Von Willebrand Disease, Type 3

Alternative Names
VWD3
Von Willebrand Disease, Type III
VWD, Type 3

Record Category
Disease phenotype

WHO-ICD
Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism > Coagulation defects, purpura and other haemorrhagic conditions

Incidence per 100,000 Live Births
0-1

OMIM Number
277480

Mode of Inheritance
Autosomal recessive

Gene Map Locus
12p13.31

Description
Type 3 Von Willebrand disease (type 3 VWD) is the most severe and rare form of VWD. It is characterized by a bleeding disorder associated with a total lack of Von Willebrand factor (VWF) in the plasma as well as in cellular compartments. This type causes a profound deficiency of factor VIII (FVIII) level in plasma. Annual incidence varies between countries. However, it revolves around 1/500,000 in countries favoring consanguineous marriages. Usually, the age of onset occurs in the neonatal period or in infancy. The bleeding anomalies are characterized by mucocutaneous hemorrhage and prolonged bleeding after surgical interventions. Affected individuals may have hematomas and hemarthrosis due to the severe FVIII deficiency.

Diagnosis can be easily made by the absence of detectable VWF levels, accompanied by the deficiency in FVIII. Patients with type 3 VWD do not respond to desmopressin therapy. Therefore, substitution therapy with purified human VWF in conjunction with FVIII is the best preventative option for affected patients.

Molecular Genetics
VWD is caused by mutations in the VWF gene. Mutations in this gene lead to intracellular retention or rapid clearance of VWF from the circulation. The level of VWF in blood group O is 25-35% lower than in non-O blood groups. Therefore, individuals with blood group O are at a greater risk for developing VWD.

Epidemiology in the Arab World

Saudi Arabia
Egypt
See Saudi Arabia > El-Bostany et al., 2008

Saudi Arabia

El-Bostany et al. (2008) recruited 43 children and adolescents from Saudi Arabia and Egypt with various bleeding disorders to assess the prevalence of inherited bleeding disorders (IBD). Their ages ranged from 1-18 years. They also included 15 matched controls. Extensive laboratory work-ups were made including complete blood count, coagulation studies and platelets functional analyses. A total of 12 patients were found to meet the criteria of VWD. Multimeric analysis, used to determine the subtype of VWD, found that four of these patients had VWD type III. The authors concluded that VWB was common in Egypt and Saudi Arabia and therefore, hematological screening ought to be considered routinely in children with family history of bruising or bleeding disorders.

References
El-Bostany EA, Omer N, Salama EE, El-Ghoroury EA, Al-Jaouni SK. The spectrum of inherited bleeding disorders in pediatrics. Blood Coagul Fibrinolysis. 2008; 19(8):771-5. PMID: 19002043.

Related CTGA Records

External Links

http://www.hog.org/handbook/article/1/7/types-of-von-willebrand-disease
http://www.nhs.uk/conditions/von-willebrand-disease/Pages/Introduction.aspx
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=166096
https://www.nhlbi.nih.gov/health/health-topics/topics/vwd

Contributors

Ameera Balobaid: 24.7.2016