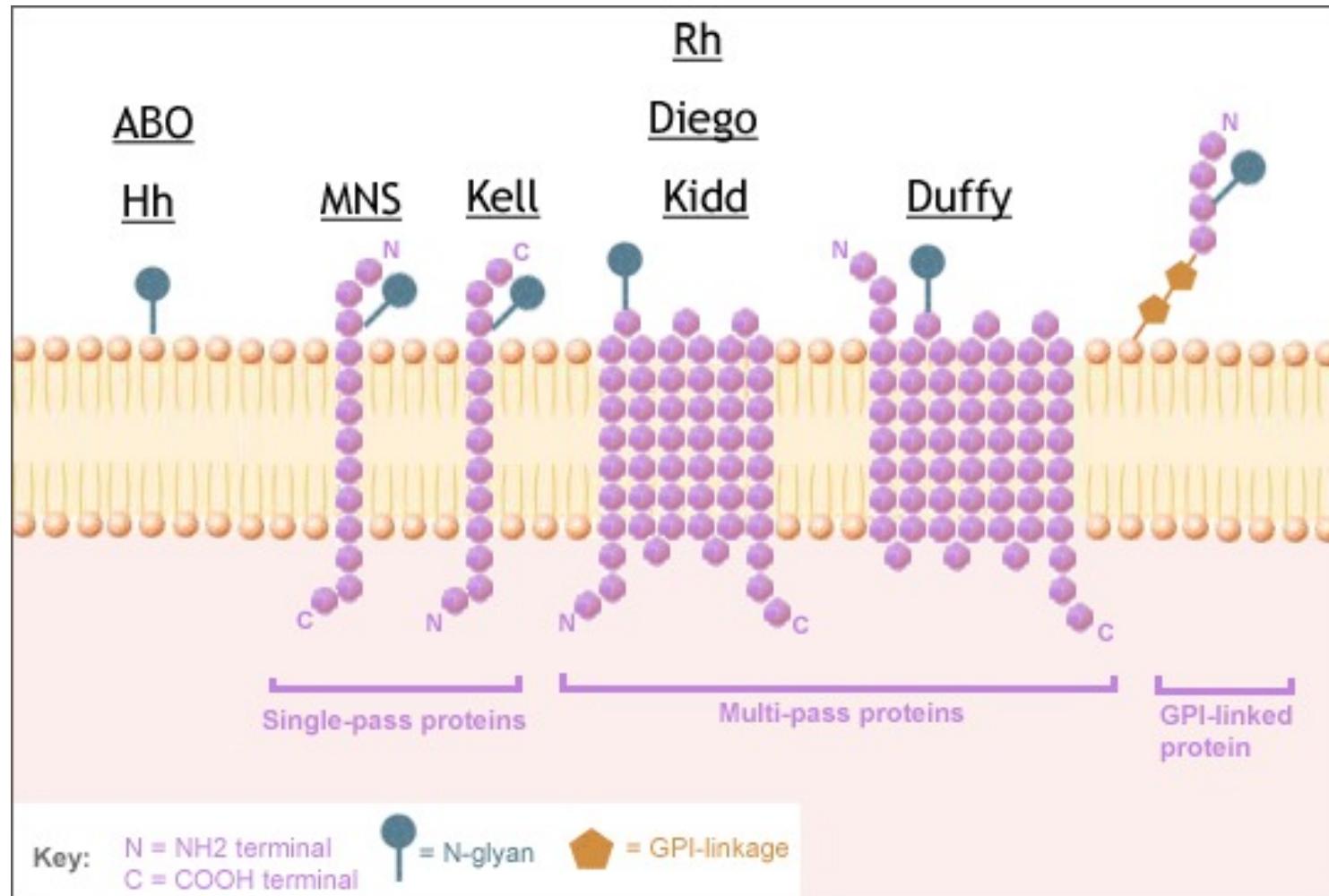


# Genetics of human blood groups



# What are blood group antigens ?

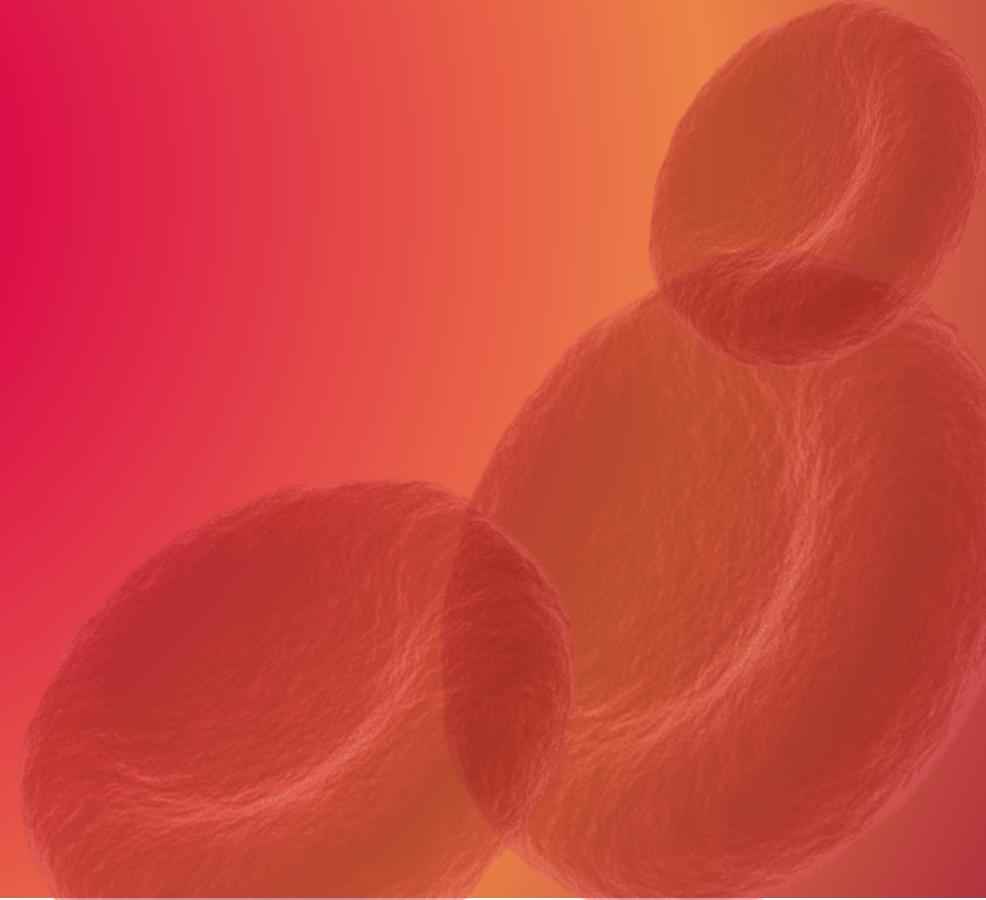


