Hemophilia in Girls and Women

QUICK REFERENCE GUIDE FOR PRIMARY CARE PROVIDERS

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Hemophilia Fast Facts [1]

Hemophilia A:
- Factor VIII deficiency
- Most common subtype
- Prevalence: 1 per 5,847 males

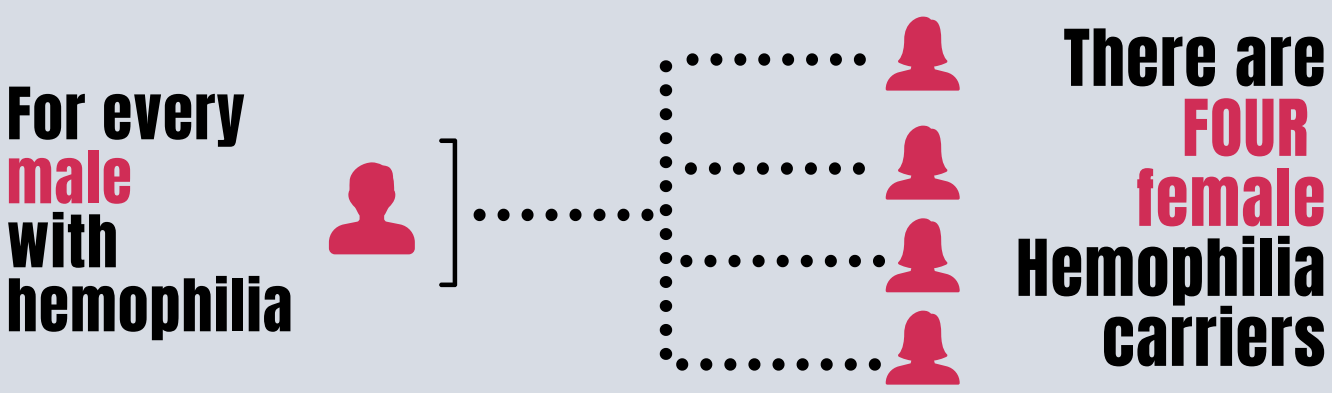
Hemophilia B:
- Factor IX deficiency
- Less common subtype
- Prevalence: 1 case per 26,316 males.

Both types follow X-linked recessive inheritance pattern

of cases have **no known family history** due to spontaneous mutation.

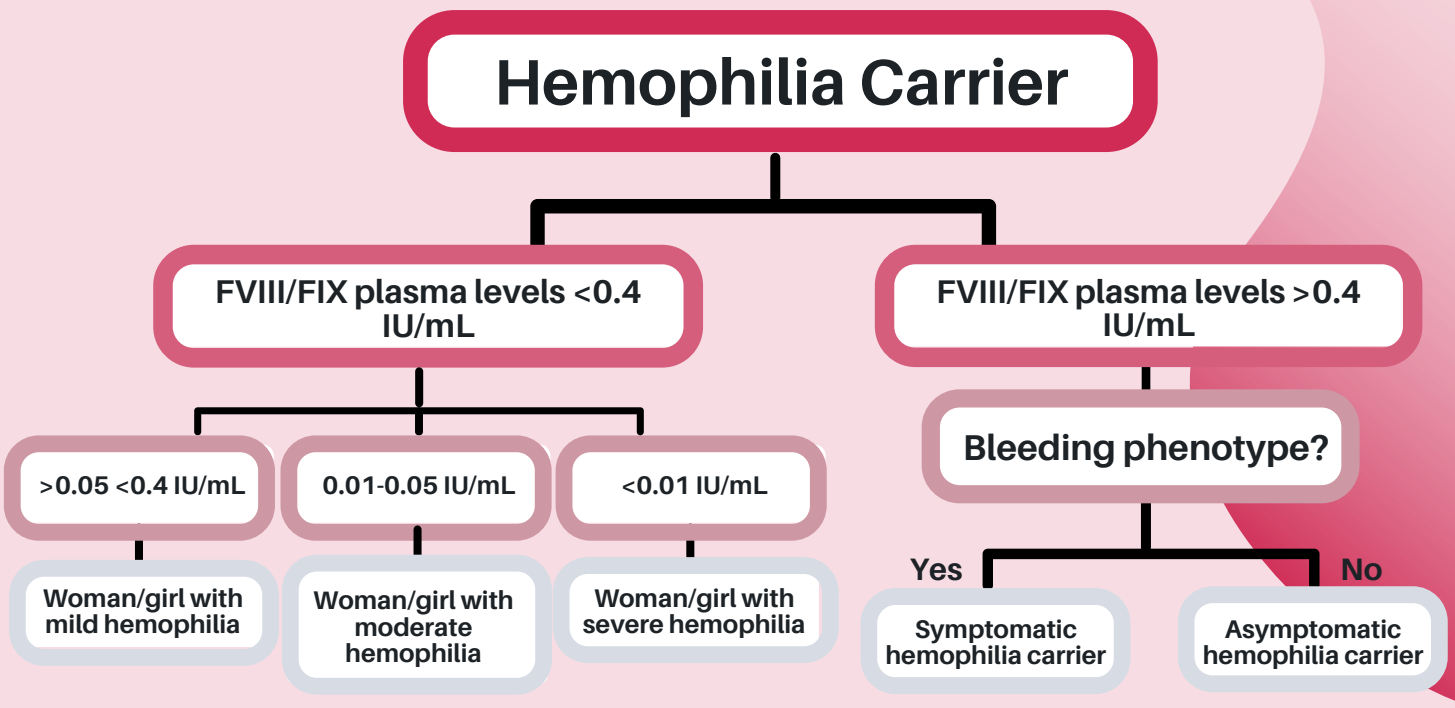
How Many Women are Affected by Hemophilia?

