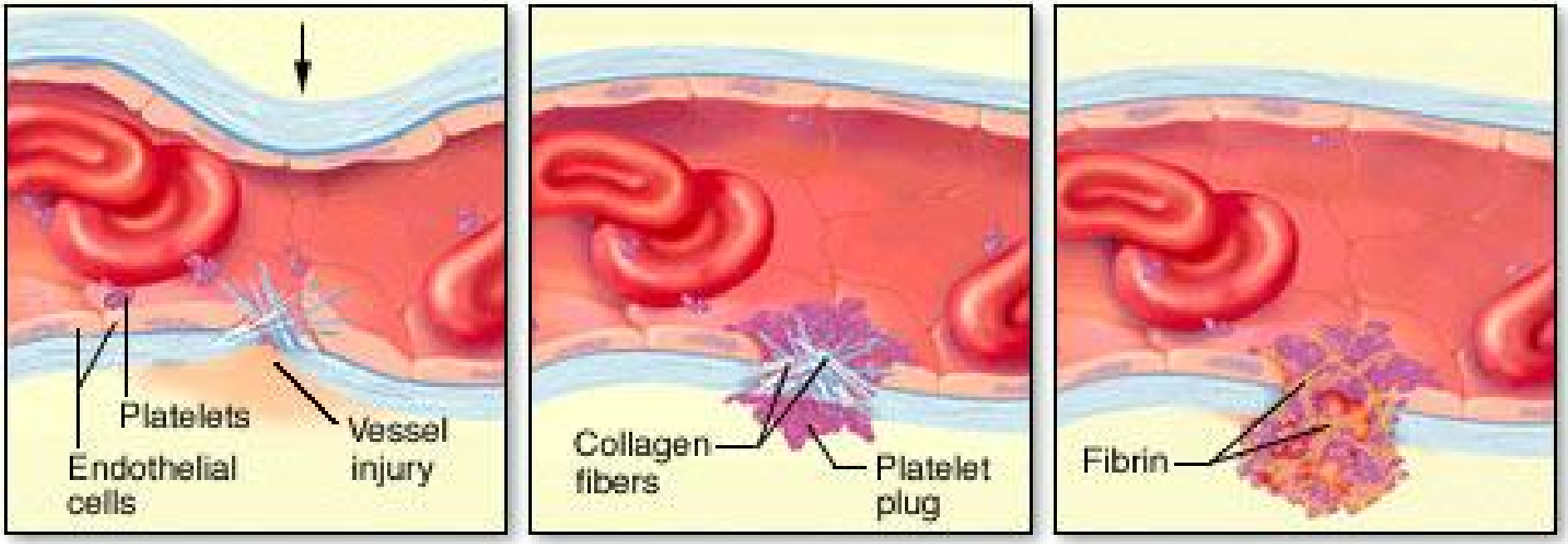
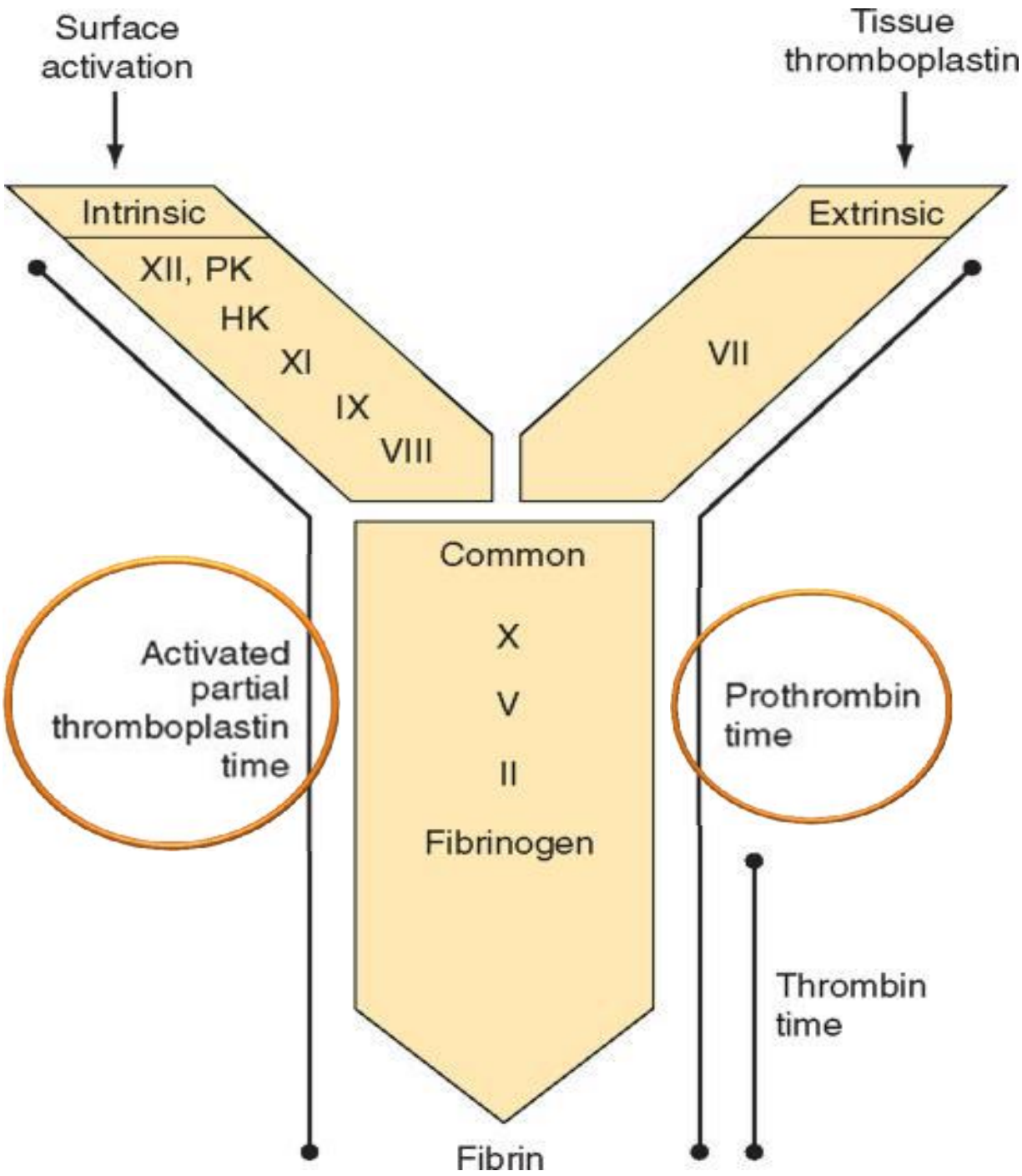
HEMOPHILIA



