

Rare Bleeding Disorders

Rare Bleeding Disorders

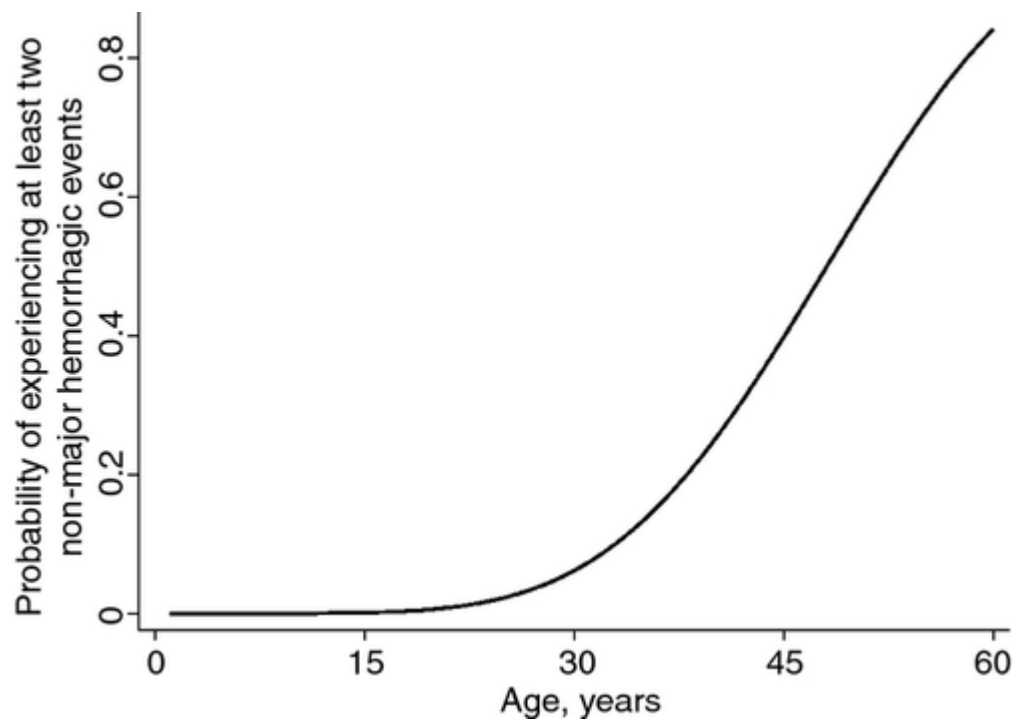
Table 1. Genetics, epidemiology and management options for rare congenital factor deficiencies

Factor	Estimated prevalence	Hemostatic level	Half-life of transfused factor	Recovery (%rise/IU/kg given)	Factor replacement therapy	Clinical-laboratory correlation
Fibrinogen						
Afibrinogenemia	extremely rare	0.50 g/l	2–4 days	0.017 g/l per mg/kg administered	Plasma-derived, pathogen-inactivated fibrinogen concentrate; cryoprecipitate; FP/FFP	Strong
Hypofibrinogenemia	rare					
Dysfibrinogenemia	rare					
Prothrombin	extremely rare	0.2–0.3 IU/ml	3–4 days	1.6%	PCCs; FP/FFP	Strong
FV	1:1,000,000	0.15–0.2 IU/ml	36 h	1%	FP/FFP	Poor
FV + FVIII combined	1:1,000,000	as for individual factors	as for individual factors	as for individual factors	FP/FFP + FVIII concentrate (chapter 6)	Weak
FVII	1:500,000	0.15–0.2 IU/ml	4–6 h	1%	rFVIIa; plasma-derived, pathogen-inactivated FVII concentrate; PCCs; FP/FFP	Poor
FX	1:1,000,000	0.15–0.2 IU/ml	40–60 h	1.5%	plasma-derived, pathogen-inactivated FX concentrate; PCCs; FP/FFP	Strong
FXI	depends on ethnicity ¹	0.15–0.3 IU/ml	40–70 h	1.8%	FP/FFP; plasma-derived, pathogen-inactivated FXI concentrate	No association
FXIII	1:2,000,000	0.1–0.2 IU/ml	11–14 days	1.7%	rFXIII; plasma-derived, pathogen-inactivated FXIII concentrate; cryoprecipitate; FP/FFP	Strong

Rare Bleeding Disorders



- Do you have a bleeding disorder?

