

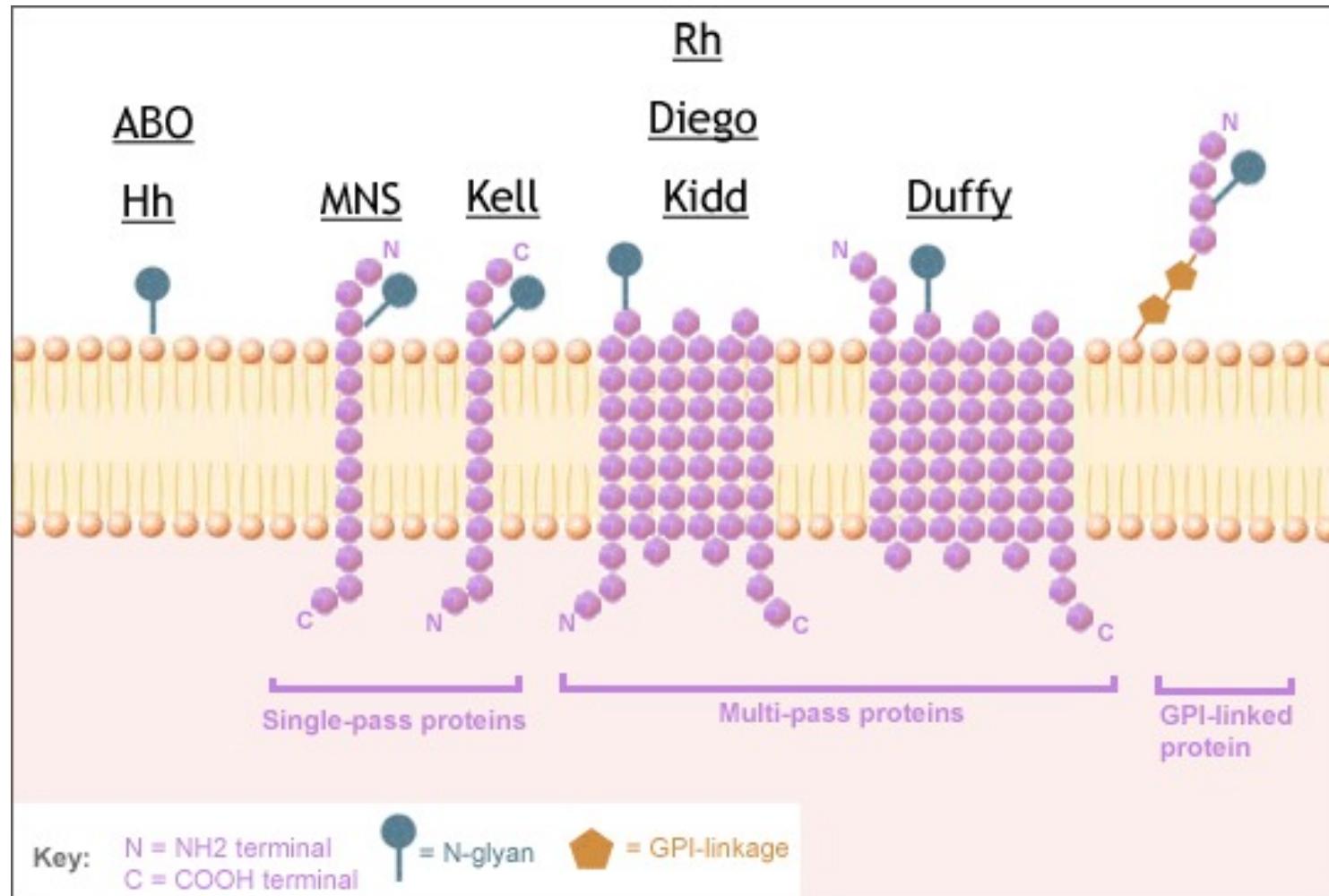
Genetics of human blood groups



IGIB
INSTITUTE OF GENOMICS
& INTEGRATIVE BIOLOGY
Genomics Knowledge Partner

Mercy Rophina
Senior Research Fellow
CSIR – Institute of Genomics and Integrative Biology (IGIB)

What are blood group antigens ?