(a) Vasoconstriction (b) Platelet aggregation (c) Clot formation



PT : 2+5+10

7

PTT : 2+5+10

8 9

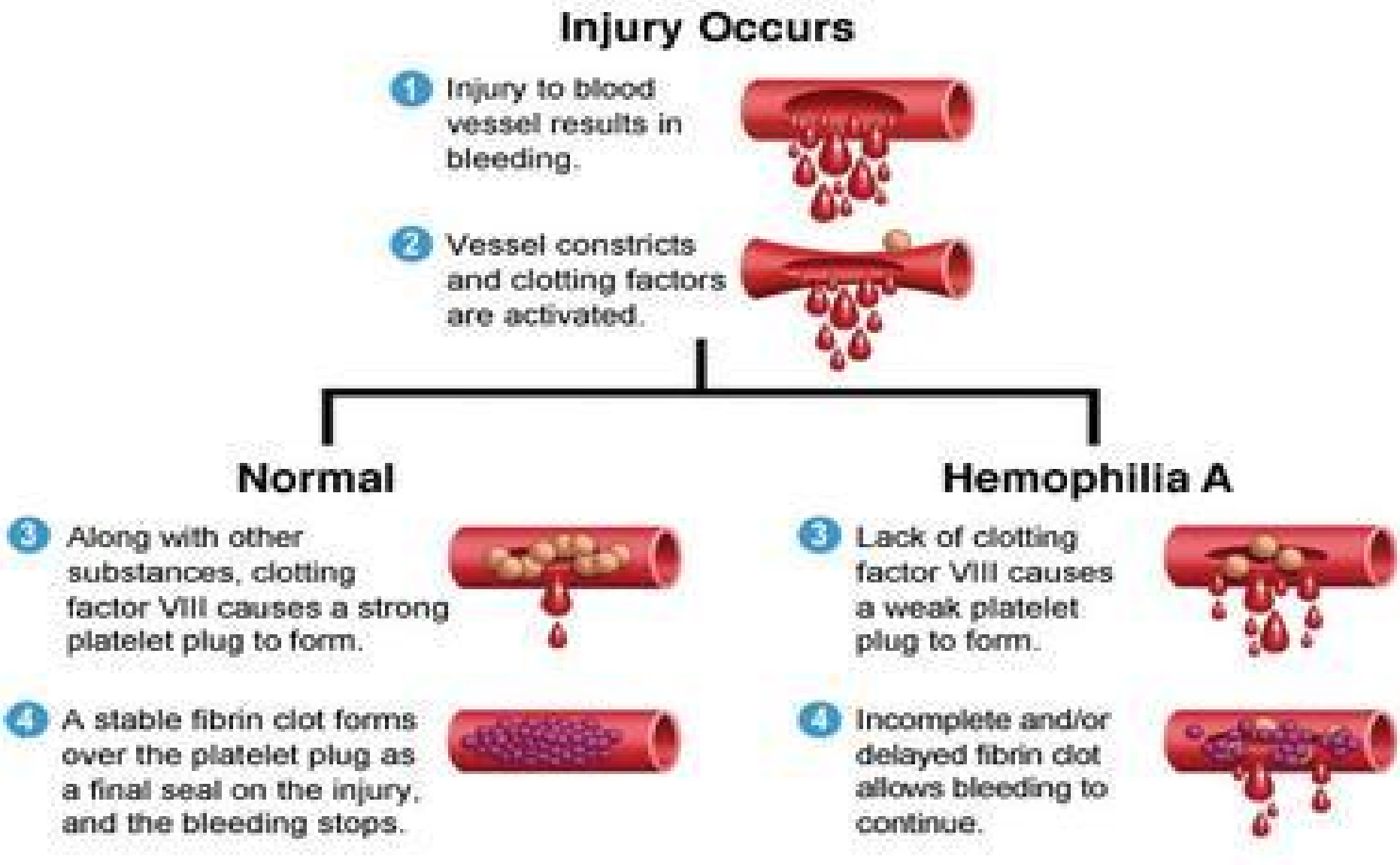
11 12

HEMOPHILIA A

- A reduction of factor VIII
- The most common *congenital disorder* of coagulation.
- Factor VIII is synthesized by:
  - ü Liver
  - ü Endothelial cells
  - ü Others: spleen, kidney and placenta.

- X- linked recessive disorder.
- Almost all patients males (only males actually have symptoms of hemophilia)
- Female hemophilia carriers with lower than normal levels of factor VIII may have bleeding problems similar to males with mild hemophilia. Some female carriers have normal clotting protein levels and do not have abnormal bleeding.

