

Genomics in Immunohematology



IGIB
INSTITUTE OF GENOMICS
& INTEGRATIVE BIOLOGY
Genomics Knowledge Partner

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“Although our scientific understanding of the functions of blood is more sophisticated than ever, we remain in thrall to its primary mystique”

*Jerome Groopman
The New Yorker*

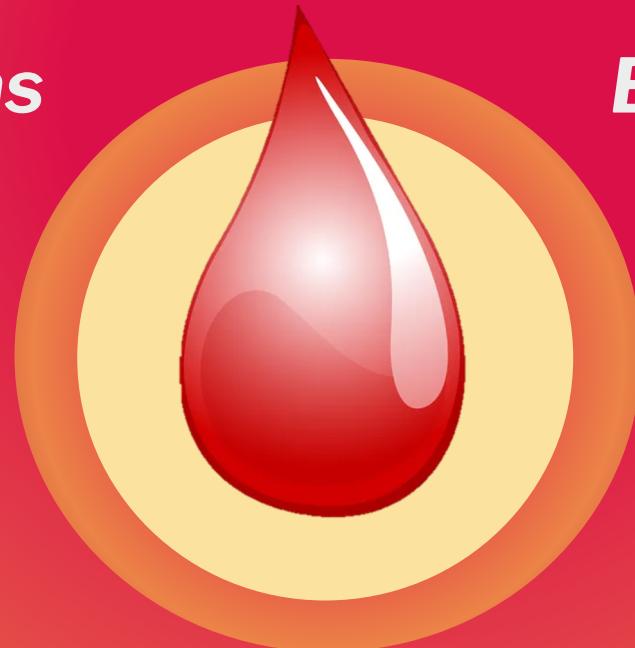
Aims & Highlights !

Human blood groups

RBC antigens

Variability of blood

History of discovery



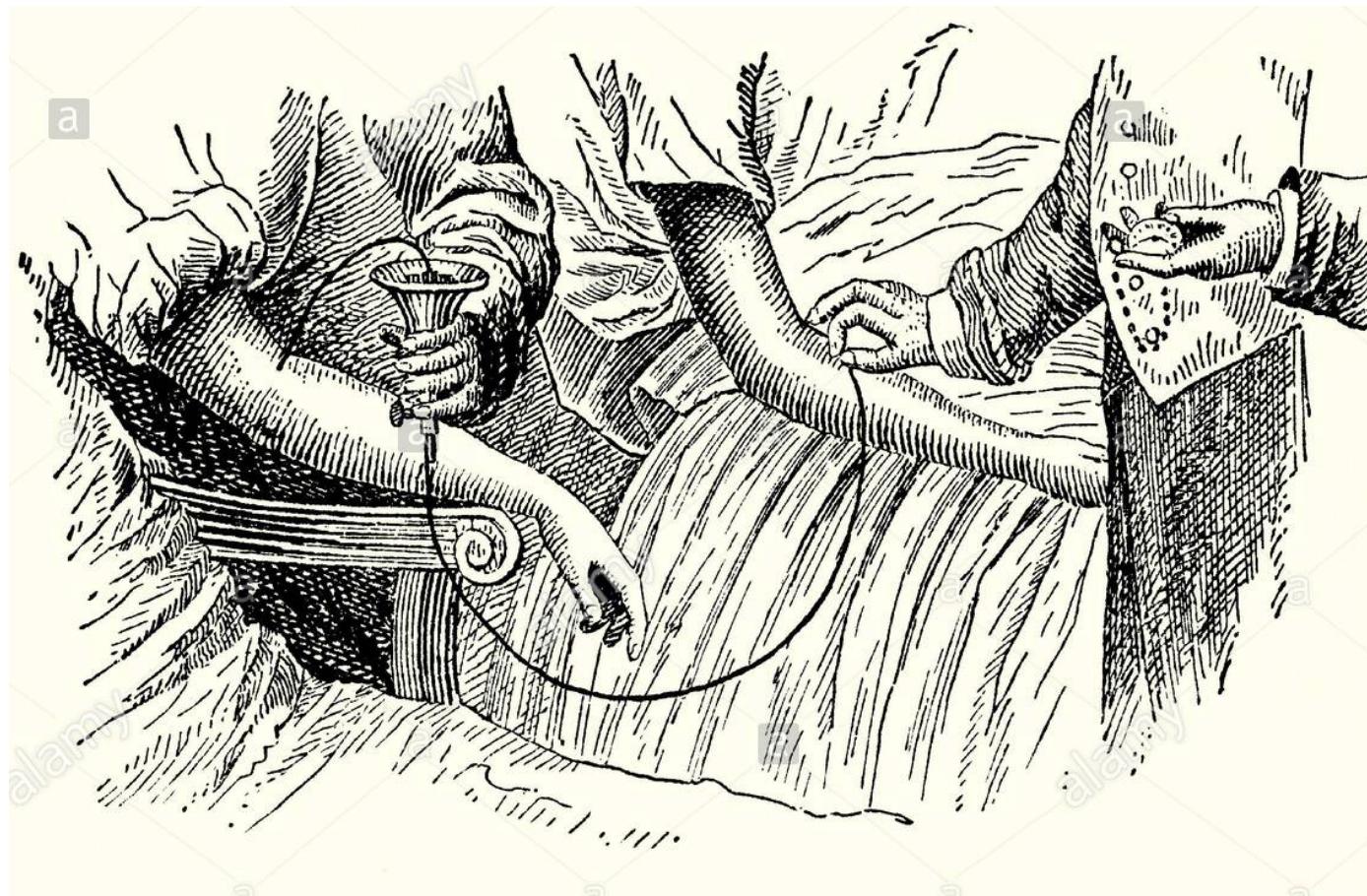
Blood group profiling

Molecular diagnostics

NGS & Future ?

