Hemophilia in focus

World Hemophilia Day 2017

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Every year on April 17, World Hemophilia Day is marked in the calendar of health professionals around the world. This theme and action day was established in 1989 by the World Federation of Hemophilia (WFH) to increase awareness of hemophilia and other congenital bleeding disorders (1). The date of April 17 was chosen to honor Frank Schnabel, the founder of WFH, whose birthday falls on the same date.

World Hemophilia Day 2017

As also outlined by the International Society on Thrombosis and Haemostasis (ISTH), the awareness day focuses on bringing attention to the challenge that too many children, men and women of the bleeding disorder community are facing worldwide: lack of access to treatment and care (2). Around the world, there is an enormous discrepancy in the level of care available to patients with a bleeding disorder.

While some are diagnosed very young and take benefit of medical care throughout their life, millions of other patients with bleeding disorders have no access to treatment and care and still suffer debilitating pain, permanent joint damage, or death. World Hemophilia Day highlights the need that treatment of *all* patients with a bleeding disorder is required and that this goal can only be achieved with the help of the community at large. Ensuring that an individual with a bleeding disorder is diagnosed early and will be cared for adequately remains a shared responsibility. "*Treatment for All is the Vision of All*" has therefore been chosen as the slogan of this year's World Hemophilia Day.

In fact, World Hemophilia Day is a critical effort since, even in Europe, patients with rare diseases and their families do not receive the attention and support hat they have deserved. Therefore, concerted activities and specific information about diagnosis, treatment options and social care of rare diseases remain mandatory to improve the awareness in the general population and among politicians. For example, the German Parliament has published in its official calendar for 2017 many anniversaries of historic events together with memorial, action and theme days. However, with regard to our field, neither the World Hemophilia Day nor the World Thrombosis Day (October 13) is listed (3).

Hemophilia research and patient care

Recent advances, challenges, and controversies in hemophilia research and patient care have been subject of regular issues and supplemental editions of *Hämostaseologie – Progress in Haemostasis* throughout. For example, results of the SIPPET study (4), which assessed the incidence of FVIII inhibitors among hemophilia patients treated with either recombinant or plasma-derived FVIII products, or the first clinical trial with emicizumab (5), a FVIIIa mimetic, which binds to FIXa and FX, thereby exerting FVIII cofactor activity, were extensively reviewed and commented in this *Journal* (6). The annual supplement of *Hämostaseologie – Progress in Haemostasis* reports on highlights of the traditional Hamburg Hemophilia Symposium under the leadership of J. Oldenburg and K. Kurnik.

Living with hemophilia

On the occasion of this year's World Hemophilia Day, the current theme issue of *Hämostaseologie – Progress in Haemostasis* is dedicated to this inherited bleeding disorder and presents six selected articles of original work that were submitted to the Journal in the course of 2016. In their retrospective study, **H. Rehm** et al. evaluated fall rates, circumstances, consequences and possible fall risk factors in a cohort of middle-aged patients with severe hemophilia (7). About one third of the patients suffered from a fall incident in the past 12 months, half of them experienced more than one incident. These figures illustrate that fall incidents are a rather frequent phenomenon in patients with severe hemophilia. The authors therefore conclude that fall risk screening and fall prevention should be included into hemophilia care.

Hemophilic arthropathy is a complex joint disorder with mechanical, inflammatory and degenerative components. Recently, it was shown that interleukin (IL)-1β plays a pivotal role in blood-induced cartilage damage (8) and that inflammation is a key mechanism in the pathogenesis of hemophilic arthropathy (9). Presenting a case report on a 65-year-old patient with mild hemophilia and multiple recent hemarthroses, **R. Wharton** et al. discuss a specific role of subchondrial bleeding and subsequent osteochondral destruction in hemophiliac arthropathy

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(10). The authors also suggest a novel potential therapeutic target for preserving joint integrity. However, this approach will require future research prior to possible clinical use.

Prevention of bleeding complications including hemophilic arthropathy remains a major objective of hemophilia care. In this context, a health promotion program at the University Medical Center in Jena offered to young patients with hemophilia deserves particular attention. **J. Sondermann** et al. (11) report on the results of an observational study, in which the effects of health counseling, adjuvant exercise therapy and school sports were assessed prospectively in a small group of children and adolescents with hemophilia A or B. Various clinical data (including bleeding complications) and selected anthropometrical parameters were evaluated. First results of this program suggest that a combination of functional prevention diagnostics and individual health counseling can be a useful practical approach to improve health knowledge, self-competence and physical fitness among young patients with hemophilia (11).

Complementary to this study, a hemophilia patient survey performed by **H. Lechner** et al. (12) is of particular interest. The investigators used a standardized questionnaire to evaluate the unmet medical needs ("*wishes and worries*") from the patient perspective among 290 hemophiliacs. The analysis was limited by a rather low response rate of 51 %. Major findings included high satisfaction with medical care, chronic pain "due to hemophilia", and worries about health, safety and availability of factors products among hemophilia patients >44 years of age (12). Offers for psychosocial support in addition to medical care were required in all age groups.

Chronic hepatitis C virus (HCV) infection in patients with congenital bleeding disorders is still a current problem causing high morbidity and significant mortality among this population. **J. Wiegand** et al. (13) performed a multicenter study, in which interferon-free antiviral regimens were evaluated in 18 patients with chronic HCV mono-infection and inherited bleeding disorders. Using sofosbuvir/ledipasvir, sofosbuvir/ledipasvir/ribavirin, or paritaprevir/ombitasvir/dasabuvir, sustained virologic response was achieved in 17 of the 18 patients without severe therapy-related side effects, demonstrating that HCV-infected individuals with inherited bleeding disorders can be effectively and safely treated with interferon-free regimens (13).

Patient registries can be powerful instruments to support medical care and research. Here, **L. Teixeira** et al. (14) report on recently developed protocols and software tools used to support the Portuguese National Registry of Hemophilia and other Congenital Coagulopathies (PorR H&CC). Technologically, the registry is based on hemo@record, a web application, which runs on a remote and secure server. The registry provides information to support clinical decision making, to assess the efficacy and safety of hemotherapy and other treatment regimens, to evaluate cost-effectiveness, and to address specific questions related to clinical hemophilia research (14).

The current issue also contains several reports on hemophilia symposia being held during the GTH Congress 2017 and organized by pharmaceutical companies. These reports are published on the "blue pages" under the responsibility of Schattauer.

Continuing medical education (CME)

Along with the educational task of *Hämostaseologie – Progress in Haemostasis*, **C. Mannhalter** (15) provides a comprehensive article on new developments in molecular biological diagnostics. This seminal contribution covers the state-of-the-art techniques of genotyping and carefully reviews inherited disorders and predispostions, forensic analyses, and the clinical relevance of distinct polymorphisms in hemostatic abnormalities.

Needless to say that this CME article is highly recommended to future participants of the GTH Course of Clinical Hemostasis under the leadership of **B. Kemkes-Matthes**.

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