PATHOPHYSIOLOGY



CLINICAL FEATURES

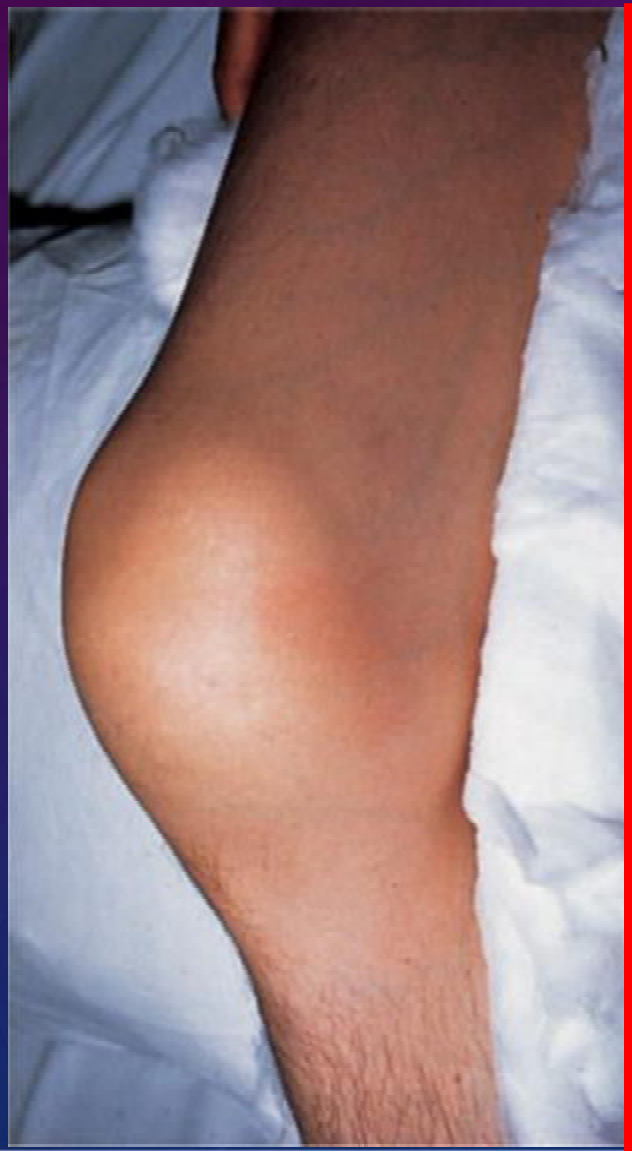
- Hemophilia *sporadic* in one - third of cases.
- Whereas severe hemophilia is usually diagnosed within the first 2 years of life (usually after 6 months), individuals with moderate and mild forms may escape diagnosis until adulthood.

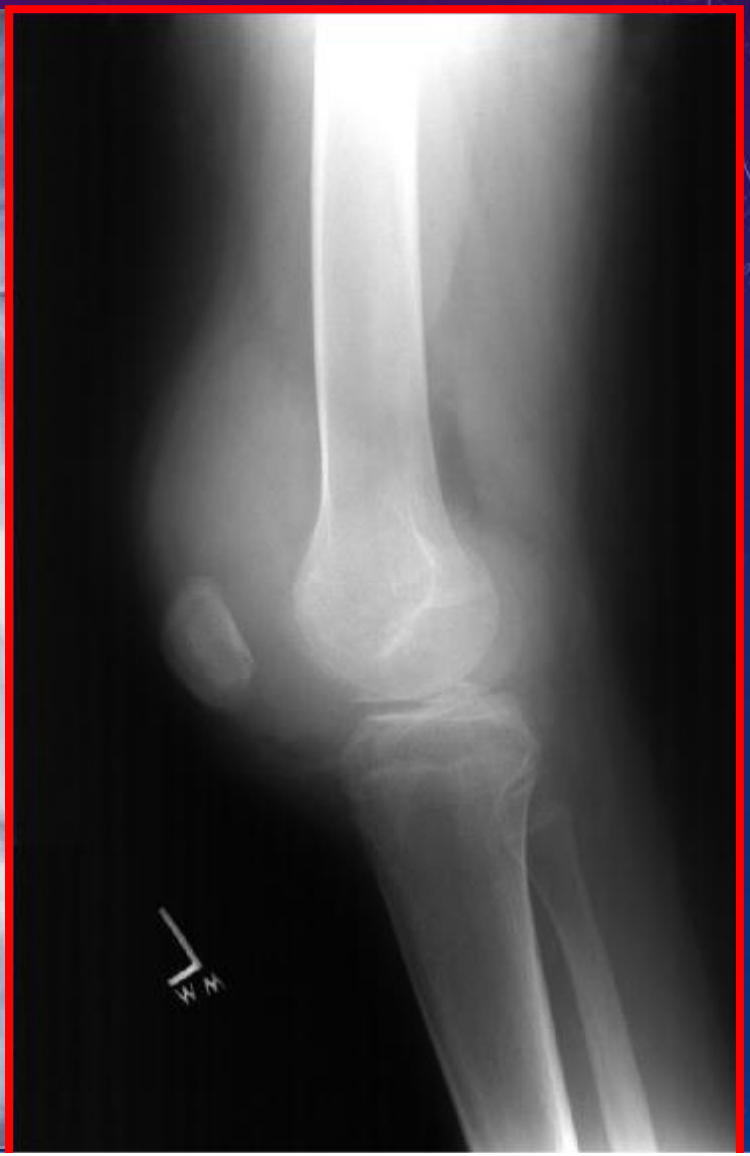
Severity	FVIII level	Clinical presentation
Severe	< 1%	Frequent spontaneous bleeding into joints, muscles and internal organs
Moderate	1-5 %	some ‘ spontaneous ’ bleeds, bleeding after minor trauma
Mild	5-40 %	Bleeding only after significant trauma, surgery

