Von Willebrand disease

Suzanne O'Callaghan

Von Willebrand disease

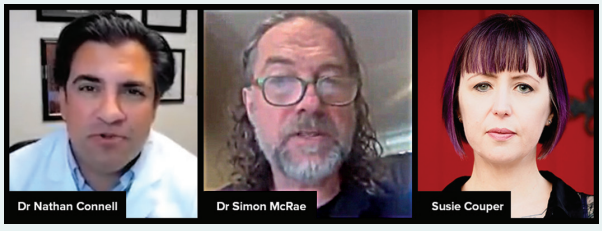
Chair ~ Susie Couper

VWD personal experience ~ Perry

VWD genetics, diagnosis and classification ~ Dr Simon McRae

VWD personal experience ~ Simoni

VWD treatment and future directions ~ Dr Nathan Connell



The von Willebrand disease session brought together some of the key players in the development of the new international clinical VWD guidelines to discuss the implications for diagnosis, treatment and care into the future.

THE PATIENT PERSPECTIVE

Setting the scene from the perspective of the patient were two compelling personal experiences from Perry and Simoni.

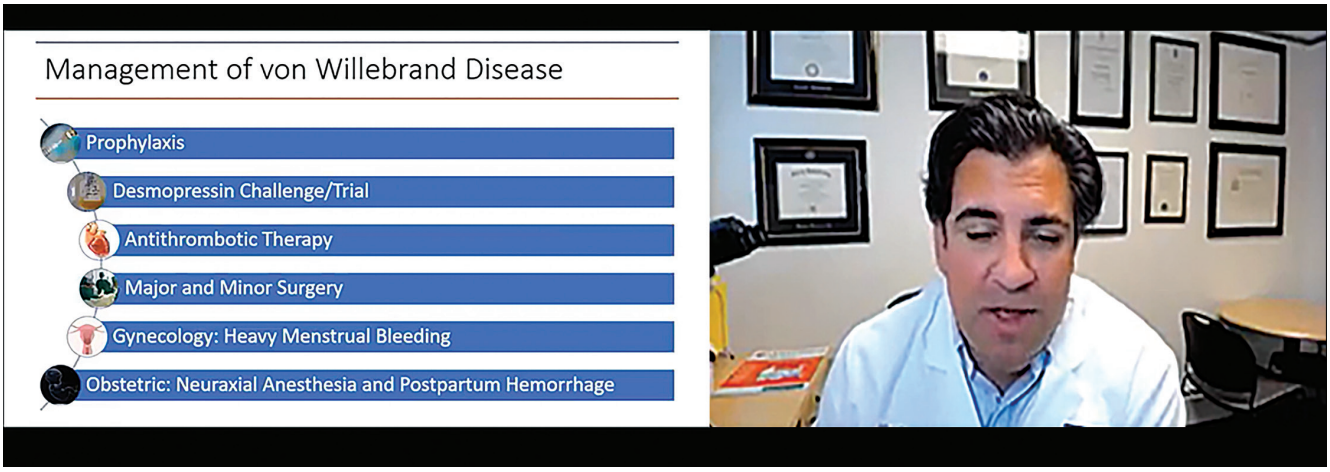
As an older man with type 2A VWD, Perry had not been diagnosed until he was in his 50s. He spoke about the problems he experienced with bleeding episodes over his lifetime before he was diagnosed and had a treatment plan – nose and mouth bleeds, lengthy bleeding after surgery. This bleeding is now much better controlled in liaison with his Haemophilia Treatment Centre.

Simoni is a young woman with type 3 VWD and was diagnosed as a young child after bruising severely. As she has grown up, she has had a number of bleeding problems, including heavy menstrual bleeding and bleeds after sports injuries. For Simoni, starting prophylaxis was a turning point for taking control of her bleeding and her quality of life.

You can read Perry and Simoni's personal stories in this issue of *National Haemophilia*.

What is exciting about the new international guidelines from a patient perspective? As a patient representative in the international panel for the clinical management guidelines, Susie Couper had three reasons to be excited:

1. VWD now has more visibility and as a result there is more understanding
2. The VWD guidelines were produced with patient involvement at every step, acknowledging the differing values of patients
3. The guidelines have opened up research priorities and put a focus on the strength of evidence.



VWD DIAGNOSIS AND CLASSIFICATION

Dr Simon McRae, now based at Launceston General Hospital in Tasmania, was a member of the international panel for the VWD diagnostic guidelines. He explained that there had been some variations in diagnostic criteria in previous guidelines and the new guidelines had been an opportunity to develop some consistency and provide decision-making tools.

The guidelines considered:

- Who should have diagnostic tests performed?
- What is the role of a structured bleeding assessment tool (BAT)?

The circumstances of diagnosis were very relevant.

A BAT would be useful if a patient was being seen at a primary care setting, eg a GP clinic, where the likelihood of them having a bleeding disorder was lower. A validated BAT could identify those who have abnormal bleeding that falls into the type of bleeding seen in a bleeding disorder such as VWD and indicate where further testing is appropriate.