Blood group antigens in the human RBC membrane

Image source : Blood Groups and Red Cell Antigens

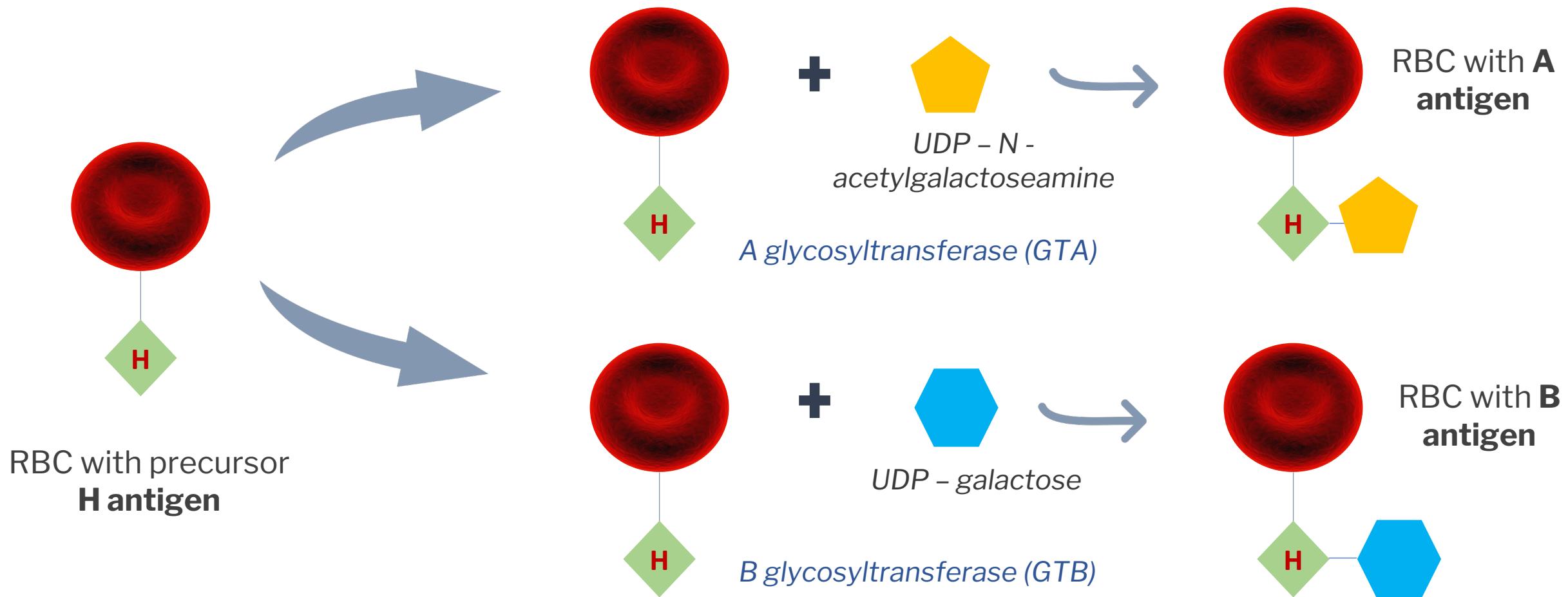
# How are these blood group antigens formed ?

