Event Report: EHC Round Table of Stakeholders on 'Women and Rare Bleeding Disorders'

About the event

On Tuesday 19 June 2018, the European Haemophilia Consortium (EHC) held its second Round Table of Stakeholders of the year on the topic of 'Women and Rare Bleeding Disorders.' The event was held at the European Parliament in Brussels and gathered patient representatives, health care professionals, members of the European Parliament (MEPs) and representatives from the pharmaceutical industry.

The event's agenda, list of speakers and presentations can be consulted online on the EHC website. Pictures from the event can be viewed on the EHC Facebook page.

The event was supported by MEP Jana Žitňanská (Slovakia / ECR) and was attended by MEPs Mrs Nessa Childers (Ireland / S&D), Dr Miroslav Mikolášik (Slovakia / EPP), Dr Cristian Bușoi (Romania / EPP) and Mrs Norica Nicolai (Romania / ALDE).

On Women and Rare Bleeding Disorders

Rare bleeding disorders (coagulopathies) is a generic term used to classify a group of rare and congenital disorders affecting the ability of an individual to produce all the necessary proteins and components for their blood to clot normally. These disorders can affect, for example, the production of coagulation factors (such as in the case of haemophilia A and B, von Willebrand Disease and other rarer bleeding disorders, such as factor (F) V, FVII and FXIII deficiencies) and platelets.

Traditionally, rare bleeding disorders have been more strongly associated with affecting males than females. This is primarily due to the fact that haemophilia, the most known of the bleeding disorders, impacts men more visibly than women. However, women are also affected as they also carry the faulty gene and can have low levels of coagulation factors resulting in bleeds. In addition, these disorders are rare and, as such, people affected by them experience issues associated with many other rare conditions, such as lack of knowledge amongst the general population and non-specialist health care professionals, difficulty in diagnosis and lack of adequate care and treatment.

Through this event, the EHC wanted to acknowledge and stress that women are also affected by rare bleeding disorders and highlight the physical and mental barriers they face. In addition, the EHC wished to start a conversation to identify actions to raise awareness and provide better support to these women. In fact, this is the focus of the newly established EHC Committee on Women and Bleeding Disorders, chaired by EHC Steering Committee Member Ms Naja Skouw-Rasmussen. Several members of this committee attended and spoke at the Round Table.

Findings and discussions

On women and rare bleeding disorders in Europe

As stated above, there is very little awareness amongst the general public and non-specialist health care professionals about the fact that rare bleeding disorders affect women. This is also reflected in data collection. For instance, it is estimated that for every recorded male with haemophilia there should be at least 1.5 actual somatic female carriers, 0.5 of whom is a carrier with low levels of coagulation factors. In

practice, this would mean that for every 100 males with haemophilia there are 50 women with low levels of coagulation factors. Yet, this is not at all reflected in existing haemophilia registries where women make up two to 10 per cent of the total recorded population. This lack of data results in a lack of understanding of how severely these disorders impact women and also prevents further research into developing adequate treatment protocols.

In 2017, the EHC carried out a survey to better understand the impact of rare bleeding disorders on women. The surveys – there were, in fact, three surveys aimed at women affected by bleeding disorders, clinicians and EHC National Member Organisations (NMOs), i.e. patient organisations – were widely circulated. The survey aimed at women affected by rare bleeding disorders proved very popular, with over 700 responses. The results showed that disorders that are rarer and less well cared for (in terms of treatment available), such as platelet disorders and rare factor deficiencies (for e.g. FV, FVII, FXIII), as well as women with unidentified disorders, are more affected on all aspects of their day-to-day life. In addition, lack of, or difficulty in diagnosis, is strongly correlated with mental health problems. The impact on reproductive life was also strongly highlighted by respondents, with 40 to 70 per cent of respondents affected by various disorders (e.g. haemophilia, von Willebrand Disease, platelet disorders and rare bleeding disorders) saying that their disorder impacted their ability to have children. Furthermore, the survey collected concrete examples on how harrowing some of the symptoms experienced by women with bleeding disorders can be. The most commonly reported symptom is heavy menstrual bleeding, which severely impacts day-to-day life and prevents regular activities, such as work and study, and also has repercussions on physical and mental health. Other impactful symptoms include nose bleeds and bruising. The survey found that women with better quality of life outcomes were those with a proper diagnosis and an adequate management plan providing both treatment and hormonal contraception to manage heavy menstrual bleeds. On this note, the EHC survey showed that in most cases, women would eventually be referred to a haemophilia treatment centre for proper diagnosis and multi-disciplinary care, but this process is often lengthy and ways should be found to speed it up.