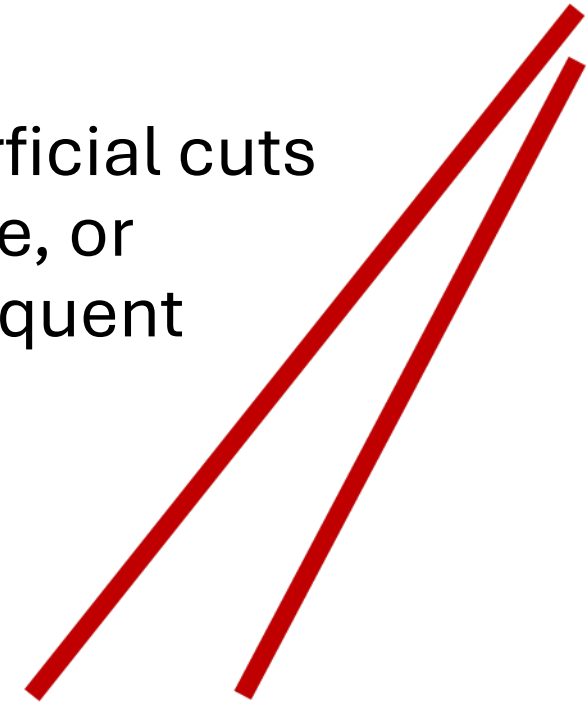


You are **not** more **fragile** than anyone else

Rare Bleeding Disorders



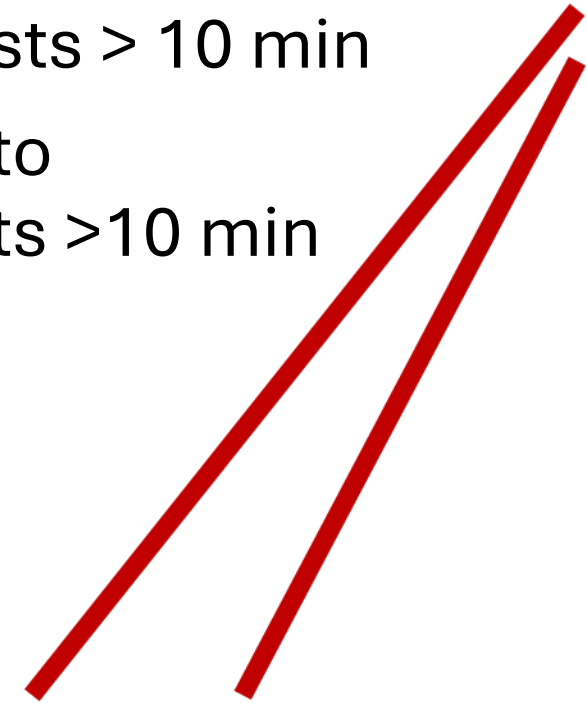
- Epistaxis: Nosebleed that causes interference or distress with daily or social activities
- Cutaneous bleeding: Bruises, five or more (> 1 cm) in exposed areas
- Minor cutaneous wound: Bleeding caused by superficial cuts (e.g. by shaving razor, knife, or scissors) that requires frequent bandage changes



Rare Bleeding Disorders



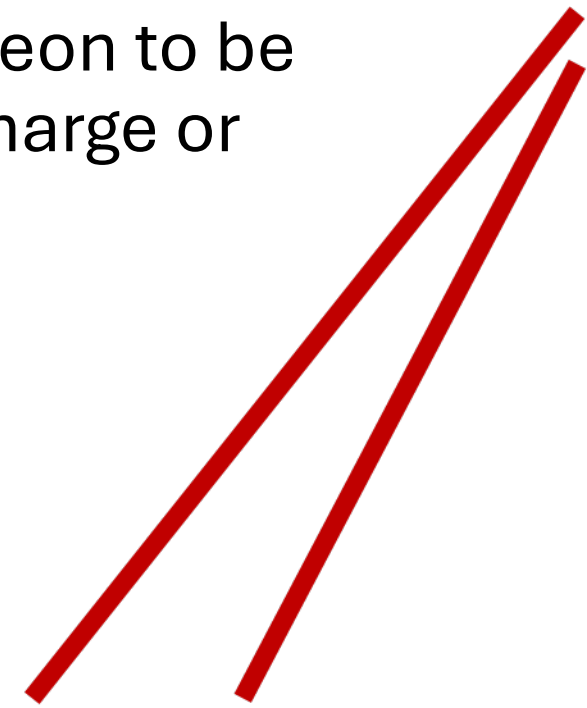
- Oral cavity bleeding
 - Gum bleeding that lasts for 10 min or longer on more than one occasion.
 - Tooth eruption or spontaneous tooth loss bleeding that requires assistance or supervision by a physician or lasts > 10 min
 - Bleeding occurring after bites to lips, cheek, and tongue that lasts >10 min



