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ORIGINAL ARTICLE

Women with bleeding disorders

Haemophilia testing of young girls in Canada: Describing the current recommendations for factor level and genetic testing and the experiences of Canadian parents

Megan Chaigneau[1] | Julie Grabell[1] | Emil Wijnker[2] | Mackenzie Bowman[1] | Paula James[1]

[1]Department of Medicine, Queen's University, Kingston, Ontario, Canada

[2]Canadian Hemophilia Society, Queen's University, Kingston, Ontario, Canada

Correspondence
Paula James, Department of Medicine, Queen's University, Room 2015 Etherington Hall, 94 Stuart Street, Kingston, Ontario K7L 3N6, Canada.
Email: jamesp@queensu.ca

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Abstract

Introduction: It is widely acknowledged that haemophilia affects women and girls, yet current testing recommendations for factor level and genetic testing vary and do not universally incorporate updated research. Canadian parents have expressed frustration at inconsistent recommendations and reported instances where delayed testing led to missed diagnosis and preventable bleeding.

Aim: Study aim was to explore and describe the practice of haemophilia-related testing of young girls in Canada.

Methods: A mixed methods study was carried out with two populations: (1) Nurses working in haemophilia care completed a survey regarding the current testing recommendations of their Haemophilia Treatment Centre (HTC), (2) Parents of obligate or potential haemophilia carriers completed a structured interview with questions about their family experience of haemophilia and testing decisions for daughters.

Results: Twenty-six survey responses were received and showed wide variation in the usual recommendations of Canadian HTCs. Different factor level testing recommendations may be given to obligate and potential carriers despite no difference in bleeding risk. Only a minority of HTCs currently recommend an early baseline factor level (< 10 years) to obligate carriers (27%) or potential carriers (15%). For genetic testing of potential carriers, 70% of HTC would approve a family request for genetic testing of a minor with specific conditions. The majority of parents interviewed felt dissatisfied with their testing experience (58%) and highlighted many issues related to delayed testing recommendations.

Conclusion: Updated, nationally affirmed testing recommendations are needed that align with research on bleeding in women and girls affected by haemophilia.

KEYWORDS
delayed diagnosis, genetic carrier testing, haemophilia, inherited blood coagulation disorders, women's health

1 | INTRODUCTION

The historical understanding of haemophilia was that men were affected while women were unaffected genetic transmitters only. Yet the different ways haemophilia affects women and girls are now clearly recognized: as individuals with haemophilia corresponding to their factor level, as symptomatic haemophilia carriers, or asymptomatic haemophilia carriers.[1] It is estimated that for every man with haemophilia there are 3–5 haemophilia carriers and approximately one-third of these women and girls will manifest below normal factor levels and have haemophilia.[2–4] These numbers are not reflected in national and international registries, highlighting issues of underdiagnosis and indicating a need to better identify all women and girls affected by haemophilia.[5] This is especially important as research clearly demonstrates that carriers of haemophilia can experience abnormal bleeding symptoms, even those with normal factor levels.[3,6] These symptoms include heavy menstrual bleeding, postpartum haemorrhage, excessive post-surgical bleeding, epistaxis, easy bruising, oral cavity bleeding and musculoskeletal bleeding.[3,4,7–10]

The first calls for systematic assessment of factor level in all haemophilia carriers to determine bleeding risk 'for safety reasons alone' date back to the 1990′s,[11,12] yet testing practices vary across Canadian Hemophilia Treatment Centre's (HTC) and many different timepoints may be suggested including: early childhood, if abnormal bleeding symptoms present, prior to surgical/dental procedures, around age of menarche, at age of consent, or prior to pregnancy.

