

# AHCDC Rare Inherited Bleeding Disorders Subcommittee Annual Report – March 25, 2010

## Subcommittee Members

Drs. John Wu and Margaret Rand (Co-Chairs), Victor Blanchette, Stephanie Cloutier, Sara Israels, Walter Kahr, Man-Chiu Poon, Linda Vickars, Irwin Walker and Rochelle Winikoff, and Ms. Caroline Tra (Nurse Representative)

## Data Manager

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## Background and Mandate

Inherited bleeding disorders other than Factor VIII and Factor IX deficiencies and von Willebrand disease can cause major health impairment and even death. Because most of these abnormalities, that encompass rare coagulation factor deficiencies and platelet function disorders, are rare, less is known about their natural history and optimal management. This Subcommittee was established with the **mandate** to:

1. better understand these disease entities
2. enhance delivery of comprehensive care
3. establish management guidelines
4. provide education to patients and health care providers
5. promote safety of product supply
6. further basic and clinical research directed at these disorders.

## 2009-2010

The Rare Inherited Bleeding Disorders Registry (RIBDR) was established in 2003 following the success of a pilot survey in 2002, to allow tracking of patients in Canada with rare inherited bleeding disorders, i.e. inherited bleeding disorders other than Factor VIII and Factor IX deficiencies and von Willebrand disease. As of March 2010, there are 1362 patients in the RIBDR, 752 with coagulation factor deficiencies and 610 with platelet function disorders. After our annual request from the Centres for validation of existing data and registration of newly diagnosed patients, updated 2010 numbers will be available on the Canadian Hemophilia Registry (CHR) website ([www.fhs.mcmaster.ca/chr/](http://www.fhs.mcmaster.ca/chr/); also accessible via the AHCDC website). The registry forms for both coagulation factor deficiencies and platelet function disorders, also available on the CHR website, will be updated as well.

Subcommittee material is posted on the AHCDC website under the Research sidebar “Rare Inh. Bleeding Disorders”: diagnostic criteria (updated in 2010) and a diagnostic algorithm for platelet function disorders, developed by Drs. Israels, Kahr and Rand; and English and French versions of the second edition of “Disorders of platelet function. An information booklet for patients, families and health care providers”, co-authored by Drs. Israels, Poon and Rand on behalf of the Canadian Pediatric Thrombosis and Hemostasis Network.

The diagnostic algorithm is being disseminated in the community: by Dr. Israels at a Laboratory Investigation of Neutrophil/Platelet Disorders Workshop at the Pediatric Hematology Update in Toronto in October, 2009, and in a presentation entitled “Evaluation of Patients with Suspected Platelet Dysfunction” at a Symposium on Platelets at the upcoming American Society of Pediatric Hematology/Oncology in Montreal in April, 2010; and by Dr. Rand in a presentation on “Demystifying Platelet Function Testing” at the Annual National Education Meeting of the Canadian Association of Nurses in Hemophilia Care in Montreal in November, 2009.

A guide for the use of the algorithm, along with further information on, for example, bleeding scores, blood films, platelet aggregation studies, etc., is being developed. As a start, details on standardized, quantitative mucocutaneous bleeding scores, both adult<sup>1</sup> and pediatric<sup>2,3</sup>, are being posted on the website as an adjunct to the algorithm; these include the bleeding questionnaires themselves and guides to their usage.

To continue to meet the needs of people living with a rare inherited bleeding disorder, the Canadian Hemophilia Society will be offering a Regional Workshop for Western Canada in November, 2010, with Dr. Wu on the Organizing Committee; this builds on the very successful National Workshop on “Living with a Rare Bleeding Disorder” that was held in November, 2008 in Montreal.

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<sup>1</sup>Bowman M, Mundell G, Grabell J, Hopman WM, Rapson D, Lillicrap D, James P. Generation and validation of the Condensed MCMDM-1VWD Bleeding Questionnaire for von Willebrand disease. *J Thromb Haemost* 2008; **6**: 2062-6.

<sup>2</sup>Bowman M, Riddel J, Rand ML, Tosetto A, Silva M, James PD. Evaluation of the diagnostic utility for von Willebrand disease of a pediatric bleeding questionnaire. *J Thromb Haemost* 2009; **7**: 1418-21.

<sup>3</sup>Biss TT, Blanchette VS, Clark DS, Wakefield CD, James PD, Rand ML. Use of a quantitative Pediatric Bleeding Questionnaire to assess mucocutaneous bleeding symptoms in children with a platelet function disorder. *J Thromb Haemost* 2010. Epub ahead of print March 17, doi: 10.1111/j.1538-7836.2010.03846.x