Rare Bleeding Disorders



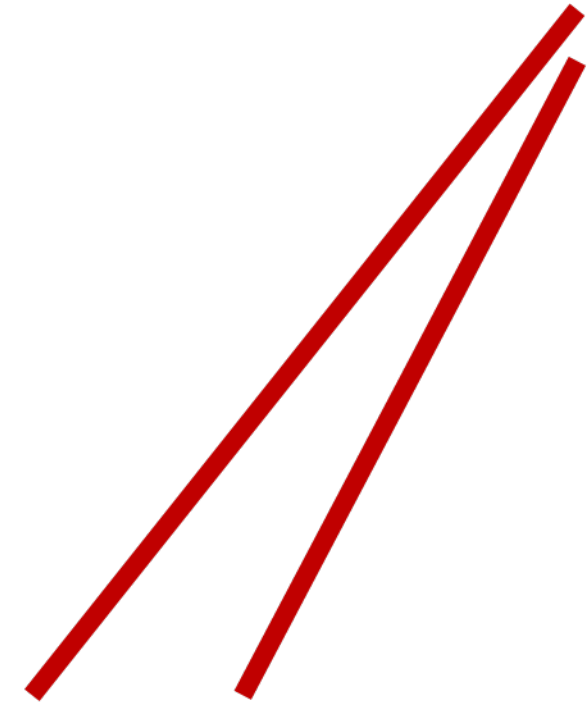
- Tooth extraction: Any bleeding occurring after leaving the dentist's office and requiring a new, unscheduled visit or prolonged bleeding at the dentist's office causing a delay in the procedure or discharge
- Surgical bleeding: Any bleeding judged by the surgeon to be abnormally prolonged that causes a delay in discharge or requires some supportive treatment



Rare Bleeding Disorders



- Menorrhagia Any bleeding that interferes with daily activities such as work, housework, exercise, or social activities during most menstrual periods






Menstrual chart and scoring system




Date of start

day	month	year

 Score

--

Towel	1	2	3	4	5	6	7	8
								
								
								
Clots/flooding <i>Clots: size</i>								

Tampon	1	2	3	4	5	6	7	8
								
								
								
Clots/flooding <i>Clots: size</i>								

Scoring system

Towels

1 point for each lightly stained towel

5 points for each moderately soiled towel

20 points if the towel is completely saturated with blood

Tampons

1 point for each lightly stained tampon

5 points for each moderately soiled tampon

10 points if the tampon is completely saturated with blood

Clots

1 point for small clots

5 points for large clots

Source: U.K. Haemophilia Society, A Guide for Women Living with von Willebrand's

Hemophilia Treatment Centers



- Provide and coordinate care (hospital/outpatient) to patients and their families. Comprehensive clinics yearly for adults and every 6 months for children
- Maintain a supervised home treatment program
- Collect key information: regional and national registries of patients with bleeding disorders; communications with state agencies; policy development to guarantee resource optimization
- Document treatment: number of episodes per year; number of absences from school or work; long-term results / musculoskeletal function
- Perform and lead basic and clinical research, including collaborative trials.

