



The PT-VWD Registry

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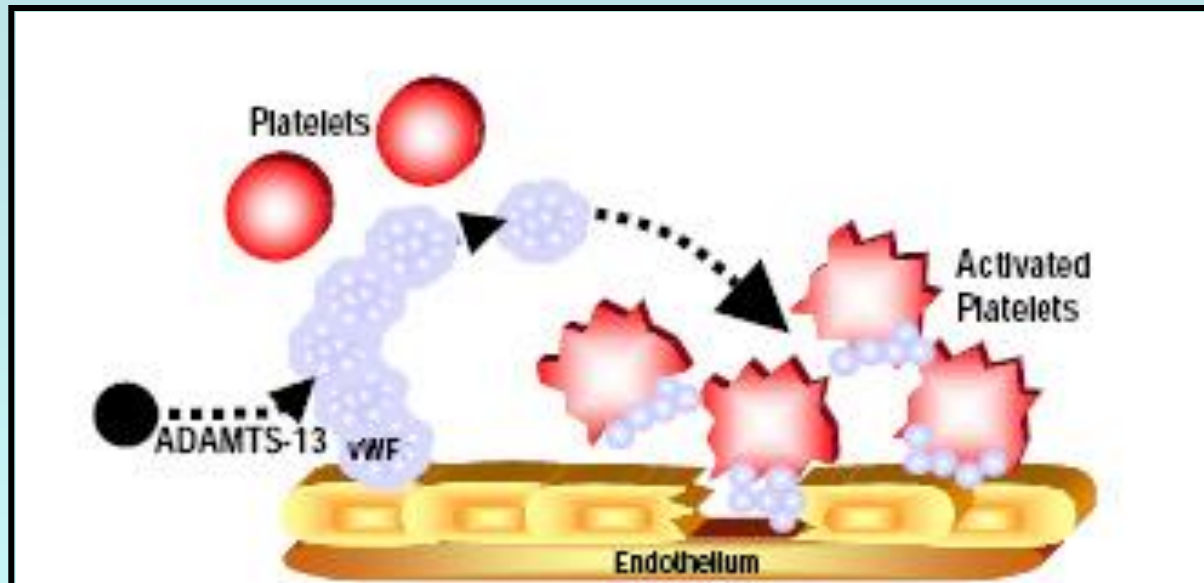
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Collaborative BScN program

Adjunct Assistant Professor

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Platelet-Type Von Willebrand Disease (PT-VWD)



Rare bleeding disorder

Platelet not VWF defect

Mild bleeding

Low plasma VWF + Low platelet count

PT-VWD

A Diagnostic Challenge

PT-VWD & Type 2B VWD

“The non identical twins”

□ Phenotypic discrimination:

- RIPA mixing studies
- Cryoprecipitate challenge
- Simplified RIPA-mix

□ Genotypic discrimination

- DNA sequence analysis:
 - Exon 28 *VWF* gene
 - Platelet *GP1BA* gene



Fresh blood sample
Issues: application/ interpretation



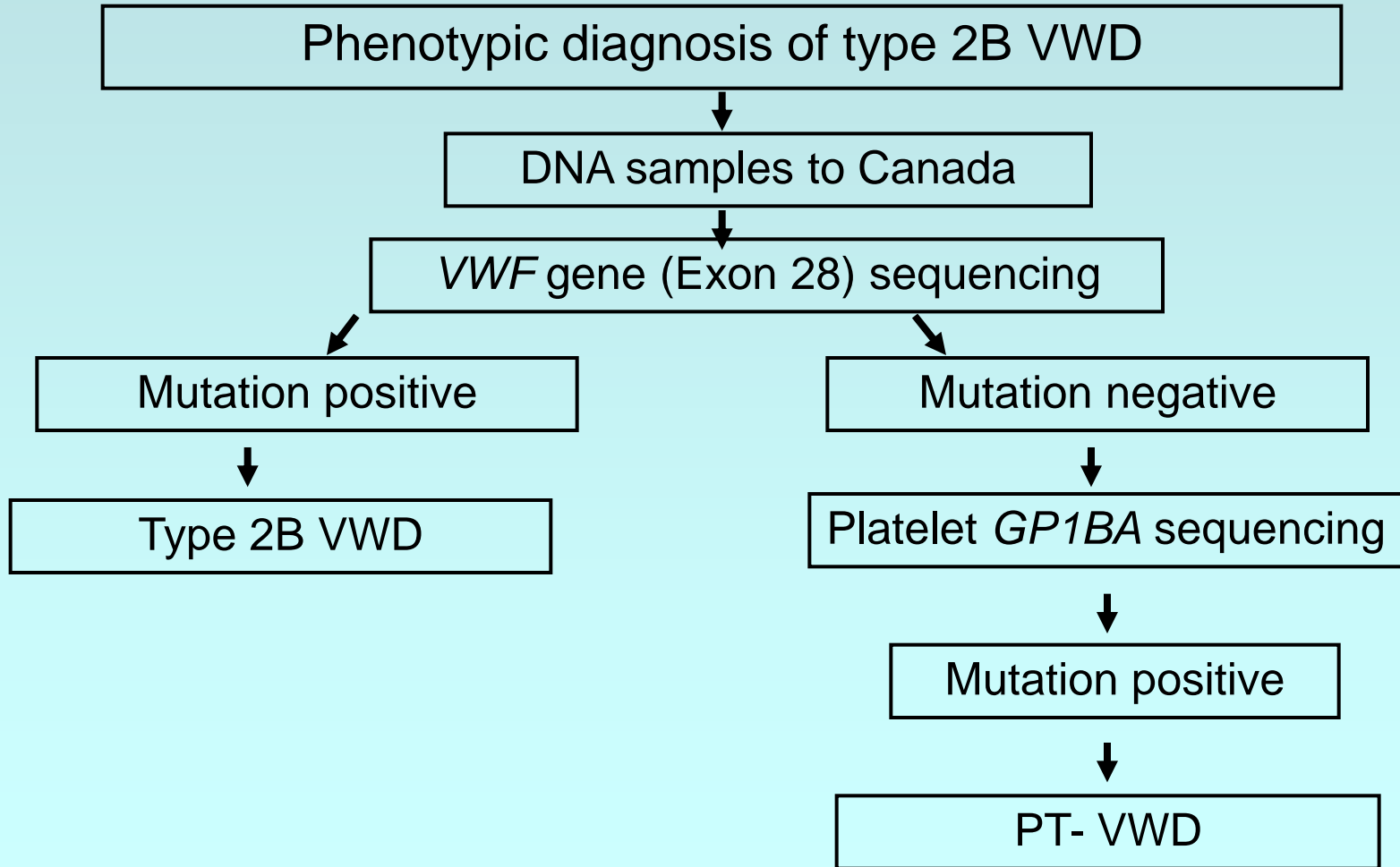
DNA sample
Conclusive evidence

PT-VWD Canadian Project

- Questions to be Addressed:
 - Is PT-VWD truly rare or under diagnosed ?
 - How many cases are being misdiagnosed among type 2B VWD cases?
- Sample source:
 - Canadian
 - International



PT-VWD Canadian Project





Platelet Type von Willebrand Disease

PT-VWD.org



Registry/Database

Reported Cases

[Click here to view table of confirmed cases](#)

Last updated August 22, 2008

About the Registry

Dears

The overall aim of the PT-VWD database/ registry project is to determine the frequency of this rare bleeding disorder in the world and to collect data about the molecular pathology, phenotype/genotype correlations as well as treatment. Therefore, we invite you to complete a form that includes the main information in order to allow us to learn about your experience with PT-VWD patients and if you would like to participate in the Canadian PT-VWD project.