Genetic testing for haemophilia has been available since the mid-1980′s for testing prior to pregnancy for family planning and identification of affected male children.[11,13] It is currently available to all potential carriers in Canada at the age of consent, and increasingly considered in adolescence when the child is able to give assent or even earlier in childhood if abnormal bleeding symptoms present. Although genetic testing of minors must be approached with caution, it is recommended when to the medical benefit of the child for assessment, monitoring or treatment purposes.[14]

In previous research, Canadian parents have reported frustration at the lack of consistency regarding testing practices nationwide and instances of testing refusal where parents were unable to obtain the testing they desired or felt that they compromised their relationship with their HTC by insisting on early testing for their daughters.[15] Additionally, parents have been shown to have a preference for earlier testing as compared to healthcare providers (HCP), motivated by a desire to do what is best for their child.[15–18]

It is almost universally recognized that when resources permit, a male-child at risk for mild haemophilia based on family history should have factor level testing in the first few years of life and often genetic testing as well. Yet despite the now established fact that one third of women and girls heterozygous for haemophilia will have mild haemophilia, it is not uncommon for a female child to have no testing (factor level or genetic testing) until much later in life. The absence of early recognition of a potential bleeding risk in young girls can have deleterious effects on the health and quality of life and delay access to appropriate care.[19] Recently, some groups have released clear updated guidelines around haemophilia related testing for women and girls such as the 'European principles of care for women and girls with bleeding disorders' which calls for systematic factor level testing in early childhood, repeated prior to menarche, and genetic testing recommended when the individual is competent to understand results, or earlier if clinically indicated.[20]

The extent to which testing recommendations have evolved in Canada is unknown, thus our study aim was to explore and describe the practice of haemophilia-related testing in young girls from the perspective of both HCP and parents in Canada. From HCP we aimed to describe the current testing recommendations being given out by Canadian HTC's and from parents we sought to describe their experiences as they made testing choices for their daughters.

2 | MATERIALS AND METHODS

We conducted a descriptive mixed methods project with two study populations, HCP and parents of haemophilia carriers/potential haemophilia carriers. A convergent parallel design was used where quantitative and qualitative aspects were conducted simultaneously, analysed separately, but then merged for interpretation to fully answer the research question.

A convenience sampling method was used for both study groups. HCP were recruited via affiliation with the Canadian Association of Nurses in Hemophilia Care (CANHC) which includes nurses from all 26 HTC's across Canada. Parent participants were recruited via a one-page flyer distributed by provincial haemophilia societies.

The quantitative aspect of the study consisted of a five-question anonymous survey sent via email to all 45 active members of CANHC. One email reminder was sent 2 weeks after initial email contact. Consent was obtained at the beginning of the survey and slightly different questions were asked depending on the type of HTC the respondent was affiliated with, adult-only or paediatric/lifespan (Appendix A). Questions were related to the usual recommendations of their HTC for factor level testing of obligate and the potential carriers. HTC's that treat paediatric patients were also asked about usual recommendations around genetic testing of a minor when the request has been made by the family. No location-related data was collected in order to preserve anonymity as many Canadian provinces only have one HTC. Survey responses were exported to Microsoft Excel and organized as percentage of respondents who selected each answer category. Results were analysed collectively and then separately based on type of HTC (adult-only vs. paediatric/lifespan). As this is a descriptive study with the aim of summarizing the current landscape of recommendations being given by HTCs, no analysis of statistical significance was done.

The qualitative aspect of the study consisted of a structured interview carried out with parents of haemophilia carriers/potential carriers. Inclusion criteria were any English-speaking parent of an obligate or potential haemophilia carrier of any age residing in Canada. There were no exclusion criteria and parents of potential carriers who had received genetic testing of their daughters with negative

TABLE 1 Current recommendations for timing of factor level testing for young girls.

Timing recommendation for factor level testing	Obligate ($n = 26$)	Potential ($n = 26$)
At least once in early childhood (< 10 years)	27% ($n = 7$)	15% ($n = 4$)
If abnormal bleeding symptoms present	23% ($n = 6$)	23% ($n = 6$)
Prior to menarche (10–14 years)	12% ($n = 3$)	8% ($n = 2$)
At age of consent	0	8% ($n = 2$)
COMBO: If abnormal bleeding symptoms present, otherwise prior to menarche	15% ($n = 4$)	15% ($n = 4$)
COMBO: If abnormal bleeding symptoms present, otherwise at age of consent or prior to pregnancy	19% ($n = 5$)	27% ($n = 7$)
Other: Discussion with family	4% ($n = 1$)	4% ($n = 1$)