The elixir of human life

Human blood – Connective tissue composed of *cells* suspended in liquid matrix called *plasma*



Tales from history

The ancient Greek Mythology

- *Blood – Ichor*
- *A magical elixir*

Loss of blood leads to death

Blood can cure illness

Average volume of blood in adults ~ **6 litres**

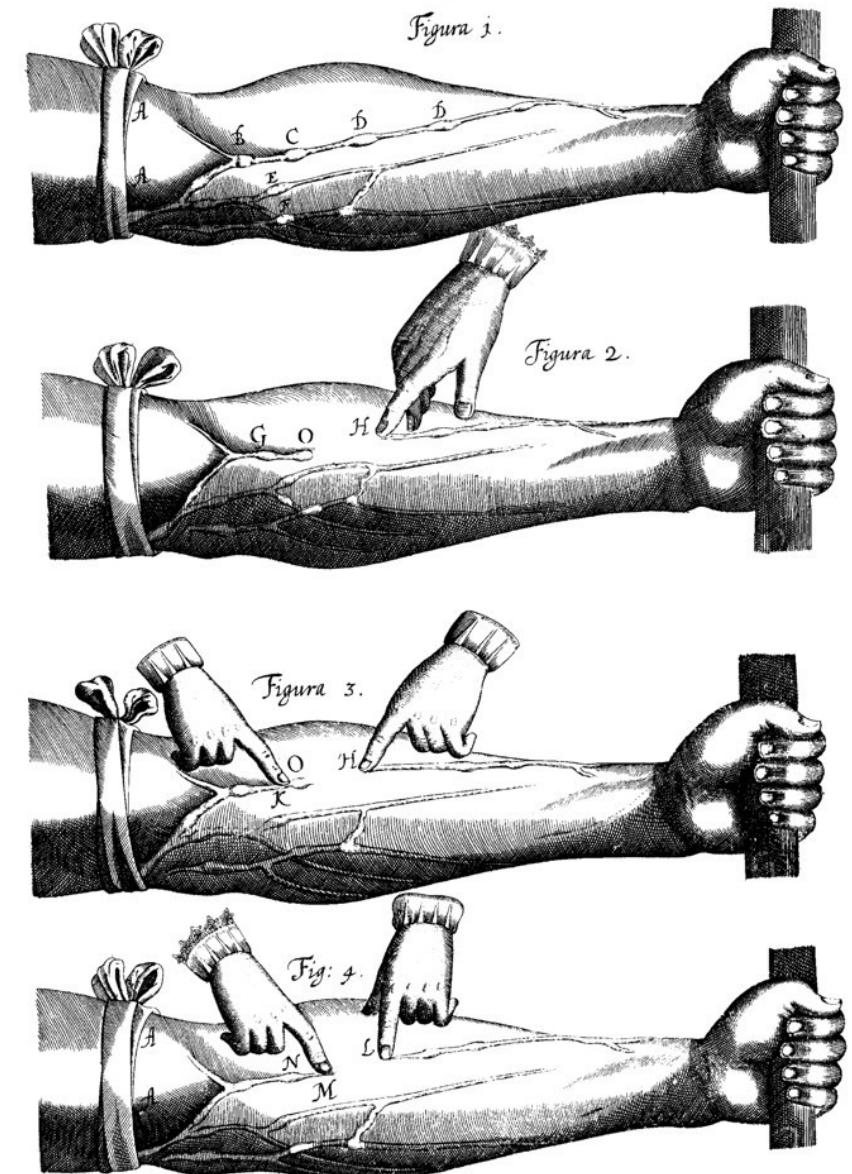
Harvey & human blood

William Harvey's discovery of blood circulation

