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# Initial Diagnosis: Understanding the Experience



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## Foreword

With recent advances in the treatment and care of people with bleeding disorders, families that receive a diagnosis are in a much better position than they would have been several decades ago.

Despite this, families still face a series of challenges during the time that their child is diagnosed with a bleeding disorder. Based on the stories that we have heard from our members, it is clear that more needs to be done to improve experiences during this time.

First and foremost, we hope that this report will provide our members with assurance that they are not alone. Many families find that having their child diagnosed with a bleeding disorder can be a very isolating and distressing experience. It is important for us to learn from each other and demonstrate that there is a community of support available.

In addition to this, we have made a set of recommendations aimed at policy makers, healthcare practitioners and safeguarding professionals. We hope that these recommendations will be taken up to improve the experience of initial diagnosis for families in the future.

Liz Carroll,

CEO, The Haemophilia Society





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## Executive Summary

This report has been compiled to provide insight into the feelings and experiences of families when they are initially diagnosed with a bleeding disorder. As well as demonstrating that parents experience an array of emotions and challenges during this time, this report also contains a set of recommendations for policy makers, healthcare practitioners and safeguarding professionals to improve this experience for families in the future.

These recommendations include amendments to current safeguarding guidelines, equality of care and consideration for girls and women, improvements to post-diagnosis information provision, greater use of community networks and peer support, and better access to emotional and psychological counselling services.

This report is based on interviews held with parents who have had a child diagnosed with a bleeding disorder. Healthcare and safeguarding professionals were also consulted during the production of this report.





## An Introduction to Bleeding Disorders

### What are genetic bleeding disorders?

Genetic bleeding disorders are a rare and complex group of conditions that cause a reduction in the blood's ability to clot efficiently. They are characterised by the deficiency or defect of certain coagulation factors or proteins in the blood. Depending on the nature of the deficiency or defect, bleeding disorders can either be severe, moderate or mild.

The most commonly recognised and diagnosed bleeding disorders in the UK are von Willebrand Disease (VWD) and Haemophilia. While VWD is caused by a defect in the concentration, structure or function of von Willebrand factor, Haemophilia A and B are caused by deficiencies in factors VIII and IX respectively.

VWD is the most common of these disorders; it is estimated that around 1 in 100 are affected. In many cases, VWD is a mild disorder so it is common for the condition to go undiagnosed. Research suggests that as many as 9 out of 10 people with mild forms of VWD are not diagnosed. In some more severe cases, however, symptoms of VWD can be similar to those associated with severe haemophilia.

Haemophilia A and B are more widely recognised, despite being less common than VWD. Haemophilia A is estimated to affect round 1: 5,000 people in the UK, while Haemophilia B is less common with only 20% of people with haemophilia estimated to have haemophilia B. People with haemophilia are predominantly male. Women who carry the defective gene are described as 'carriers' of the condition. However, women may experience symptoms and can also be diagnosed with mild haemophilia.

Both types of haemophilia have the same symptoms, but they are treated differently depending on which clotting factor is deficient. The severity of these conditions depends on the amount of the clotting factor missing, whereby the lower the factor level, the more severe the condition.

### How are people with bleeding disorders treated?

Recent developments have made a significant improvement to the way that bleeding disorders are managed.

For VWD, treatment options include desmopressin (DDAVP), VWD replacement therapy and tranexamic acid. DDAVP may also be used for the treatment of mild haemophilia A, but not for mild haemophilia B. For both moderate and severe haemophilia A and B, prophylaxis with replacement factor VIII or IX is the most common treatment option, but other agents that help clotting are also available.

These treatments act to replace the deficient or faulty clotting factor or help clotting through assisting or bypassing certain functions. The aim of this is not only to stop bleeding episodes and prevent potential long-term damage, but also to prevent the bleeding episode occurring at all.



While the treatment for people with bleeding disorders has improved considerably over the past several decades, diagnosis continues to pose challenges. The presentation of a symptomatic infant or child in a primary care setting can cause suspicion of non-accidental injury. As a result, safeguarding procedures introduced to protect vulnerable people from abuse or harm are often used to investigate these cases. Often, this delays the blood tests which confirm the presence of a bleeding disorder.

### **How is care provided for people with bleeding disorders?**

Specialist haemophilia centres across the UK are responsible for providing care for people with bleeding disorders. These centres provide services such as treatment monitoring, clinical advice, and support for home therapy programmes. These services are provided by specialist consultants, clinical nurse specialists and physiotherapists.

Some haemophilia centres are classed as Comprehensive Care Centres (CCCs) and provide wider care for people with bleeding disorders. As they have more resources, they tend to deal with more complex treatment and care issues. As well as coordinating treatment for the patient's bleeding disorder, CCCs provide services such as genetic counselling and testing, 24-hour advice, counselling and support, physiotherapy and occupational therapy, social worker support and welfare advice, specialist services, involvement in clinical research and trials, education programmes, and co-ordination for home delivery services.