Hemophilia Treatment Centers

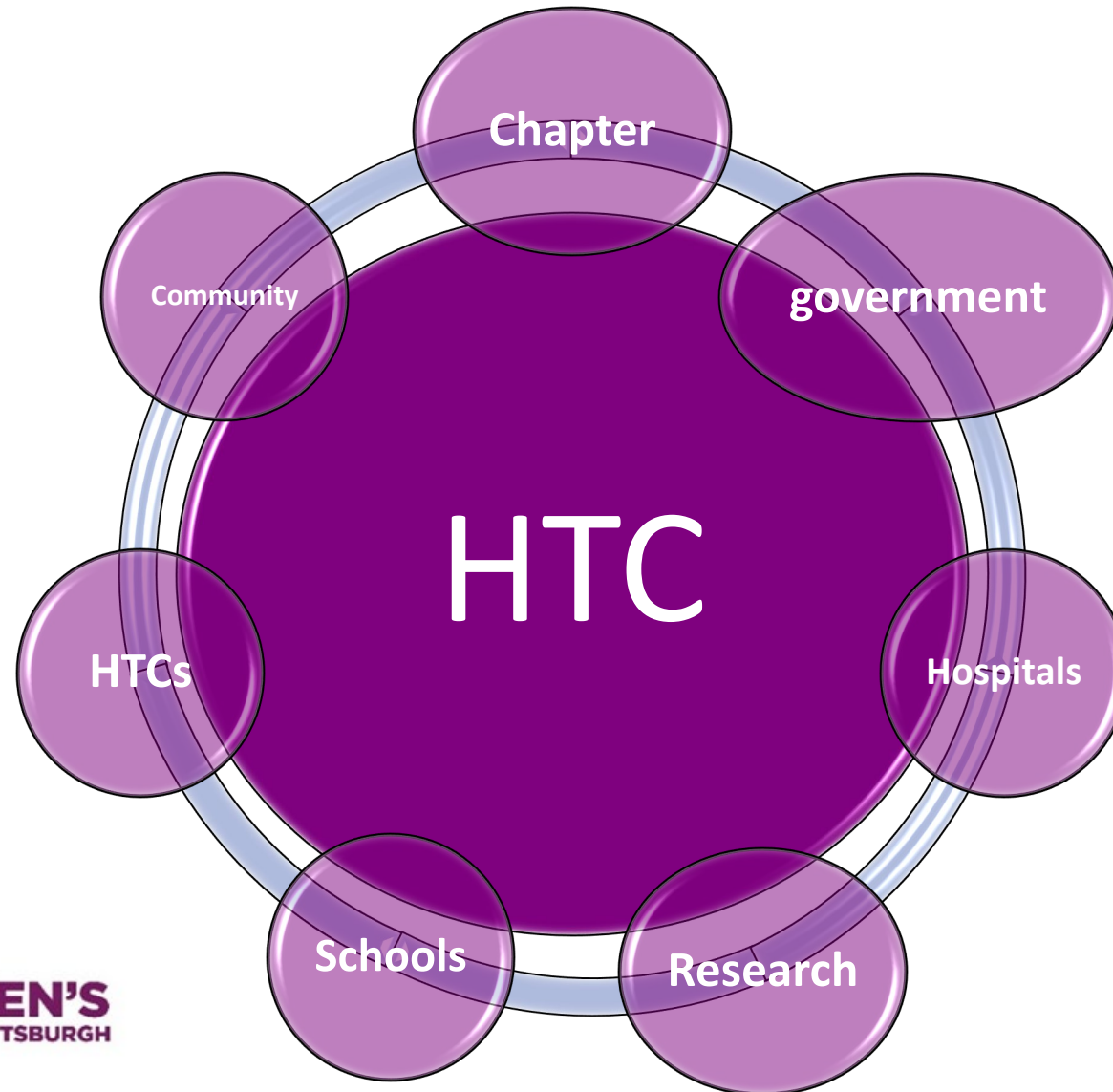


- A CDC study of 3,000 people with hemophilia showed that **those who received care at an HTC were 40% less likely to die of a hemophilia-related complication compared to those who did not** receive care at a treatment center
- Patients who received care at an HTC were **40% less likely to be hospitalized for bleeding complications**
- ~70% of people with hemophilia in the US receive care at a HTC

NHF Directives

Continuation of the hemophilia treatment center (HTC) model of care
Health care disparities between various communities (based on race, ethnicity, gender, education, income, etc.)
Access to research and mechanisms (ways) to conduct research
Access to specialized care
Mental health (depression, anxiety, emotional impact, substance use disorders, etc.)
Pain management
Treatment for all BDs (gene therapy, non-factor replacements, etc.)
Specific treatment for other RBDs (non-hemophilia, non-VWD)
Women's health and access to care
Treatment of other chronic disorders and issues affecting PWIBD (e.g. heart disease, diabetes, etc.)
Treatment of the aging population with BDs
Joint disease management

Vital HCWP neighborhood



Complex diseases need comprehensive care

Multidisciplinary health care professionals required for hemophilia care:

- Hematologists
- Physical therapists
- Nurses
- Social workers and other mental health professionals
- Genetic Counselors
- Other specialists such as dentists, dental hygienist, orthopedic surgeons, Gastroenterologist, Anesthesiologist, Infectious diseases, OB/GYN, Adolescent Medicine



Hemophilia Treatment Centers provide integrated comprehensive care

EMERGENCY ROOM - ADVOCATING FOR YOURSELF

Triage



- 1) Individuals with bleeding disorders should be triaged urgently as delays in administering appropriate therapy, such as infusion of factor concentrate, can significantly affect morbidity and mortality.
- 2) Consultation with the patient's primary provider of bleeding disorder care, in most cases a hematologist, is strongly advised. If this provider is unavailable, consultation with a bleeding disorders provider from the closest hemophilia treatment center is recommended. Administration of clotting factor replacement to the patient should not be delayed waiting for a consultation.

EMERGENCY ROOM - ADVOCATING FOR YOURSELF Assessment



Treatment for a suspected bleeding episode is based on clinical history. Physical exam findings may be normal in the early phases of most bleeding episodes associated with an underlying bleeding disorder. Spontaneous bleeding is common in those with severe disease (baseline factor levels $<1\%$). When in doubt, administer clotting factor replacement therapy immediately.

Treatment decisions should be based on the suspicion of a bleeding-related problem, not the documentation of one.

