NAME OF PROJECT:

Prospective evaluation of bleeding phenotype in PT-VWD to support evidence based diagnosis and management

Subcommittee: Platelet Physiology

- Person responsible (Chair / Principal Investigator):
  - Paolo Gresele/ Maha Othman

- Aim / Mandate of the project:
  - Background:
    - Only 53 patients have been reported with PT-VWD worldwide. Most of these patients; included in the PT-VWD registry pt-vwd.org; are currently inaccessible with limited follow up. Information about the disease is still lacking with respect to: bleeding phenotype, severity of bleeding symptoms, phenotype-genotype correlations, standardized platelet aggregation based testing and the need for routine DNA analysis for diagnosis. PT-VWD remains under and sometime misdiagnosed. Correct identification is critical to the treatment decision and life-threatening bleeding can occur if not diagnosed/treated appropriately, particularly at times of hemostatic challenges such as surgery, pregnancy and childbirth. Moreover, there is evidence to suggest that abnormalities beyond the bleeding condition may be associated with the abnormally hyper-responsive platelet GPIb alpha. Over the years it has proven extremely difficult to capture the required information about these patients and worldwide cooperation is critical to support evidence-based diagnosis and management.
  - Objectives:
    - To gather detailed information about the clinical and laboratory phenotypes of patients with PT-VWD
    - To evaluate the use of appropriate laboratory methods for diagnosis including DNA analysis.
    - To gather detailed information about the current treatment practices including challenges and complications of platelet transfusion.
    - To investigate systematically the bleeding pattern during pregnancy and the occurrence of pregnancy associated complications beyond haemostasis.
    - To generate evidence to support a clear diagnostic plan as well as treatment guidelines.

- Methodology (in very brief, not more than 1 paragraph):
  - A structured online survey will be designed to capture information about diagnosis and treatment of PT-VWD patients
- Presentation of data at ISTH SCC meetings
- Publication at JTH
- List of questions are attached

- Inclusion / recruitment criteria (if applicable):
  - The survey will be posted on ISTH website and will be sent to all physicians and specialists who are potentially dealing with bleeding/platelet disorders

- Year of starting:
  - Sep 2014

- Annual report of project:
  - June 2015/2016

- Year of completion (expected):
  - Sep 2016