

# Genomics in Immunohematology



**IGIB**  
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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*

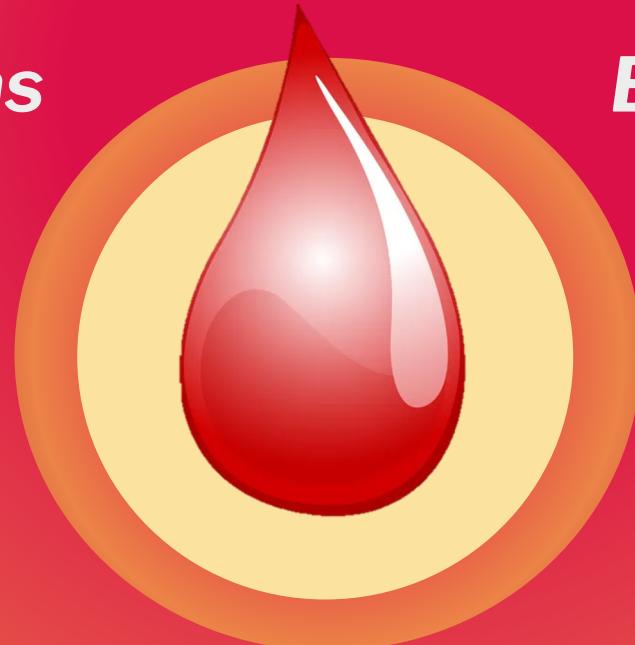
# Aims & Highlights !

