MEDIBEATS
The Medical Bulletin

In Syndrome

HEMOPHILIA

Introduction :

- It is an inherited X-linked disorder of coagulation.

Etiology :

- Hemophilia is usually an inherited condition and is caused by the deficiency of clotting factors in the blood.
- It is almost always due to a defect or mutation in the gene for the clotting factor.
- Both hemophilia A and B are inherited via an X-linked recessive pattern where 100% of females born from affected fathers will be carriers, and none of the males born will be affected.
- Female carrier mothers have a 50% chance of having affected males and a 50% chance of having carrier females.

Epidemiology: 1 in 10000 live births.

Types :

- Hemophilia A – due to factor VIII deficiency.
- Hemophilia B – due to factor IX deficiency.

Symptoms :

- Start in childhood and continue for rest of life.
- Recurrent bleeding into the joints. Bleeding into soft tissues of arms and legs can result in compartment syndrome.
- After IM injections, intramuscular hematomas may develop.
- Bleeding from mucous membranes.
- Hematuria
- Intracranial bleed (2nd most common cause of death).

Signs :

- Hemarthrosis causes bond and joint destruction. Knee joint most commonly affected.

Diagnosis :

- Prothrombin time - Normal.
- Activated partial thromboplastin time – Prolonged.
- Clotting time – Prolonged, Bleeding time – Normal.
- Diagnosis is confirmed by demonstrating specific factor (VIII or IX) deficiency by the specific assay.

Treatment:

- To avoid trauma or falls.
- Replacement of specific factor is must when bleeding occurs.
- For minor bleeding, factor VIII concentrates should be raised to 25% of normal, for severe bleeding it should be raised to about 50%.
- For major surgery, factor VIII should be raised to 100%.
- Desmopressin is given IV, which can elevate factor VIII levels.

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