APPROXIMATE FREQUENCIES OF BLEEDING AT DIFFERENT SITES

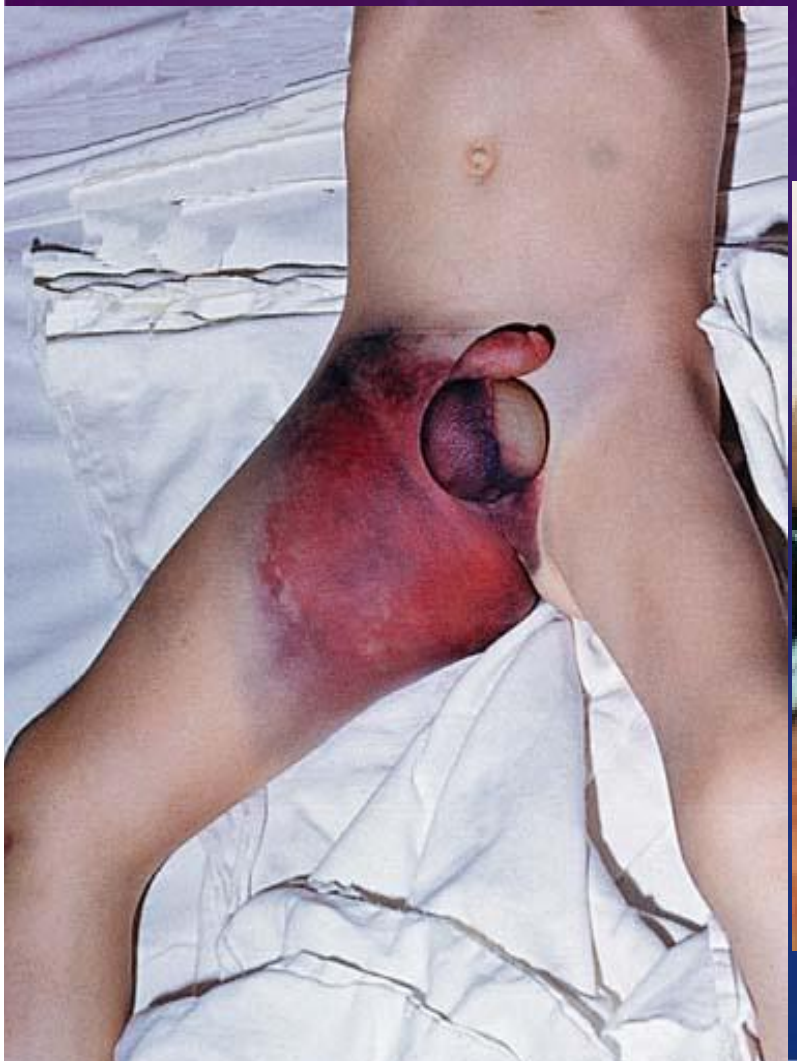
Site of bleeding	Frequency
Hemarthrosis	70-80%
Muscle	10-20%
Other major bleeding	5-10%
CNS bleeding	<5%

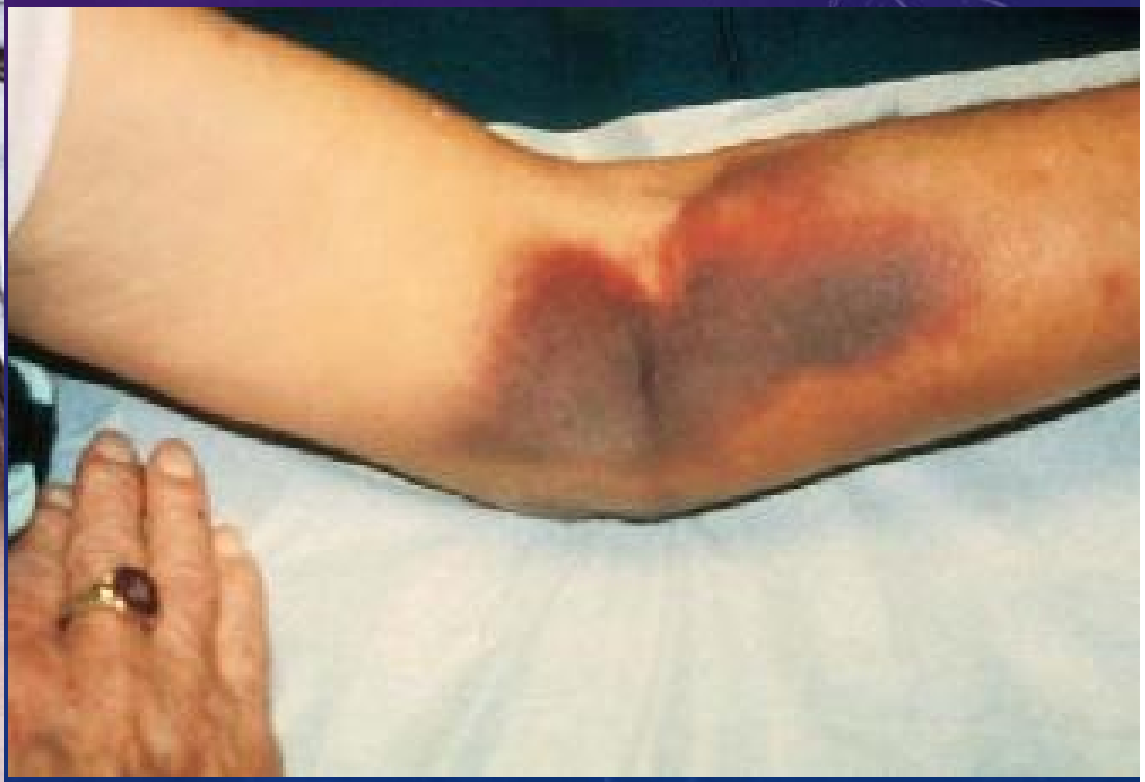
- Individuals with severe hemophilia experience recurrent hemarthroses in large joints .



