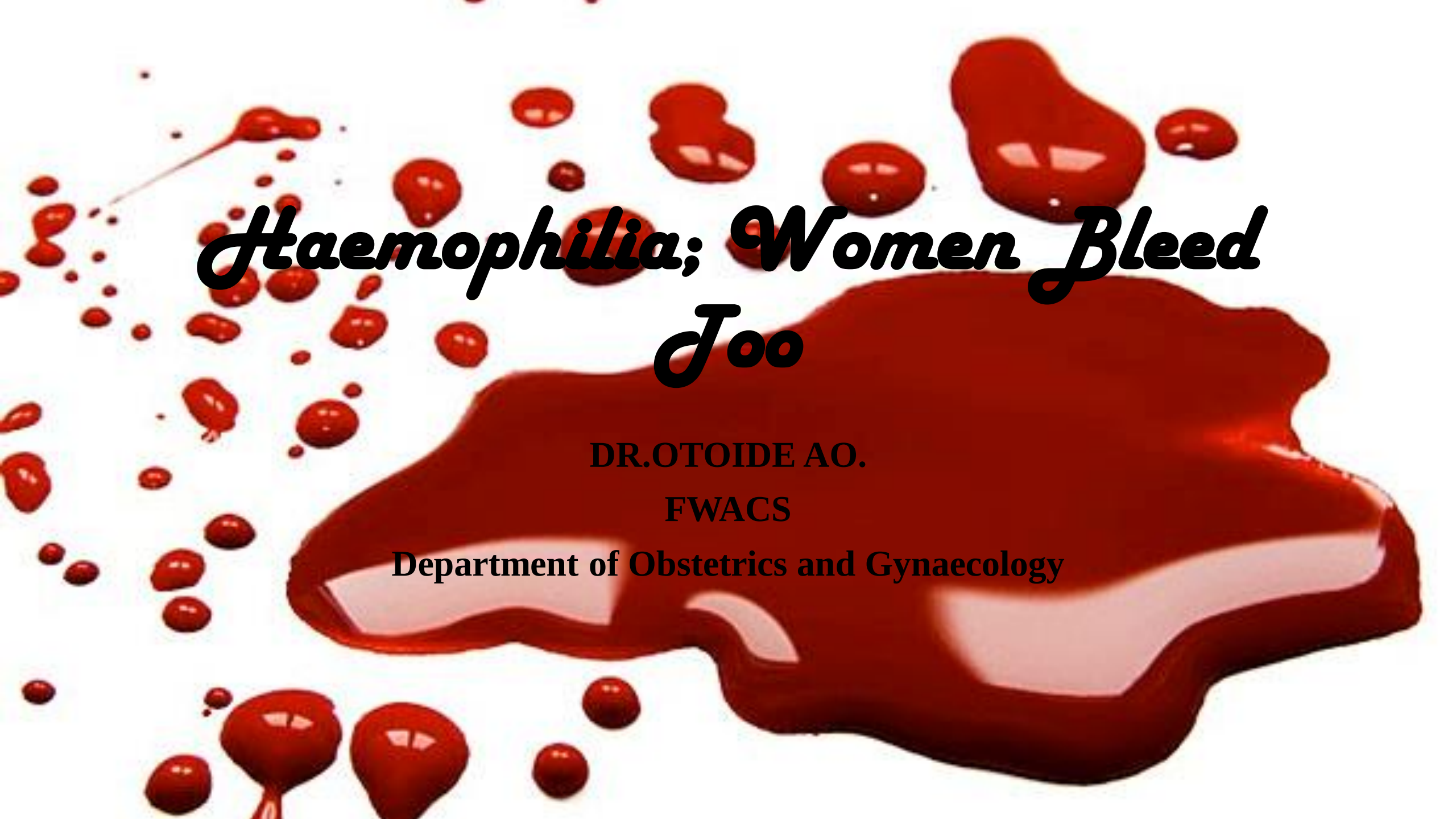

Haemophilia; Women Bleed Too

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SYNOPSIS

1. INTRODUCTION
2. EPIDEMIOLOGY
3. AETIOLOGY
4. CLINICAL PRESENTATION
5. DIAGNOSIS/INVESTIGATIONS
6. TREATMENT
7. COMPLICATIONS
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INTRODUCTION

- Haemophilia is a genetically inherited bleeding disorder **primarily** affecting males.
- Females can also have haemophilia but it is much rarer.
- It is a coagulation disorder caused by mal-functional, absent or low levels of protein clotting factor; Factors VIII and IX.

