Von Willebrand disease

Suzanne O'Callaghan

VWD

Chair ~ *Dr Chee Wee Tan*

Personal story ~ *Adam*

Personal story ~ *Susie*

VWD diagnosis ~ *Dr Geoffrey Kershaw*

VWD clinical management ~ *Dr Nalini Pati*

PERSONAL STORIES

The von Willebrand disease (VWD) session began with Adam's story of his personal journey with type 3 VWD. He has always been very physically active, but as he has grown older, he has become more aware of the impact of bleeds from injuries on his long-term health, as well as experiencing gastrointestinal bleeds, and is now looking at ways to manage this better into the future. He has learned to self-infuse and has considered employment options to support working from home to help manage bleeds. His advice to others with VWD:

- Don't ignore a bleed (as he has done, being male)
- Get a good GP and train them; and a good gastroenterologist
- Explore flexible employment options.

He finished with the comment that VWD is a disorder, not a disease – a view held by many in our community.

Susie followed with her experiences of living with type 1 VWD. Susie's story is published separately in this issue of *National Haemophilia*.

VWD DIAGNOSIS

Diagnosing VWD is complex and specialised. Dr Geoffrey Kershaw gave a remarkably accessible presentation on the key aspects of laboratory testing in an area that is very scientific. He explained that different regions of the von Willebrand factor (VWF) structure have different functions: binding to collagen or FVIII (factor 8) or platelets through glycoprotein 1b (Gp1b). These different functions relate to the laboratory tests available.

Testing and results can be affected by many variables. For example, an individual's VWF levels can increase because of stress, physical exertion and inflammatory illnesses and the sample must be kept at a specific temperature range for its quality to be maintained.

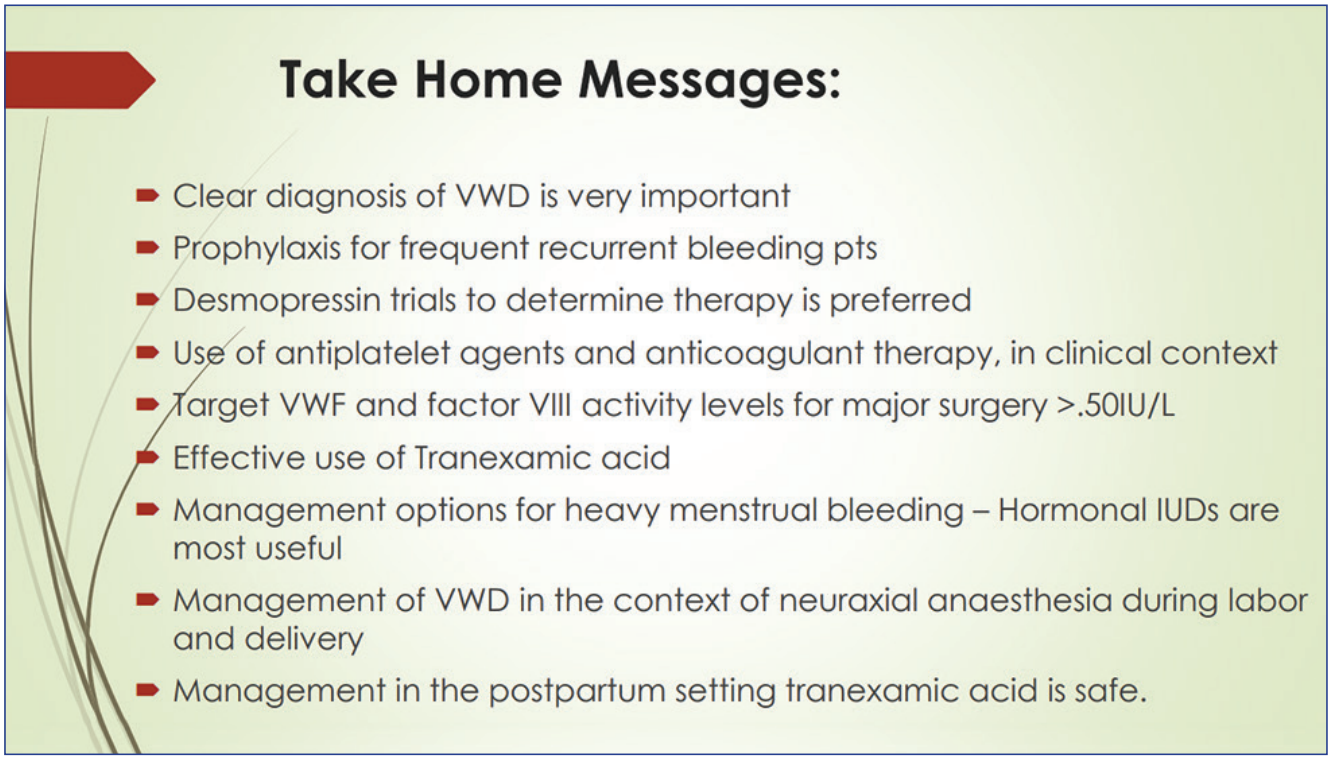
Von Willebrand Disorder (VWD):

- Most common inherited bleeding disorder.
- Arises from deficiencies or defects in von Willebrand factor (VWF).
- VWF has two primary functions/roles:
  - 'carries' FVIII, and protects/stabilises FVIII:C function.
  - permits adhesion of platelets to sites of vascular damage.
- VWF appears in plasma as a multimeric protein (low to high molecular weight or 'small' to 'large' size; higher MW = greatest adhesive function).

VWD CLINICAL MANAGEMENT

In a presentation on the medical issues, Dr Nalini Pati highlighted some of the factors affecting a patient's journey to appropriate diagnosis and treatment.

A lack of awareness among general practitioners and other health professionals in the wider community



and a wide variation in presentation means that VWD diagnosis often occurs late. He noted that treatment needs to be personalised but that treatment options in Australia are currently limited.

There was a consensus in the session that current challenges include:

- Need more data on Australian experience
- Need Australian VWD clinical guidelines
- May be multiple factors at play, not just VWF
- No recombinant product available in Australia
- Would be good to have a long-acting VWF product
- Better education for GPs, dentists, allied health to identify VWD and refer appropriately.

This was discussed in the final plenary, with a recommendation from Assoc Prof Chris Barnes that VWD needs to be included in the Australian Bleeding Disorders Registry (ABDR), along with education of health professionals in the wider community.

Suzanne O'Callaghan is HFA Policy Research and Education Manager

WANT TO KNOW MORE?

Many of the Conference presentations are now available on the HFA website.

View the presentations:
haemophilia.org.au/conf23