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COMMUNITY FOCUS

"You're only a carrier" – women and the language of haemophilia

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- Women with haemophilia and factor VIII activity >40% are called carriers, but this label does not reflect their experience of heavy bleeding, pain and impaired quality of life
- The term 'carrier' should be used only in the context of inheritance
- Person-first terminology is needed for women with haemophilia

Women who have the gene variant for haemophilia are labelled solely as 'carriers' unless they have a factor VIII activity of ≤40%. This term, which describes an individual who can pass on a disorder but are themselves unaffected, reflects a legacy that extends from the 18th century to the present day. There is strong evidence that women labelled as carriers experience heavy periods, joint damage, pain and impaired quality of life. The label 'carrier' does not recognise this burden and is associated with guilt, stigma and difficulty accessing care. People living with a long-term disorder should now be described using person-first terminology and it is common to see the term 'people with haemophilia'. The term 'carrier' should be limited to its application in genetics and not used as a catch-all label for women with haemophilia.

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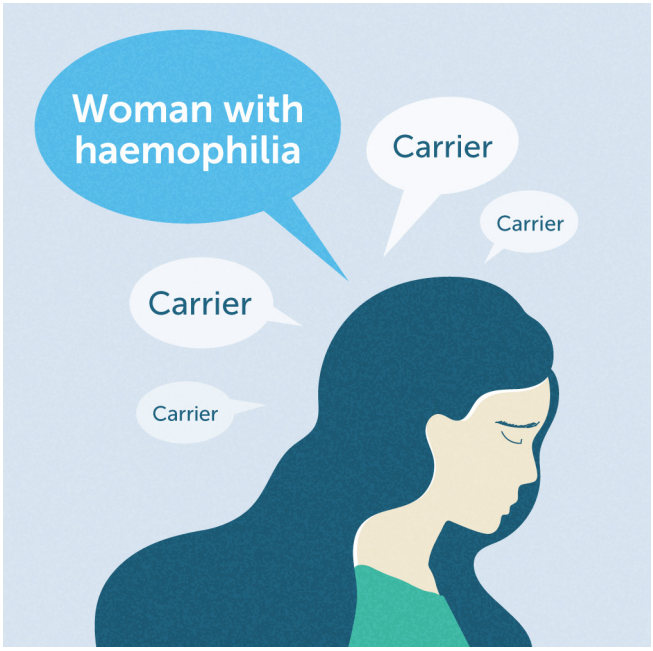

The language used to describe women who have and are affected by the gene variant for haemophilia impacts the care they receive. Proposals to change this are welcome but using the label 'carrier' remains problematic

Keywords: *Haemophilia, women, carriers, genetics, terminology as topic*

Women who have a gene variant for haemophilia are labelled a 'carrier'. Historically, this has been interpreted largely in the context of the risk of their sons developing a clinically apparent bleeding disorder and the implications for tracing affected family members, taking little account of the personal impact of living with the effects of the gene variant. Inappropriate use of the term 'carrier' is perpetuating

this. It is time to limit the use of this word to its rightful application and, in other contexts, to adopt an alternative that accurately reflects a woman's experience of having haemophilia.

HOW HAS HISTORY DESCRIBED WOMEN WITH HAEMOPHILIA?

In 1803, John Conrad Otto's landmark description of a family affected by a 'hemorrhagic disposition' noted that 'females are exempt' [1]. Women figured prominently in John Hay's 1813 description of a family affected by haemophilia without mentioning any bleeding tendency on their part [2]. Symptoms in women were soon recognised – heavy menstrual bleeding was described in 1857 [3], though only as a marker of sons with haemophilia, and in 1884 it was observed that symptoms did not emerge in girls until the onset of menstruation 'or marriage produces a crisis that will excite it' [4]. That was the year that Queen Victoria's son Leopold, arguably the most famous person with haemophilia, died following a traumatic haemorrhage. Media coverage of Victoria's role as an unaffected carrier and her son's as unfortunate victim of incurable illness cemented the image of a male-only bleeding disorder in the public mind [5].

By the start of the 20th century, sufficient cases of women with a bleeding disorder (not necessarily haemophilia, since other disorders had not been recognised) had been reported that they merited a section in the definitive 1911 account of haemophilia, Bulloch and Filde's contribution to the *Treasury of Human Inheritance* [6]. However, the authors dismissed the conflicting medical opinion, concluding there was a lack of adequate evidence to support the view that women could bleed, other than as a rare occurrence. Such cases did not meet the diagnostic criteria for haemophilia and any resemblance was 'superficial' and 'slight or atypical' compared with 'well-marked haemophilia' in males.