*Human blood groups*

*RBC antigens*

*Variability of blood*

*History of discovery*



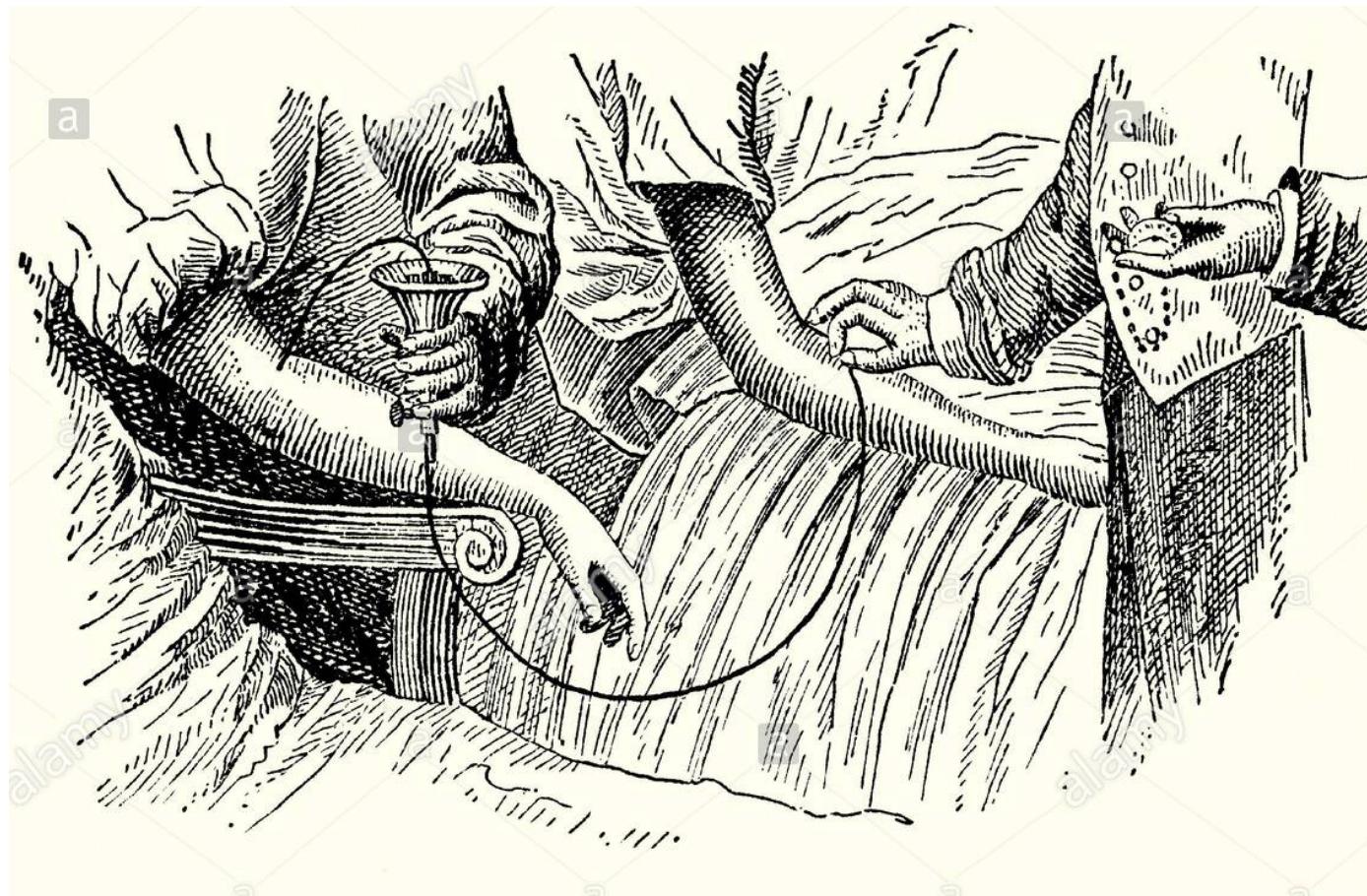
*Blood group profiling*

*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**

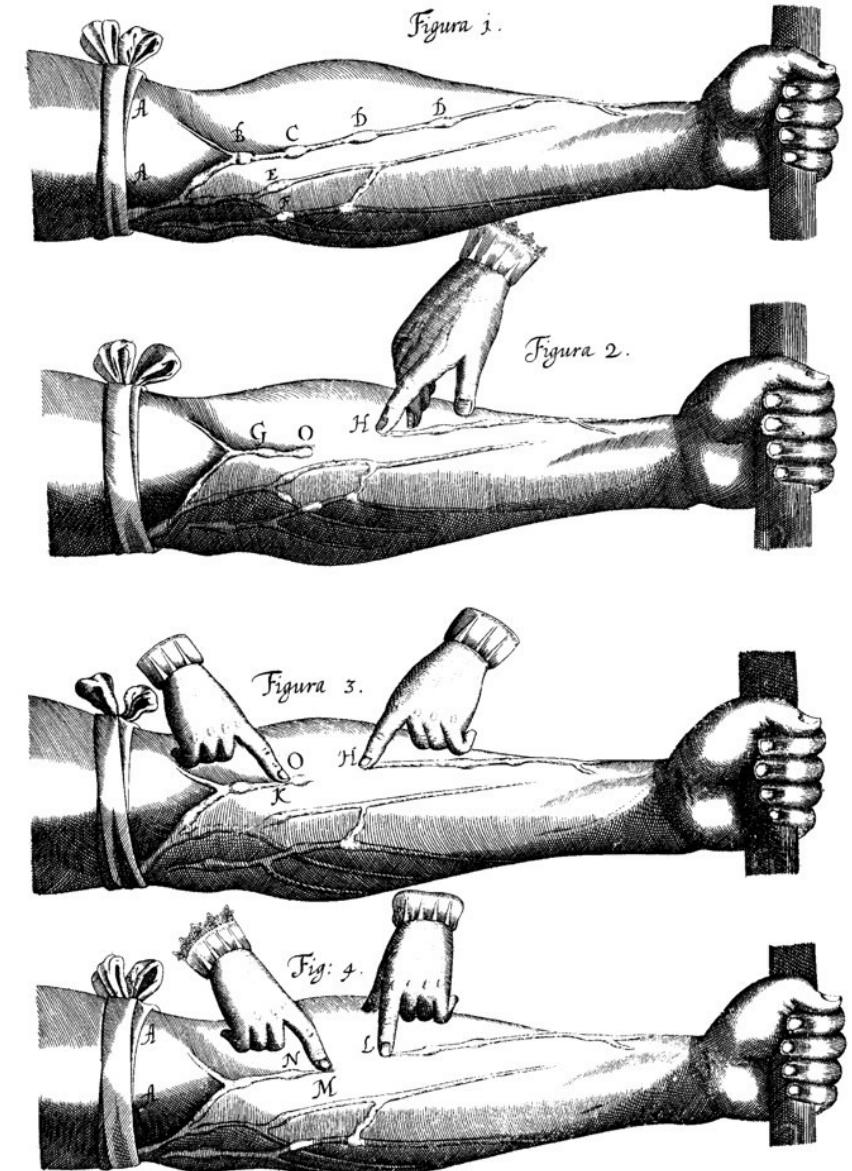