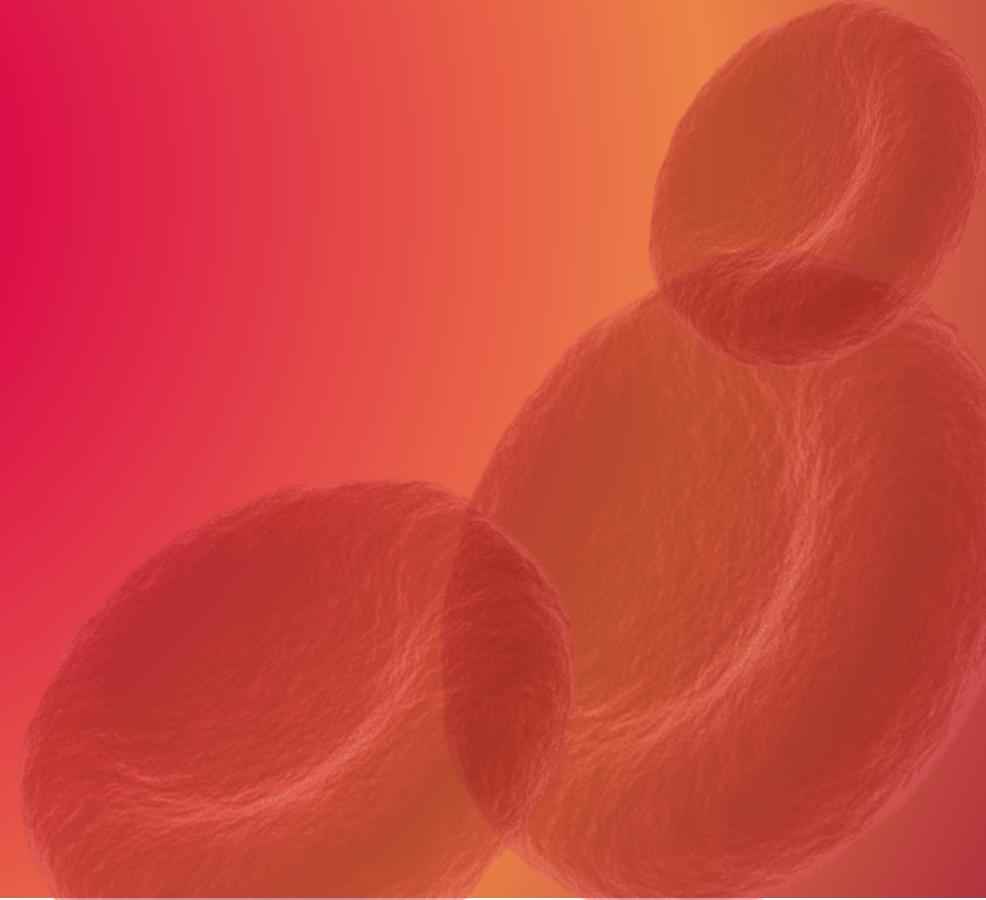


Blood group antigens in the human RBC membrane

Image source : Blood Groups and Red Cell Antigens

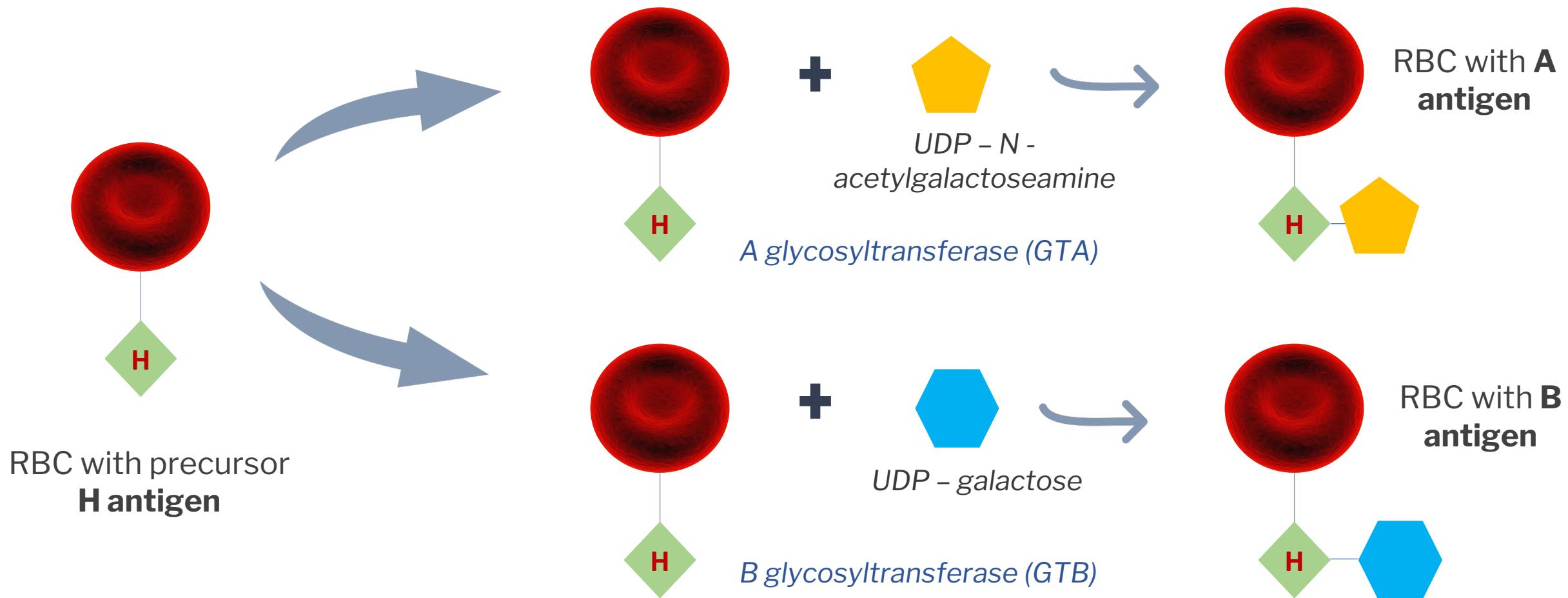
How are these blood group antigens formed ?

