National University – Sudan

Faculty of Medical Lab. Sciences

Advanced haematology

MLS- HEMA-324

Batch 9 – 3rd year (sem. 6)

von Willebrand Disease

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von Willebrand Disease

Objectives

- 1- define the von Willebrand disease.
- 2- List the causes of the von Willebrand disease.
- 3- Outline the pathophysiological aspects of the von Willebrand disease.
- 4- List the clinical features of the von Willebrand disease.
- 5- Diagnose the von Willbrand disease.

von Willebrand Disease (vWD)

- It is one of Hereditary coagulation abnormality "Autosomal inherited bleeding disorder".
- It arises from a qualitative or quantitative deficiency of VWF > a protein that is required for platelet adhesion.
- vWF > synthesized in : endothelium & Megakaryocytes (α-granules of plts).
- vWF & FVIII??

- *von Willebrand disease is the most common inherited disorder of primary hemostasis.*
- The prevalence of vWD mutations may be as high as 1 to 2% of the population.
- **However,** the prevalence of *clinically evident vWD is much lower.*

Classification of vWD

- **Type 1:**
- This is by far the most common (≥70% of cases of clinical vWD).
- There is a decrease in the concentration of vWF in the plasma (quantitative defect).
- **Type 2:**
- There is a qualitative defect in vWF.

- **Type 3 (rare):**
- In this type, there is a total or near-total absence of vWF in the plasma.
- The patients have markedly decreased factor VIII levels (???).

- Classification of vWD:
  - Type 1 Partial quantitative deficiency
  - Type 2 Qualitative deficiency
  - Type 3 Total quantitative deficiency

Clinical Features

- Most cases present with the typical picture of a primary hemostatic defect:
- - mucocutaneous bleeding (epistaxis, bleeding gums),
- - easy bruising, and …
- - immediate bleeding from cuts, incisions, and dental extractions.
- Most patients have a mild to moderate bleeding tendency.

Laboratory Diagnosis

- The most important diagnostic tests for vWD are:
- - the *bleeding time*,
- - *quantitative assay of vWF concentration*,
- - *platelet aggregation*, and …
- - *agarose gel electrophoresis for the high-molecular*-weight multimers of *vWF*.
- Plt counts ??
- PT ?? PTT ??

- *vWF* levels vary over time in a given individual; - they are increased with estrogen therapy,
- - during stress,
- - with liver disease and …
- - during pregnancy (as does the level of the FVIII).

- If the vWD is strongly suspected but the assay shows a normal level, the test should be repeated at another time.
- If the patient is a pregnant woman, the test is repeated several weeks after delivery.

- vWF levels vary with the blood group; they are lowest in patients with blood group O, higher in patients with groups A, B, or AB.
- Care should be taken when diagnosing vWD in people with blood group O.

Thank You