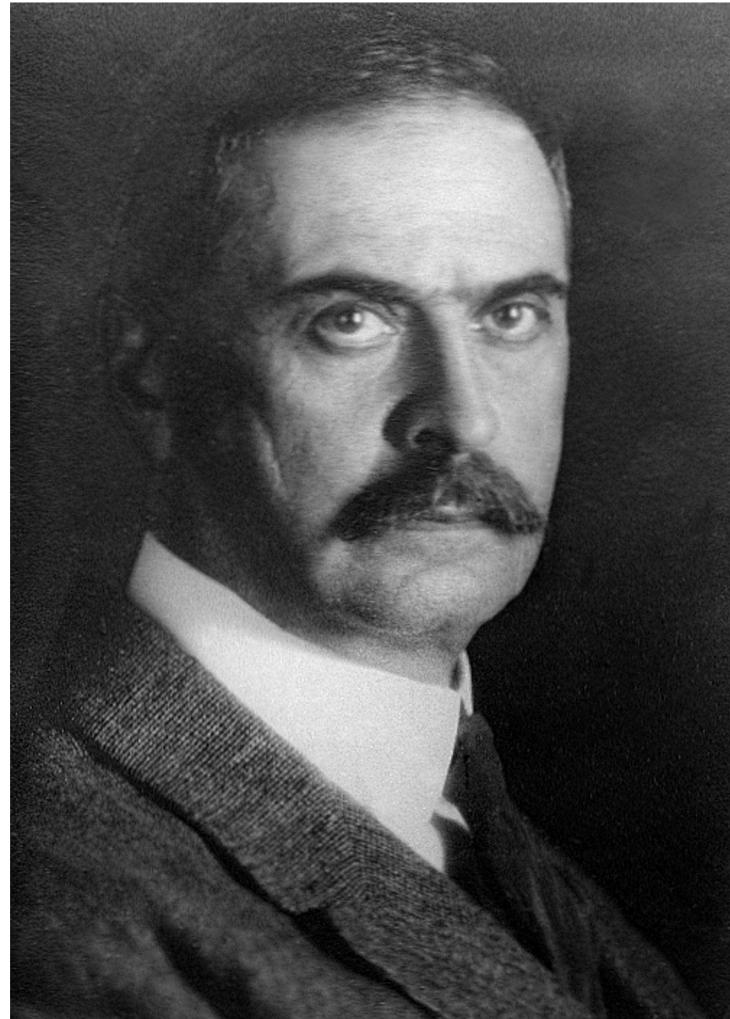
English Physician

Significant contributions in
anatomy and physiology

Venous blood flowed to
the heart, body's valves in
the veins maintained the
one-way flow



Human blood is not the same !



Karl Landsteiner

Austrian – American biologist, Physician and Immunologist

Father of blood transfusion

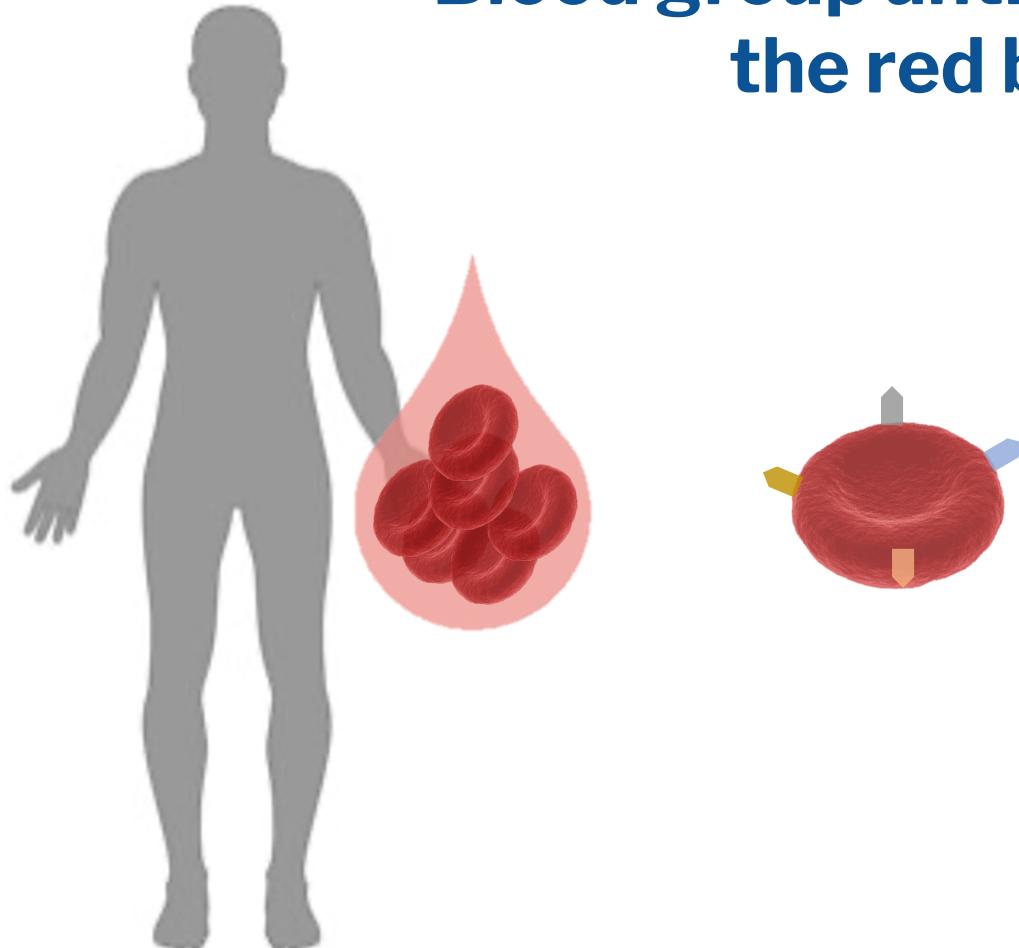
“Agglutination of Normal Human Blood”