By the mid-1950s, there was no doubt that women with a bleeding disorder experienced significant bleeding but sexism prevailed [7]. In 1956, two US physicians described an analysis of antihaemophilic activity in '19 presumptive and 8 possible carriers' [8]. They found a low level in one woman and commented:

> *"Many authors have described minor hemorrhagic phenomena in these heterozygous conductors. However, such women are of course likely to be acutely aware of bleeding phenomena, and may exaggerate otherwise unremarkable symptoms".*

Such attitudes influenced those who followed. In 1983, an analysis of the UK haemophilia database described approximately 5,000 registered patients as if all were male, suggesting either no women were registered or they were unrecognised [9]. The 2012 World Federation for Hemophilia (WFH) management guideline included a section on carriers as a 'special management issue' [10]. It stated 'Hemophilia is an X-linked disorder that typically affects males, while females are carriers', 'Most carriers are asymptomatic' and 'A few carriers may have clotting factor levels in the hemophilia range — mostly in the mild category', adding that 'menorrhagia and bleeding after medical interventions are the most common manifestations among carriers with significantly low factor levels'. While the WFH 2020 management guideline pays full regard to the clinical and personal significance of being a haemophilia carrier [11], it continues to refer to women with haemophilia almost exclusively as 'carriers'. The medical profession has left unchallenged the notion that women are unaffected by haemophilia for many years, reflecting a culture of sexism that has permeated the management of bleeding disorders for decades [12,13].

WHAT'S IN A WORD?

In an era where personal autonomy is encouraged and people are supported in being active partners in their care [14], good professional practice mandates the use of person-first language [15]. Using an adjective to label a person or to put a medical diagnosis before their status as an individual is disliked by most people. For example, a survey of 971 people with epilepsy and their carers found that 87% and 93% respectively favoured the phrase 'That person has epilepsy' over 'They're epileptic' or 'They're an epileptic person', rating the latter terms disliked/intensely disliked [16]. The person-first term was felt to distinguish the identity of the individual from their condition, so that epilepsy was only one of their attributes; it suggested they might have some mastery over it; and the word 'epileptic' was considered old-fashioned and associated with negative perceptions of epilepsy.

NHS England has published guidance on the use of language, particularly in the context of diabetes care but with general applicability [17]. It points out that words, phrases and descriptions are potentially problematic, whatever the intention of the user, and states that health professionals should use person-first terms to avoid labelling a person as their condition. Some people do consider the term 'diabetic' acceptable but it is important to ask before describing them as

such. This reflects a long-term trend. A survey of language used by five leading medical journals between 1976 and 2015 found that the use of person-centric words grew significantly in all but one publication, with an overall average increase of 19% [18].

WHY IS 'CARRIER' A NEGATIVE TERM FOR WOMEN WITH HAEMOPHILIA?

Early descriptions of family trees used the word 'carrier' in its true sense of an individual 'who carries and is capable of passing on a genetic mutation associated with a disease and may or may not display disease symptoms' [19].

There is no dispute about the relevance of the second part but certainly a lack of awareness of its impact. Women labelled as a haemophilia carrier often have lower levels of factor VIII activity compared with unaffected women and may experience increased bleeding symptoms such as easy bruising, bleeding after minor surgery and oral bleeds [20]. The prevalence of heavy menstrual bleeding is 40%–50% [21]. Like their male counterparts, they can experience joint damage: among those with low or unknown factor activity (but not activity ≥40%), the risk of a diagnosis of a joint disorder or hospital admission for a joint disorder is two to three times greater than in the general population [22]. The impact of living with these signs and symptoms is associated with impaired quality of life scores for pain and general health compared with unaffected controls [23]. If they were men, many would be diagnosed as having mild, moderate and even severe haemophilia [24,25].

By contrast, it is not relevant to refer to the risk of genetic transmission other than in the context of inheritance. For example, it is clearly important to identify family members who might pass on a haemophilia gene variant to their offspring so that they can be offered counselling; it is therefore meaningful to describe women in this context as carriers. But there is no purpose in calling a woman a carrier when she seeks medical advice about heavy menstrual bleeding. In an era when annualised bleeds approaching zero are considered achievable rates in people with haemophilia [26], having 12 major, predictable and avoidable bleeds per year is unacceptable. Yet women labelled as carriers have described dismissive attitudes from caregivers who did not believe they could have excessive bleeding and trivialised their concerns, symptoms or requests for haemophilia-related care [27,28]. One Canadian survey found that 40% of 70 health professionals believed that a label of carrier contributed to diagnostic delay; obstacles to accessing care included lack of awareness among health professionals (73%), stigma associated with vaginal bleeding (29%), and the women's focus being on male family members with haemophilia rather than on themselves (20%) [29].

