

Haemophilia gene therapy snapshot survey

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In May-June 2024 HFA conducted a short community survey to ask people affected by haemophilia about what they want from their treatment and their thoughts on gene therapy in particular.

With a range of new and innovative haemophilia treatments becoming available, it is important to understand what people with haemophilia and their parents, partners and family would like to see as outcomes of the treatment. What would result in a better quality of life for them?

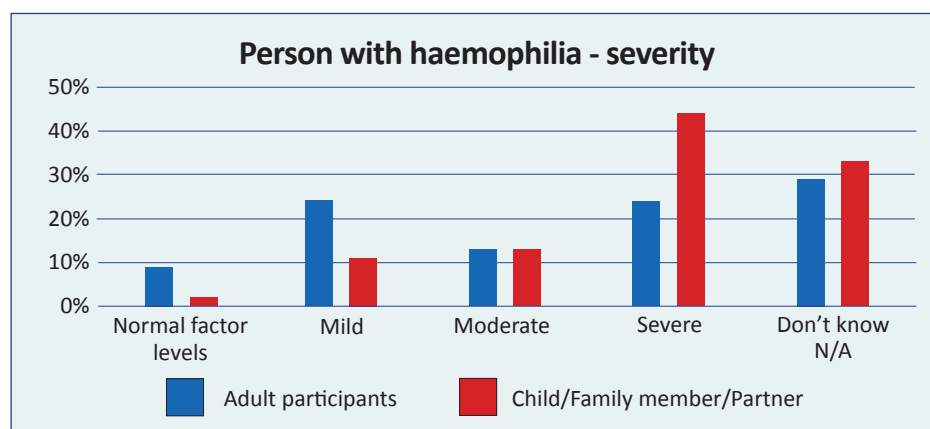
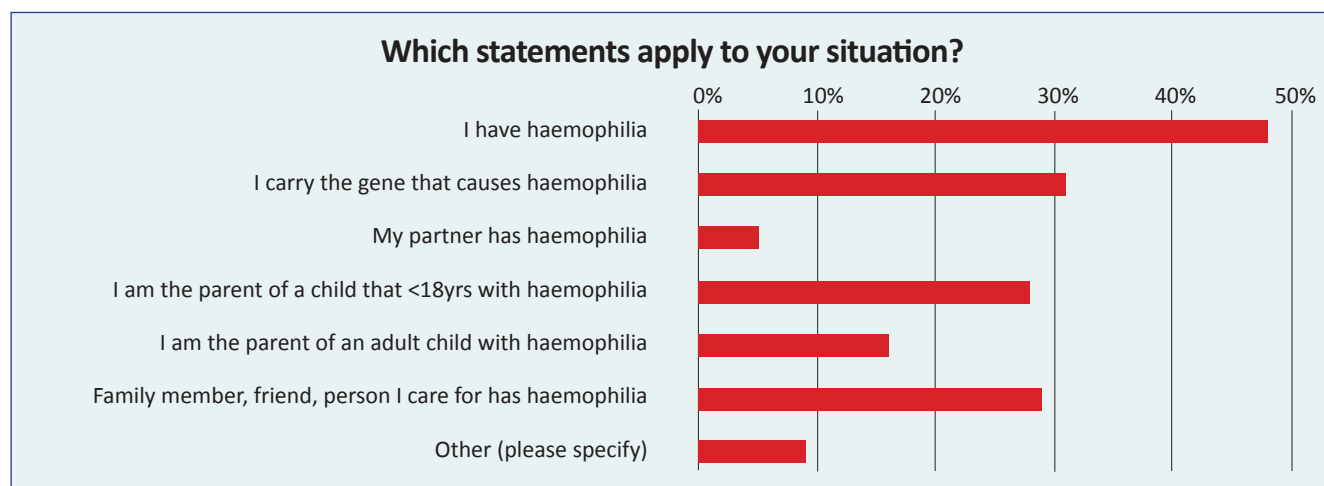
Our thanks to all who participated.

WHO COMPLETED THE SURVEY?

- 45 people (19 female/24 male/2 preferred not to say) completed the survey.
- All age groups from 18 years and over were represented. 53% were 31-54 years old.
- 69% lived in a capital city; 4% in the rural/urban fringe of a capital city; 24% from a regional/rural remote area.

How were they affected by haemophilia?

In many cases both they and their child/family member were affected by haemophilia.



