Rare Disease Day 2023

Rare Disease Day is celebrated worldwide on 28 February to raise awareness about the experience of people with rare diseases. It is also an effort to work globally towards equity in social opportunity, healthcare and access to diagnosis and therapies for people living with a rare disease.

The 2023 theme of **Rare is many. Rare is strong. Rare is proud** reminds us how important it is to come together and connect as a community. This is an opportunity to share personal stories and acknowledge the challenges for our community members who live with a rare disorder.

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WHAT IS A RARE DISEASE?

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people.

Around 8% of Australians (2 million people) live with a rare disease.

About 80% of rare diseases are genetic.[1]

Haemophilia is considered rare. Approximately:

- 1 in 6,000 males has haemophilia A
- 1 in 30,000 males has haemophilia B

Researchers are still gathering data on how many females are affected by haemophilia.

Some bleeding disorders are very rare.

For example, factor X (10) deficiency only affects 1 in a million people.

Type 3 VWD is the rarest form of von Willebrand disease, occurring in 1 in 500,000 people in countries like Europe and the USA.[2]

PERSONAL STORIES

In this issue of *National Haemophilia*, Adam tells his story of living with Type 3 VWD. He explains what it was like to grow up with a severe and rare form of VWD and what he has learned about becoming independent, participating and being active.

Sharing personal stories is an important way to help people with rare diseases feel connected. It can also help the wider community to understand better what it is like to live with a rare disease.

You may have seen some other personal stories about living with very rare bleeding disorders on our social media platforms in the lead up to Rare Disease Day, for example:

- Belinda's story (factor X deficiency) - https://tinyurl.com/FI-Belinda

Our thanks to Adam, Belinda and our other community members with rare bleeding disorders for sharing their personal stories.

Many people with rare diseases speak of feeling isolated. They may never have met another person with their condition. If they are the first in their family with the condition, it may have taken a long time for them to be diagnosed. Women and girls with haemophilia also talk of not being believed because of the common assumption that only males have haemophilia.

TREATMENTS

Treatment development is another issue. When rare diseases are very rare and numbers are small, this can mean that the development of new and highly effective treatments is slow. There may even be no treatment that specifically targets that condition. For example, while there has been great excitement around the world about the novel therapies developed for haemophilia and the difference they make to reducing bleeds and quality of life, there is not yet a specific clotting factor concentrate that is suitable to treat factor V (5) deficiency and fresh frozen plasma may be used for treatment instead.

HOW CAN YOU HELP?

You can help to raise awareness by sharing the stories of people with bleeding disorders through your personal networks.

Do you have a story you would like to share yourself? Visit the SHARE YOUR STORY section on the HFA website to tell us more - www.haemophilia.org.au/shareyourstory .

For more information on Rare Disease Day, visit www.rarediseasedayaustralia.com.au.

REFERENCES

1. Australian Government. Department of Health. What we're doing about rare diseases. https://www.health.gov.au/health-topics/chronic-conditions/what-were-doing-about-chronic-conditions/what-were-doing-about-rare-diseases Accessed 21 February 2023
2. Orphanet: the portal for rare diseases and orphan drugs. https://www.orpha.net/ Accessed 21 February 2023