*What is the role of genetics behind this ?*



# The ABO story - Illustration

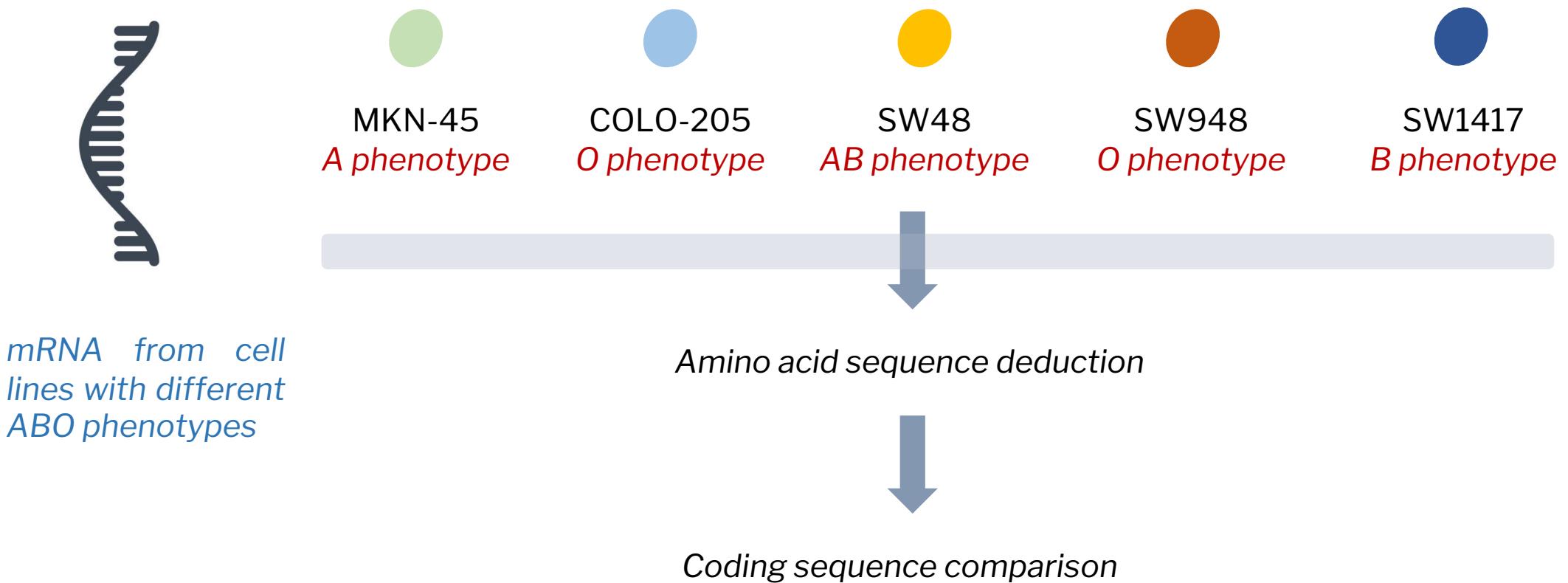
How are these blood group antigens formed ?



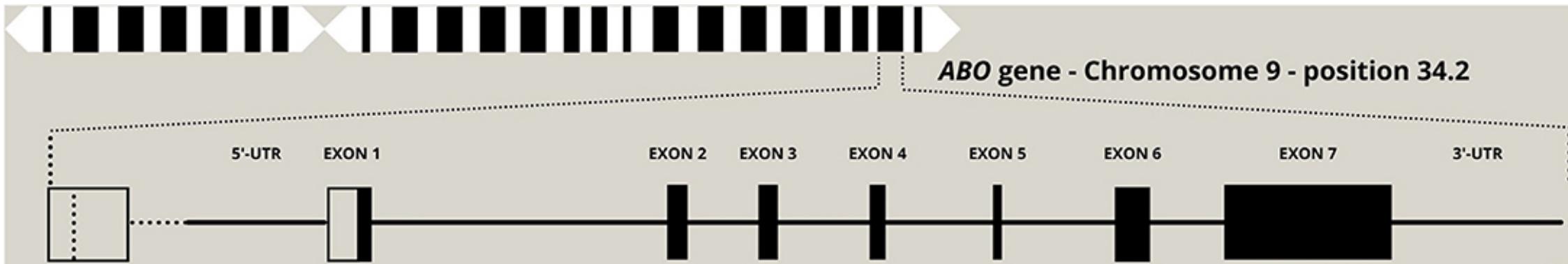
# ABO story – Genetic view

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

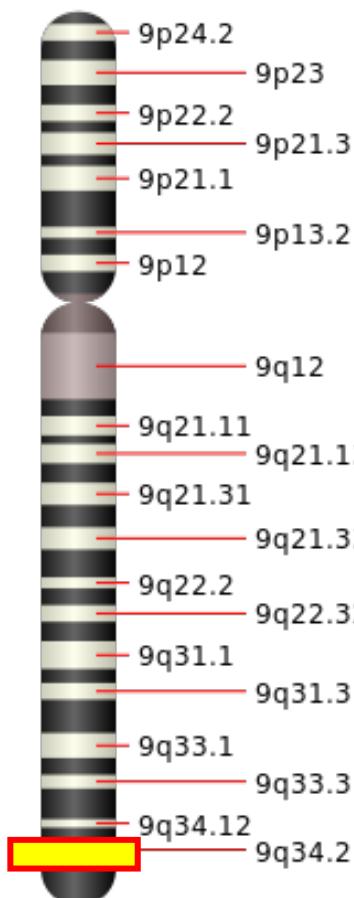
**B allele** – Produces B transferase enzyme

