Editorial

Progress in Hemostasis (Part 1): Improved Management of Inherited Platelet Disorders: Reality or Illusion?

Werner Streif[1]

[1] Department of Pediatrics 1, Medical University of Innsbruck (MUI), Innsbruck, Austria

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Platelets are key drivers of hemostasis. Low platelet counts, dysfunction in platelet adhesion, and aggregation lead to increased bleeding tendency. Inherited platelet disorders (IPDs) form a highly heterogeneous group of rare diseases with variable bleeding tendency. IPDs may be associated with other signs and symptoms often referred to as "syndromic." The underlying genetic defect may prone patients to develop hematopoietic diseases such as leukemia. Over the last decade, accumulating knowledge in genetics has led to the detection of many "new" platelet disorders. However, still many patients with a well-described platelet dysfunction remain undetected until severe bleeding occurs.

Genetic testing for IPDs has been proven as a game changer. With the introduction of high-throughput sequencing, a more accurate diagnosis of IPDs has become available. In this issue, **Gebetsberger et al.**[1] update the molecular-diagnostic approach to support clinicians in medical decision-making and to counsel affected patients and their families. This includes strategies how to handle unexpected findings such as predisposition to malignancies.

In the next issue of this journal, we will shed special light on light transmission aggregometry that has been accepted as the gold standard test for diagnosing platelet function defects for more than 50 years. We will undergo the difficult task to appraise critically its value in the era of point-of-care platelet testing in the operating room and refined molecular genetic testing. Recently published guidelines and personal experience with different methods will deepen presented up-to-date knowledge.

Thrombocytopenia-absent radius (TAR) syndrome, is a very rare form of hereditary thrombocytopenia associated with a bilateral radial aplasia due to microdeletion on chromosome 1 and a single nucleotide polymorphism. The genotypic and phenotypic description of TAR by **Strauss and Schulze et al.**[2] more than 20 years ago essentially improved diagnosis and therapy of TAR syndrome, with a defined genetic background leading to strengthened self-empowerment of these patients. Authors report in this issue on the current understanding of TAR syndrome and discuss related patients' issues resulting from networking between affected individuals and families.

Despite the relevant progress in the diagnosis of IPDs, prevention of excessive bleeding and controlling of bleeding episodes are still poorly standardized and widely depend on local experience. Minimal standards are summarized in guidelines published on the AWMF webpage (AWMF S2K guideline #086–004; www.awmf.org). **Bargehr et al.**[3] provide not only an update but complete recommendations and add specific counseling of typical clinical scenarios.

Thrombopoietin receptor agonists (TPO-RAs) are a promising approach to treat IPDs with thrombocytopenia. Successful treatment depends on the specific subtype of the disorder and the severity of thrombocytopenia. In the next issue of this journal, we will present carefully collected published information and personal experience with TPO-RAs. We will show how a sustained increase of platelet count coincides with less bleeding symptoms, leading to beneficial effects on health-related quality of life.

Already several years ago, knowledge gaps in the management of patients with IPDs have induced the initiation of a network of the GTH. To fill these gaps, this THROMKIDplus network has decided to establish a patient registry with associated biomaterial banking for children. In the next issue of this journal, Andres et al. will present this registry designed as a retrospective-prospective, multicenter observational study currently comprising 24 sites in Germany, Austria, and Switzerland. The launch is planned for the second half of 2023.

In summary, these articles provide current evidence for the real-world use of diagnostic tests and treatment options in IPDs. Presented articles include information from literature as well as personal communication within the Working Group THROMKIDplus during the meeting of the "Ständige Kommission Pädiatrie" of the GTH held in September 2022 in Igls near Innsbruck (Austria), expanding and updating information from published guidelines and reviews.

Address for correspondence
Werner Streif, MD, Department of Pediatrics 1, Medical University of Innsbruck (MUI), Anichstrasse 35, A – 6020 Innsbruck, Austria (e-mail: werner.streif@i-med.ac.at).

Georg Thieme Verlag KG, Rüdigerstraße 14, 70469 Stuttgart, Germany

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