The objectives of the PT-VWD Registry are:

- To document patients with Platelet- type VWD worldwide.
- To characterize genetic abnormalities responsible for this phenotype

Home

About PT-VWD

- What is Platelet type von Willebrand disease?
- Clinical Features
- Laboratory Findings
- 2B or not 2B?
- Treatment of Platelet type VWD
- Molecular Characterization of Platelet typeVWD
- Platelet GPIBA gene
- Platelet GPIB/IX/V complex
- Platelet GPIBA polymorphisms
- Nomenclature

Registry / Database

Canadian Project

Registration

Links

- > AHDCDC
- > BSS Database
- > CHS
- > ISTH website
- > Scientific subcommittee on von Willberand factor
- > Sheffield database
- > Queen's University Molecular Hemostasis Laboratory
- > American Society of Hematology

pt-vwd.org



PT-VWD Registry

- 18 families (44) cases: 31 F 13 M
- 10 families: G 233 V
- 4 families: G 233 S
- 3 families : M 239 V
- 1 family: 27 bp deletion

Data from the PT-VWD Project

Total cases : 86

Cases analysed in Canada (49):

37 cases from across Canada

8 cases from Brazil

3 cases from Switzerland

1 case from Boston, USA

Cases analysed outside Canada (37):

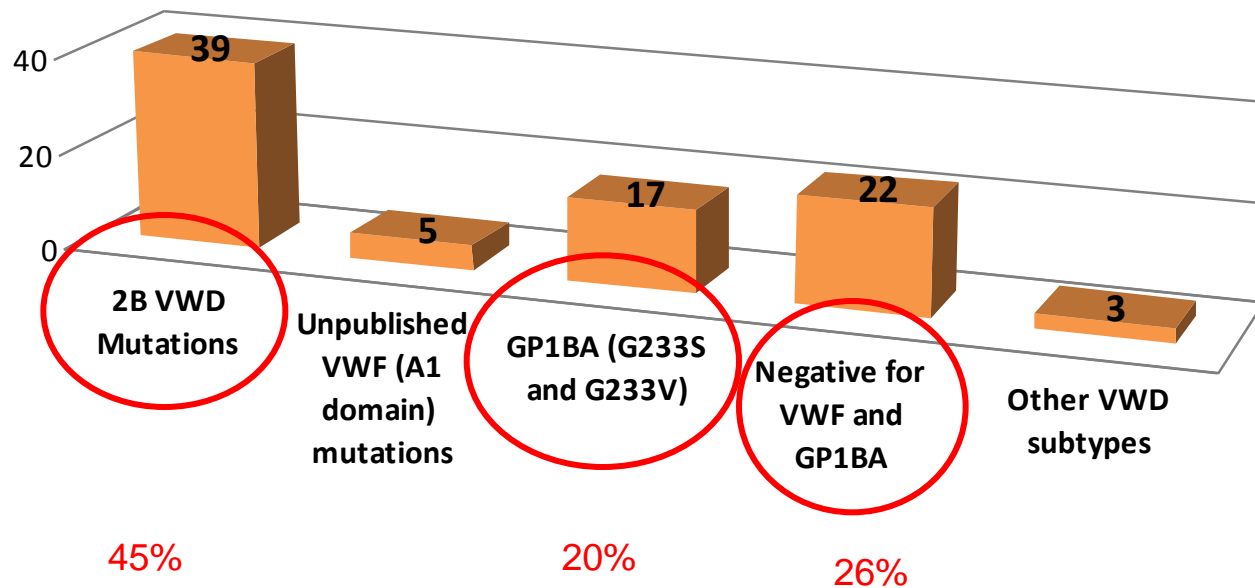
16 cases from Australia

15 cases from UK

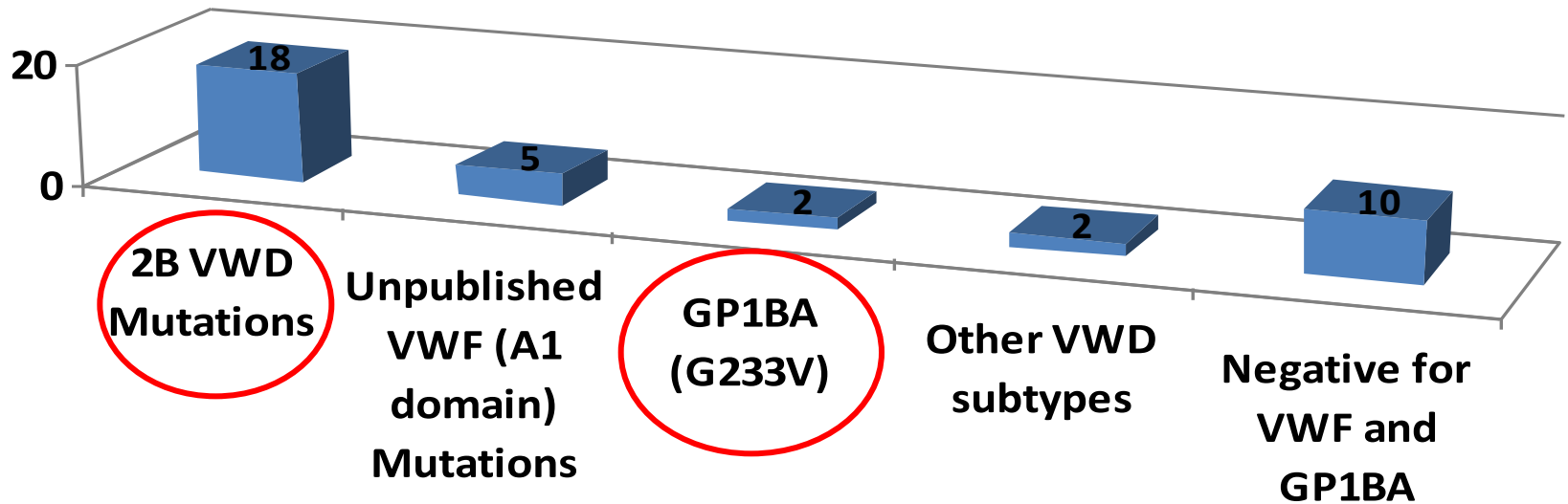
6 cases from Argentina

Data from the PT-VWD Project