results (i.e., not carriers) were still included as interview questions were related to experiences around testing decisions. The recruitment flyer instructed interested participants to contact study staff by phone or email, after which full study details were sent by email. Each participant underwent a structured interview via telephone or Microsoft Teams (conducted by M.C.) with consent obtained prior. Interview questions were related to the experience of haemophilia in the family, testing decisions, interactions with HCP including testing recommendations, and parental opinion on optimal timing of haemophilia-related testing in young girls (Appendix B). Parental responses to questions were transcribed in real-time with longer answers read back to participants to ensure accuracy of recorded written responses. Results were imported into Microsoft Excel and shown as the percentage of total participants in each response category. For longer responses regarding beliefs, testing experiences and decision-making factors, 10 key findings deemed highly relevant by the research team were selected. As interviews were not recorded and no transcripts were made, thematic analysis of these longer answers was not possible.

Ethics approval was granted by the Queen's University Health Sciences and Affiliated Teaching Hospitals Research Ethics Board prior to study commencement.

3 | RESULTS

3.1 | Current testing recommendations across Canadian HTCs

A total of 26 responses to the survey were received, six from adult-only HTC's and 20 from paediatric or lifespan HTCs. Overall there was wide variation in the usual recommendations of HTC's on the timing of factor level testing for young girls (Table 1). Respondents were able to select more than one testing timepoint if that most accurately reflected the advice of their HTC resulting in the 'COMBO' answer categories.

For obligate carriers, the most frequent recommendation was for factor level testing 'at least once in early childhood' (27%) followed closely by 'if abnormal bleeding symptoms present' (23%). For potential carriers, the most frequent recommendation for factor level testing was 'if abnormal bleeding symptoms present otherwise at age of consent or prior to pregnancy' (27%), followed closely by singular advice to seek testing 'if abnormal bleeding symptoms present' (23%).

When results were separated by HTC's that treat children (paediatric or lifespan, $n = 20$) or HTC's that do not (adult-only, $n = 6$), the number of adult only responses appears low yet there are only six adult-only HTCs in Canada. Similar to the joint findings, a wide variation in testing recommendations was observed with a noticeable difference in the advice given to potential carriers. Another key finding is the different advice given to obligate and potential carriers by adult-only HTCs, where some adult-only HTCs do recommend factor level testing in early childhood to obligate carriers but no adult-only HTCs give this recommendation to potential carriers (Table 2).

Survey responses on the topic of genetic testing for potential carriers showed close to a majority of HTCs (45%) would agree to a request by the family for genetic testing of a minor if the patient was experiencing abnormal bleeding symptoms and the family understood full implications (Table 3). One-quarter of respondents (25%) would approve a genetic testing request with no conditions apart from the family understanding the full implications, while another quarter (25%) state their HTC would refuse a genetic testing request until the child is old enough to give consent.

3.2 | Experience of Canadian parents

A total of 14 parents were interviewed from four different provinces in Canada (two Western, one Central, one Eastern). There was a close to even split of obligate and potential carriership (42% vs. 58%), with the majority of families affected by haemophilia A (79%) and a smaller minority haemophilia B (21%) consistent with disease demographics (Table 4).

Most families were dissatisfied with the outcome of their testing experience (58%) with the remaining families having either mixed or positive experiences. Of the three families who were satisfied with their experience, two were families of potential carriers of haemophilia A whose daughters had factor level testing at 0–4 years of age at the recommendation of the HTC. In the third family who reported a positive experience, the mother was diagnosed with mild haemophilia A in adulthood and subsequently received testing (both factor level and genetic testing) of her two children, one male and one female who were in early adolescence. In the three families who reported mixed experiences (two obligate, one potential), families initially agreed to delay testing at the recommendation of their HTC but then subsequently

TABLE 2 Recommendations on factor level testing for young girls by type of treatment centre.