What is the role of genetics behind this ?



The ABO story - Illustration

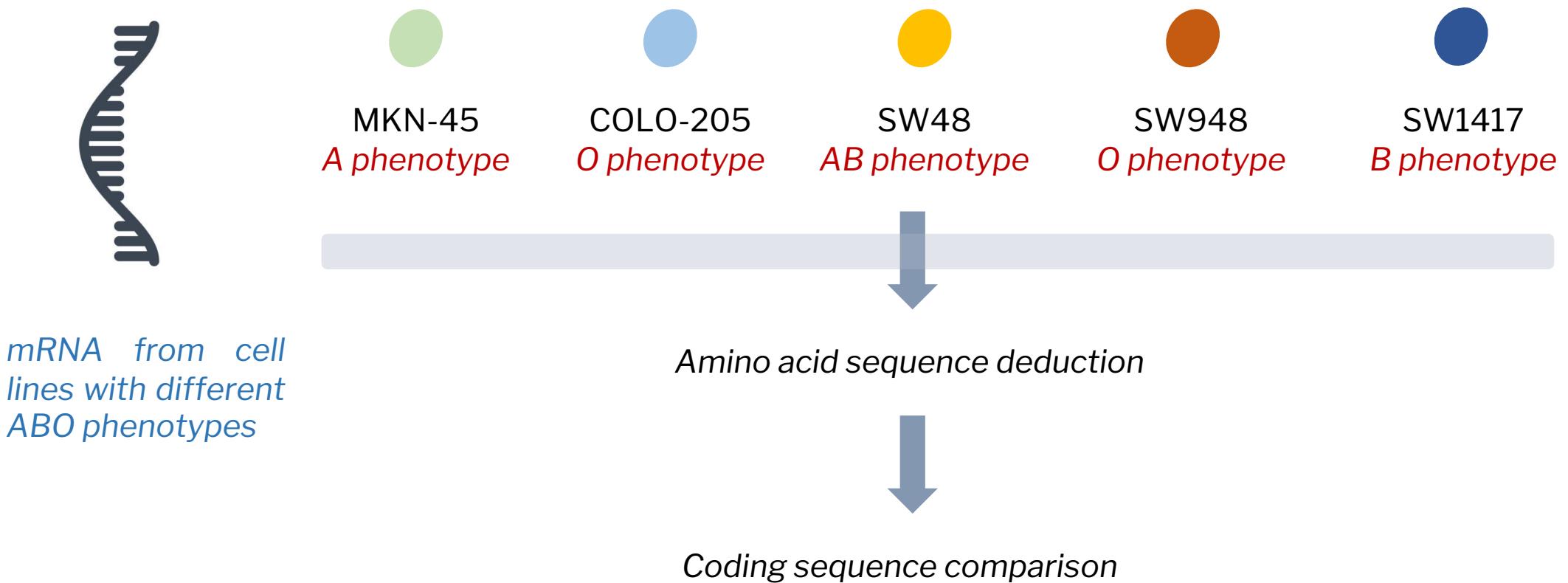
How are these blood group antigens formed ?



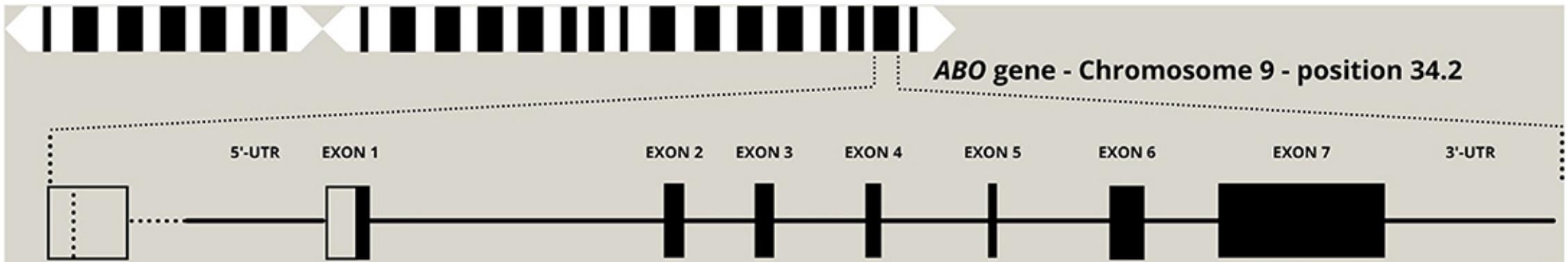
ABO story – Genetic view

How are these blood group antigens formation regulated ?