The term carrier also serves to remind women of their role in the occurrence of haemophilia in their children. They describe feelings of guilt and sorrow about having passed on the variant to their children and report hearing similar sentiments from their mothers [30,31]. Fear of passing haemophilia to a child is frequently a factor in a woman's decision not to have children; therefore, this is a highly sensitive issue with implications for the individual and her family [32].

Of course, men with haemophilia pass the variant on to their daughters but they are never labelled a carrier – with one exception: those who undergo gene therapy are reminded that they continue to be genetic carriers. This is an appropriate use of 'carrier' and it should be used in that way for women.

HAS THE HAEMOPHILIA COMMUNITY RESPONDED?

The Scientific and Standardization Committee (SSC) of the International Society on Thrombosis and Haemostasis has proposed a change to the labels used to describe women with haemophilia [33]. This will mean – eventually – that health professionals and researchers will cease to call them carriers but instead use language that recognises their experience as people with a bleeding disorder.

The SSC has been developing and consulting on its proposals since 2017 and recommends two categories to describe women and girls diagnosed as haemophilia carriers, defined by their factor level. Among those with levels <40 IU/ml, there are three groups labelled in a way that corresponds to the male equivalent: women with mild (>0.05 to <40 IU/ml), moderate (>0.01 – 0.05 IU/ml) or severe haemophilia (<0.01 IU/ml).

The second category is women and girls with a factor level of ≥40 IU/ml. It defines two groups: one with a bleeding phenotype, the other without. The proposed labels for these people are symptomatic and asymptomatic haemophilia carrier. The SSC recommends that the term 'haemophilia carrier' should be reserved for use in discussions regarding genetic counselling, reserving 'symptomatic carrier' or 'haemophilia' for when the focus is on bleeding concerns.

While the SSC acknowledges that the label 'carrier' 'has hampered diagnosis, management, and research', by persisting with the term 'symptomatic carrier' it is helping to perpetuate the problem. Recognition of the power of language to affect the care that women with

haemophilia receive is vital step and is to be welcomed. But if that care is to improve in both specialist and non-specialist settings for women who have or may have the haemophilia gene variant – including general practice and gynaecology, often gatekeeper services for accessing specialist care – the broad use of the carrier label also needs to change. Referring to a person as a carrier has nothing to do with their symptoms but contributes, in the SSC's words, to 'marginalisation' and a 'poor relationship' with health professionals.

IF NOT CARRIER, THEN WHAT?

The bleeding disorders community has changed its use of language over the years. It was once common in the scientific media to see the term 'haemophiliac'; now it is rare unless used by a person with haemophilia to describe themselves. Instead, we use the term 'person/people with haemophilia', or PWH, when describing males. That should be the preferred term for women with haemophilia when their sex is not relevant. In other cases, if a specific person-first term is needed, the obvious example would be 'woman with haemophilia' – a title that embraces all the implications of living with a bleeding disorder, not just those associated with blame and denial. Future iterations of the WFH management guideline should also incorporate this first-person language.

New terminology changes little unless it is supported by action. All women who may have the haemophilia gene variant and all daughters of fathers with haemophilia should be offered measurement of factor VIII/IX activity. They should be described as having a mild, moderate or severe phenotype and women levels <40% should be added to a haemophilia register. This is an organisational step that should increase access to care. However, it should not detract from the primacy of management of bleeding symptoms and improving quality of life in women with haemophilia.

ACKNOWLEDGEMENTS

The authors have advised no interests that might be perceived as posing a conflict or bias. There was no external funding support for this work.

SC drafted the manuscript. SC and KK reviewed and finalised the manuscript.

The authors would like to thank Kathryn Jenner, Technical Editor, Haemnet, for her contribution to the manuscript.

This paper does not contain any studies involving human participants or animals performed by any of the authors.

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HOW TO CITE THIS ARTICLE:
Chaplin S, Khair K. "You're only a carrier" – women and the language of haemophilia. *J Haem Pract* 2021; 8(1): 128-132. *https://doi.org/10.2478/jhp-2021-0015*