# Harvey & human blood

## William Harvey's discovery of blood circulation

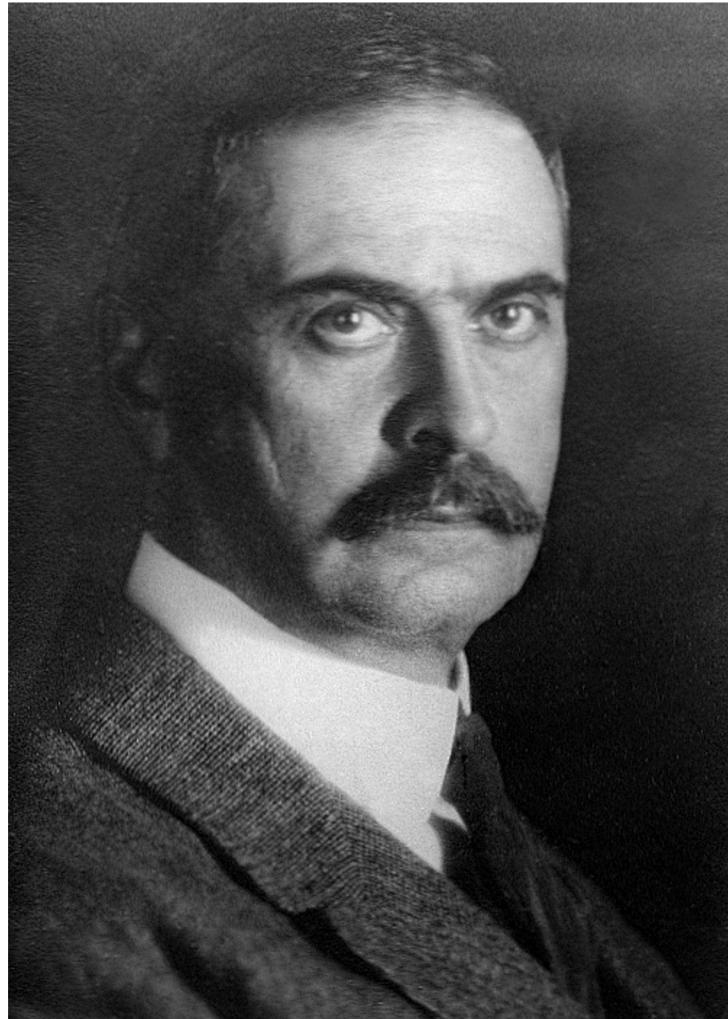
English Physician

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

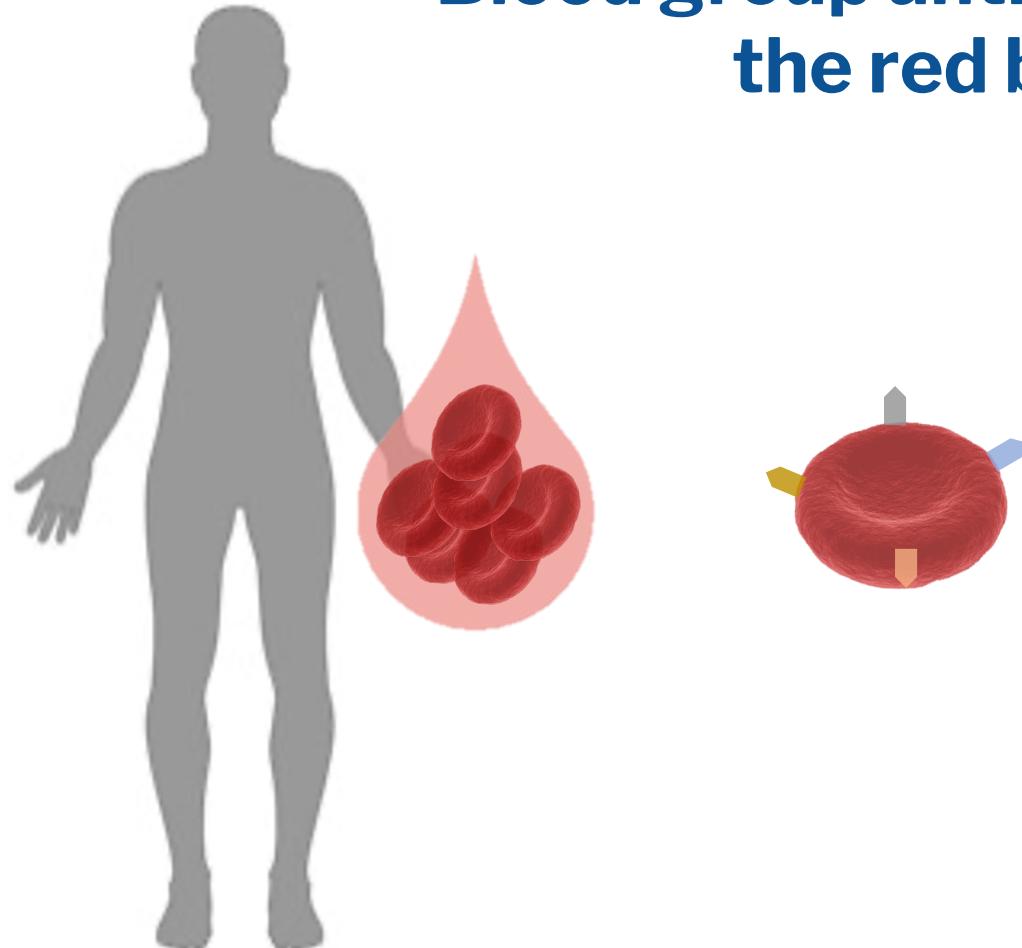