What is the molecular mechanism behind this ?



ABO story – Genetic findings



3 major alleles

A allele – Produces A transferase enzyme

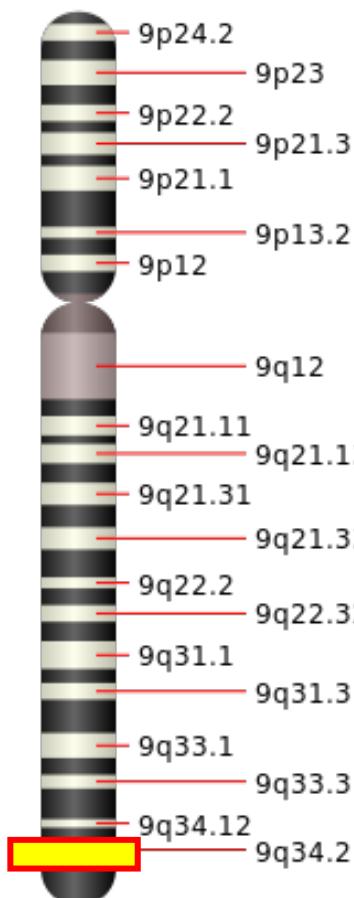
B allele – Produces B transferase enzyme

O allele – Lacks both enzymatic activity

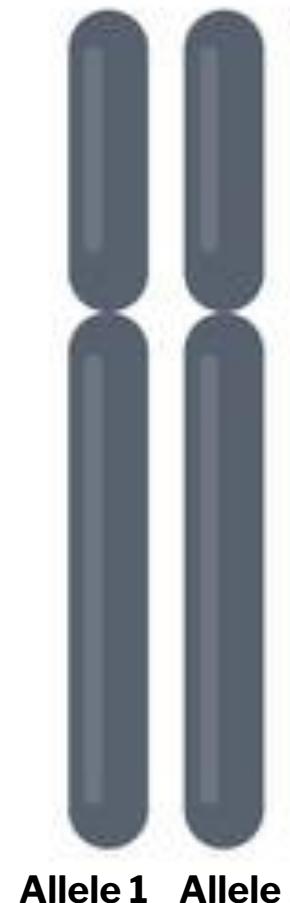
How to identify and categorize these alleles ?