**O allele** – Lacks both enzymatic activity

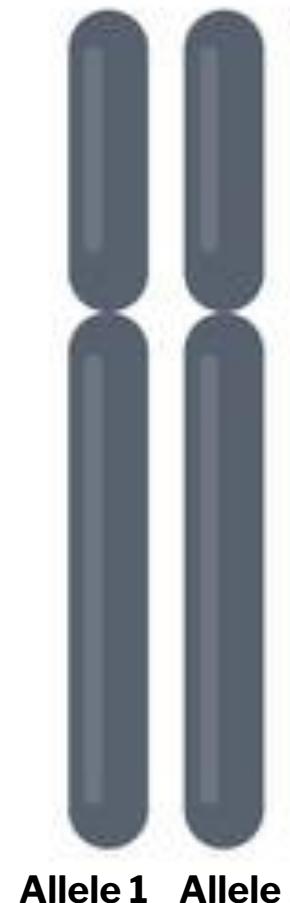
**How to identify and categorize these alleles ?**

# ABO story – Genetic findings

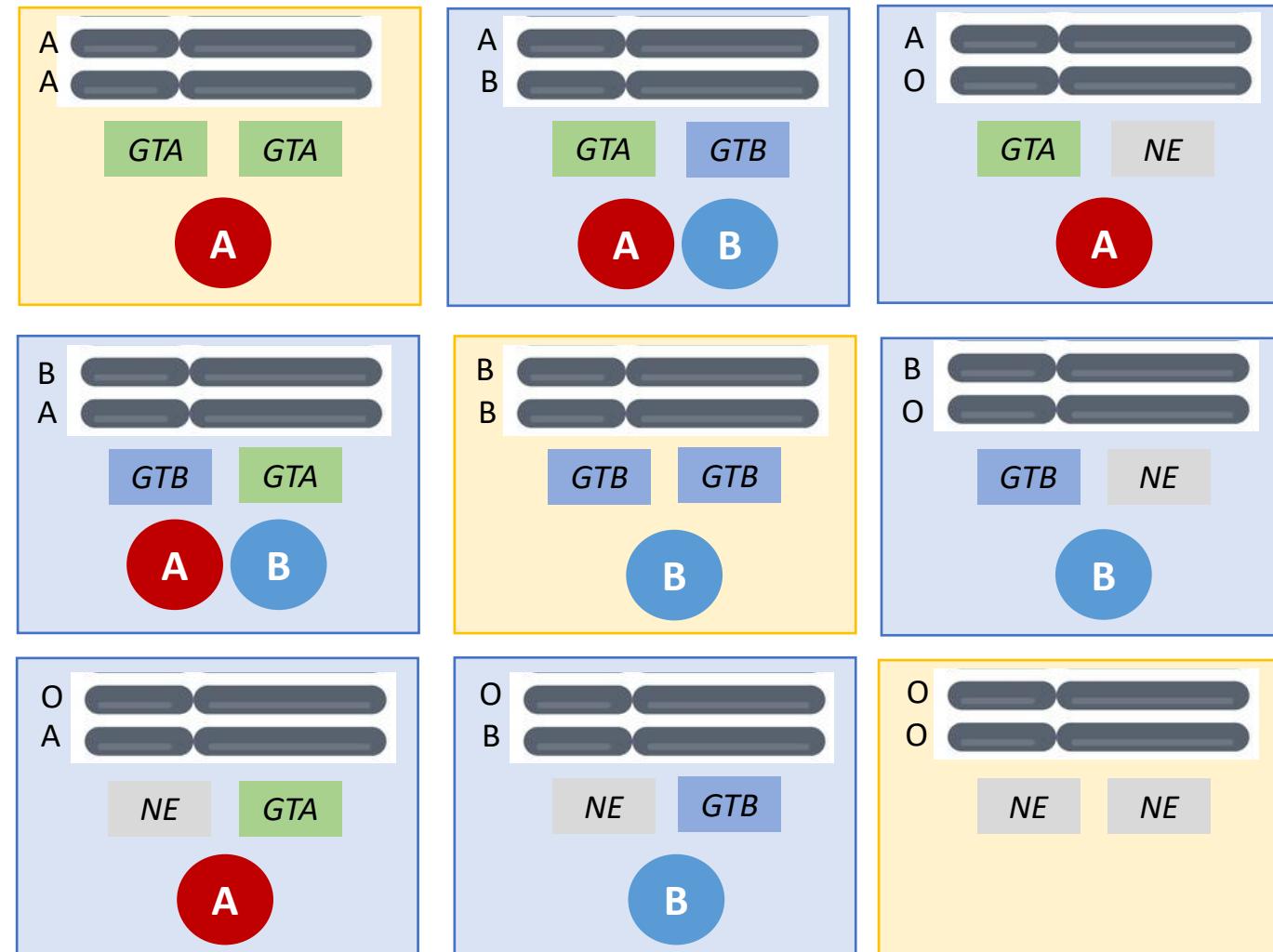
Chr 9



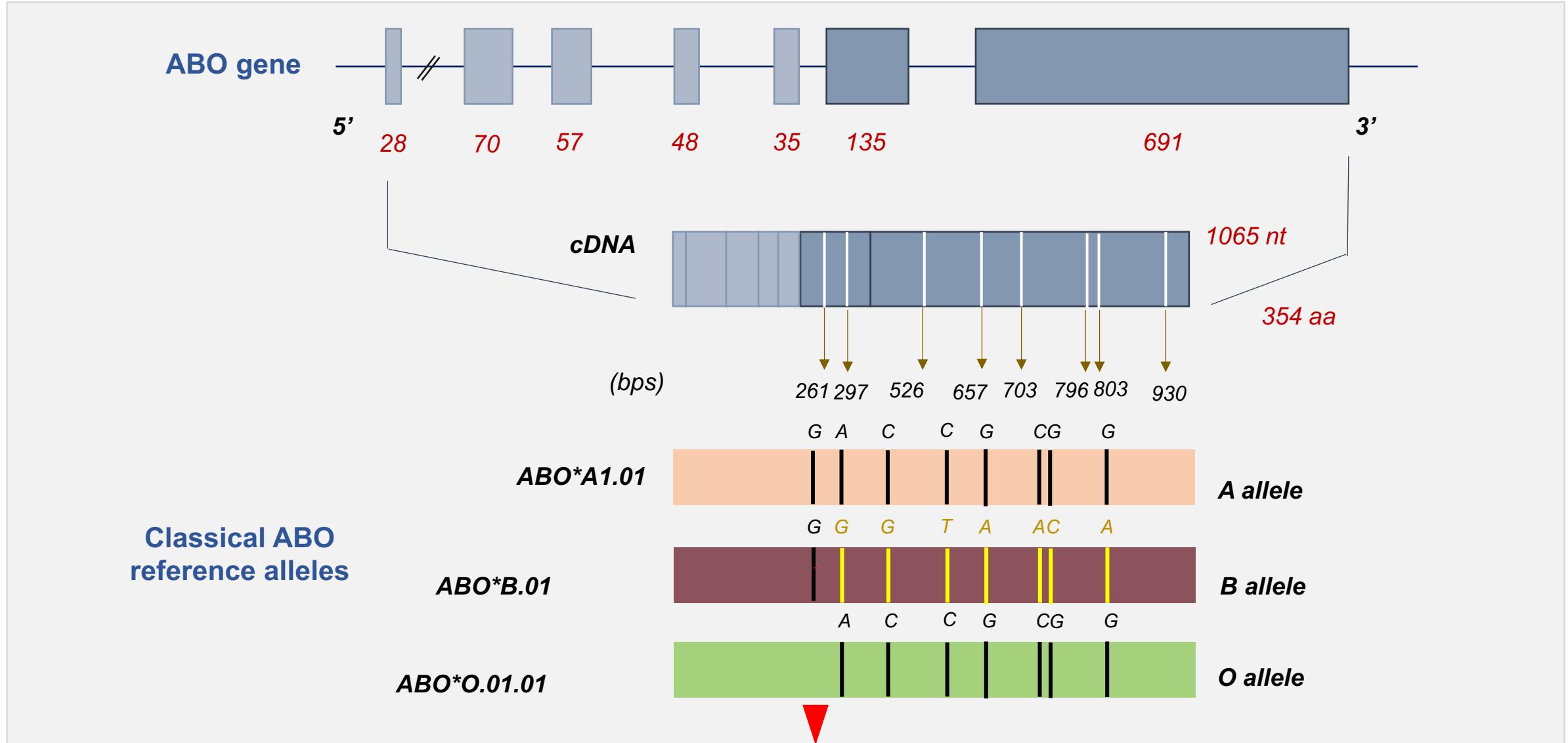
Diploid nature



ABO allele combinations



# ABO story – Genetic findings



# Human blood groups – The gene play

1900

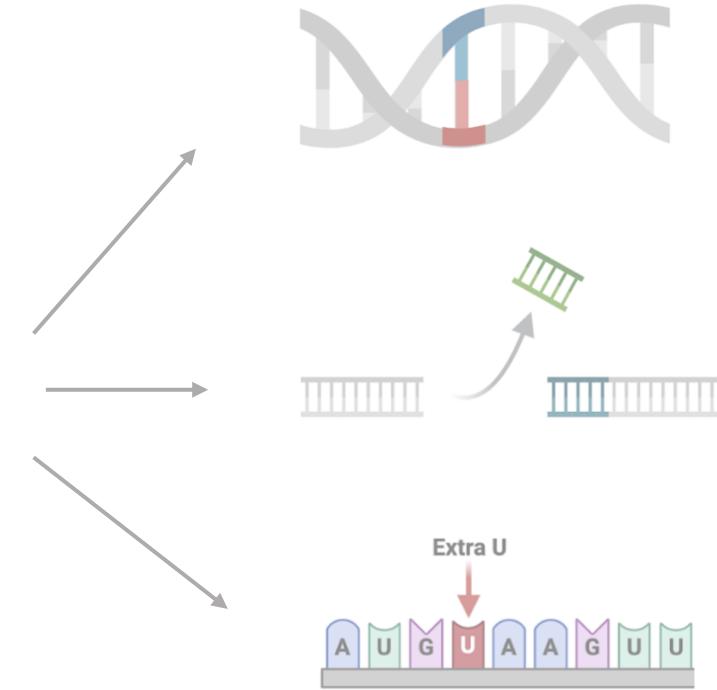