### “Agglutination of Normal Human Blood”

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 19<sup>th</sup> century

**Mother of a still born child** needed blood transfusion

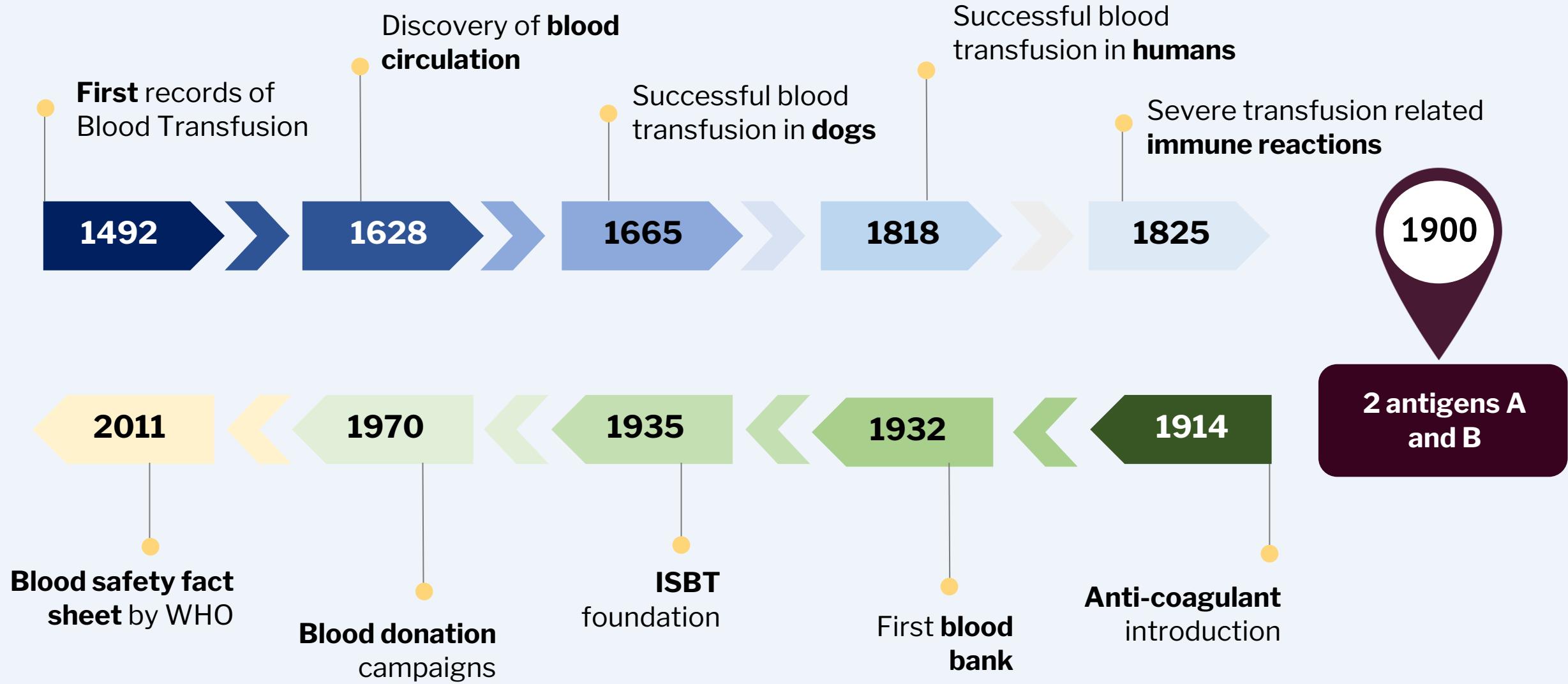
**ABO compatible** blood was transfused from patient's husband