Timing recommendation for factor level testing of Obligate Carriers:	Adult-only ($n = 6$)	Paediatric or Lifespan ($n = 20$)
At least once in early childhood (< 10 years)	33% ($n = 2$)	25% ($n = 5$)
If abnormal bleeding symptoms present	17% ($n = 1$)	25% ($n = 5$)
Prior to menarche (10–14 years)	0	15% ($n = 3$)
At age of consent	0	0
COMBO: If abnormal bleeding symptoms present, prior to menarche	17% ($n = 1$)	15% ($n = 3$)
COMBO: If abnormal bleeding symptoms present, at age of consent, prior to pregnancy	33% ($n = 2$)	15% ($n = 3$)
Other: Discussion with family	0	5% ($n = 1$)
Timing recommendation for factor level testing of Potential Carriers:	**Adult-only ($n = 6$)**	**Paediatric or Lifespan ($n = 20$)**
At least once in early childhood (< 10 years)	0	20% ($n = 4$)
If abnormal bleeding symptoms present	0	30% ($n = 6$)
Prior to menarche (10–14 years)	0	10% ($n = 2$)
At age of consent	17% ($n = 1$)	5% ($n = 1$)
COMBO: If abnormal bleeding symptoms present, prior to menarche	17% ($n = 1$)	15% ($n = 3$)
COMBO: If abnormal bleeding symptoms present, at age of consent, prior to pregnancy	67% ($n = 4$)	15% ($n = 3$)
Other: Discussion with family	0	5% ($n = 1$)

TABLE 3 Recommendations around genetic testing: Paediatric or lifespan HTC only ($n = 20$).

Question: What is the usual response of your HTC when a family asks for genetic testing of a potential carrier under 18 years?	
Yes, as long as family understands full implications	25% ($n = 5$)
Yes, but only if patient is experiencing abnormal bleeding symptoms and family understands full implications	45% ($n = 9$)
No, genetic testing must wait until child can give consent	25% ($n = 5$)
Other: Discussion with family	5% ($n = 1$)

Abbreviation: HTC, Haemophilia Treatment Centre.

regretted that decision, leading to mixed feelings. In two of the families, daughters would go on to be diagnosed with mild haemophilia after delayed testing occurred close to the age of menarche. In the third family, the daughter experienced a major traumatic injury before any baseline testing had occurred leading parents to wish they had done testing earlier.

The number of daughters in each family, current age of daughters and age at first factor level testing was collected. Seventy percent of daughters were tested prior to age 10 years. The three oldest daughters captured (age 21–50 years) all had their first factor level test in adulthood with families feeling dissatisfied with their experience and wishing they received testing sooner. One daughter experienced a delayed diagnosis of mild haemophilia that did not occur until her mid-20's while the other two had their first factor level test prior to pregnancy as that was the recommendation at the time. Contrastingly, the four youngest daughters captured (age 0–5 years) all received their first factor level test at 0–4 years of age. Two of the families felt dissatisfied with their experience as they reported pushback from HTC when requesting early factor level testing, while the remaining two families had been recommended early factor level testing by their HTC and felt satisfied with their experiences.

Close to half of the daughters captured (45%) had a parent-reported factor level below normal, with another 20% reported as 'borderline'. When parents were asked their opinion on when haemophilia carriers should have testing done, parents universally responded 'as soon as possible', 'earlier' or 'as early as family would like.'

The primary result of responses regarding beliefs about haemophilia testing and decision-making factors was that families make these choices with the goal to protect and promote the health and safety of their daughters. Many families cited updated research showing girls can have haemophilia as underpinning their decision to have early-baseline factor level testing, citing it as a very logical decision in light of that information. Although promoting health and safety was consistent across all participants, secondary results were varied and were often related to each family's unique lived experience with haemophilia. A list of 10 key findings from parent experiences are outlined in Table 5.

4 | DISCUSSION

It is now increasingly recognized that all carriers of haemophilia should have early baseline factor level testing, with genetic testing

TABLE 4 Results of parent interviews.