Uncovering the **genetic background** of human blood group systems

1986

51

Unique genes



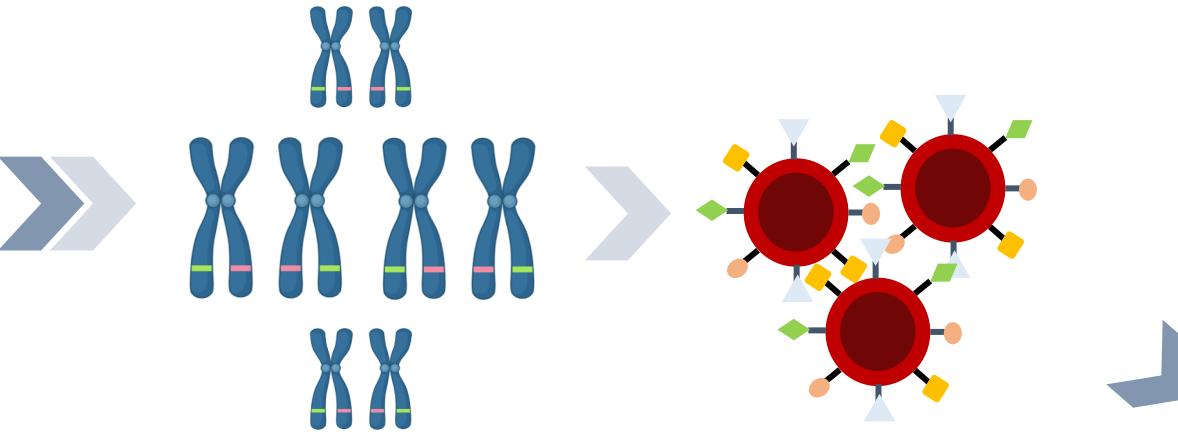
Variable expression of Antigens

Variation in Blood Phenotype

# Pieces of the puzzle

51

Blood  
group  
genes

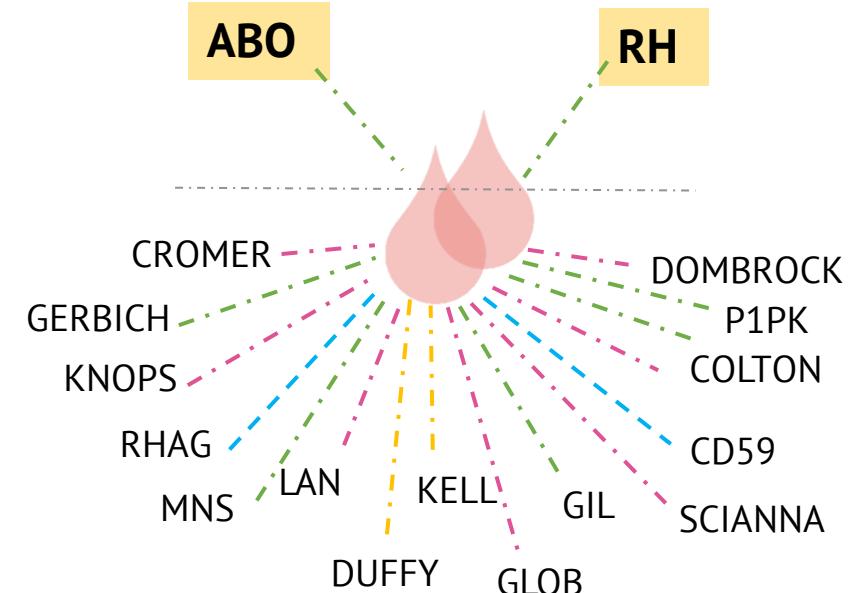


1873  
genetic  
variants

349  
blood  
antigens



634  
blood  
phenotypes



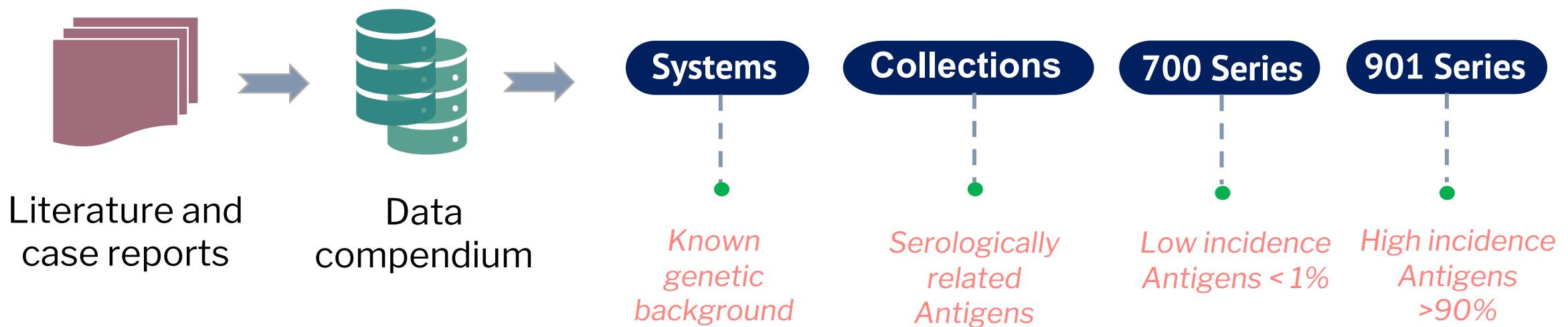
44  
BG systems