**Severe adverse reactions  
encountered**

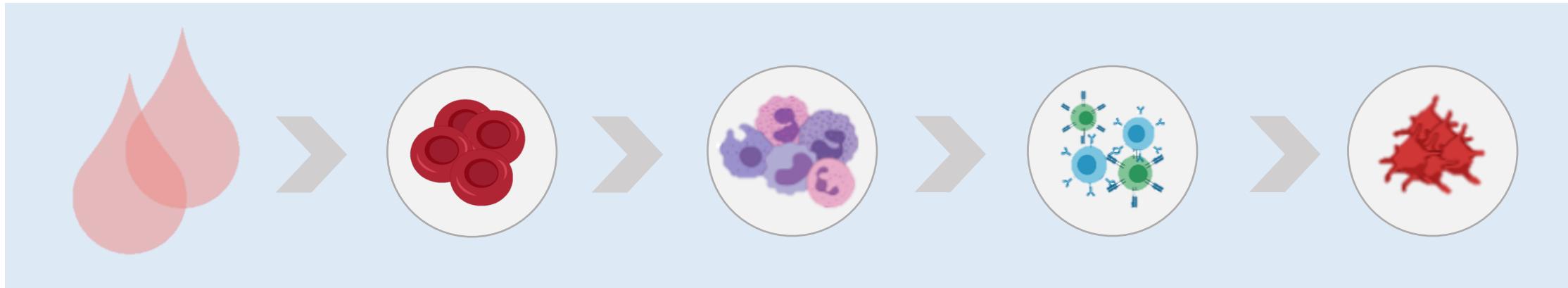
RH factor in human  
blood discovered

# 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

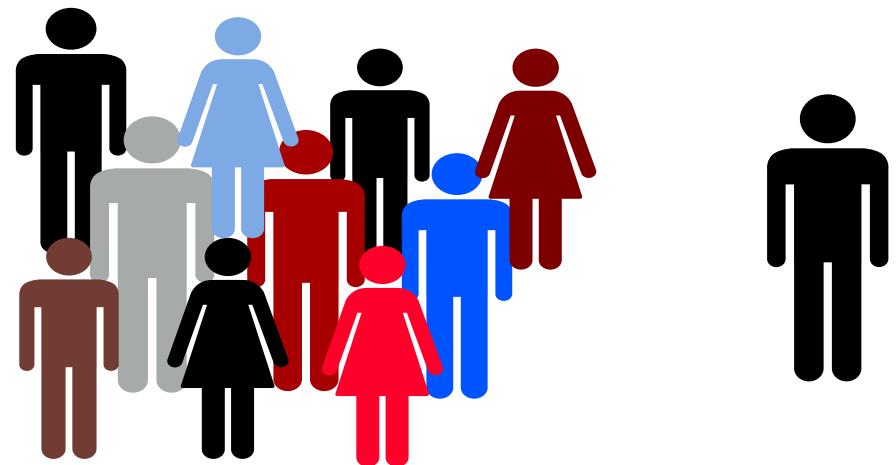
# Prevention is better !