## Before Diagnosis: Identifying a Problem

As a condition that is present at birth, the majority of severe bleeding disorders are diagnosed within the first few months of infancy. In situations where the parents have a known family history of genetic bleeding disorders, it is common for the child to be diagnosed during pregnancy or at birth. In these cases, parents tend to have greater awareness of bleeding disorders and their implications, often because they have older relatives that have lived with one. As a result, feelings of confusion, anxiety and shock are less common among these parents, although they can feel an element of guilt.

Louise and Gavin's son, Reggie, was diagnosed with severe haemophilia A during Louise's pregnancy. As Louise knew that she was a carrier, she was able to inform her hospital and was subsequently assigned to a haematologist who stayed with her throughout her pregnancy and labour.

*"It would have been a much bigger shock had we not known about the presence of bleeding disorders in the family. As we did, everything went as smoothly as we could have hoped."*

It is estimated that one third of people have no known family history of bleeding disorders. This could be because of a lack of diagnosis or due to a new genetic mutation in the mother or child. Parents in these instances can have a more complicated experience prior to diagnosis. This stems largely from the fact that it can take a matter of months or years for the child to show symptoms of their condition and for these to be identified as being caused by a bleeding disorder.

For children that go undiagnosed at birth, it is often only once they become mobile that symptoms of their bleeding disorder appear. Symptoms may be more or less obvious depending on the type and severity of their condition. As such, a child can have an undiagnosed bleeding disorder for up to a year and sometimes longer if the condition is milder.

During this undiagnosed period, many parents start to notice unusual bruising and bleeding that, in most cases, occur without any clear explanation. These bruises and bleeds tend to be caused by relatively minor incidents. For example, they may be bruises obtained from playing with siblings, or excessive bleeding experienced after a small cut or scrape such as an inoculation.

Parents tend to find that it is only in hindsight that these events are recognisable as symptoms of a bleeding disorder; at the time they seem strange but not necessarily alarming. Eventually, parents often take their child to their GP or to A&E when these symptoms fail to subside, or because the child experiences a more significant injury where bruising or bleeding seems to be unexpectedly excessive.

Anna-Leigh, whose son Louie was diagnosed with severe haemophilia A when he was 8 months old, had noticed bleeding prior to the injury that encouraged her to take Louie to A&E. When Louie was 2 months old he bled for five days after a minor cut on his finger. He similarly bled through his plasters and clothing after having his inoculations.

It was when he was 8 months old that Louie rolled onto a soft toy when sleeping that he acquired a significant bruise on his stomach. After this, Anna-Leigh decided to take him to A&E.

The experience of parents prior to their child being diagnosed with a bleeding disorder therefore differs based on whether they are aware of their family history or not. For those that are, the experience tends to be relatively straightforward. For those that are not, it can be a much longer and more distressing time period characterised by confusion and concern for the child's wellbeing.







## Engaging with the Healthcare System

Parents that have a known family history of bleeding disorders are generally referred to a haemophilia centre prior to their child's birth or shortly after. Once they are part of this system, parents tend to find that they receive high quality care and advice. Notwithstanding this, some parents also find that the information provided can be fairly basic, and that they would have found more meetings with specialists as well as meetings with other families more useful. Parents therefore require better access to other families in the haemophilia community who can provide advice and support.

For parents that end up in an A&E setting with no known family history of bleeding disorders and a concern regarding unexplainable bruising and bleeding in their child, the majority are confronted with child safeguarding procedures. For many parents, this is considered the most emotionally traumatic aspect of their child's diagnosis. In addition to feelings of anxiety and fear towards child abuse investigations, parents can also feel like they are being interrogated. This can cause them to feel alienated, alone and vulnerable. A greater consideration of bleeding disorders is therefore necessary within safeguarding assessments, so as to avoid situations where families are put through undue emotional trauma.

When Adele took her son to A&E after he acquired some concerning bruises, the pair were taken into a treatment room for assessment. Once safeguarding procedures were invoked, Adele and her son were separated.

*"It was total interrogation. My husband was very upset and angry when Jack was taken away from us. I had to calm him down because there was that threat hanging over us"*

At 2am, a nurse came to tell them that they were no longer being investigated for child abuse. Instead of providing assurance to the pair, the nurse merely said;  
*'You're off the hook, there's something wrong with his blood'.*

It was only when more tests were run the next morning that a proper diagnosis was received.

