European Haemophilia Consortium (EHC)
Rue de la Loi 28
1040 Brussels, Belgium

Report on the Lived Experience with Factor X Deficiency:

Factor X Deficiency (FXD) is a rare bleeding disorder resulting in reduced plasma coagulation FX activity and potentially life-threatening symptoms. Despite challenges in patient representation at national and international levels, it is crucial to continue efforts to support this underserved and underrepresented group. As part of the ERIN work, we spoke to three international patients and caregivers with FXD to discuss their challenges and experiences.

1. Specific Challenges Faced by Patients and Families

FXD presents numerous challenges, significantly impacting patients and their families differently based on geographic location and healthcare access.

- **U.S. Patients:**
  - The parents have FX levels of approximately 50-56%.
  - Now aged 27 years, their son was diagnosed shortly after birth. Mother had no diagnosis and he suffered bleeding complications during birth. He then suffered major intracranial bleeding at five weeks old, leading to severe damage.
  - 2 years later, their daughter was born and diagnosed at birth, she received a port-a-cath at two days old. While diagnosis was prompt, she went on to experience complications with managing periods and faced a reluctance in her HTC to reassess her treatment plan.
  - Their Grandmother was subsequently diagnosed in her 60s due to bleeding caused by a lumpectomy.
- **Western European Patient:**
  - Male patient diagnosed after a tonsillectomy which resulted in being hospitalised for three weeks.
  - He suffered prolonged bleeding since childhood, frequent nosebleeds, painful bruising and inflammation of joints. Despite experiencing joint pain for years, which was initially dismissed as unrelated to FXD, with on-demand treatment he reported that the pain and stiffness in his joints improved.
  - He reports a significant impact on the quality of life growing up – being unable to participate in school sports due to high injury risk, fear and uncertainty at what was wrong.
- **Eastern European Patient:**
  - Mother suffered multiple miscarriages and prolonged severe bleeding but was undiagnosed so considered this as her 'normal'.
  - Her son was diagnosed after an intracranial haemorrhage at birth; he diagnosis was confirmed two weeks later.
  - Family have suffered from the limited information, awareness and support for FXD.

2. Navigating the Healthcare System: Learnings and Barriers

European Haemophilia Consortium (EHC)
Rue de la Loi 28
1040 Brussels, Belgium

Patients' experiences with the healthcare system vary widely based on location and healthcare infrastructure:

- **U.S. Patients:**
  - Early diagnosis was efficient, but paediatric treatment inconsistencies exist and continue to exist.
  - There are still delays in optimising treatment plans and this is leading to unnecessary bleeding and complications for patients.
  - There is a lack of joined-up thinking and treatment planning between haematologists and other departments in the hospital – pain management specialists, physiotherapy, gynaecological services etc.
- **Western European Patient:**
  - Diagnosis delayed until adulthood, highlighting a lack of education and information and missed opportunities for screening.
  - Patients are facing difficulty in getting clinicians to recognise joint pain as an FXD symptom.
  - There is limited access to disorder-specific support – nationally there is not an identity, availability of resources and education for FX patients.
- **Eastern European Patient:**
  - Families feel that they are faced with needing to advocate tirelessly and constantly educate and convince healthcare professionals about FXD.
  - There is a lack of local patient support; patient organisation outreach is present but lacks resources specific to the needs of this group.
  - A serious need for HCP education is essential to improving FXD care.

3. Advice for Others with FXD

- Patients need to understand that they should present with symptoms that are impacting their quality of life – despite the mantra 'it was normal for me'.
- Push to seek early diagnosis and proactive treatment.
- Patients need to continue to advocate if their treatment is not adequate or meeting their needs.
- Engage with patient communities, such as online support groups, for shared knowledge and experiences.
- Push for recognition of subclinical bleeding symptoms, including joint stiffness and unexplained, painful bruising.
- Work closely with Haemophilia Treatment Centres (HTCs) or specialists to ensure access to appropriate therapies.

4. Strengthening the Broader Patient Community

To improve patient outcomes and support networks, the broader community must:

- **Increase innovation and access to therapies** to provide better management options.

European Haemophilia Consortium (EHC)
Rue de la Loi 28
1040 Brussels, Belgium

- **Develop understandable educational resources** to ensure patients are well-informed about their condition and available treatments.
- **Empower patients to self-advocate** and engage proactively with clinicians to ensure a preventative rather than reactive treatment approach.
- **Collaborate with NMOs** to improve FX patient identification and representation within larger networks like the EHC.
- **Ensure data collection for FX patients** to represent real-world experiences, including serious and subclinical bleeding symptoms.

Conclusion

Factor X Deficiency remains a complex condition with varying levels of recognition, treatment, and support worldwide. While progress has been made in some regions, significant gaps persist in terms of diagnosis, access to care, and patient representation. By fostering education, innovation, and advocacy, all stakeholders and patient communities can work together to improve the quality of life for individuals living with FX Deficiency. This is a commitment of the ERIN work as part of EHC.