# 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'.

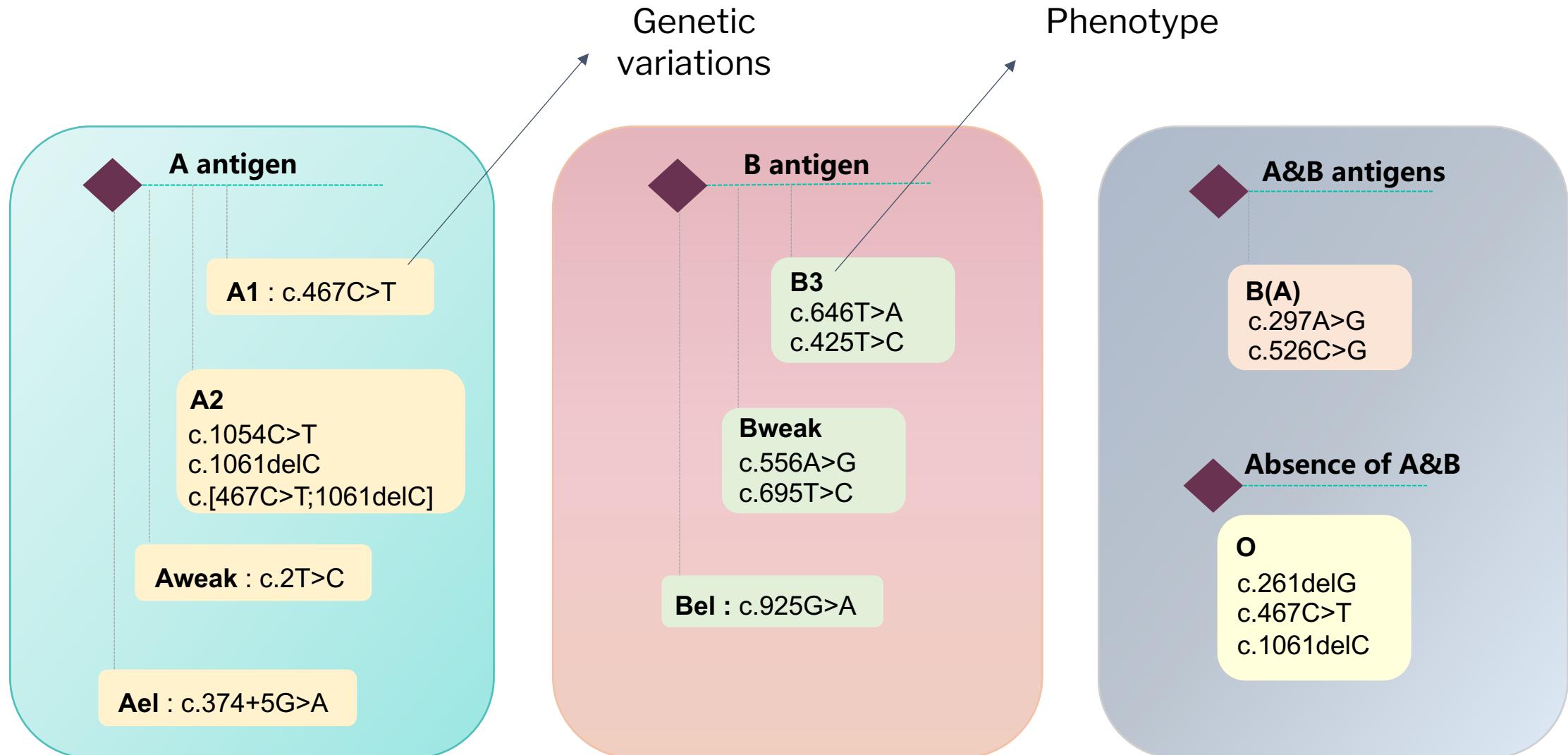
This screenshot shows the ERYTHROGENE search engine for blood group genes. The header features the ERYTHROGENE logo and the text 'Updated!'. Below the header, it says 'The search engine for blood group genes'. There is a search bar with the placeholder 'Gene, variant, etc.' and radio buttons for 'All', '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 logo with a stylized DNA helix and the text 'Antigen Atlas & Typing'. The main content area contains a large green button with the text '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 <b>A and B</b>	ABO BG system	<b>13 phenotypes</b> A1, A2, A3, Ael, <b>A1</b> -C.467C>T, Am, Aweak, <b>Aweak</b> -C.21>C Ax, B, B3, <b>B3</b> -C.646T>A, Bweak, Bel, <b>B3</b> -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