**Genetic mutation frequency (all regions)
in 86 cases with phenotypic diagnosis as
2B VWD**



Genetic mutations in Canadian cases with phenotypic diagnosis as 2B VWD: N = 37



Canadian Haemophilia Registry and Rare Bleeding Disorders Registry

http://www.fhs.mcmaster.ca/chr/pdf/09/CHRVWD090413.pdf - Windows Internet Explorer

http://www.fhs.mcmaster.ca/chr/pdf/09/CHRVWD090413.pdf

File Edit Go To Favorites Help

http://www.fhs.mcmaster.ca/chr/pdf/09/CHRV...

2 / 2 100% Sign Find

CANADIAN HEMOPHILIA REGISTRY

Von Willebrand Disease, All Ages, April 13, 2009

STATS CANADA - AGE GROUPS

Report Date: Monday, April 13, 2009

Selection Prov: * Inhib.: * HIV: * HCV: *
 Centre: All Centers Gender: * PtStatus: Active or Pending Level: *

Category	Gender	Count	(0-4)	(5-9)	(10-14)	(15-24)	(25-34)	(35-44)	(45-54)	(55-64)	(65-74)	(75-84)	(85+)
Von Willebrand Disease													
VWD 2B	F	39	0	2	3	8	8	4	7	3	3	1	0
	M	41	1	1	3	7	7	7	6	5	2	1	1
	U	1	0	0	0	0	0	1	0	0	0	0	0
	subtotal:	81	1	3	6	15	15	12	13	8	5	2	1
VWD 2M	F	15	0	2	2	1	3	3	2	0	0	1	1
	M	16	1	5	2	6	0	1	0	0	1	0	0
	subtotal:	31	1	7	4	7	3	4	2	0	1	1	1
VWD 2N	F	14	0	0	1	3	1	1	3	4	1	0	0
	M	13	1	0	1	2	4	0	1	1	1	1	1
	subtotal:	27	1	0	2	5	5	1	4	5	2	1	1
VWD 3	F	44	1	6	0	8	6	7	8	2	2	4	0
	M	33	4	2	2	9	2	4	5	1	2	2	0
	U	12	0	0	0	0	1	2	2	4	2	1	0
	subtotal:	89	5	8	2	17	9	13	15	7	6	7	0
VWD unk	F	1	0	0	0	0	0	1	0	0	0	0	0
	M	4	0	1	0	2	1	0	0	0	0	0	0