ABO story – Genetic findings

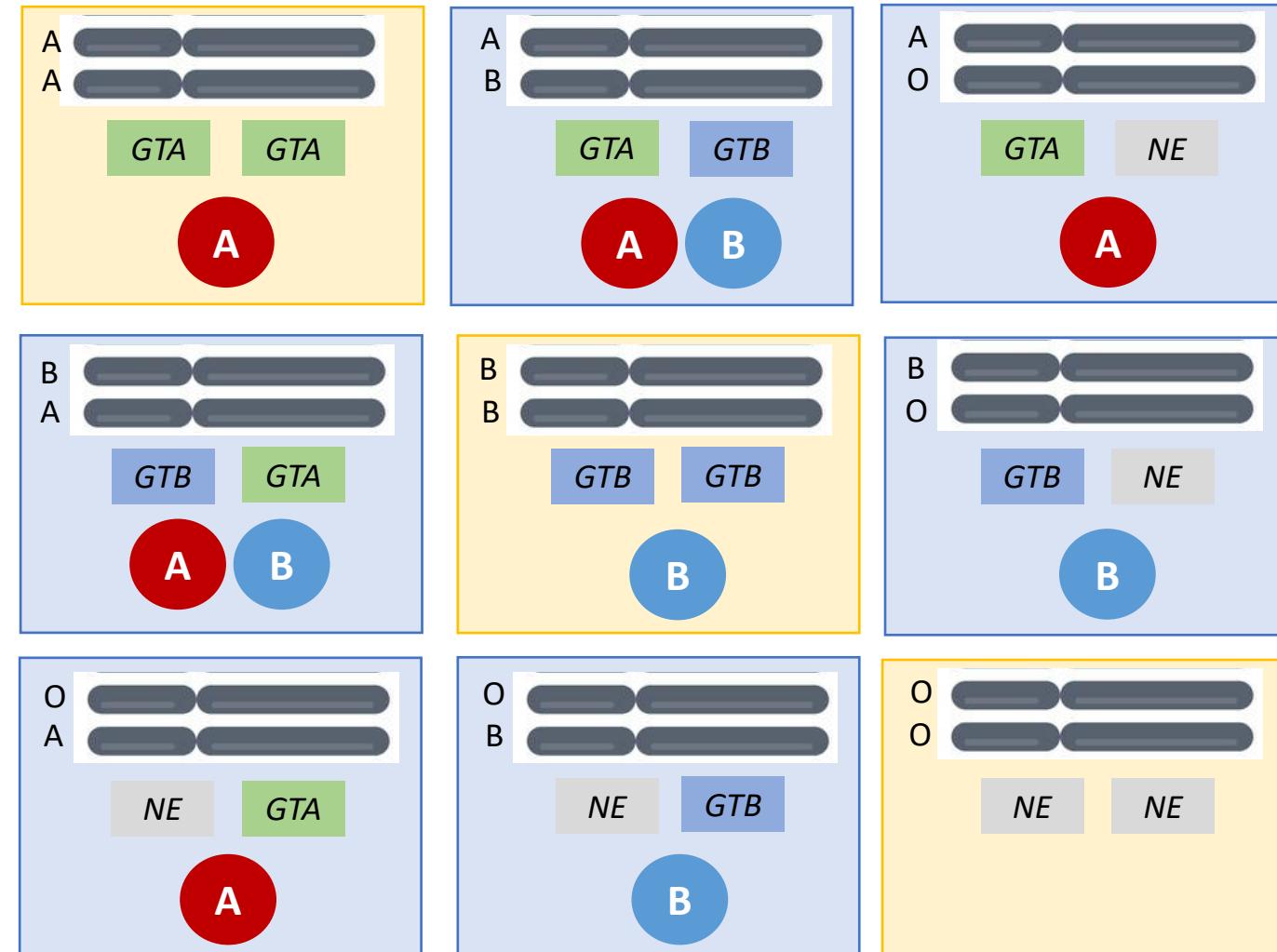
Chr 9



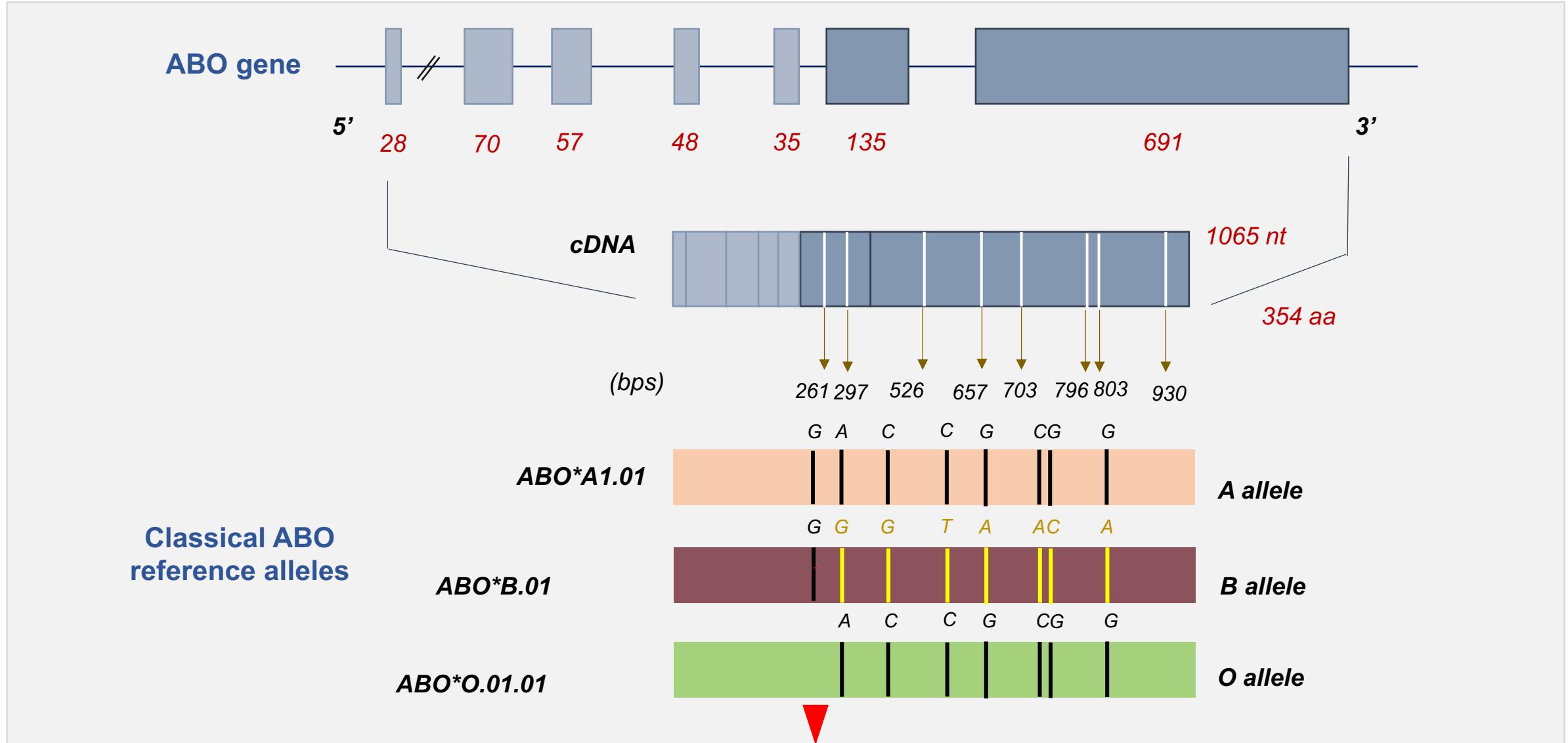
Diploid nature



ABO allele combinations



ABO story – Genetic findings



Human blood groups – The gene play

1900

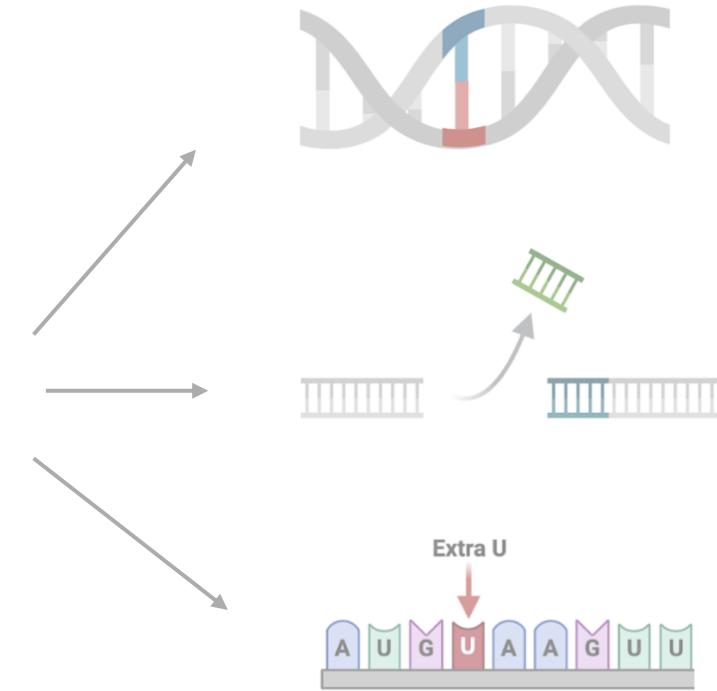


Uncovering the **genetic background** of human blood group systems

1986

51

Unique genes



Single Nucleotide

Deletions

Insertion