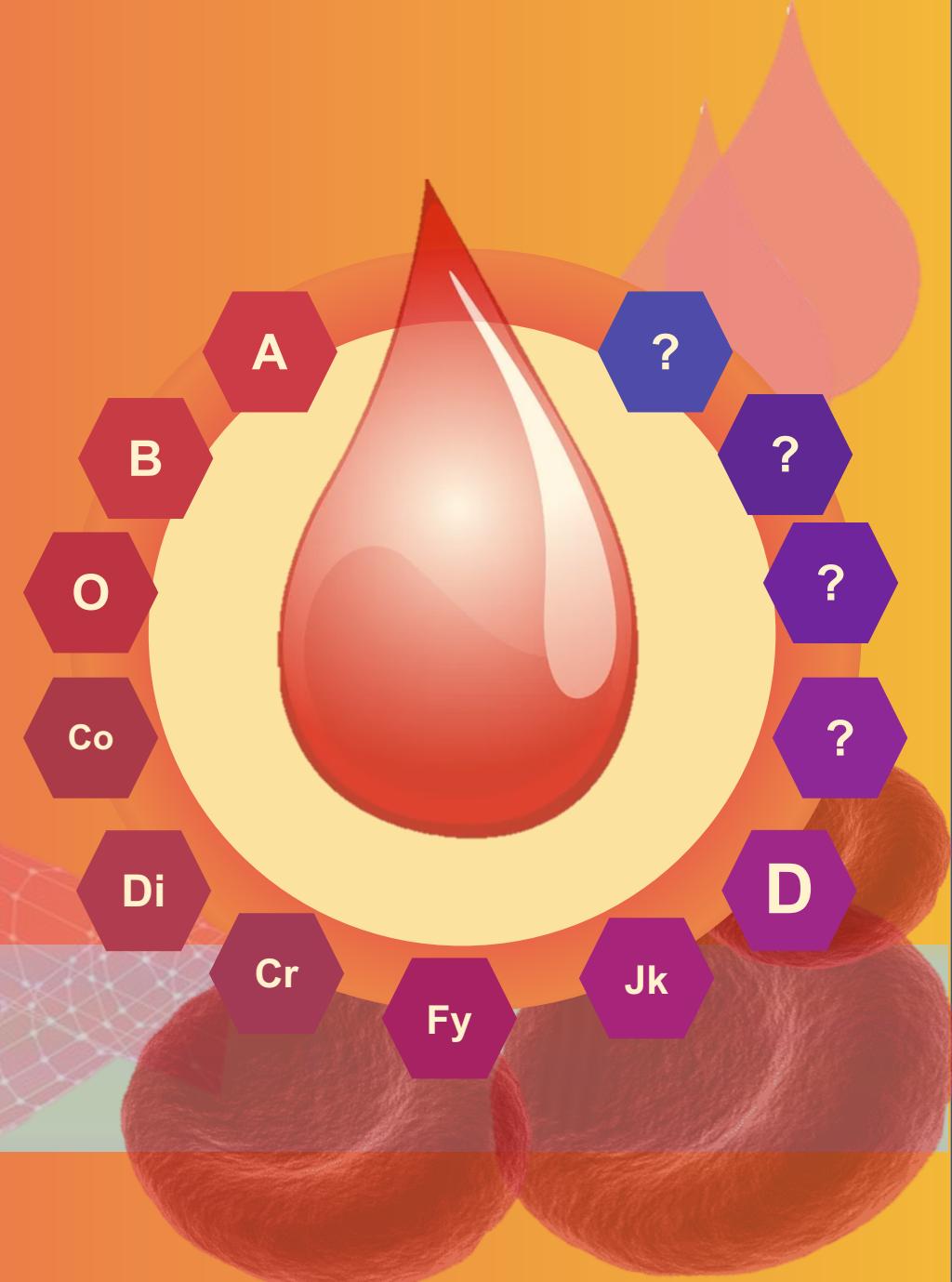
You



# Genetics of human blood groups



Mercy Rophina  
CSIR-IGIB



# Genetics of human blood groups



Mercy Rophina  
CSIR-IGIB

