Paul-Ehrlich-Institut

Haemophilia

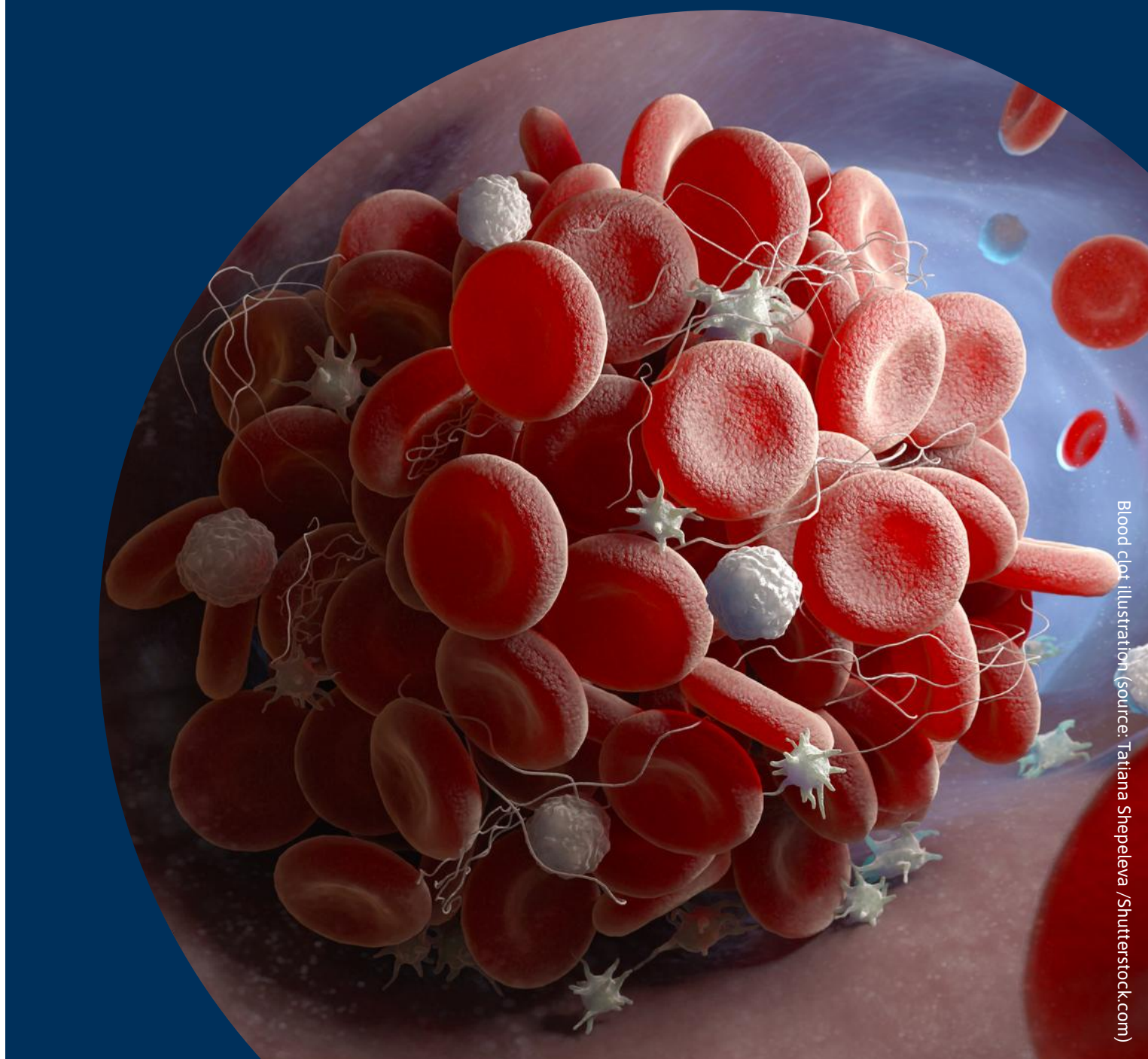
is a hereditary disease that leads to a blood clotting disorder.
In the two most well-known forms of haemophilia (A and B), the blood lacks one of the proteins required for blood coagulation (coagulation factors).

Haemophilia A

- lack of coagulation factor **VIII**
- approx.
  1 in 5,000
  men are affected.

Haemophilia B

- lack of coagulation factor **IX**
- approx.
  1 in 25,000 to 30,000
  men are affected.



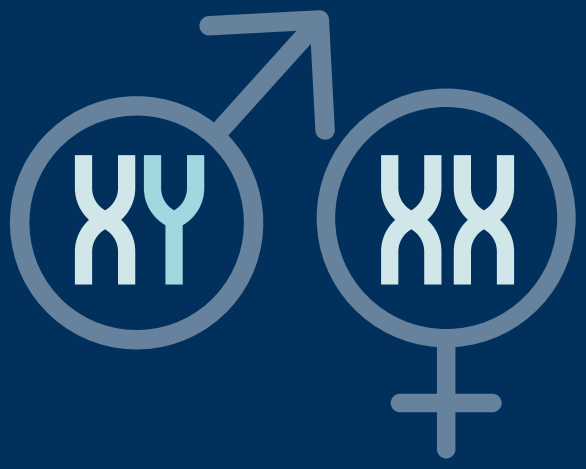
Blood clot illustration (source: Tatiana Shepeleva /Shutterstock.com)

Paul-Ehrlich-Institut

Coagulation Factor Gene on the X Chromosome

The genes for the formation of coagulation factors VIII and IX are located on the X chromosome.