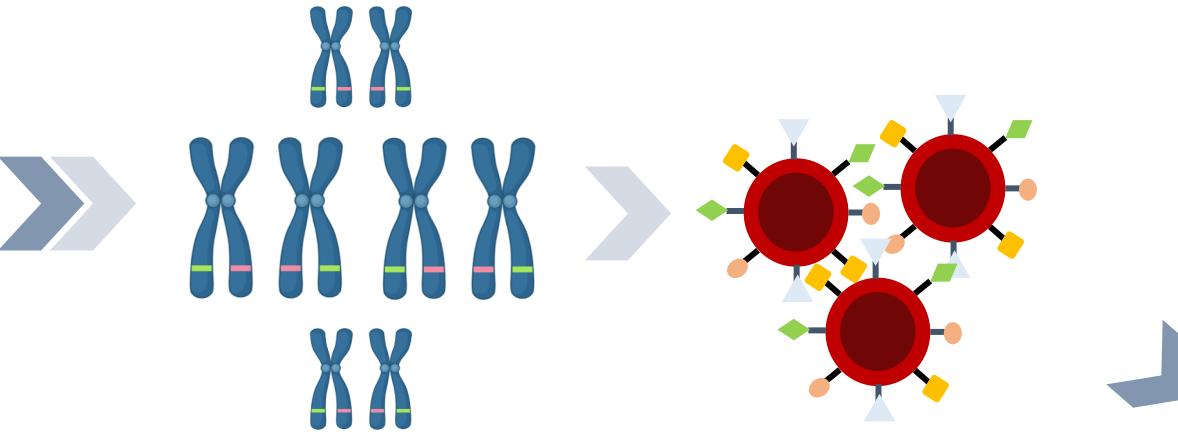
Variable expression of Antigens

Variation in Blood Phenotype

Pieces of the puzzle

51

Blood
group
genes

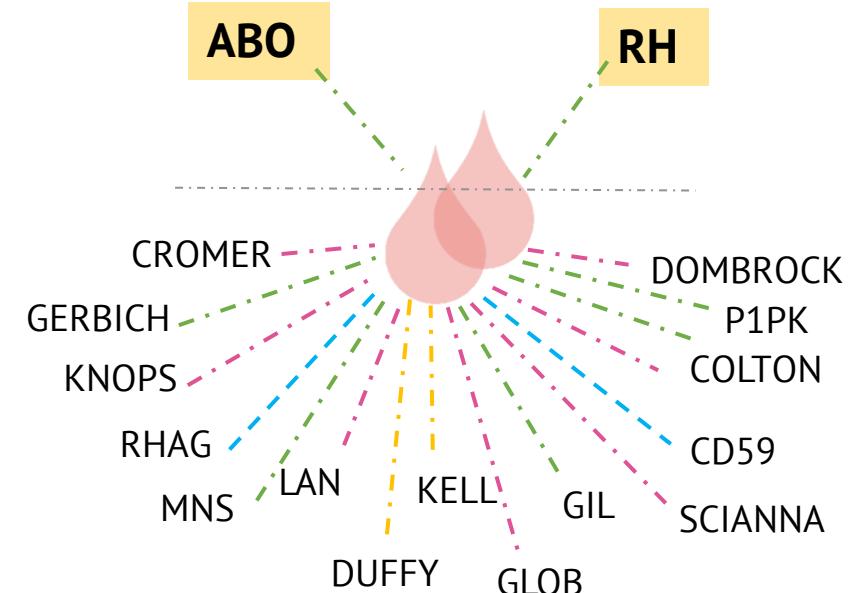


1873
genetic
variants

349
blood
antigens



634
blood
phenotypes



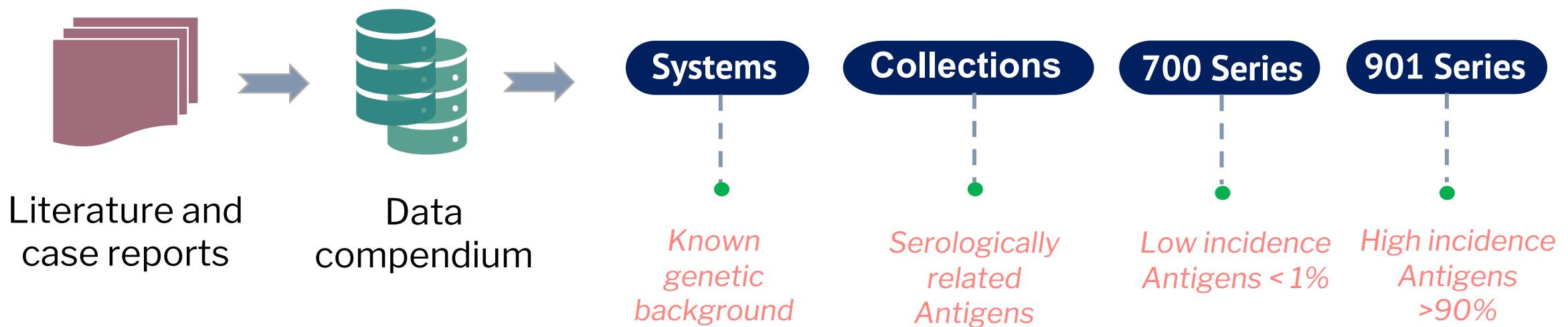
44
BG systems

Blood group antigen genetics

Resources & repositories

Blood Gene Allelic Variations Maintenance & Nomenclature

A **systematic maintenance** of discovered antigens along with their genetic background



This screenshot shows the homepage of the International Society of Blood Transfusion (ISBT) Red Cell Immunogenetics and Blood Group Terminology website. The header includes the ISBT logo and navigation links for Home, ISBT Academy, and Working Parties. The main content area features a red banner with the text 'Red Cell Immunogenetics and Blood Group Terminology'.