PREFERRED TREATMENT OUTCOMES NOW

When asked what they would like haemophilia treatment to achieve for them **NOW**, people completing the survey described both impacts on their quality of life and the way this would be achieved – with treatment and how treatment is delivered and supported.

The most common preferred outcomes were preventing bleeds, avoiding injections/infusions and living a normal life. They often had a realistic appreciation of current gene therapies and their limitations – that gene therapy may not work for all or result in normal factor levels or have an effect that is sustained permanently. Some women and parents of children who are both currently ineligible for gene therapy were hoping for access. Some respondents talked generally about ‘better quality of life’.

Treatment outcomes

- Reducing symptoms, preventing bleeds, stable normal factor level
- Easier and less painful administration; no needles/injections/infusions; oral/tablets
- Longer effect in the body
- Normal life, participating in activities, travel, sport
- Improves impact of complications - arthritis, HIV, work, education, pain
- Not so many hospital visits/stays; quicker recovery
- Cure
- Safety, no inhibitors, no other side effects, eg liver
- Better quality of life
- Happy with current treatment outcomes.

Treatment delivery

- Access to new treatments, including gene therapy
- Easy access to medical advice, testing, multidisciplinary care with bleeds.

‘No more needles every two weeks. No more bruising. Less hospital visits’

‘As someone with Moderate that only has factor from trauma, approx 1-2 times a year, the ability to not have to go to the hospital for treatment, allowing me to travel and not feel tied down.’

‘To fix the gene responsible so my son can undertake normal activities like sport.’

PREFERRED TREATMENT OUTCOMES IN THE FUTURE

When asked what they would like their treatment to achieve **IN THE FUTURE**, people responding to the survey had an opportunity to give their wish list and their hope that future gene therapies would be an effective and safe cure. Some were looking for a germline gene therapy that would mean the gene alteration and haemophilia would not be passed onto future children.

Treatment outcomes

- Cure
- No bleeds, normal factor levels, prevent joint damage
- Easier much less frequent administration; no needles/injections/infusions; oral/pills
- Normal healthy longer life, participating in activities, sport
- Safe, no side-effects
- Long-term effectiveness
- Recovery from joint and muscle damage
- Having children safely; not passing haemophilia on to children.

Treatment delivery

- Better resourcing for HTC multidisciplinary care, support, referral, home testing
- Access to gene therapy
- Affordable genetics services to support family planning.

‘Longer acting factor replacement. Pill form rather than injection. A gene therapy cure that lasts.’

‘To CRISPR it out of my and my children’s genetic structure.’

‘To be able to play sport like other kids. No more needles. No need to worry about bleeds.’

‘Repairing existing joint damage for patients with a history of bleeds. Research into some truly novel approaches (other than joint replacement/fusion) to heal or regenerate.’

‘Informed treatment options. Holistic support. Continuity of specialist care.’



Photo: PEXELS/Gustavo

GENE THERAPY – BENEFITS AND CONCERNS

Participants were asked what they thought the main benefits of current gene therapies would be for them or their child/partner/family member. Their focus was on avoiding the frequency of painful treatments and the better quality of life that could result from no bleeds – perhaps being able to live a ‘normal’ life like people without haemophilia. Some hoped for a cure that would mean permanent normal factor levels and no longer having haemophilia. However, for others this was tempered with the understanding that it may not work for an individual or be a complete cure.

Perceived benefits

- No need for prophylaxis, frequent treatment
- Living a normal life, better quality of life, easier travel, no bleeds
- Cure: no longer having haemophilia
- Single long-acting treatment
- Potentially normal factor levels
- Beneficial if it works
- Preventing joint/muscle damage
- Less anxiety about periods, childbirth

‘Single injection/infusion. Get rids of the burden on further injection/infusion.’

‘Being “healed” from haemophilia for life.’

‘A life without needles and treatment. A more normal way of living.’

‘Better outcome for joints and muscles with longer, higher factor level. No more use of medication for a long time hopefully (regeneration for the injection places), no medication on vacations, no problems with these on custom controls and security checks.’

Their most common concerns by far were with safety and efficacy.

Concerns

- Safety, side-effects - known and unknown
- How long the effect will last
- Failure/might not work
- Can’t have it again
- Eligibility
- Waiting for more advanced gene therapy
- Cost
- Requirements: no alcohol, contraception
- Requirements: appointments, travel
- Not a complete cure
- Not reversible
- Effect on future children
- Access: regional, rural, remote

‘Triggering other health issues by manipulating genes.’