If the patient or the parent of a patient suspects that occult bleeding is occurring, administer clotting factor replacement. Patients often are instructed to carry with them appropriate factor replacement dosing guidelines as advised by their treating

EMERGENCY ROOM - ADVOCATING FOR YOURSELF

Diagnostic Studies



- Clotting factor replacement therapy should be given before any diagnostic studies are performed to evaluate a suspected bleeding problem, especially in the case of head trauma or suspected intracranial hemorrhage. For routine joint bleeding, no radiographic studies are indicated.
- For patients with BD who have illnesses or disorders that necessitate an invasive procedure (lumbar puncture, arterial blood gas, arthrocentesis, etc.) or surgery, factor replacement therapy or bypass therapy must be administered in the emergency department prior to the planned procedure or surgery. In this situation, consultation with a hematologist is strongly recommended.
- For an individual with known BD, routine laboratory studies (PT, aPTT, factor levels), are not indicated in the treatment of a routine bleeding episode unless requested by the patient's hematologist. Treatment should not be delayed waiting for test results which may take several hours. In some cases, screening assays such as the aPTT and factor activity assays will not be accurate.



Bleeding

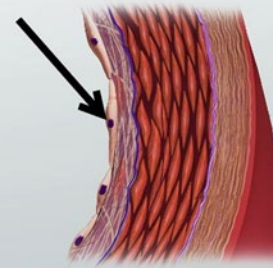
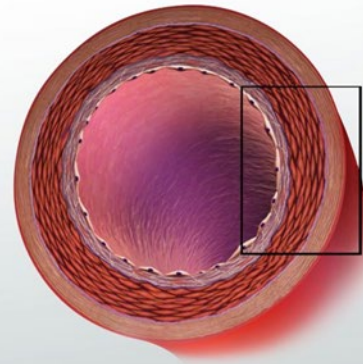


“Fixing “ the problem

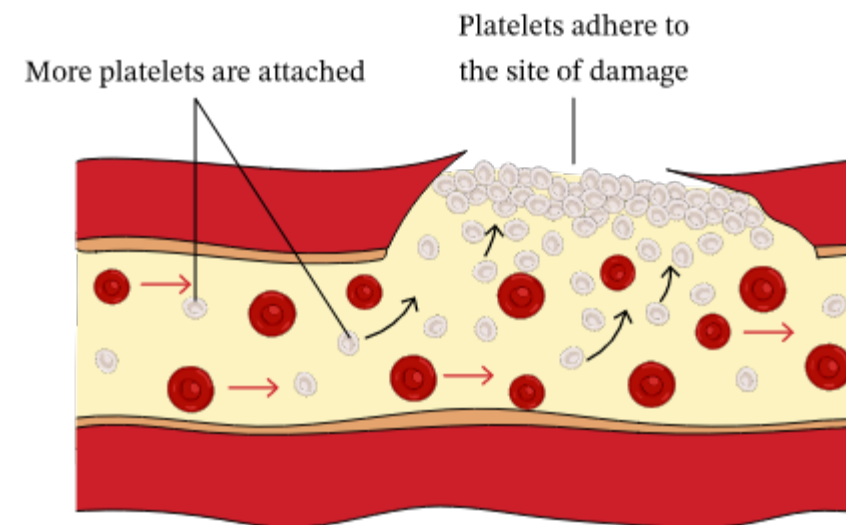
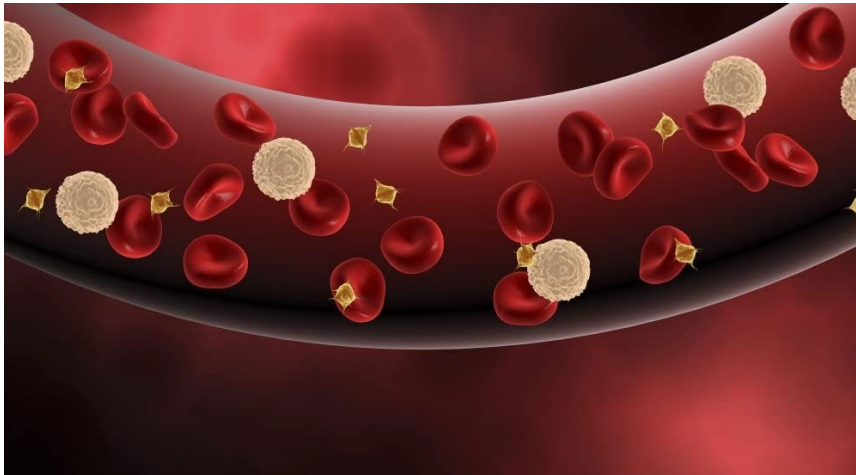
WHAT ARE ENDOTHELIAL CELLS?