INTRODUCTION

- Factor VIII deficiency is known as Haemophilia A and Factor IX deficiency is known as Haemophilia B or Christmas disease and it is rarer than Haemophilia A
- Acquired Haemophilia is a separate non-inherited condition. It is much rarer than congenital haemophilia and has an autoimmune-related aetiology with no genetic inheritance pattern

INTRODUCTION

- It is classified as mild, moderate and severe based on the levels of factors VIII and IX.
- Women with Haemophilia may be carriers, symptomatic carriers or mild haemophilia
- Moderate or severe haemophilia is very rare in females.

EPIDEMIOLOGY

- Haemophilia A is found in 1 in 5000 males, whereas the incidence of Haemophilia B is found in 1 in 30,000 males.
- It has been described that for one man with haemophilia, 2.7 to 5 potential carriers can be found in the family and 1.56 of them are actual symptomatic carrier.

EPIDEMIOLOGY

- A third of carriers are symptomatic
- About 30% of women report heavy menstrual bleeding but only about half of them seek medical attention
- Of those who seek medical attention 15-30% have a bleeding disorder.

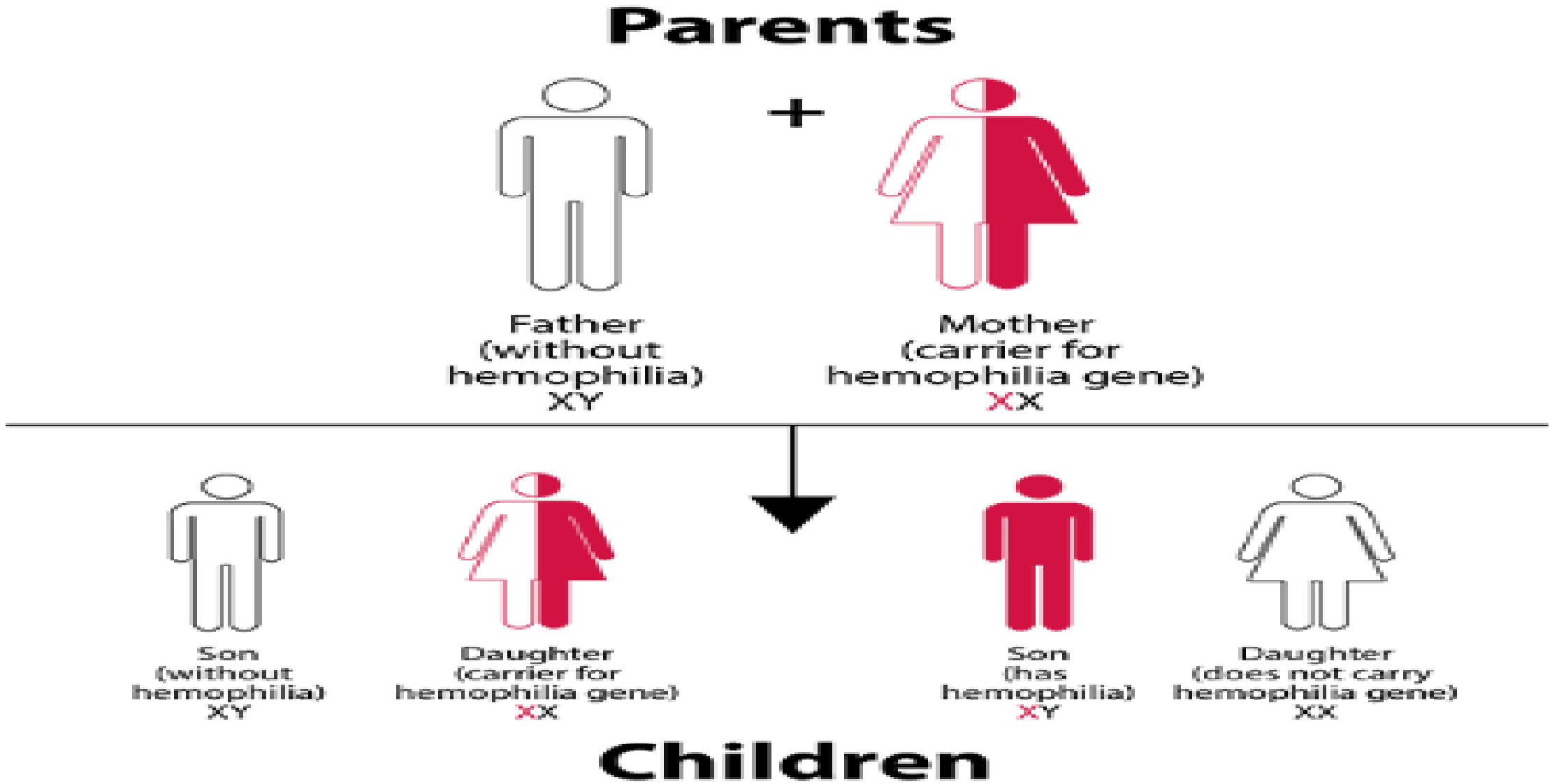
EPIDEMIOLOGY

- Postpartum Haemorrhage (PPH) affects 6% of all deliveries and it is the leading cause of maternal death worldwide.
- The commonest causes of PPH are, TONE (uterine atony), TRAUMA (laceration, hematoma, inversion, rupture etc), TISSUE (retained products of conception or placenta, morbidly adherent placenta), THROMBIN (coagulopathy).

