion Phenotype



A 17 Fortis ASSOCIATE

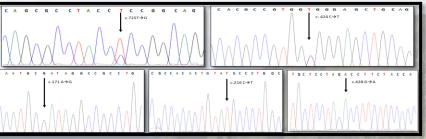






MOLECULAR FINDINGS:

FUT1: FUT1*01N.09/FUT1*01W.23 (Weak H Ag on RBCs), FUT2: se^{171,216,428}/
se^{171,216,428} (non-secretor), ABO: ABO*A1.01/ ABO*A1.01 (A Ag on RBCs)







You ask what is the use of classification, arrangement, systemization? I answer you: orde and simplification are the first steps toward the mastery of a subject-the actual enemy is the unknown.

(Thomas Mann)

DISCUSSION					
Levine P. Vox Sang. 1961.	First A _h Case.				
H.M. Bhatia. Vox Sanguinis 1962.	Incomplete expression of genes.				
Gerard G. Am J Hum Genet 34:937- 947, 1982	Reunion Island case series of 42 H- deficient phenotype individuals.				
Le Pendu J. Am J Hum Genet. 1983.	Proposed to call this variant weak H phenotype as Réunion.				
Fernandez-Mateos P, Cailleau A, Henry . Fernandez-Mateos P, Cailleau A, Henry S, et al. Vox Sang. 1998.	First study to elucidate Molecular Work-up of a Réunion case.				
Dhiman Y. et al. Transfus Apher Sci. 2023	First Indian Réunion Study that addresses challenges with classification of Para Bombay and Réunion Phenotypes.				

Conclusion: This case highlights the importance of integrating molecular diagnostics with serology to accurately classify the Réunion phenotype, thereby ensuring precise blood group identification and safe transfusion practices.