endothelium

they form a one-cell-thick



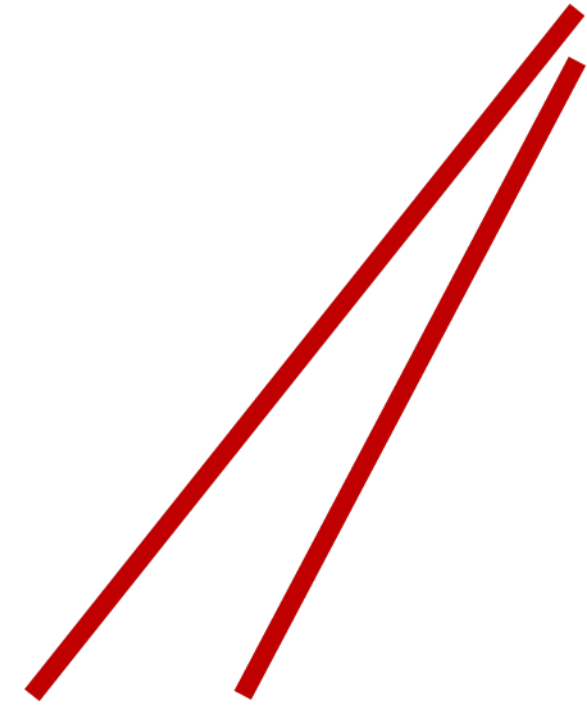
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The Basics



- Where is the bleed?
- How did it happen?
- What are you feeling?
- How much are you bleeding?

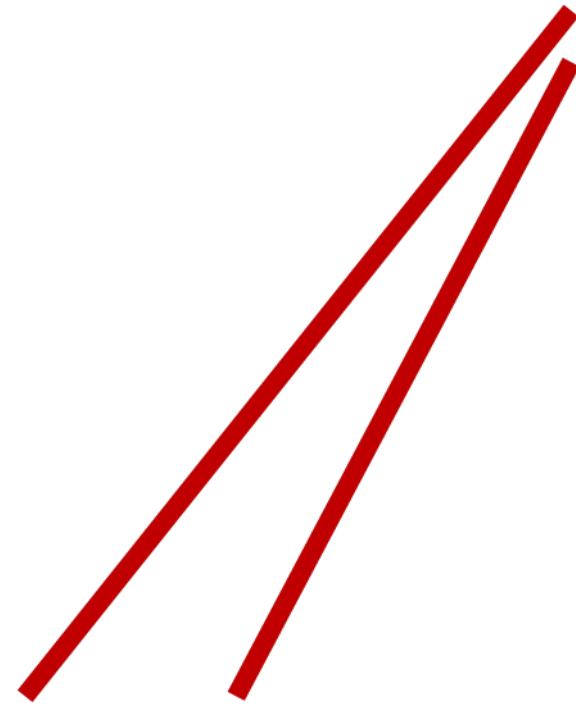


The Basics



- Local hygiene
- Ice
- Compression
- Elevate
- Control the pain

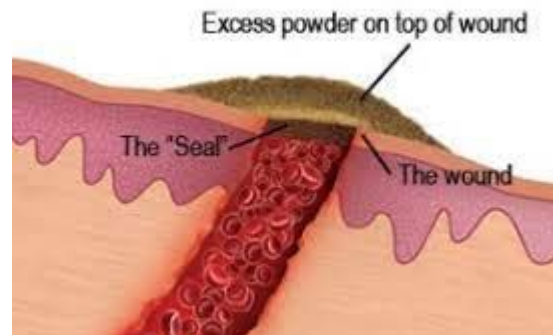
I C E



SKIN/soft tissue - home

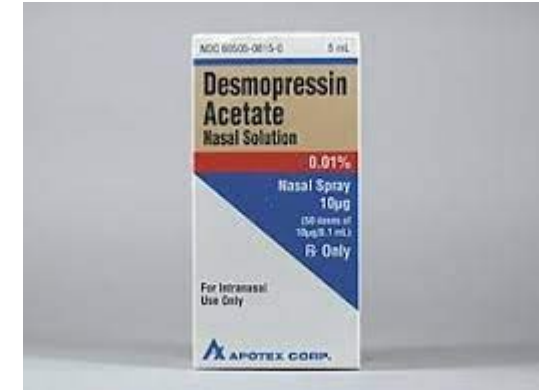


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 **THE HEMOPHILIA CENTER**
of Western Pennsylvania