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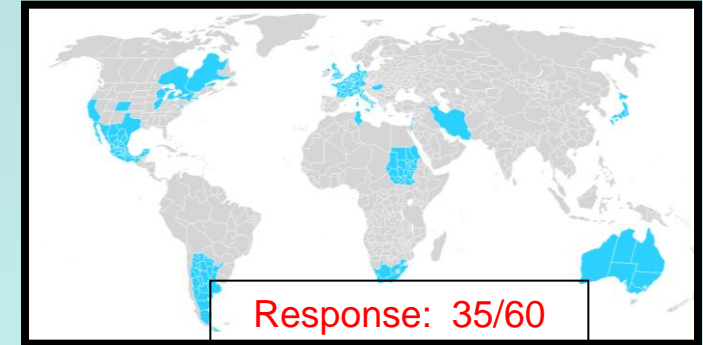
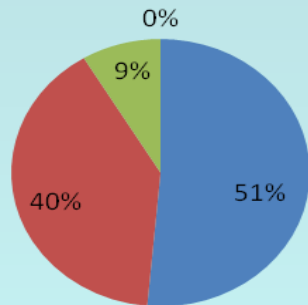
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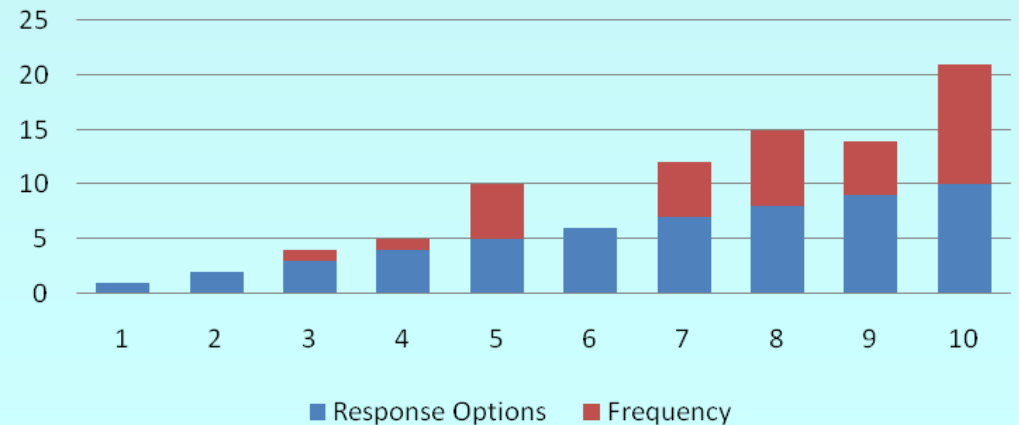
PT-VWD World Wide Survey

Witnessed or Diagnosed Cases of PT-VWD: N = 35

■ 0 ■ 1-5 ■ 5-10 ■ 10+



Importance of discriminating between 2b VWD and PT-VWD: N=35



Conclusion

- PT-VWD is misdiagnosed among 2B VWD
- PT-VWD should be included among RBD-Platelet function defects
- Genotyping is available and relatively easy
- Challenge remains to estimate the true incidence of PT-VWD:
 - Rarity of the disease
 - Insufficient collaboration with samples/ data

Acknowledgments



David Lillicrap

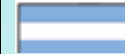
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Canadian Hemophilia Society
Help Stop the Bleeding



Alexander Hamilton
Year 3 Nursing Student



leisure & tourism

Southampton University 2003

PhD defence



Our Research Group
Pathology & Molecular medicine , Queen's
Farwell 2005



Canadian Hemophilia Society Research summit Feb 2007



My Canadian start and one of the achievements along the road



FAMILY



Canada Or World Wide Participation DNA Samples or Data

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