The sera of healthy humans has an **agglutinating effect**, not only upon animal blood cells, but frequently upon blood cells from other individuals as well; the question is whether this phenomenon is due to inherent individual differences or to the effect of influenced damages

1930 – Nobel prize in Physiology or Medicine

Basic variability of human blood

Blood group antigens are surface markers on the red blood cell membrane



- Blood group antigens are either sugars or proteins, and they are attached to various components in the red blood cell membrane.
- People belong to different blood groups, depending upon the surface markers found on the red blood cell

Discovery of divergence



Mid 19th century

Mother of a still born child needed blood transfusion

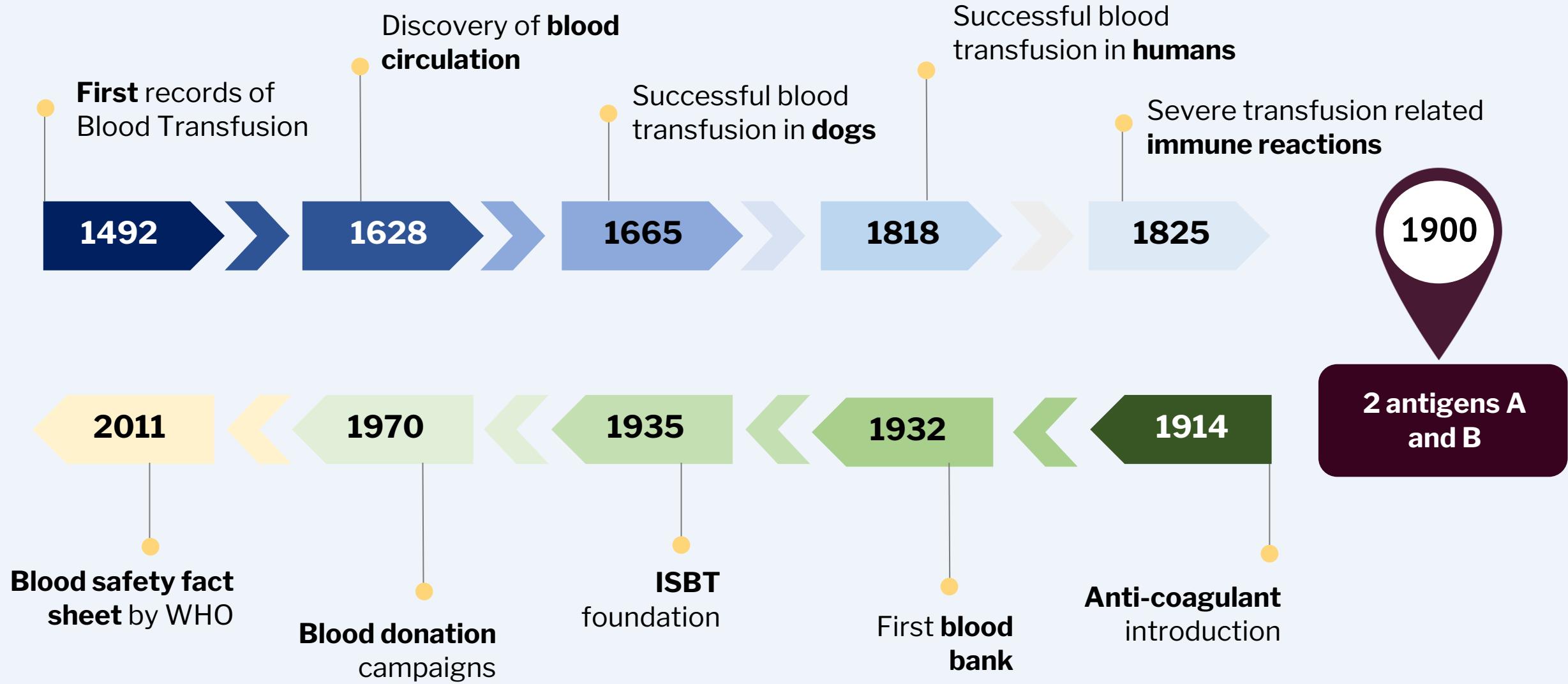
ABO compatible blood was transfused from patient's husband