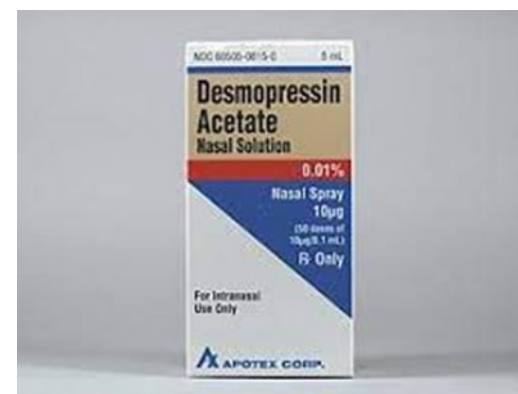
SKIN/Soft tissue - Doctor



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MOUTH - Home



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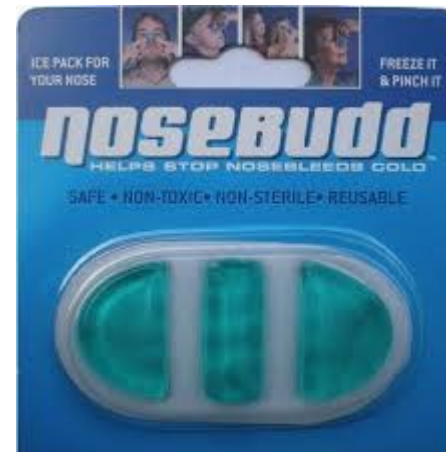
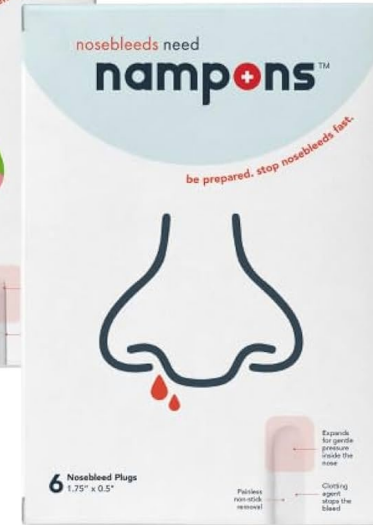
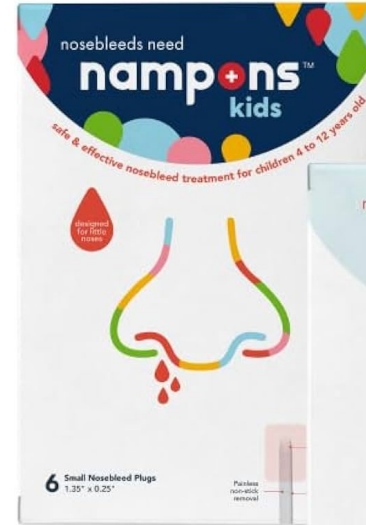
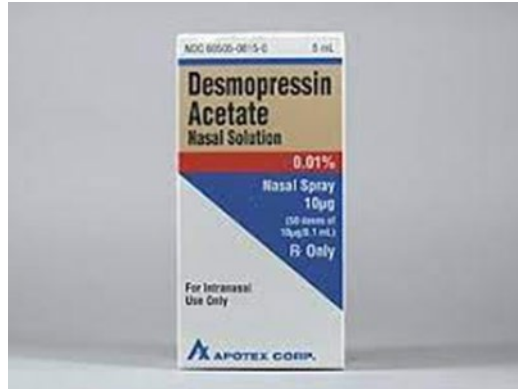
MOUTH - Doctor



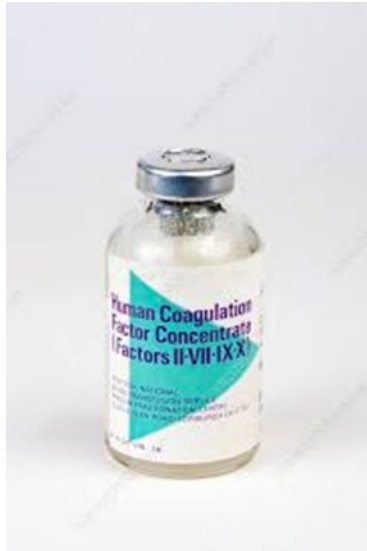
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Nose - home



NOSE - Doctor



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Menstrual



Birth control pills

Blood Products

Plasma derived (no reported blood born disease transmission since early 80's)

Recombinant products (made in the lab)