Men have one X and one Y chromosome, women have two X chromosomes.



3D illustration X chromosome (source: Anusorn Nakdee/Shutterstock.com)

Haemophilia occurs mainly in men because women usually have an intact version (allele) of the coagulation factor gene located on the second X chromosome, which is sufficient for adequate blood coagulation.

Paul-Ehrlich-Institut

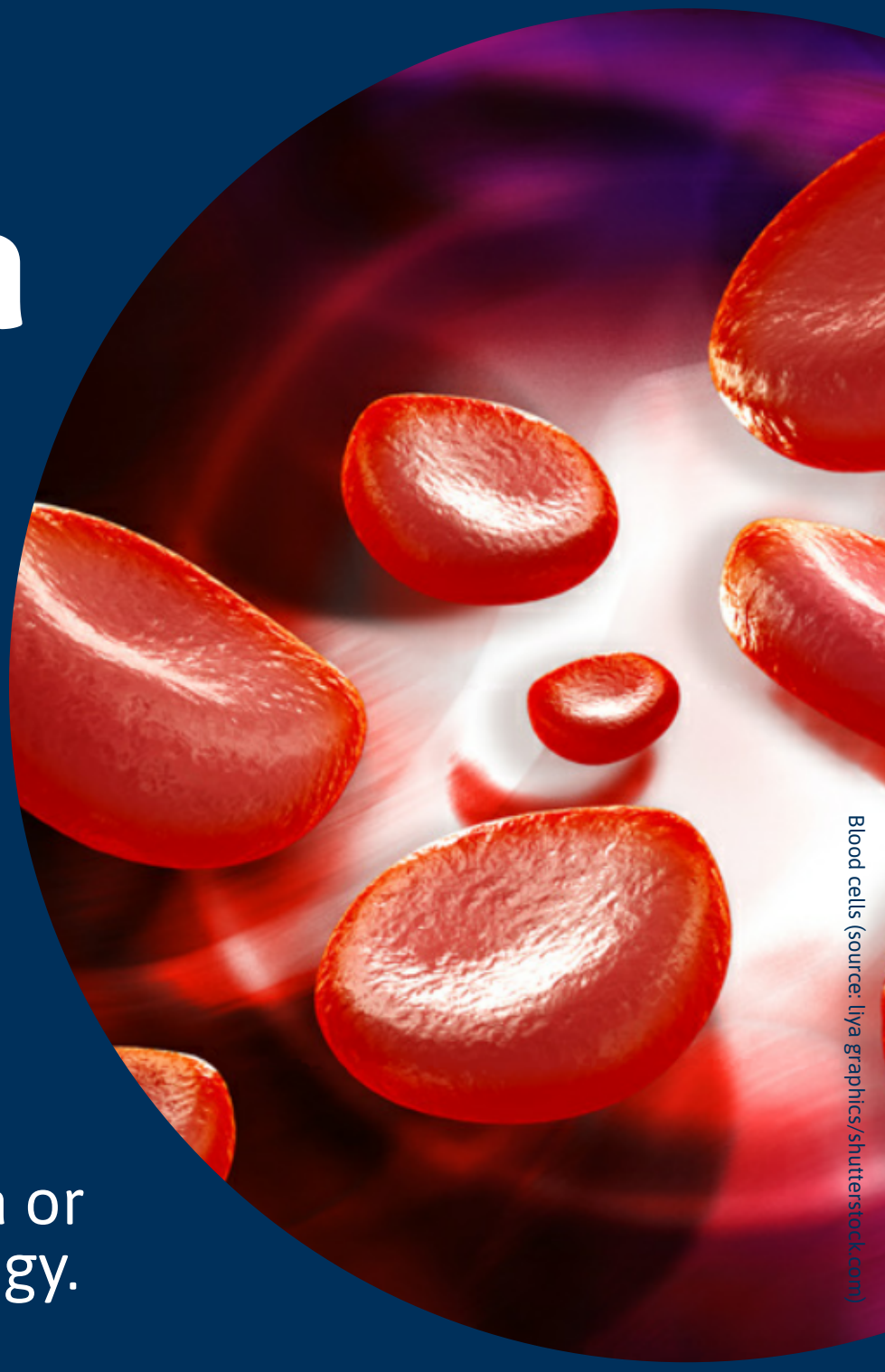
Multiple Treatment Options for Haemophilia

Treatment is primarily carried out by substitution of the missing coagulation factor protein.

Lifelong therapy is required.

The coagulation factor required for treatment is produced

- using human plasma or
- through biotechnology.



Blood cells (source: liya graphics/shutterstock.com)

Gene therapeutics are a newly authorised treatment for severe forms of haemophilia.

Paul-Ehrlich-Institut

Gentherapeutics for Haemophilia

Gene therapeutics have now been authorised for the treatment of severe haemophilia A and severe and moderate haemophilia B.

The aim of the gene therapies is to introduce a functional coagulation factor gene into some of the patient's liver cells via a **single** intravenous administration in order to provide sufficient amounts of the coagulation factor in the blood.

This treatment prevents bleeding or reduces bleeding episodes.

Syringe and vial (source: cottonbro studio/pexels.com)