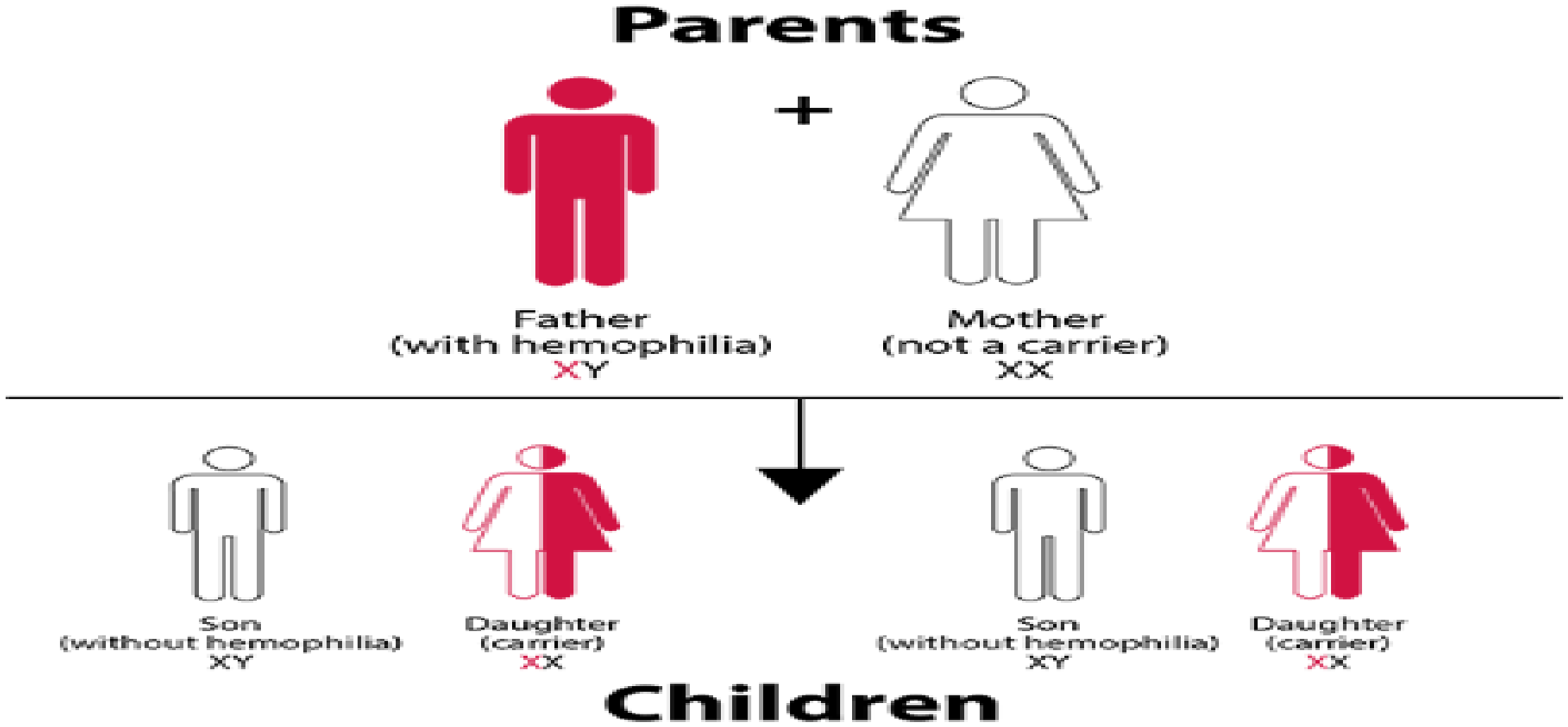
EPIDEMIOLOGY

- The incidence of PPH in women with inherited bleeding disoders is greater than that of unaffected women.
- It is known that these women have a maternal mortality rate nearly 10 times that of unaffected controls

AETIOLOGY


Parents
Father (without hemophilia) XY
Mother (carrier for hemophilia gene) XX
Son (without hemophilia) XY
Daughter (carrier for hemophilia gene) XX
Son (has hemophilia) XY
Daughter (does not carry hemophilia gene) XX
Children

AETIOLOGY


Parents
+
Father
(with hemophilia)
XY
Mother
(not a carrier)
XX
Son
(without hemophilia)
XY
Daughter
(carrier)
XX
Son
(without hemophilia)
XY
Daughter
(carrier)
XX
Children

AETIOLOGY

- In some cases, female carriers of haemophilia can have low levels (<50%) of either factor VIII or factor IX and may experience bleeding symptoms. Female carriers who have bleeding symptoms are considered to have mild haemophilia.
- These were previously classified as symptomatic carriers.