Than cure ...

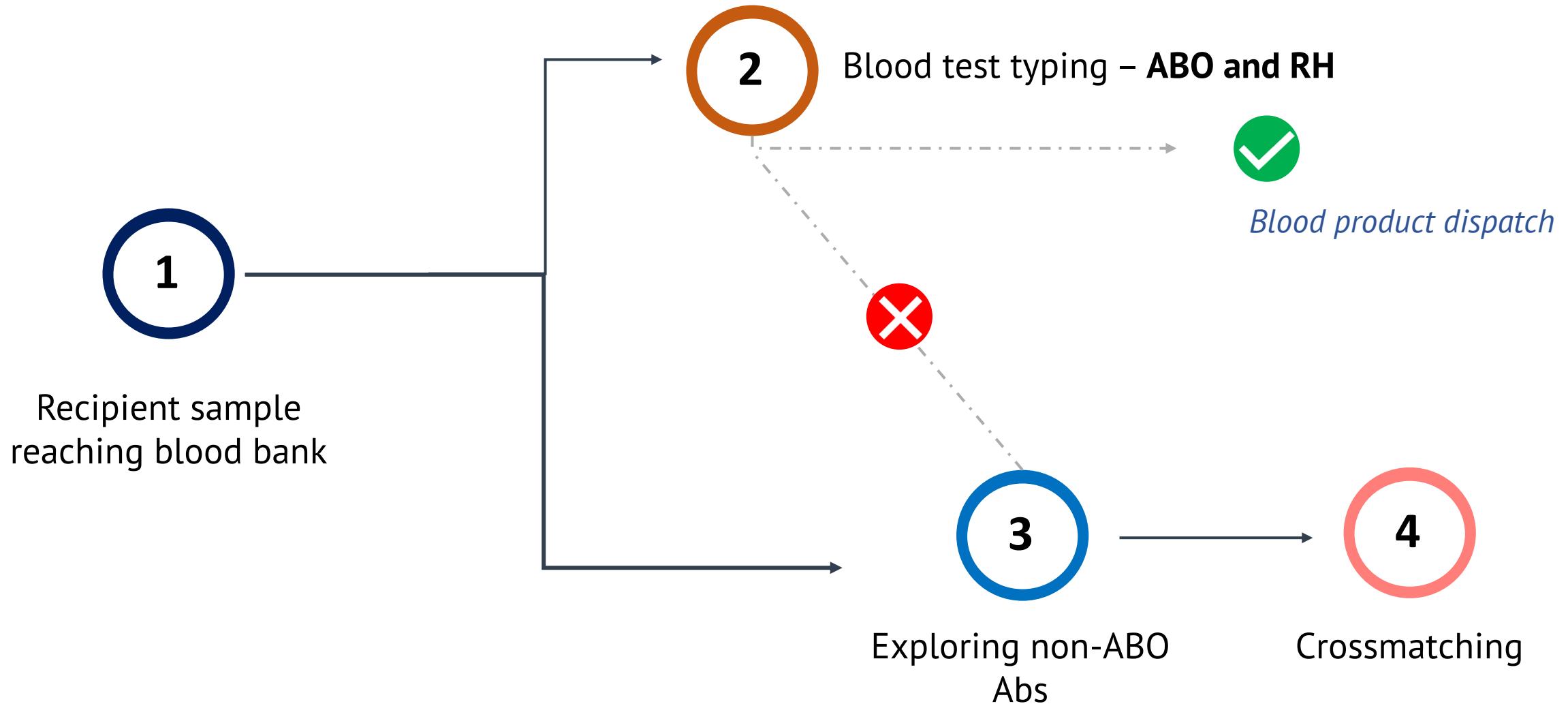
“Selecting compatible donor blood components for transfusion, thereby preventing harm to the recipient and maximizing post transfusion RBC survival”