The issue of **gender inequality** in the health care system was widely discussed. It was highlighted that despite the first recorded girl dying from a menstrual bleed in the 1920s, there is still little knowledge amongst medical professionals about the severity of these disorders and how negatively they impact women's quality of life. For example, it was highlighted that health care professionals are often reluctant to accept that haemophilia carriers have serious bleeding episodes. Statistics show that women arriving at an emergency department usually wait 30 minutes longer than men when they have abdominal cramps, because this symptom is considered 'normal' for them. Also, some health care professionals do not wish to talk about intimate issues related to bleeding episodes, such as the menstrual blood flow or the number of sanitary protection products used. In addition, pain management is often inadequate, again because the symptom (i.e. menstrual pain) is considered normal in women. This is particularly negative as it leads to lack of adequate treatment even when the treatment is available for male counterparts. Gender inequality also exists when discussing the terminology used to describe the same condition affecting women and men. For example, a woman with 25 per cent of coagulation factor will be called a carrier while a male will be called a person with haemophilia. The terminology downplays the severity of the condition and the need for the female to be offered medical support. In addition, it was also discussed that women, and in particular young girls, are often not included in clinical trials. This has several effects: on the one hand, it reinforces the idea that medicinal products are aimed at males, but also contributes to the lack of data on the use of these products in women, who may need them during more critical times such as pregnancy, delivery and post-partum. Finally, it was noted that women often face problems with regard to reimbursement as their insurances do not acknowledge the existence of haemophilia in women.

The event discussions also focused on the issue of **stigma** associated with the disorder, which contributes to feelings of guilt and helplessness in affected women. This feeling will prevent women from talking about their condition, seeking help and support, as well as receiving a diagnosis and adequate medical care. This can result in some women not getting tested early on and only discovering they're impacted by a bleeding disorder during pregnancy. In fact, literature reviews suggest that up to 19 per cent of

haemophilia carriers find out about their disorder during pregnancy. This situation can lead to difficulties during pregnancy. Besides facing the usual pregnancy hurdles, mothers-to-be, will have to process information about how this condition will impact their own health and that of the child they are carrying. Furthermore, being a carrier of haemophilia, or having any other rare bleeding disorder, increases the risks of miscarriage and bleeding, such as an intracranial bleed for the baby during pregnancy and delivery. These situations of lack of information are often the result of families not wanting to disclose or discuss this information due to fear or shame. There is also an element of some families not wanting to go to the haemophilia treatment centres, as the place is associated with the loss of family members and similar sombre experiences. It is therefore a challenge to reach the patients even when information is available, as they may not wish to be informed for fear of stigma. Participants noted that many women affected by rare bleeding disorders felt very isolated and stressed the importance of having 'safe' places where they could share their story and receive accurate information about the management of their condition.

On medical issues

As noted above, the lack of data on clinical practices makes it difficult for health care professionals to care for these women, in particular during pregnancy. Speakers highlighted the importance of genetic testing and genetic counselling, which should be performed by a multidisciplinary team whenever women with rare bleeding disorders decide to have children. Women should be given information on the risks associated with pregnancy and delivery, both for them and for their child. As noted above, women with rare factor deficiencies and platelet disorders are at a higher risk of miscarriage, as well as spontaneous bleeding both in the mother and child. Therefore it is very important that these women are made aware of measures that can minimise the risk of bleeds. Genetic diagnosis during pregnancy can often be quite difficult to perform and it is usually invasive, which increases the risk of bleeds and a miscarriage, all of which increases stress in mothers-to-be. This is why it is very important that women are diagnosed before pregnancy so that they can make informed decisions with regard to child bearing.

In addition, it was noted that joint damage resulting from bleeds, which is something that is closely monitored in males with haemophilia, is virtually not monitored in females and therefore we lack data as to whether female patients are also affected by joint damage. Again, this is due to the fact that women are not perceived to be impacted as much by the bleeding disorder as males. This is something that also needs addressing.

Amongst the recommendations made by participants to ensure the improvement of women with rare bleeding disorders, it was noted that it was very important to ensure that young doctors join the field of haematology, which is often not perceived as an exciting field of work. The uptake of young doctors in the field was seen as vital to ensure adequate treatment and care for people affected by rare bleeding disorders.

On the impact of rare bleeding disorders on everyday life

The event featured two personal accounts from members of the EHC Committee on Women and Bleeding Disorders. These personal stories summed up much of the topics discussed above, in particular they stressed the personal struggle in coming to terms with living with a chronic and not well-recognised condition. Speakers also discussed that the important role of an informal carer for someone affected by a rare condition is still not well understood and often stigmatised. Finally, speakers noted the importance of a support system, which can be provided by patient organisations in terms of a safe space to meet peers and discuss common issues.

Conclusions

During the event we learned that women affected by rare bleeding disorders are still not recognised and that the stigma surrounding their condition prevents them from talking freely about their diagnosis. Participants agreed that in this regard there is a need for a European effort to raise awareness about these conditions, so that affected individuals can openly discuss their health status and seek better diagnosis, treatment and social acceptance.

It was also agreed that the lack of data on, and inclusion of women in clinical studies is a major shortcoming for providing adequate treatment and care and, in particular, to tailor treatment and care to the needs of each woman's life-stages. Participants agreed that more should be done to improve this.

Clinicians present at the event noted that health care professionals should join forces to ensure that there is more education about the impact of rare bleeding disorders in women and awareness is raised and more collaboration to ensure adequate in all of Europe.

The meeting closed with the impetus to have all stakeholders do much more in this area and to that end, the EHC announced that it will organise a European Conference solely dedicated to women and rare bleeding disorders in 2019, where discussions and concrete work will continue.