

## What does the haemophilia community think about genetic screening?

### Warwick Medical School researchers report their project findings

As genomic medicine advances, screening the whole population for genetic conditions is now possible. However, little is known about what people living with bleeding disorders think about screening, nor the social and ethical issues it raises.

In 2017, researchers Drs Felicity Boardman, Rachel Hale and Philip Young began to explore this topic, using interviews and a survey. With The Haemophilia Society's support, 22 people were interviewed and 415 people completed a screening attitudes survey, all of whom had a bleeding disorder or were relatives of someone who did.

#### Mixed views

The results showed a mix of perspectives towards both screening and bleeding disorders. A minority of participants described bleeding disorders as debilitating and burdensome; however most considered them manageable, particularly when well controlled. The most obvious differences in attitude were between older adults with haemophilia (many of whom had been exposed to blood-borne viruses) and younger participants, many of whom are benefiting from treatment improvements, particularly prophylaxis.

#### Concerns around screening

Both survey and interview participants were quite critical of the idea of population screening for bleeding disorders, the major concern being that it might lead to pregnancy terminations. Instead, they emphasised the need for early diagnosis and treatment, with 77% of survey participants believing that

newborn screening was the most acceptable screening programme. The prospect of pregnancy or pre-pregnancy screening generated less support, at 59% and 57% respectively.

Given the negative attitudes towards termination, it was initially surprising that pregnancy screening was viewed slightly more favourably than pre-pregnancy. However, further analysis revealed that this was because participants believed the information could assist preparation for the birth of a child with haemophilia, rather than inform a termination decision. Indeed, 90% of survey participants regarded termination for haemophilia as ethically unacceptable.

Overall, bleeding disorders were largely viewed as liveable conditions. Screening was supported when used for information, although fears emerged about the accuracy and quality of that information, particularly when the general public were viewed as largely ignorant of bleeding disorders. High quality information, early diagnosis, education and access to treatment (especially prophylaxis) were considered the most important goals of any screening programme, rather than prevention per se.

#### What will happen to the results?

These results will be prepared for a report to the UK National Screening Committee and also distributed to academics, researchers and clinicians, giving them an overview of the main concerns that families affected by bleeding disorders have about screening.