Suzanne A, 2019

## Pre-transfusion testing A preventive strategy



# Routine practice to deal the variability

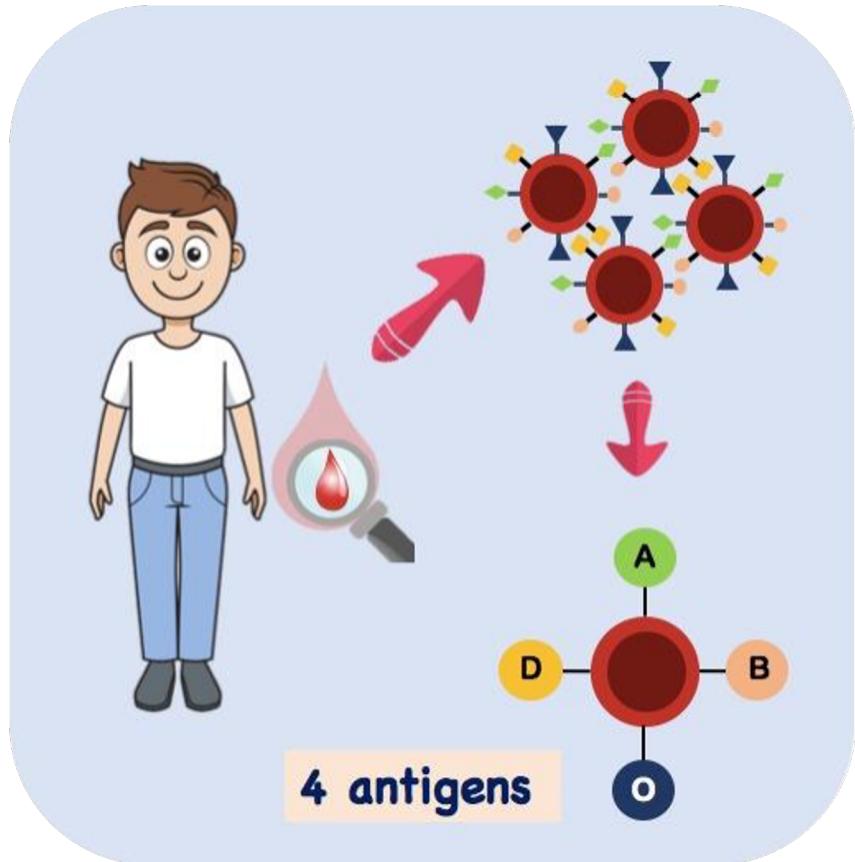


# Blood groups beyond the horizon

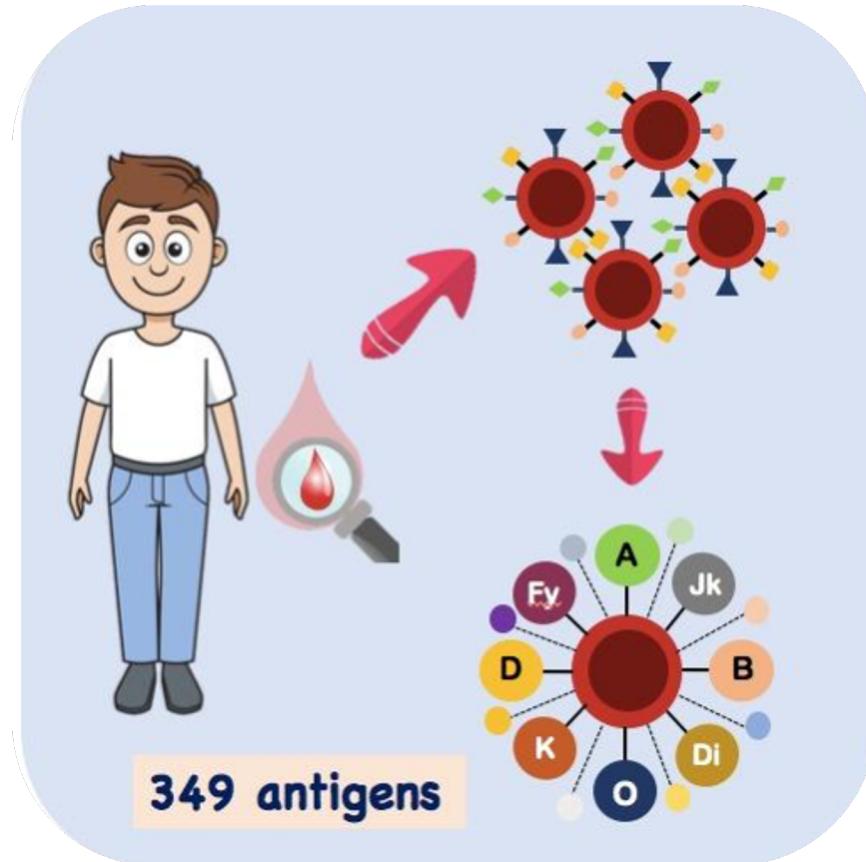


# Blood variability – The hidden truth

Expectation