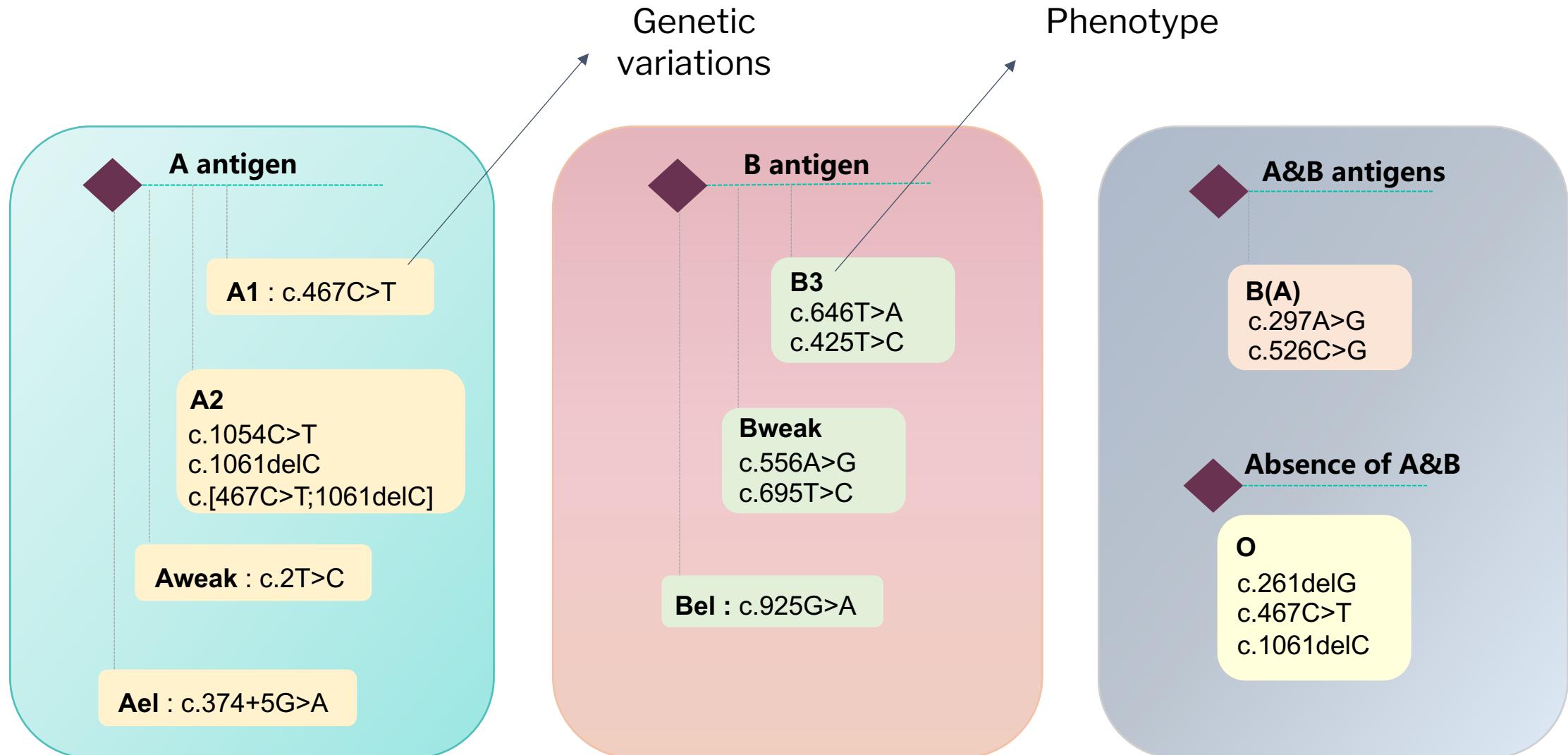
This screenshot shows the ERYTHROGENE search engine website. The header features a DNA helix icon and the text 'ERYTHROGENE Updated!'. Below the header, it says 'The search engine for blood group genes'. There is a search bar with the placeholder 'Gene, variant, etc.' and radio buttons for 'All', 'Reference', and '1000 Genomes'. At the bottom, there is a note: 'Search examples: ABO, RHD, ABO*A2, c.261delG, p.Trp16Cys, 1136'.

The Human RhesusBase

This screenshot shows the Antigen Atlas & Typing website. The header features a green and blue graphic of a blood cell. The main text reads 'Antigen Atlas & Typing'.

Blood Gene Allelic Variations

The ABO story



Blood Gene Allelic Variations

The ABO story

Blood gene	Variants	Antigens	BG system	Phenotypes
ABO gene	149 variants (SNVs, Indels)	2 antigens A and B	ABO BG system	13 phenotypes A1, A2, A3, Ael, A1 -C.467C>T, Am, Aweak, Aweak -C.21>C Ax, B, B3, B3 -C.646T>A, Bweak, Bel, B3 -C.425T>C AB, CisAB

International Society of Blood Transfusion

Red cell immunogenetics and blood group terminology

Summary

- Expression of RBC antigens are genetically controlled
- Blood groups follow **Mendelian pattern of inheritance**
- As of date, **51** genes are associated with blood groups (human blood group genes)
- More than **1800** genetic variations have been reported from blood genes
- Over **600** phenotypes have been genetically very well characterized
- Currently there exist a total of **44** human blood group systems and **2** erythroid specific transcription factors

Upcoming courses ...

- How genetic findings of blood groups improved blood group profiling ?
- Genomics in advancing the future of blood group profiling
- From genomes to populations – Population specific blood group registries

Thank

You