If a patient has been referred to a haematologist or has a strong family history, they are more likely to have a bleeding disorder and a BAT is not needed to decide about testing. However, a BAT can be useful to help define their particular bleeding pattern.

Other areas that the guidelines clarified were:

- The value of newer assays or tests that measure the platelet-binding activity of VWF (von Willebrand factor) more accurately, such as VWF:GPibM, VWF:GPibR etc. Most laboratories in Australia already use these.
- What VWF assay level defines a diagnosis with VWD? The 30-50% VWF level has been contentious in decisions about VWD diagnosis and diagnosis will depend on the person's bleeding history. There may be other factors contributing to bleeding; but it is also important to be wary if a person has not yet had bleeding challenges, eg surgery, particularly males.

The guidelines also considered questions such as differentiating between type 1 and type 2 and when VWD genetic testing is useful (ie, for type 2A, 2B or 2N and for family planning in type 3).

What are the implications of the new diagnostic guidelines for Australia?

Dr McRae pointed out that the guidelines won't change diagnostic practice in most Australian settings and are unlikely to change patient management for individuals. They are likely to lead to a review of current diagnoses in the ABDR and there may be a need to retest patients where insufficient information is available. This will take some time. However, an important outcome for patients will be standardising of the wording for individual diagnoses, including on the ABDR patient card.

He also recommended a national network approach for genetic testing and some functional assays. Genetic testing funding is not always available nationally, making it difficult in some cases to work out how to fund the tests that are required for accurate and timely diagnosis.

VWD TREATMENT AND FUTURE DIRECTIONS

Dr Nathan Connell gave an update on the latest developments in VWD treatment, based on the new international VWD guidelines. Dr Connell was Vice-Chair of the international VWD guidelines Scoping Group and explained that the Group was an international partnership to develop evidence-based clinical management guidelines to promote better health outcomes, quality of life and health equity.

He discussed some of the key clinical management issues covered by the guidelines.

Prophylaxis is recommended for type 3 (severe) VWD, but conditionally – a shared decision-making process between the doctor and the patient:

- Prophylaxis reduces risk of bleeding episodes, hospitalisations, and heavy menstrual bleeding.
- The value of prophylaxis to individual patients will depend on how often they experience bleeding episodes.

The recommendations for heavy menstrual bleeding:

- Different options for women who do not wish to conceive vs women who wish to conceive.
- Focus on hormonal therapy (hormonal contraceptive therapy or levonorgestrel-releasing IUD) or tranexamic acid over desmopressin.
- Women might need to use multiple options at once if their heavy menstrual bleeding is not well controlled with the initial therapy.

- Importance of a multidisciplinary approach, with haematologist, gynaecologist and patient to manage both the bleeding disorder and gynaecological complications, which may or may not be related to VWD.
- Include assessment and treatment of iron deficiency and anaemia.

For people with mild and moderate VWD, what are the 'moments that matter', the triggers to get in contact with their Haemophilia Treatment Centre (HTC)?

- If they are planning any surgery or invasive medical procedures, they should connect with their HTC.
- Managing any bleeding, particularly heavy menstrual bleeding in women.
- Need to plan in advance for pregnancy.

RESEARCH PRIORITIES

Both Simon McRae and Nathan Connell pointed to a real need for evidence-based research to answer a range of questions about VWD, for example:

- Bleeding risk as people with VWD age
- Are there predictors for who will bleed and who will not, particularly those in the 30-50% VWF range
- Evaluating different bleeding assessment tools in males and children
- Research into hormonal contraceptive therapies for women
- Large randomised controlled trials on prophylaxis for VWD
- Use of plasma-derived vs recombinant VWF concentrate for prophylaxis.

We were grateful to all the presenters for sharing their expertise and their personal stories. The expert presentations from Simon McRae and Nathan Connell were immensely valuable and gave a concise and accessible overview of priority issues in VWD diagnosis and treatment. Having the personal perspectives of Susie, Perry and Simoni alongside their presentations grounded the discussion about the guidelines in real-life experience – and highlighted just why the process of developing international guidelines has been so important.

Suzanne O'Callaghan is HFA Policy Research and Education Manager

Women and girls

Jaime Chase

Women and Girls

Chair, introduction and personal story ~ Sharron Inglis

Understanding, recording and reporting bleeding symptoms in girls and women ~ Jaime Chase, Joanna McCosker

New clinical approaches in managing women and girls with bleeding disorders across the lifespan ~ Dr Mandy Davis

Gynaecological issues for women and girls with bleeding disorders ~ Dr Angela Dunford

Sport and exercise for girls and women ~ Hayley Coulson

The session regarding women and girls commenced with a personal story by Sharron Inglis, an Australian community leader, and raised some interesting questions about society's perspective on women with bleeding disorders. Advocacy, support and education were highlighted as her most important messages.

Heavy periods can negatively impact on physical, emotional and social quality of life and reduce work capacity - so how can we fix this?

Joanna McCosker and Jaime Chase discussed the importance of normalising the conversation around periods as they commence and as a woman moves through her lifespan to menopause.