Reality



Data source : International Society of Blood Transfusion

# Beyond the ABO horizon

75%

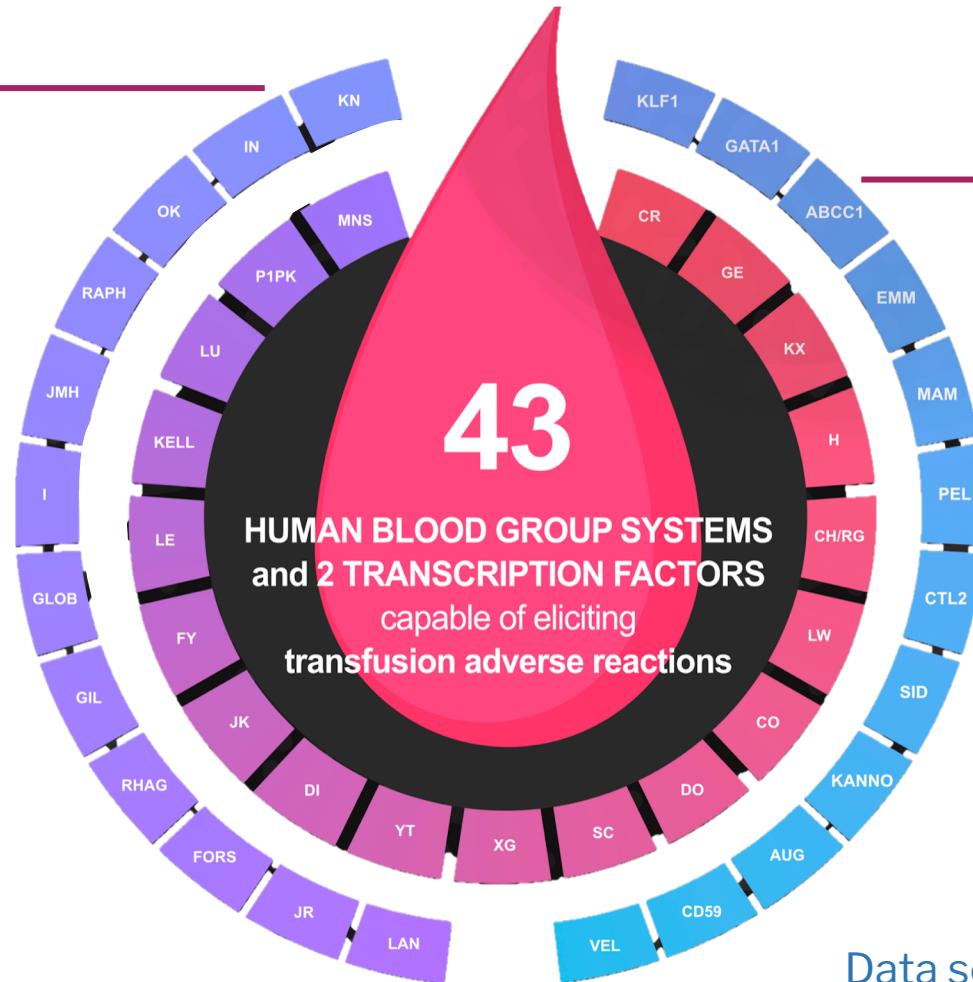
61 out of 81 fatalities associated with **Hemolytic transfusion reactions**

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