**Severe adverse reactions
encountered**

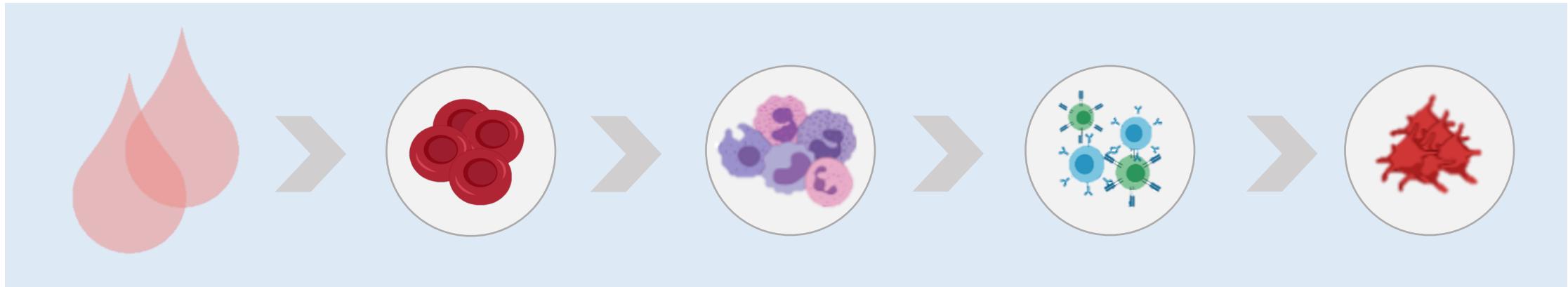
RH factor in human
blood discovered

Human blood group studies



Data source : American Red Cross

The vital fluid of life



Clinical use of human blood

- Blood transfusion is an essential part of modern health care.
- Around **118.5** million units of donated blood are collected globally every year
- An estimated **5 million** people need blood transfusions each year throughout the U.S.
- Global blood shortage - **100 million units** of blood per year

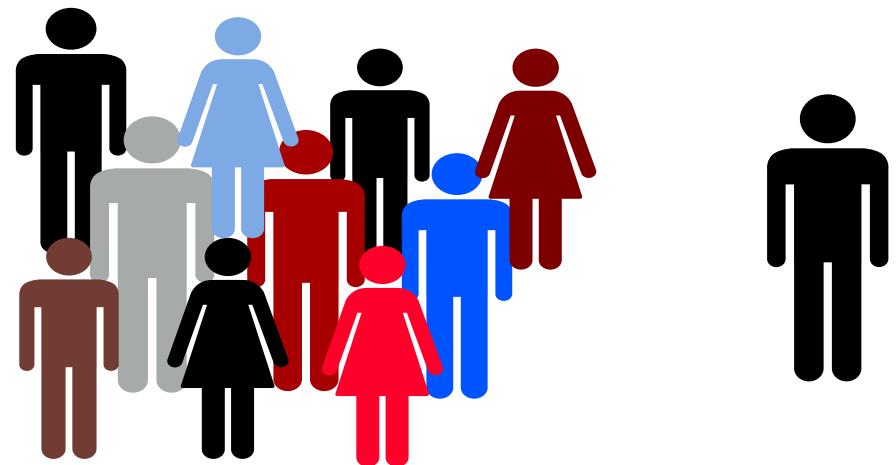
Prevention is better !

Than cure ...

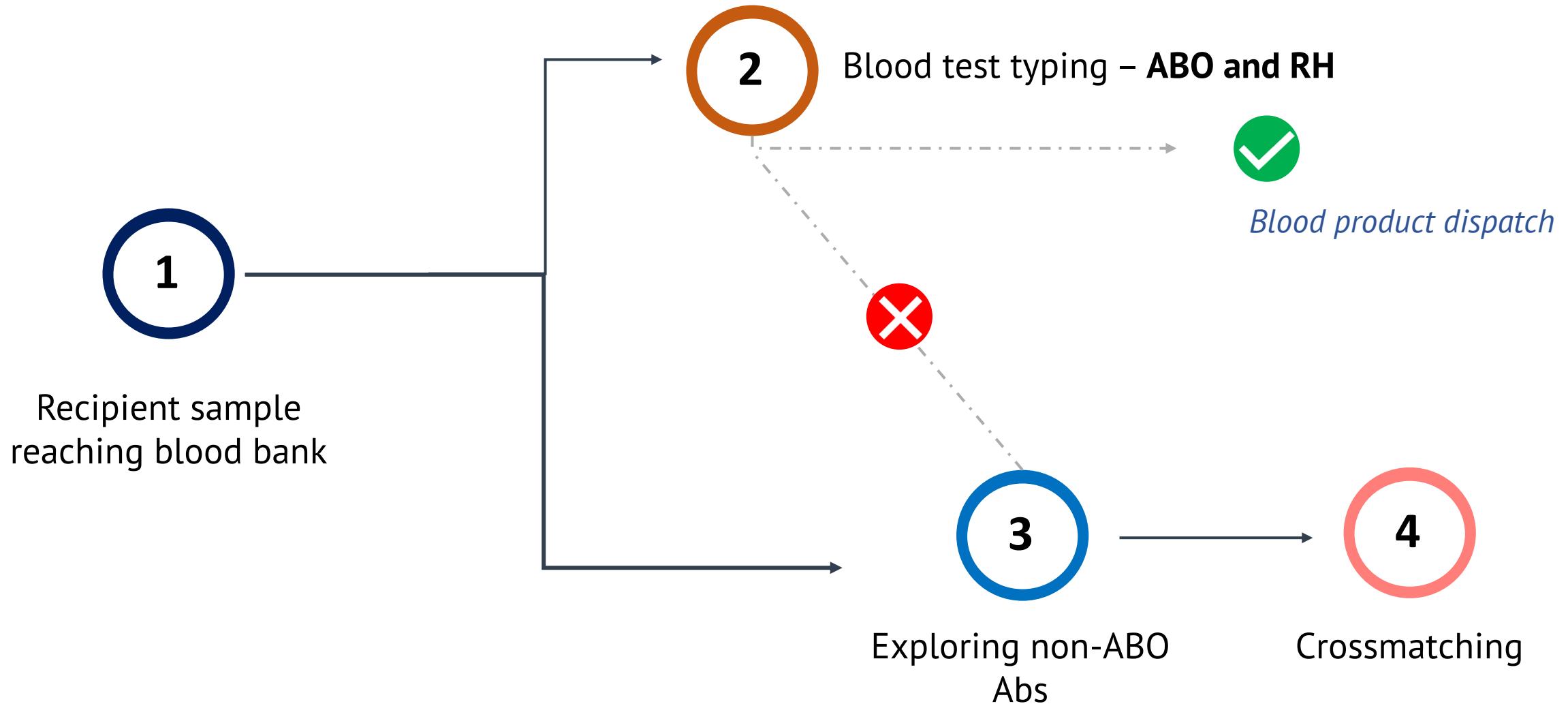
“Selecting compatible donor blood components for transfusion, thereby preventing harm to the recipient and maximizing post transfusion RBC survival”

