

Thursday, October 18, 2012
11:00 am
in LSC3

Life Sciences Centre
2350 Health Sciences Mall



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“Modifiers of the von Willebrand factor phenotype: join the queue”

von Willebrand disease is the most prevalent inherited bleeding disease in humans. The genetic pathology responsible for the bleeding phenotype is highly variable, with forms of the disease resulting from mutations showing recessive, dominant, co-dominant and complex inheritance patterns. While the causative mutations resulting in the dominant and recessive type 2 and 3 forms of VWD are now well characterized, the pathogenesis of the common disease variant, type 1 VWD, remains poorly defined.

Type 1 VWD is manifest by a mild/moderate quantitative reduction of functionally normal VWF. Pedigree analysis and twin studies have shown that up to 70% of the variance in plasma VWF levels is genetically determined. Further analysis has indicated that the ABO blood group genotype represents 30% of this genetic influence.

Initial genotyping studies in ~500 type 1 VWD index cases has identified putative mutations in the VWF gene in ~65% of cases, although the primary pathogenic nature of these variants remains unclear in many instances. From these studies, and prior linkage analysis in which the type 1 phenotype segregates with the VWF gene in only ~50% of kindred, there is growing evidence of locus heterogeneity and an impact from several (?many) other genetic loci in determining plasma levels of VWF.

In this seminar, the evidence supporting the presence of an increasing number of genetic modifiers of the VWF phenotype will be presented. More specifically, new data relating to a novel clearance mechanism for VWF will be reviewed.

This Seminar is sponsored by:



Host: Dr. Ed Pryzdial, Clinical Professor Pathology and Laboratory Medicine & Centre for Blood Research



Refreshments will be served 10 minutes before the seminar
Seminar information: 604 822 7407