AETIOLOGY

- It is also possible for a female to have a father with haemophilia and a mother who is a carrier, and thereby inherit an affected X chromosome from both parents. Such daughters would have haemophilia, which may be mild, moderate or severe.
- Mild haemophilia don’t typically cause spontaneous bleeds, it causes so few symptoms that it is not diagnosed until adolescence or adulthood. Moderate and severe haemophilia are typically causes for more frequent and more severe bleeding compared to mild haemophilia.

AETIOLOGY

- In chromosomal disorders in which females carry only one X chromosome. If these females inherit the X chromosome that carries the haemophilia gene, they will have haemophilia.
- Skewed x-inactivation occurs when the affected X with the haemophilia gene is more active than the unaffected X. When this occurs, females can have low factor levels.

CLINICAL PRESENTATION

- Heavy menstrual cycles (40-50% of carriers) such as:
  - Bleeding for more than 7 days from the time bleeding begins before it completely stops.
  - Flooding or gushing of blood that limits daily activities such as housework, exercise, or social activities.
  - Passing clots that are bigger than a quarter.
  - Changing a tampon or pad, possibly even both, every hour or more often on heaviest day(s).
  - Being treated for anemia or being told you are “low in iron.”

CLINICAL PRESENTATION

- Internal organ bleeding is often life threatening. And commonly occurs as and during;
- Postpartum hemorrhage (1PPH;13-22%, 2PPH; 9-20%)
- Haemorrhagic ovarian cysts
- Insertion of intrauterine devices
- Surgical abortion

CLINICAL PRESENTATION

- They may also experience heavy bleeding after surgeries; Caesarean section, myomectomy, dental surgery etc.
- Two-thirds of hysterectomies are as a result of heavy uterine bleeding.
- Experiencing prolonged bleeding episodes such as might occur as a result of:
  - Frequent nose bleeds (longer than 10 minutes);
  - Bleeding from cuts or injury (longer than 5 minutes); or
  - Easy bruising (weekly, raised, and larger than a quarter in size).

DIAGNOSIS/INVESTIGATIONS

- Diagnosis is made by;
- Personal history of bleeding
- Family history of bleeding and the inheritance pattern of those affected
- Genetic testing of haemophilia A and B patient as well as carriers
- Screening of other at-risk family members, especially women

DIAGNOSIS/INVESTIGATIONS

- Clotting profile
- Factor VIII
- Factor IX
- Choronic villus sampling (10-13 week, 1% risk)
- Amniocentesis (after 15 weeks, 0.3% risk)
- Fetal free cell DNA or USS for sex determination

TREATMENT

- Multidisciplinary approach
- Treatment could be prophylactic or symptomatic
- Hormonal therapy, such as oral contraceptives, for treatment of heavy periods
- Antifibrinolytic drugs which prevent blood clots from breaking down, mostly used for mucous membrane bleeding
- DDAVP (FVIII deficiency only) to increase FVIII levels when given
- Factor infusion to treat bleeding episodes

TREATMENT

- Women with factor levels less than 50% should receive treatment with factor replacement at the time of delivery
- The World Federation of Haemophilia strongly recommends pathogen-safe plasma-derived or recombinant concentrates over cryoprecipitate or fresh frozen plasma

TREATMENT

- Non-steroidal anti-inflammatory drugs (NSAIDs should be avoided.
- Tranexamic acid may be used to prevent excessive bleeding.
- Desmopressin is not preferred at the time of delivery, as safe administration of desmopressin requires fluid restriction, once a woman is no longer receiving intravenous fluids, a short course of desmopressin may also be used to prevent excessive bleeding in haemophilia A carriers.

CONCLUSION

- Management of women with haemophilia is challenging as the diagnosis is often missed due to misconceptions that the disease affects only males, poor knowledge on normal and abnormal menstruation on both the patient and the doctors side, and lack of or expensive diagnostic tests in low resource settings.

CONCLUSION

- Women often experience life threatening yet preventable hemorrhages, and even when the diagnosis is suspected or confirmed, treatments are often unavailable.
- Its imperative that we let the world know that women bleed too…


Thank
you