NAME OF PROJECT: Diagnosis of inherited platelet disorders on a blood smear
Subcommittee: Platelet Physiology

- Person responsible (Chair / Principal Investigator): Shinji Kunishima
- Design: Questionnaire survey and inter-laboratory validation exercise
- Aim/Objective/Rationale (Needs assessment / Reason):

  Inherited platelet disorders (IPDs) are rare but more frequent than previously thought. Recent investigations have identified many causative genes for these disorders, but clinical diagnosis is still challenging. This is because diagnosis of IPDs often requires assays that are available only in a few specialized centres and/or significant amounts of freshly collected blood samples. For patients who live in emerging countries, there is very limited access to the needed diagnostic facilities. However, many IPDs, i.e., thrombocytopenias and platelet function disorders, are associated with defects of platelet components (such as adhesive receptors and cytoskeletal proteins) that can be detected by immunofluorescence analysis of the blood smear. Blood smears can be shipped even over large distances and immunofluorescence analysis could efficiently drive the diagnostic process to recognize different IPDs. Whenever needed, diagnosis can be finally confirmed by targeted molecular analysis.

- Objective:

  1) To get insight into the use and perceived utility of immunofluorescence studies
  2) To carry out an inter-laboratory validation exercise

A questionnaire survey will be performed to ask different laboratories whether they use immunofluorescence analysis for the diagnosis of IPDs and which forms of IPDs are diagnosed or excluded by this technique. Moreover, information on the evaluation of the potential utility of immunofluorescence for IPD diagnosis will be asked for. Then a inter-laboratory validation exercise will be organized on several established disorders, i.e., MYH9 disorders, Bernard-Soulier syndrome, and Glanzmann thrombasthenia, by sending fixed and unstained peripheral blood smears to participating laboratories to obtain sensitivity and specificity of peripheral blood smear analysis.

- Methodology (Data expected to collect, sample size and statistical analysis):

  Analysis of the literature will be performed to select IPDs for initial questionnaire survey list.
Sample preparation for the workshop will be performed at the main organizer laboratories.

- **Study population (Inclusion, exclusion, eligibility) (patient population; recruitment of participating institutions/physicians and subjects; minimum number needed; expected number):**

  The initial questionnaire survey is mailed to the members of SSC Platelet Physiology and Platelet Immunology and to external head of laboratories expert in platelet studies. The inter-laboratory validation exercise will be performed involving laboratories (at least 10), which participated in the initial survey.

- **Expected timeline:**
  
  - Project stage/set up: planning
  - Launch: ISTH Berlin meeting, 2017
  - Duration: 2 years
  - Finalization/analysis: 2019
  - Reporting: ISTH Melbourne meeting, 2019

- **Expected outcomes (ie. publications):**
  
  - Publication type (SSC Communication, Guidance document or original article):
    - SSC communication (survey)
    - Original article (inter-laboratory validation exercise)
    - Guidance

- **Description of project set/up and management, needed infrastructure and resources (summary):**

  Drs Paolo Gresele (Chairman) and Shinji Kunishima (Co-Chairman) lead this project. External experts, Drs Andreas Greinacher and Alessandro Pecci, will join this project.

- **Possible references:**


An opinion paper regarding this project is in preparation (not a Platelet Physiology SSC product).