‘Will it be difficult to qualify, will it last and what are the side effects.’

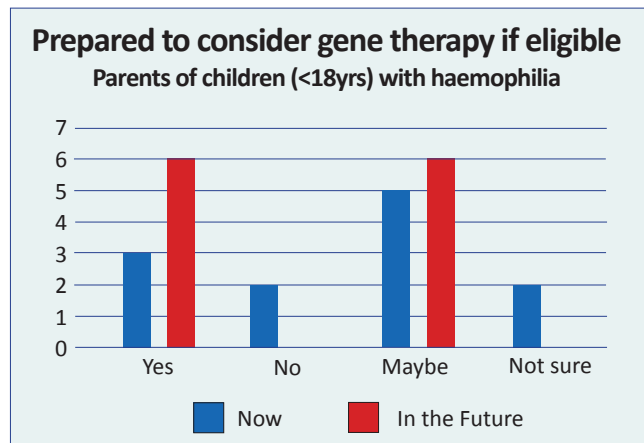
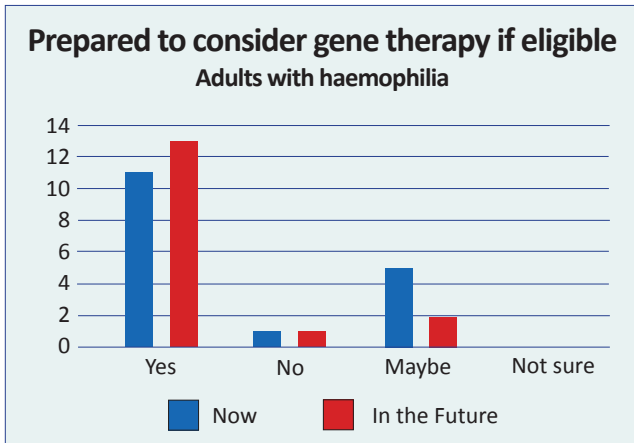
‘Seems problematic, potential steroid reliance, no alcohol use, may not work.’

‘The first year during treatment with multiple visits to the hospital and potential increased travel costs. Long term data. We are a young couple who would hopefully like to start a family and hence we are navigating when to have children vs when gene therapy becomes available.’

ATTITUDE TO GENE THERAPY

Would they consider having gene therapy now or in the future if they were eligible?

Parents of children were more doubtful than adults with haemophilia, but both assumed that treatment efficacy and safety would have improved in the future.



Adults with haemophilia	Parents of children
GENE THERAPY NOW	
<p><i>‘Having a single treatment is great and Hemlibra does not bring my factor 8 levels to normal.’</i></p> <p><i>‘Needs perfecting.’</i></p> <p><i>‘Even a small improvement is better than none.’</i></p>	<p><i>‘Until it’s proven to be safe, would not be comfortable with him receiving this therapy.’</i></p> <p><i>‘Not enough research yet and Hemlibra works fine for us atm.’</i></p> <p><i>‘Need to know that it has been tested long term, and has proof of being safe and beneficial to the patient.’</i></p>
GENE THERAPY IN THE FUTURE	
<p><i>‘Because it hopefully would increase quality of life as long as the risk to benefit ratio was acceptable.’</i></p> <p><i>‘I’d prefer not to have haemophilia.’</i></p> <p><i>‘Once it’s reliable, I’d jump at it.’</i></p>	<p><i>‘To cure him.’</i></p> <p><i>‘Hopefully by then it’s robust and thoroughly tested.’</i></p> <p><i>‘I think it’s a viable treatment at any age to improve outcomes.’</i></p>

ACCESS

Final comments from participants underlined their concerns about access and eligibility: making gene therapy available free-of-charge to the patient and enabling access for females and people with lesser severity.

‘Access for mild to moderate patients - often with significant morbidity.’

‘We need gene therapy to be available to females as well. I have a daughter with severe haemophilia B who is a sufferer just like the males. She has

intravenous factor 9 injections just like the boys and has done her entire life. She normally doesn’t get offered the same treatment options like gene therapy just because she is female.’

‘I think gene therapy will be a wonderful thing. I really hope we are able to get funding for people with haemophilia to be able to access it.’

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