**Recommendation 1: NICE guidelines on when to suspect maltreatment in under 18s should be amended to be clearer on when to consider an undiagnosed bleeding disorder in cases of unexplained bruising. Language in the guidelines should be changed to describe bleeding disorders, haemophilia and VWD, rather than "causative coagulation disorders".**

In addition to distress caused by safeguarding procedures, diagnosis can also be delayed or hindered by a lack of clinical awareness of bleeding disorders and their symptoms. Parents are often turned away from A&E after being told not to worry, while others may receive an incorrect diagnosis. As a result, parents can often feel like they need to be pushy, forceful and persistent in trying to get doctors and nurses to admit their child for more comprehensive testing. This is particularly true when the child is a girl, who are often assumed to be asymptomatic carriers. In around 1/3 of cases, however, girls can have factor levels that mean they have mild haemophilia, and very rarely their levels will be more significantly reduced.



Laura, whose daughter Penelope has mild haemophilia A, found it was difficult to get doctors and nurses to listen to her concerns.

*"I found that because she was a girl, she was not necessarily taken seriously. She is always referred to as a carrier. Whilst I appreciate she is a carrier, 'symptomatic carrier' would be a better clarification."*

Laura feels that the lack of consideration given to girls with bleeding disorders has had an impact on Penelope's care and treatment.

**Recommendation 2: Girls with bleeding symptoms should receive parity of support and care to boys, and should be referred for further care and tests as required.**

Once blood tests are conducted, a diagnosis is usually received relatively quickly. After this, a haemophilia specialist is usually called to explain the nature of the bleeding disorder and what it means for the child. This can be distressing for all parents, regardless of whether they have a known family history of bleeding disorders. For those that have no history, the diagnosis in and of itself tends to mean very little as parents have often never heard of the condition. They therefore find that the information provided by doctors and nurses does little to ease their anxiety, finding that it can sometimes make the condition sound worse than it really is.

Keith found that the shock of receiving his son's diagnosis meant that he found it difficult to comprehend what it meant.

*"We felt like we were in limbo. We had the diagnosis, but didn't know what it was."*

In addition to this, Keith found that the information booklets provided by the hospital made them panic about the condition. It was the experiences of other families who had been through the same process that was more insightful and useful for Keith and his family.

Laura also found that the post-diagnosis information was inadequate.

*"It would have been beneficial to have received any information to help us understand the condition more; such as what to expect and what to look out for."*

**Recommendation 3: Parents of newly diagnosed children should be provided with better information on what the diagnosis of a bleeding disorder means for them and their child.**

Improvements to information provided by specialists are necessary, so that parents do not feel so overwhelmed and confused by their child's diagnosis. In addition, better use of community networks would provide newly diagnosed families with valuable links and peer support, therefore combatting feelings of isolation and loneliness.

**Recommendation 4: Parents of newly diagnosed children should be introduced to other families who have been through similar situations so that they can receive peer support.**



## An Experience with Social Services

James and his wife, Lucy, had their son, Scott, diagnosed with severe haemophilia A when he was 7 months old. They had taken him to A&E after he had fallen from a bed and banged his nose. Once they were at the hospital, the doctor noticed a small faded bruise on Scott that James and Lucy could not explain. Following this, child protection services were called and the family were not allowed to leave the hospital.

*"Strangely, as well as a feeling of profound fear and anxiety, I felt guilt. As if in some way I should have been able to prevent this horrible event from occurring."*

Following blood tests, it was confirmed that Scott had haemophilia and that the family were no longer being investigated.

After receiving the diagnosis and a brief explanation of the condition by a haemophilia specialist, James and his family headed home. Despite being told that they were no longer under investigation, James received a call from social services to say that they were demanding a home visit. If they were unwilling to comply, the family were threatened with police intervention.

The visit was explained as a procedure to ensure that Scott would be adequately cared for in light of his diagnosis, but James and Lucy felt that the process was unnecessary and intrusive.

*"We felt completely isolated and alone despite being surrounded by concerned family and friends."*

While Scott now receives the necessary care and treatment, the family's experience with social services was distinctly traumatic.

*"Instead of being investigated by social services, we should have been visited by another family who had gone through the same ordeal."*

*"It was right and proper that the HTC staff were acting in a professional and clinical way, but there should have been a separate counselling service that ran alongside the HTC service. It took half a year for us to be offered any psychological support."*



## What is the Ideal Scenario?

From the parents that were interviewed for this report, there were certain aspects of the diagnosis process that were highlighted as being particularly problematic. Issues were identified with current safeguarding procedures, the limited awareness of bleeding disorders among non-specialist healthcare professionals, the lack of information provided to parents at the time of diagnosis, and the lack of access to counselling services.

For parents that experienced investigation due to maltreatment concerns, many took issue with the fact that blood tests had not been conducted before this investigation. While parents understand the need for these safeguarding procedures, more clinical awareness about the signs and symptoms of bleeding disorders may help to circumvent unnecessarily distressing investigations.

Anna-Leigh said that social workers should consider bleeding disorders when investigating unexplained bruising.