Type of haemophilia in family (*n* = 14)	Haemophilia A	79% (*n* = 11)
	Haemophilia B	21% (*n* = 3)
Type of haemophilia carriership in family (*n* = 14)	Obligate	42% (*n* = 6)
	Potential	58% (*n* = 8)
Satisfied/dissatisfied with the outcome of testing experience and information received (*n* = 14)	Satisfied	21% (*n* = 3)
	Dissatisfied	58% (*n* = 8)
	Mixed	21% (*n* = 3)
Number of daughters in family (*n* = 14)	One	9
	Two	4
	Three	1
Total number of daughters captured: n = 20		
Current age of daughter(s) (*n* = 20)	0–4 years	20% (*n* = 4)
	5–10 years	15% (*n* = 3)
	11–15 years	35% (*n* = 7)
	16–20 years	15% (*n* = 3)
	21–30 years	5% (*n* = 1)
	30–50 years	10% (*n* = 2)
Age when daughter first had factor level testing (*n* = 20)	0-4 years	40% (*n* = 8)
	5-10 years	30% (*n* = 6)
	11–15 years	15% (*n* = 3)
	16–20 years	0
	21+ years	15% (*n* = 3)
Parent-reported factor level category (*n* = 20)	Below normal	45% (*n* = 9)
	Normal	25% (*n* = 5)
	Borderline	20% (*n* = 4)
	Unsure	5% (*n* = 1)
	Waiting on results	5% (*n* = 1)

generally performed later in adolescence/adulthood but considered earlier in certain circumstances.[20,21] Recommendations for early factor level testing for all carriers date back more than 30 years due to the increased risk of abnormally low levels, yet our results show this knowledge has not been translated into practice in Canada.[11,12] Collectively, our results demonstrate only 27% of HTCs recommend an early baseline factor level test (<10 years of age) for obligate carriers which decreases to 15% for potential carriers. Different recommendations for factor level testing are being given to obligate and potential carriers, despite there being no evidence of a difference in bleeding risk or likelihood of low factor levels with the two different types of carriership.[22]

A possible explanation for these findings is the failure to separate assessment of bleeding risk from assessment of carriership.[22] When genetic status is certain, as is the case with obligate carriers of haemophilia, parents and HCP may feel more comfortable proceeding with early factor level testing as part of bleeding risk assessment. Whereas when genetic status is uncertain as is the case with potential carriers, it has been suggested that parents and HCP may be more hesitant to proceed with early factor level testing for fear of establishing carrier status.[22,23] Additionally the historically pervasive and persistent belief that only men are affected by haemophilia, along with systemic sexism in the management of all women with bleeding disorders, likely contribute to why early assessment of bleeding risk with factor level testing is still not the universal standard of care for young girls in Canada.[24]

Our results highlight the importance of adult Haematologists/HTC's in disseminating information about bleeding risk assessment and factor level testing in young girls. For many men affected by haemophilia, this will be their primary source of information regarding testing recommendations for their obligate carrier daughters. Yet our results show that for 67% of adult-only HTCs, testing recommendations centre on the presence of abnormal bleeding symptoms in the child which is a determination that parents reported is difficult to make, particularly when there is no connection to a paediatric HTC such as with fathers of obligate carrier daughters.

Our results indicate increasing acceptance of genetic testing in minors, particularly in the context of abnormal bleeding symptoms, as almost 75% of survey respondents said their HTC would agree to a family request for genetic testing under those circumstances. Increasing evidence of abnormal bleeding, particularly heavy menstrual bleeding, in girls who are carriers of haemophilia but have normal factor levels outlines the argument for genetic testing of potential carriers as a means of assessing bleeding risk, as opposed to for carrier identification alone.[25]

Results of our parent interviews highlighted many previously documented findings in the literature such as: promoting health as the primary parental motivation for testing decisions,[15,16] difficulty differentiating normal and abnormal bleeding symptoms,[15,26] and the impact of delayed testing on diagnosis, symptom recognition, treatment and quality of life, particularly for heavy menstrual bleeding.[3,4,19,27,28] Our finding that trauma associated with blood draws was reported at many different testing timepoints, both in early childhood and adolescence, is noteworthy as fear that the blood draw will be traumatic for a very young child can be a rationale for delayed testing. A previous study also reported the impact of family history, particularly delayed diagnosis and untreated bleeding in women, which led women to advocate for testing of their female family members.[26] The perceived parental benefit of knowing a child's haemophilia carriers status, both positive and negative results, has also been previously documented with positive effects on both the parent and child.[16]

Limitations of our study include a possible selection bias as families who have strong feelings about testing in young girls due to previous negative experiences may have been more likely to respond to the recruitment call. Additionally, as only English-speaking participants were involved in the study, results may not be representative of the French-speaking parts of Canada.