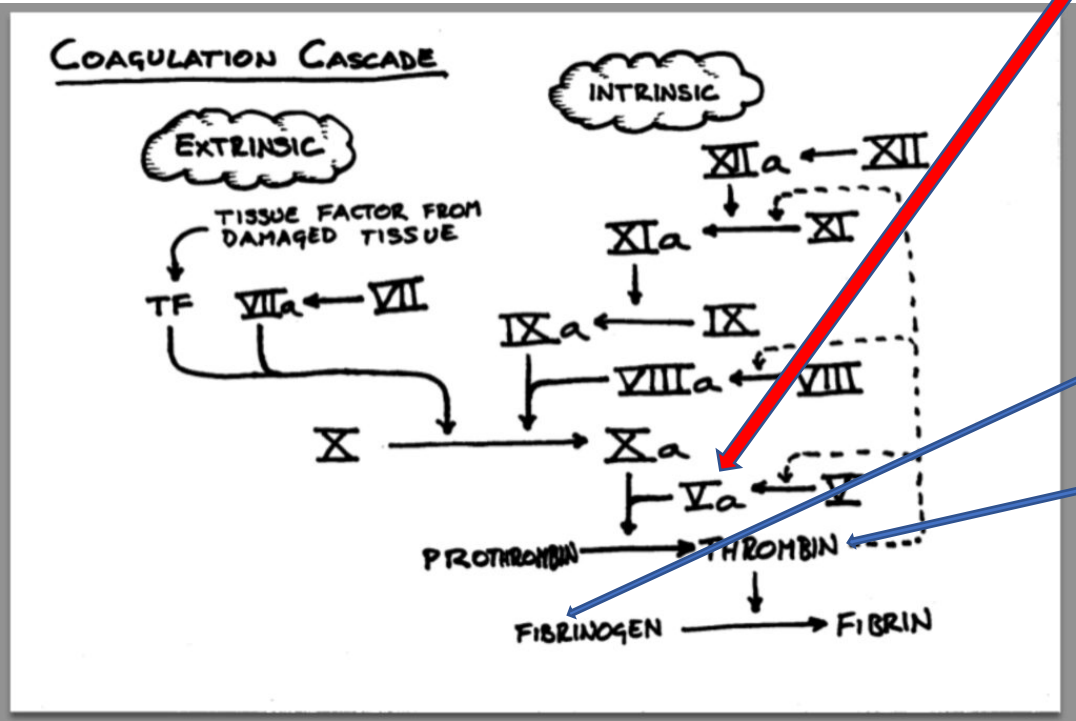
What is “rebalancing”



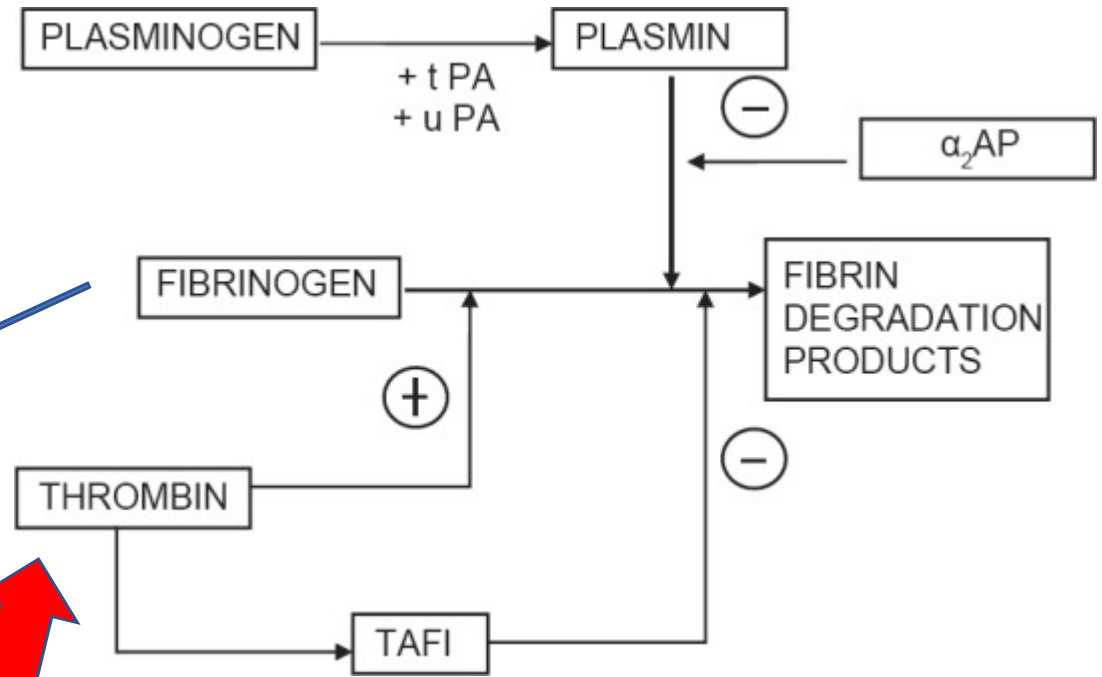
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GENE THERAPY



Protein C, S



Antithrombin





Rare Bleeding Disorders

Yes, you are a Zebra



NORD[®]
National Organization
for Rare Disorders



NEVER ALONE



<https://wfh.org/>



NATIONAL
BLEEDING DISORDERS
FOUNDATION

[The National Hemophilia Foundation Has a New Name |
National Bleeding Disorders Foundation](#)



HCCPA

PennState



Thomas Jefferson
University

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TOWER HEALTH
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