*"It's not wrong to get social services involved, but it needs to be done differently."*

In a similar vein, the lack of awareness in non-specialist care settings can also cause delays in diagnosis. Again, parents often reported that they do not expect all healthcare professionals in primary care settings to be experts in bleeding disorders, but that they should have enough awareness to be able to recognise potential signs and symptoms.

In almost all cases, parents interviewed for this report found that the most useful information that they obtained during the period of initial diagnosis came from other families that had gone through the same experience. This information was deemed to be more realistic and practical than the information provided by healthcare services. As such, parents suggested that better use of the patient community would help the parents of a newly diagnosed child see that bleeding disorders are manageable.

In addition to the need for greater awareness, feelings of shock and anxiety meant that many parents felt that they would have benefitted from emotional and psychological support.

*"It would have been beneficial to have a face-to-face meeting to discuss the diagnosis. Even in terms of emotional support, it would have been beneficial to have someone to speak to."*

As such, better access to counselling services is needed to help parents through what is commonly considered the most difficult part of having a child diagnosed with a bleeding disorder.

**Recommendation 5: Access to post diagnosis counselling services should be improved so that parents can receive psychological support.**



## Recommendations

### Recommendation 1:

NICE guidelines on when to suspect maltreatment in under 18s should be amended to be clearer on when to consider an undiagnosed bleeding disorder in cases of unexplained bruising. Language in the guidelines should be changed to describe bleeding disorders, haemophilia and VWD, rather than “causative coagulation disorders”.

### Recommendation 2:

Girls with bleeding symptoms should receive parity of support and care to boys, and should be referred for further care and tests as required.

### Recommendation 3:

Parents of newly diagnosed children should be provided with better information on what the diagnosis of a bleeding disorder means for them and their child.

### Recommendation 4:

Parents of newly diagnosed children should be introduced to other families who have been through similar situations so that they can receive peer support.

### Recommendation 5:

Access to post diagnosis counselling services should be improved so that parents can receive psychological support.





## Our Services

### Newly Diagnosed Family Weekend

*"Having a baby or young child diagnosed with a bleeding disorder can leave parents feeling overwhelmed and worried about their child's care and future health. Some parents may be prepared for the diagnosis because it runs in the family but for others, the diagnosis comes with disbelief and a lot of concern. In both cases, parents can feel isolated and in need of information and support."*

The Haemophilia Society organises regular events aimed at families who have recently had a child diagnosed with a bleeding disorder. The Newly Diagnosed Weekends are filled with activity and information based sessions with physiotherapists, paediatricians and haemophilia nurses. Parents also have the opportunity to meet the Society's Youth Ambassadors, who help them to see that their children will be able to lead fun and active lives.

*"We hope that parents have the opportunity to gain the knowledge and skills they need to help their child through childhood. We hope that the weekend will help them to gain confidence in managing the impact of a bleeding disorder."*

*"While parents may come to these weekends feeling fear, shock, guilt, mistrust, annoyance, concern and worry, we hope that they leave feeling empowered, reassured, resilient, comforted and relieved."*

Rose Ozdemir  
Services Coordinator, The Haemophilia Society

The Haemophilia society also provide information and support through community events and conferences, a network of local groups, its website, Facebook page and groups, as well as information booklets and factsheets that are available in print and on the website.



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## The Haemophilia Society – For everyone affected by a bleeding disorder

We are the only UK-wide charity for all those affected by a genetic bleeding disorder; a community of individuals and families, healthcare professionals and supporters.

For more than 65 years we have campaigned for better treatment, been a source of information and support, and raised the awareness of bleeding disorders.

We want to ensure that everyone affected by a bleeding disorder:

- Lives the best life that they can
- Never feels alone or isolated
- Feels empowered and confident

We do this by:

- Raising awareness about bleeding disorders
- Providing support at all life stages
- Influencing and advocating on policy and access to treatment

More than 30,000 men, women and children in the UK have a diagnosed bleeding disorder, and the number rises every year. Membership of The Haemophilia Society is free and open to all.

Our peer support through local groups around the UK, global family network, and online community, offers friendship and a listening ear when needed, as well as enabling people to share their views and experiences. By bringing people together for information and support at events tailored to all life stages, we amplify their voices to reduce isolation and influence government, welfare and health care policy.

Our community are at the heart of everything we do – we work collaboratively with members and health professionals to ensure we make decisions influenced by their valued input and direction.

As bleeding disorders are rare, many people will never encounter The Haemophilia Society; we are largely invisible beyond the communities we serve. So we have to work doubly hard to raise both awareness and understanding of bleeding disorders and vital funds needed to give those affected the services they deserve and need to live the best life they can.

To find out more, or to become a member for free, visit our website at [haemophilia.org.uk](https://haemophilia.org.uk) or call us on **020 7939 0780**.



# THE HAEMOPHILIA SOCIETY

## Acknowledgements

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President: Baroness Meacher

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