TABLE 5 Key findings from parent experiences.

1. Parents seek testing to promote the health and safety of their daughters
• Many families centre their testing decisions on updated knowledge of how haemophilia affects women and girls and the desire to promote the health of their child
2. Important role of adult haematologists/HTCs in disseminating recommendations for testing in girls
• Fathers with haemophilia may have no connection to paediatric HTCs and will rely on their adult team for advice and recommendations on testing for their obligate carrier daughters
3. Families have difficulty differentiating normal versus abnormal bleeding which may lead to delays in diagnosis
• Testing recommendations that centre on the presence of abnormal bleeding symptoms places the responsibility on parents to make this determination, particularly challenging for families with no or little connection to a paediatric HTC
4. Family history of bleeding symptoms and/or delayed diagnosis of haemophilia in females may affect testing beliefs
• Families with a history of bleeding symptoms in women and girls affected by haemophilia and associated negative experiences (i.e., symptom dismissal, inadequate treatment, delayed diagnosis, decreased quality of life) may be strong advocates for early testing, both factor level and genetic
5. Negative impact of delayed testing recommendations in HTCs with limited resources or for patients in rural areas
• In HTC's with limited resources/for patients in rural areas, delayed or symptom-dependent testing recommendations can lead to delays in diagnosis and treatment due to care barriers (i.e., limited appointment availability, long testing turnover times, geographical barriers)
6. Negative impact of delayed testing for girls with heavy menstrual bleeding
• For girls with heavy menstrual bleeding, delayed testing affects access to specialized gynaecology clinics with impact on adequate treatment plans, and subsequent effects on missed schooling, quality of life, health-related sequelae such as iron deficiency
7. Trauma of blood draws across all ages
• Children across all ages (0–16 years) may experience trauma related to blood draws

5 | CONCLUSION

The current landscape of haemophilia-related testing recommendations for young girls in Canada varies widely with no clear consensus on the optimal timing of factor level or genetic testing. Our results show there are likely many girls in Canada, both potential and obligate carriers, who have been inadequately screened for bleeding risk and have not had a baseline factor level test completed. Results of our parent interviews show the negative consequences of delayed recommendations and inadequate screening. Updated, nationally affirmed testing recommendations are needed that align with research on bleeding in women and girls affected by haemophilia, such as the recently published 'European principles of care for women and girls with inherited bleeding disorders'.[20] This consensus document calls for a systematic approach to testing of all females affected by haemophilia, with baseline factor level testing in early childhood, repeat factor level testing before menarche, and genetic testing for potentially affected individuals when they are competent to understand results with considerations for testing in childhood when clinically relevant.[20] Nationally recognized recommendations such as these will assist greatly in ensuring adequate screening, yet a concerted effort is also needed to identify the women and girls already missed. Next steps could include targeted proactive haemophilia carrier screening projects by pedigree analysis such as the one recently completed in Belgium.[29] Additionally, both healthcare professionals and non-medical organizations such as haemophilia societies are needed to increase awareness of updated testing recommendations and the necessity of bleeding risk assessment for all women and girls affected by haemophilia.

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CONFLICT OF INTEREST STATEMENT

P.J. has funding from Bayer and consultancy with Star/Vega, Band/Guardian, Roche, Biomarin. M.C., J.G., E.W. and M.B. have no interests to declare.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

ETHICS APPROVAL STATEMENT

Ethics approval was granted by the Queen's University Health Sciences and Affiliated Teaching Hospitals Research Ethics Board prior to study commencement. Informed consent was obtained from all participants prior to commencing study.

ORCID

Megan Chaigneau https://orcid.org/0000-0002-8001-4351
Mackenzie Bowman https://orcid.org/0000-0003-3265-3789

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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