Suzanne A, 2019

Pre-transfusion testing A preventive strategy



Routine practice to deal the variability

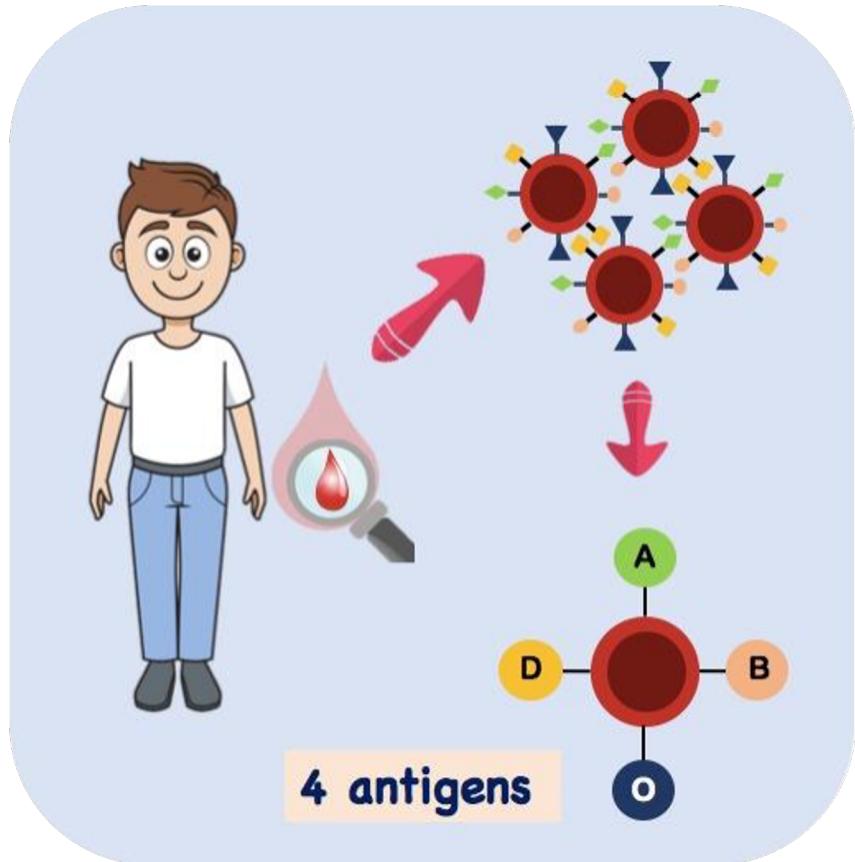


Blood groups beyond the horizon

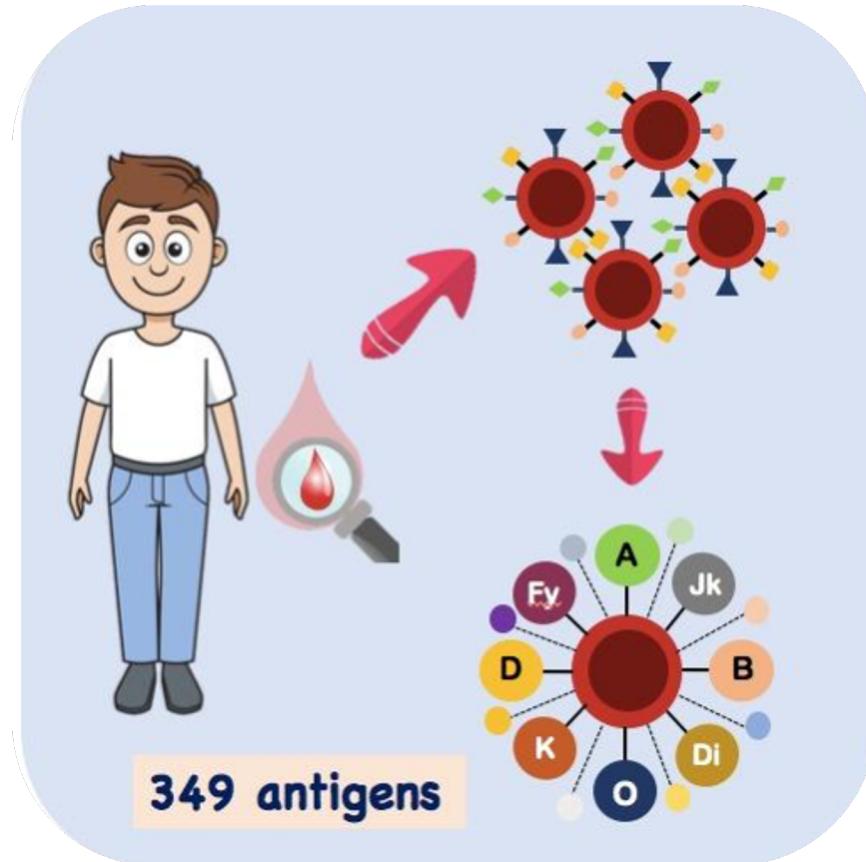


Blood variability – The hidden truth

Expectation



Reality



Data source : International Society of Blood Transfusion

Beyond the ABO horizon

75%

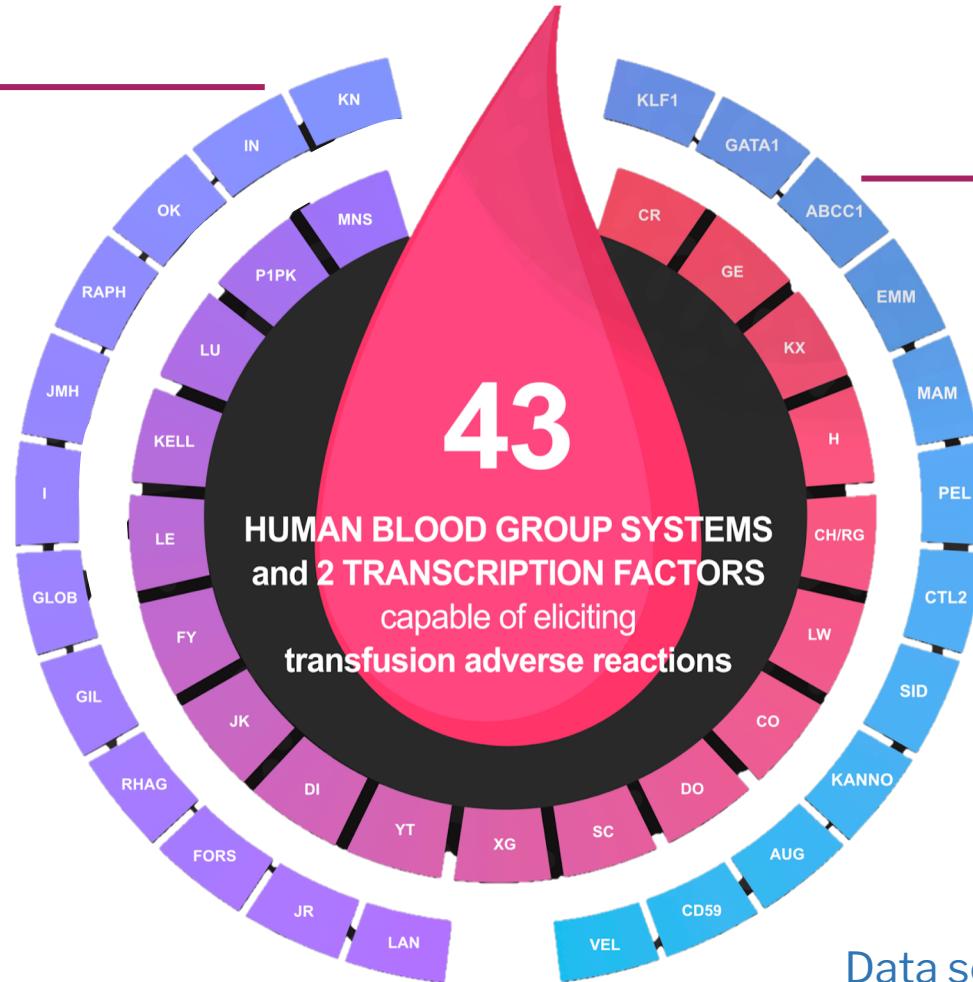
61 out of 81 fatalities associated with **Hemolytic transfusion reactions**

21%

13 out of 61 fatalities associated with adverse transfusion reactions caused by **non-ABO HTRs**

25%

20 out of 81 fatalities associated with **quality** of various donated blood products



Data source : FDA Annual Report, 2019

Global blood search



The World Responded to Help Save Girl with Rare Blood

- Zainab Mughal, a toddler fighting cancer
- Zainab's blood was extremely rare
- Difficulty in finding compatible donors

"Zainab's blood is extremely rare because she is missing a common antigen that most people have on their red blood cells."

