AHCDC Rare Inherited Bleeding Disorders Committee
Annual Report – March, 2015

Rare Inherited Bleeding Disorders (RIBD) Committee Members
Drs. John Wu and Margaret Rand (Co-Chairs), Victor Blanchette, Stephanie Cloutier, Sara Israels, Walter Kahr, Man-Chiu Poon, MacGregor Steele, Irwin Walker and Rochelle Winikoff (Nurse Representative: Vanessa Bouskill)

RIBD Data Manager
Ms. Taneya Hossain

RIBD Committee 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 Committee 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.

2014-2015

The Rare Inherited Bleeding Disorders Registry (RIBDR) was established in 2003 to help determine the numbers of patients with these disorders in Canada; patients with **deficiencies of the rare coagulation factors** *(fibrinogen, factor II, factor V, factor VII, factor X, factor XI, factor XIII, combined deficiencies of factors V and VIII, or combined deficiencies of the vitamin K-dependent coagulation factors [factors II, VII, IX and X])* and patients with **platelet function disorders** *(including deficiencies of membrane glycoproteins, disorders of storage granules, and familial thrombocytopenias)* are tracked.

This truly national registry continues to be a tremendous success thanks to the participation of all the Centres across Canada. As of March 2015, there are 1849 patients registered, 1014 with coagulation factor deficiencies and 835 with platelet function disorders (see Figure at end of report); this is a 4% increase from the 2014 numbers. In May, the updated 2015 numbers will be posted on the Canadian Hemophilia Registry (CHR) website (www.fhs.mcmaster.ca/chr/) that is also accessible via the AHCDC website. Our annual requests to the Centres for validation of existing data and registration of newly diagnosed patients will be sent out before the 2015 AHCDC AGM. These requests will be sent out by our new RIBD Data Manager, Taneya Hossain, who has taken over the position from Dewi Clark.

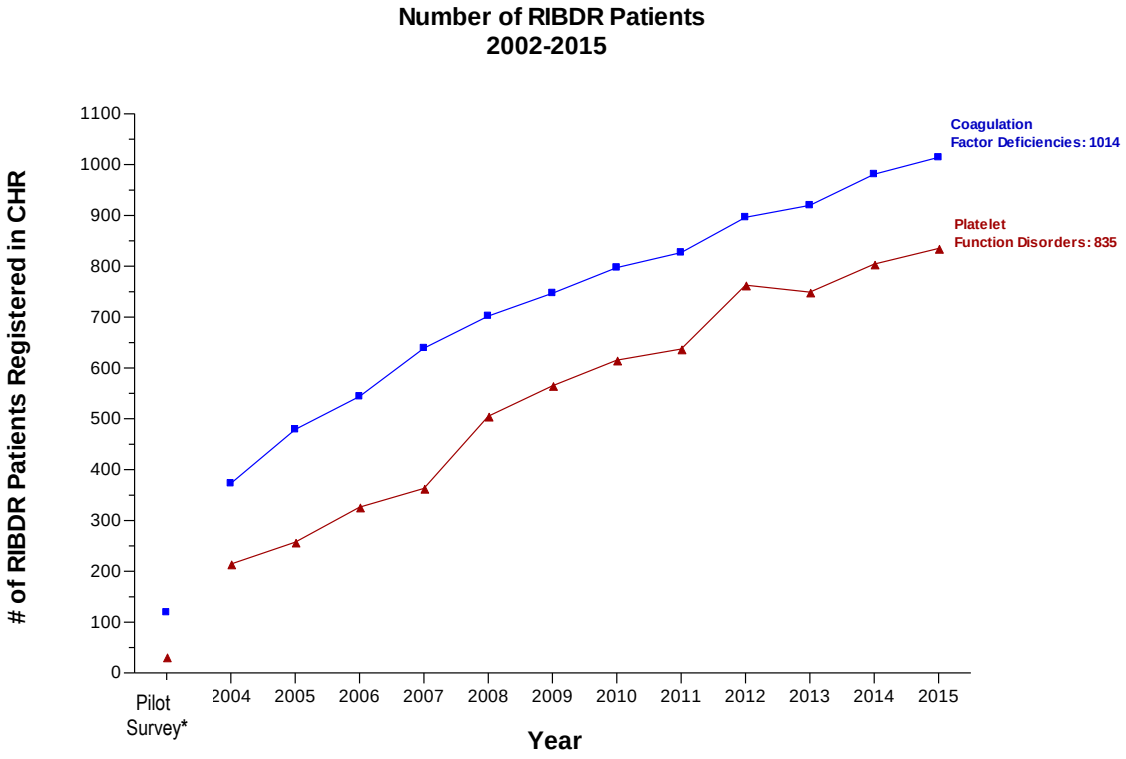
Committee material continues to be posted on the AHCDC website under the "Rare Inherited Bleeding Disorders" tab of the Research sidebar: diagnostic criteria, and our diagnostic algorithm for platelet function disorders and the review article in which it was published *(Israels SJ, et al. Pediatr Blood Cancer 2011;56:975-983).* An appendix to the algorithm with details of standardized, quantitative mucocutaneous bleeding scores for use in adults and children, specifically, the bleeding questionnaires, guides to their use, scoring keys and published validation papers, is available on the website (Appendix 1). Appendix 2 provides an updated listing of licensed diagnostic facilities for mutational analyses of platelet disorders. We will continue to develop further appendices on, for example, blood films, platelet function testing, etc, as adjuncts to the algorithm.

Also posted are the English 3rd edition and the newly completed French 3rd edition of *"Disorders of Platelet Function. An information booklet for patients, families and health care providers / Les Troubles de la Fonction Plaquettaire. Brochure d'information à l'intention des patient, de leur famille et des prestateurs de soins de santé"*, co-authored by Drs. Sara Israels, Man-Chiu Poon and Margaret Rand on behalf of the Canadian Pediatric Thrombosis and Hemostasis Network.

With the recent availability of a new product for factor XIII deficiency, the time is right for an in-depth investigation of the large national cohort of factor XIII deficient patients. Building on a Working Group survey of the 49 Canadian patients from 11 HTCs on their clinical manifestations, laboratory findings and treatment, a study proposal *"A Detailed Description and Prospective Investigation of the Canadian Cohort of Congenital FXIII-Deficient Patients"* to gain detailed insight into our unique national cohort was developed by Dr. John Wu as sponsor on behalf of the AHCDC, and has been awarded funding.

With the licensing of a fibrinogen concentrate and the post-licensure requirement, a retrospective Canadian multicenter study of the safety and efficacy of the concentrate for routine prophylaxis, treatment of bleeding and surgery in a- and hypofibrinogenemic patients is being initiated through individual HTCs. As in the past, our Canadian HTCs are uniquely suited for this endeavor which has the potential to generate exciting new knowledge in this area.

Finally, a proposal, with Dr. Margaret Rand as investigator and Drs. Shannon Jackson, Stephanie Cloutier, Paula James and Man-Chiu Poon as co-investigators, to investigate platelet function in patients with von Willebrand disease type 2B, including our unique Canadian 'Montreal Platelet Syndrome' patients, was submitted and has been awarded funding.



*Factor VII and XIII deficient patients and patients with Glanzmann thrombasthenia only