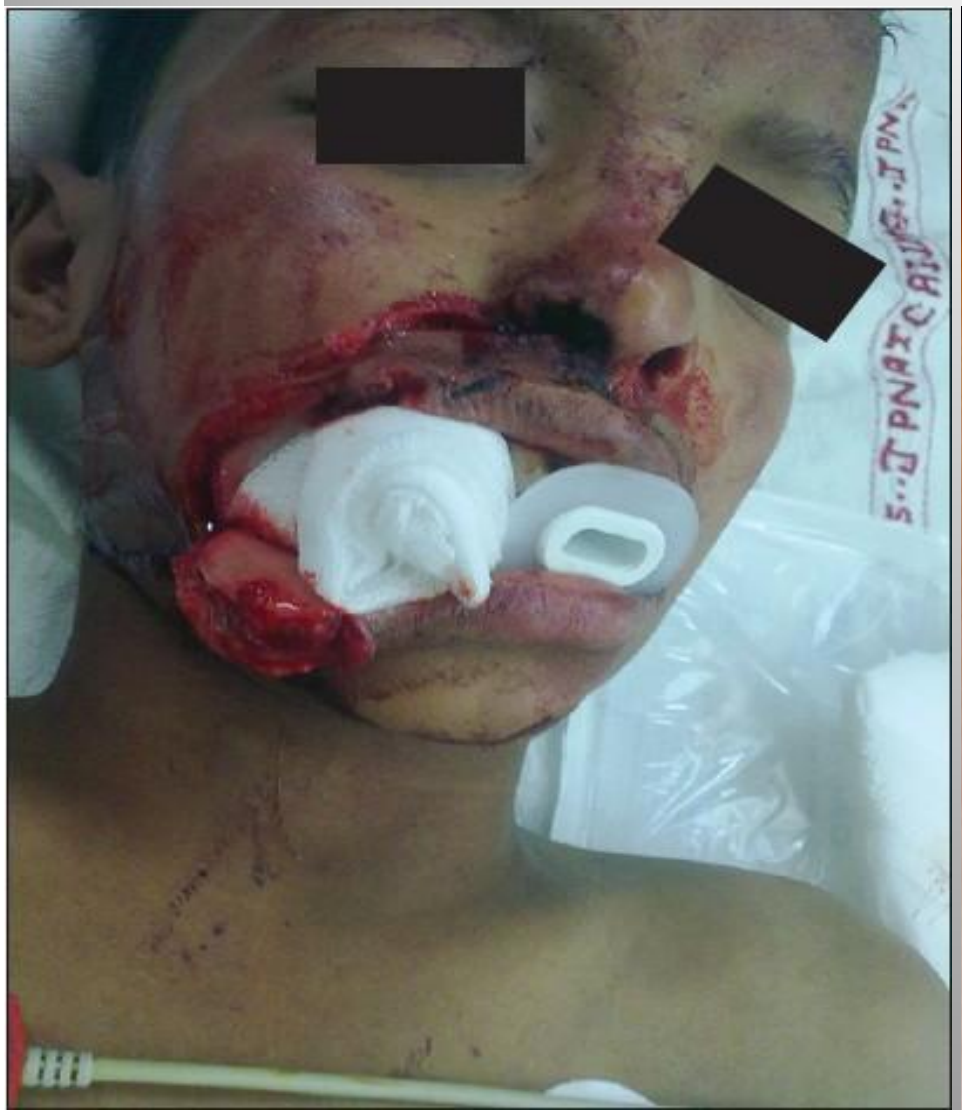


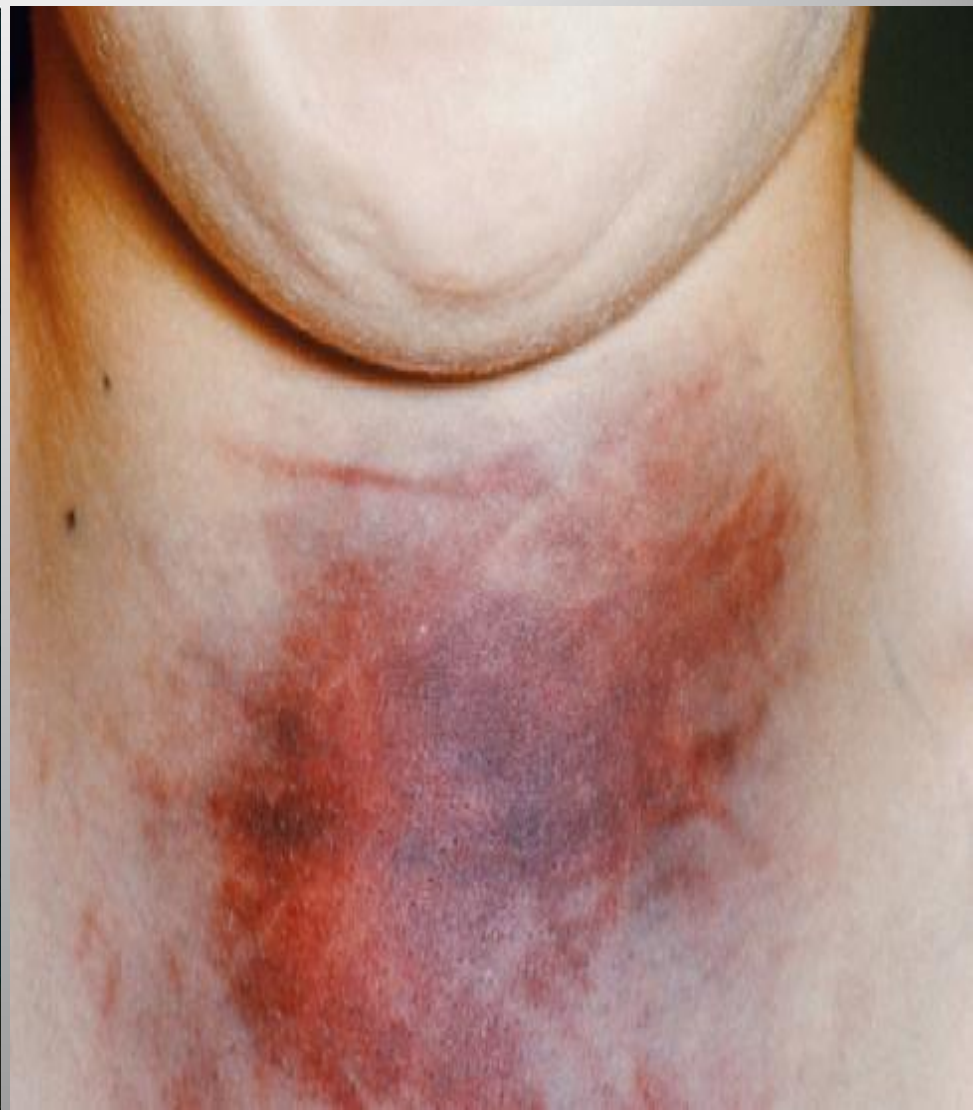
- Muscle hematomas (calf, psoas muscles) are also characteristic of hemophilia.