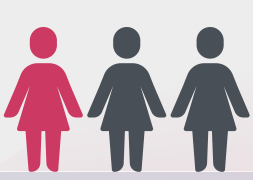
True prevalence of hemophilia carriers is not known but is estimated that



Estimated prevalence numbers are not reflected in international and national registries indicating high number of unidentified and undiagnosed hemophilia carriers.[2]

How Hemophilia can Affect Women and Girls





1 in 3 of all carriers will manifest below normal factor levels (FVIII/FIX <0.40 IU/mL) [3,4]

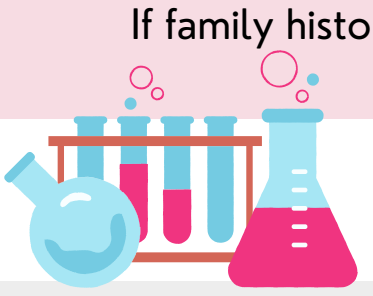
Even carriers with **normal** factor levels can experience **abnormal** bleeding symptoms [5]

Abnormal Bleeding Symptoms

- Heavy Menstrual Bleeding
- Frequent epistaxis
- Easy bruising
- Excessive post-surgical bleeding
- Oral cavity bleeding ex. excessive bleeding post-dental extractions
- Post-partum hemorrhage including delayed post-partum hemorrhage
- Joint issues, both hemarthrosis and sub-clinical joint bleeding

Diagnostic Tests

If family history of hemophilia present, consider referral to hematologist for further evaluation and diagnostic testing.



Factor Level Testing

- Factor VIII or Factor IX, depending on family history

<0.40 IU/mL	• Abnormal and diagnostic of hemophilia of corresponding severity (mild, moderate, severe), • Likely abnormal bleeding symptoms
0.40-0.60 IU/mL	• Indicative of hemophilia carriership with normal factor level • Possible abnormal bleeding symptoms
>0.60 IU/mL	• Does not rule out hemophilia carrier-ship or • Possible abnormal bleeding

Genetic Testing

- Molecular diagnostic testing for Hemophilia A and Hemophilia B (including carriership)
- Provides absolute confirmation of carrier status
- Healthcare providers can find additional information at the Canadian National Inherited Bleeding Disorder Genotyping Laboratory (https://www.nibdgl.ca/)

Bleeding Assessment

- Can be done by healthcare provider via Bleeding Assessment Tool (ex. ISTH- BAT)
- Can be done by patient via Self-BAT available at www.letstalkperiod.ca
  - If final bleeding score is abnormal, indicative of abnormal bleeding phenotype

Additional Testing

- Consider CBC and Ferritin in presence of abnormal bleeding symptoms

Management

Treatment options for women affected by hemophilia depend on disease and symptom severity and may include:

Tranexamic Acid

- Antifibrinolytic agent used to prevent breakdown of blood clots in the mucocutaneous parts of the body
- Can be used in women with Hemophilia A, Hemophilia B and symptomatic carriers with normal factor levels
- Useful for heavy menstrual bleeding, post-partum bleeding, epistaxis, pre- and post-dental procedures or other minor procedures

Desmopressin

- Synthetic hormone that can significantly increase Factor VIII level and activity
- Not used for Hemophilia B (FIX deficiency) but can be used in women with Hemophilia A (FVIII deficiency) if prior responsiveness has been determined
- Occasionally used in symptomatic hemophilia carriers with normal factor levels but significant bleeding symptoms

Factor Replacement Therapy

- Replaces the missing clotting factor and can be used in women with hemophilia

Gynecologic Treatment for Heavy Menstrual Bleeding

- Hormone therapy via oral contraceptive pill, vaginal ring, dermal patches
- Intrauterine device (IUD)
- Tranexamic acid
- Endometrial ablation
- Hysterectomy
- Factor replacement (in **rare** cases)

References:

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