The antigen is called "Indian B" (Inb)



Here starts the whole new unexplored field of human blood groups !

February 25, 2020 – Orlando, Florida

Hemovigilance – Global overview



Global status report on blood safety and availability 2021

171 countries registered under WHO Global Database of Blood Safety (GDBS)

Significant differences in patterns of blood use

High income

*Cardiovascular,
transplant surgery,
massive trauma
and malignancies*

Low & Middle income

*Pregnancy related
complications,
severe infantile
anaemia*

- **743** cases of adverse reactions associated with antigen incompatibilities/mismatch
- **44%** of the above cases were associated with **non-ABO** antigen incompatibilities/mismatch

Data source : WHO, Blood safety, 2021

How do we achieve better success rates in transfusion procedures?

Betterments in the blood group profiling techniques



Deeper understanding on the molecular underpinnings of blood group antigen formations

Upcoming courses ...

- Genetic background of human blood group antigens
- Evolution of human blood grouping techniques – The past, present & future
- Genomics in advancing the future of blood group profiling
- From genomes to populations – Population specific blood group registries

Thank

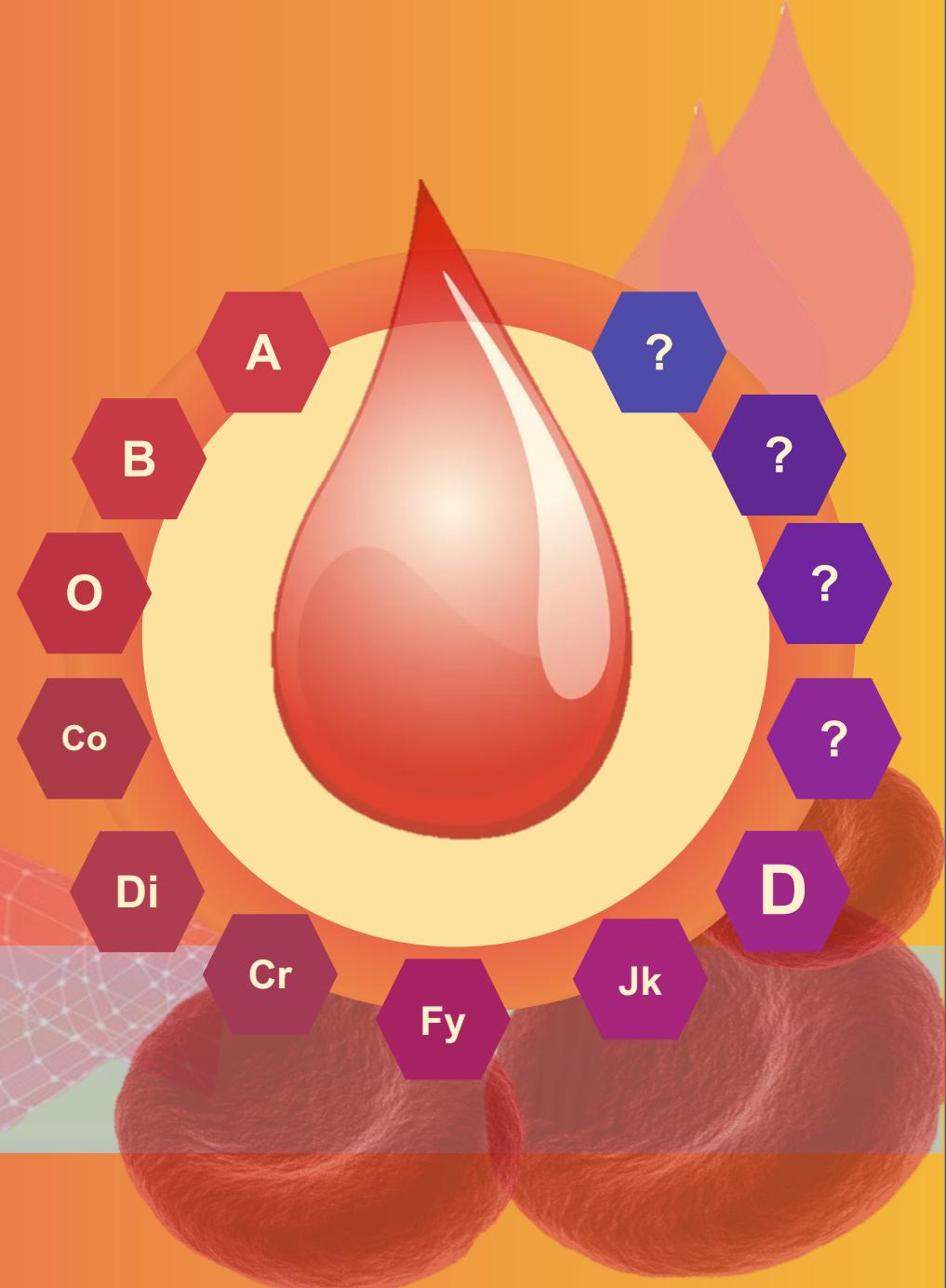
You



Introduction to Genomics in Immunohematology



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