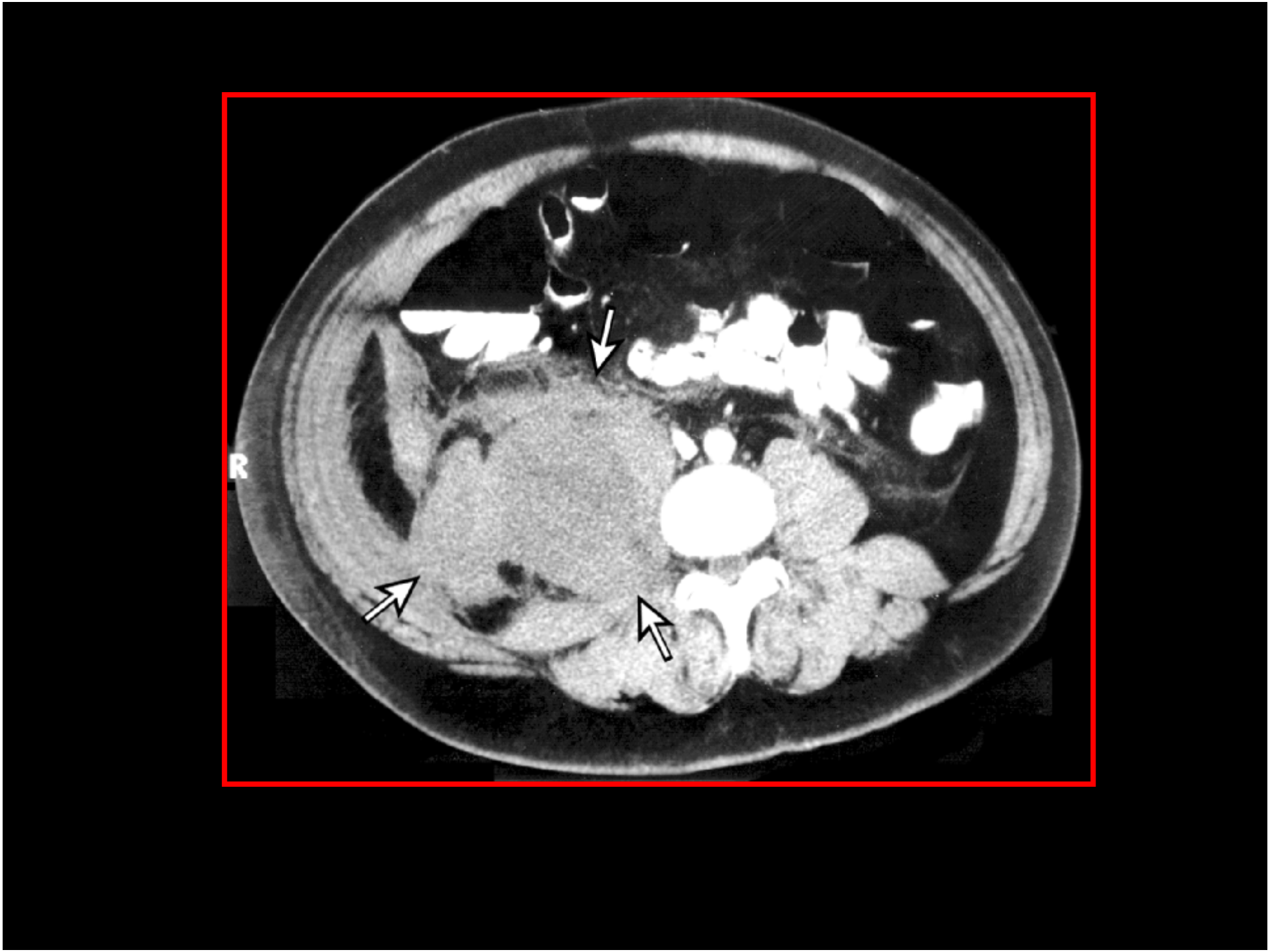


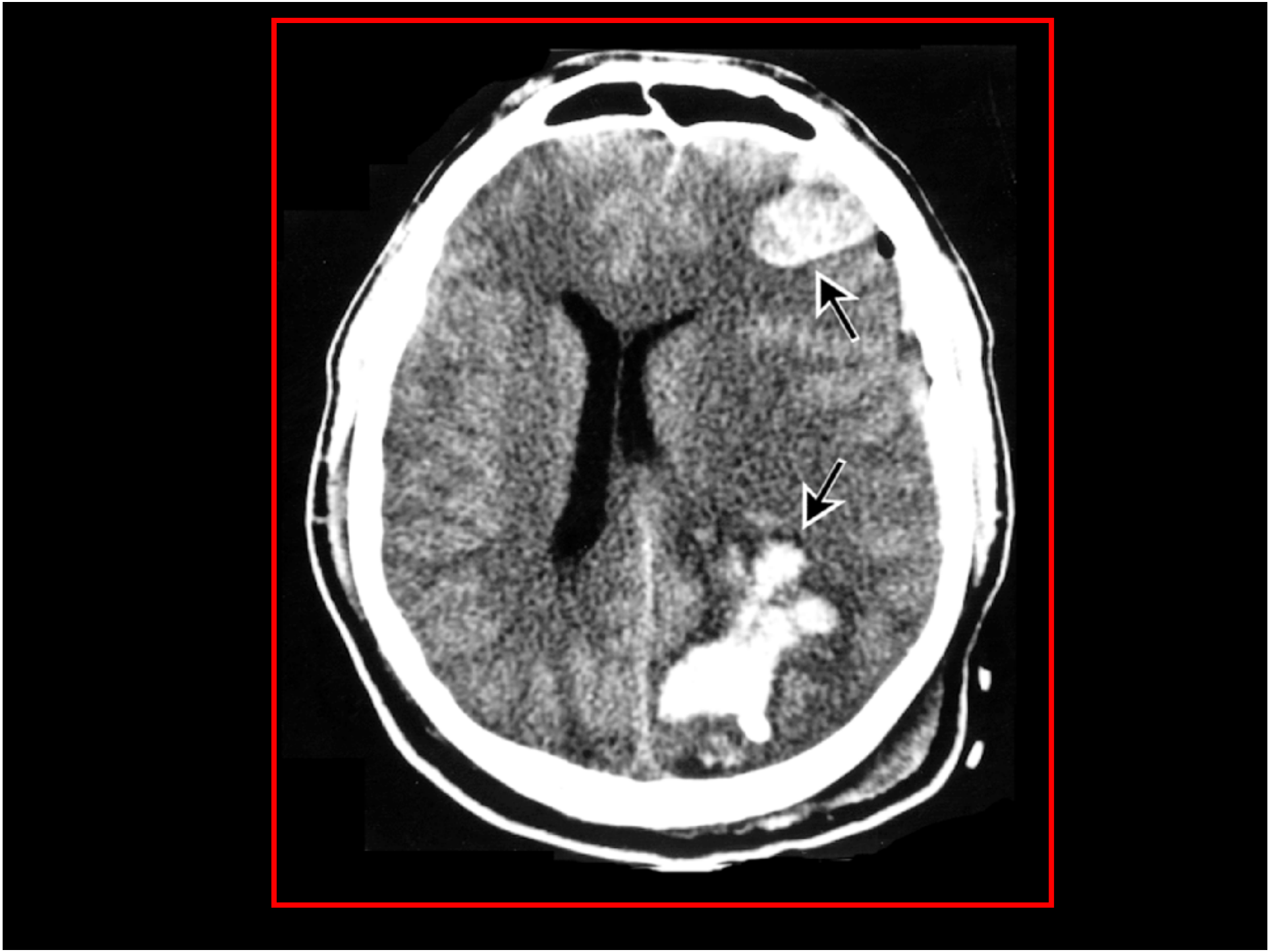


- Bleeding can occur at any site: oropharyngeal bleeding, hematuria, hematemesis and melena is less common.
- CNS bleeding is uncommon but can occur after a slight head injury and remains a significant cause of death.









- Surgery and open trauma invariably lead to dangerous hemorrhage in the untreated individual with hemophilia.

Soft tissue bleeds and bruising
- no functional impairment
- tenderness, but no severe pain
- no factor needed

Neck swelling: EMERGENCY
- potential airway compromise
- treat with a major dose of factor

Deltoid/forearm bleed and bruising
- routine factor dose
- major factor dose if a compartment syndrome is suspected

Iliopsoas bleeds
- flexed hip
- pain, inability to extend the leg on the affected side
- treat with a major dose of factor

Buttock bleeds
- pain
- with/without swelling
- routine factor dose
- major factor dose if the leg on the affected side exhibits tingling or swelling

Thigh/calf bleeds
- pain
- with/without swelling
- impaired mobility
- routine factor dose
- major factor dose if compartment syndrome is suspected

HEMOPHILIA

(Inherited Blood Disorder
Factor VIII, Classic, or Type A)

- No Cure

- Avoid Injury & Meds That Promote Bleeding

- Good Nutrition

- Good Dental Hygiene

- IV Administration Of Deficient Clotting Factor

Intracranial Hemorrhage

Prolonged Nosebleeds

Bruises Easily

Warm, Painful, Swollen Joints With ↓ Movement

GI Hemorrhage

INVESTIGATIONS

- Prolonged APTT
- Prothrombin time and thrombin time are normal.
- Normal platelet count, and bleeding time.
- FVIII clotting activity are low, with all other factors normal.
- Von Willebrand factor (VWF) antigen and activity are normal.

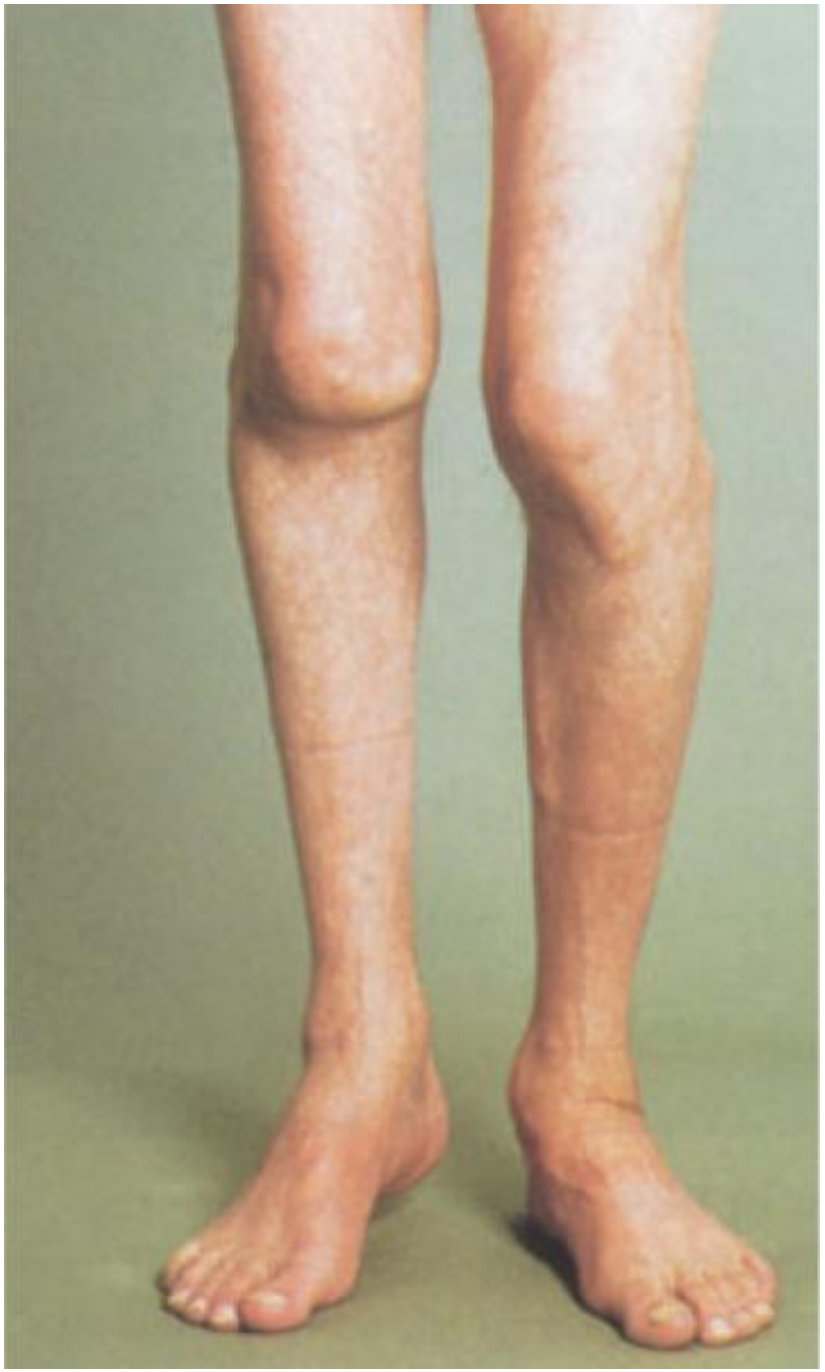
TREATMENT

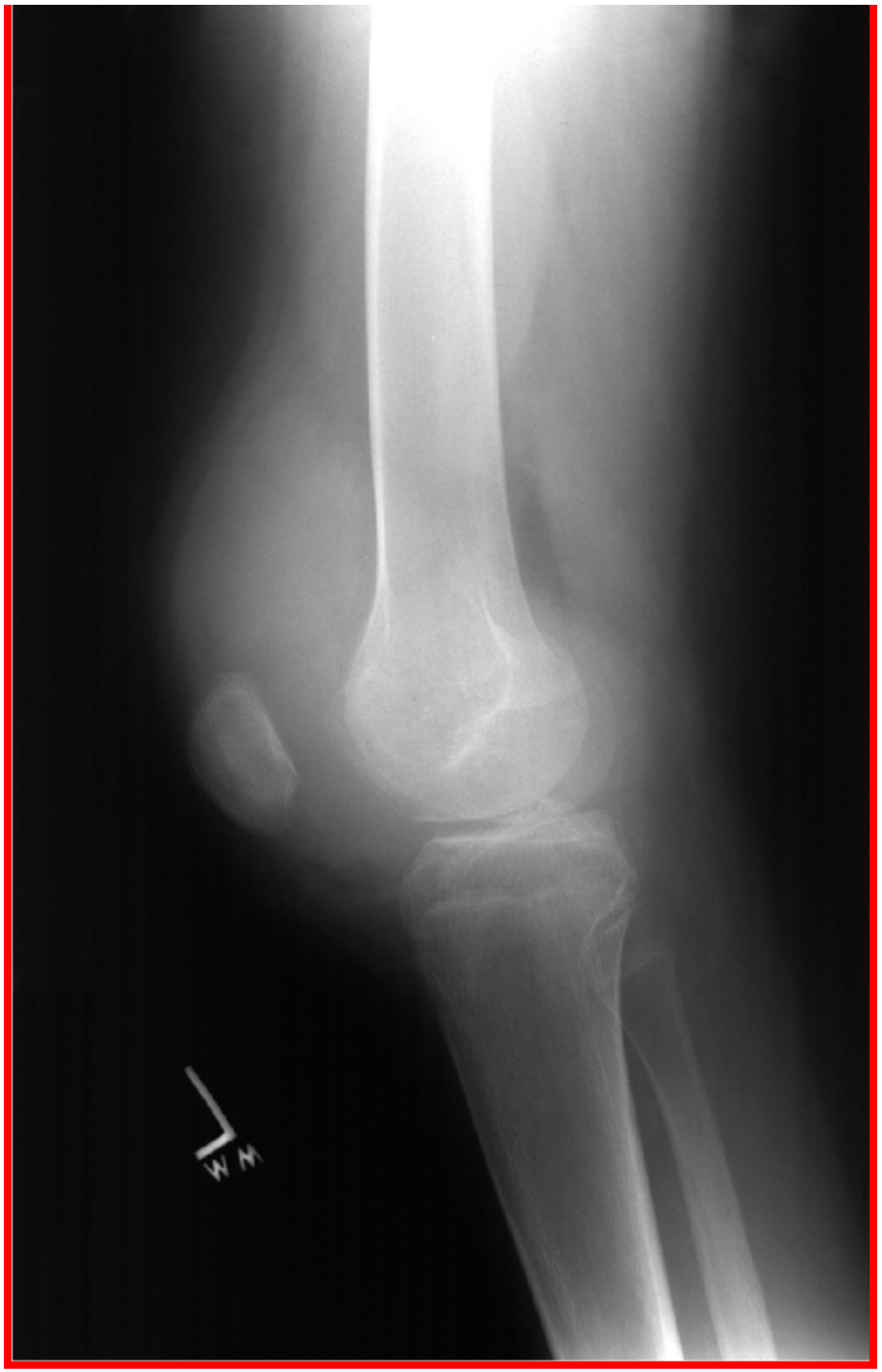
- Resting of the bleeding site.
- Once bleeding has settled, the patient should be mobilized and physiotherapy used to restore strength to the surrounding muscles.
- In individuals with a basal factor VIII level of ≥10% it may be possible to raise the level 3-5 fold with desmopressin. This is often sufficient to treat a mild bleed or cover minor surgery such as dental extraction.

- Intravenous infusion of factor VIII concentrate.
- FVIII dose (IU) = (Target FVIII levels – FVIII baseline levels) x body weight (kg) x 0.5 unit/kg
- The FVIII half-life of 8–12 h requires injections twice a day to maintain therapeutic levels.

COMPLICATION

- Synovial hypertrophy, destruction of the cartilage and secondary osteoarthrosis .
- Transmission of infection.
- Anti-factor VIII antibodies:
  - Factor VII
  - High dose factor VIII
  - Immune tolerance induction
  - plasma - derived prothrombin complex concentrate





HEMOPHILIA B (CHRISTMAS DISEASE)

- **Definition: reduction of the plasma factor IX level.**
- **This disorder is clinically indistinguishable from hemophilia A but is less common.**
- **Presentation: (less severe)**
- **Treatment?**