

Identifying mental health needs in extremely rare inherited bleeding disorders (ERBD): A narrative review

 Open Access | Volume 13 (2026): Issue 1

Amy Owen-Wyard, Stacey McGeown, Emma Hallard, Helen Tate

ABSTRACT

Background:

Extremely rare inherited bleeding disorders (ERBDs) collectively represent a meaningful population requiring lifelong, multidisciplinary management. Despite the significant physical morbidity associated with factor deficiencies and platelet function disorders, the psychological burden of ERBDs is poorly characterised. Diagnostic delays, limited treatment options, and structural and interpersonal stigma contribute to substantial mental health challenges for patients and families.

Aim:

To synthesise existing evidence on the psychological and psychosocial impacts of ERBDs, identify gaps in mental health provision, and outline implications for clinical practice, service development, and policy.

Methods:

A narrative review was conducted to explore the psychosocial and mental health needs of individuals living with ERBDs. Relevant peer-reviewed literature published up to March 2026 was identified and subjected to thematic analysis, facilitating the identification of recurrent themes pertaining to psychological distress, quality of life, social functioning, stigma, caregiver burden, and access to support services.

Results:

Available evidence exploring psychosocial outcomes in ERBDs was limited. Seven primary studies were identified that examined psychological wellbeing, mental health, health-related quality of life, or psychosocial experiences in individuals with ERBDs and similar bleeding disorder populations. People with ERBDs can experience anxiety, depression, low self-esteem, trauma, and social isolation. Recurrent bleeding, chronic pain, and prolonged diagnostic pathways may compound psychological distress. Available evidence highlighted that routine mental health screening is inconsistently incorporated into ERBDs care pathways.

Authors:

AMY OWEN-WYARD

EquiPath Analytics Limited, Worcester, UK; Herefordshire and Worcestershire Health and Care NHS Trust, Worcester, UK
Email: amy@epalimited.com

STACEY MCGEOWN

EquiPath Analytics Limited, Worcester, UK; Queen's University Belfast, Northern Ireland

EMMA HALLARD

EquiPath Analytics Limited, Worcester, UK; Black Country Healthcare NHS Foundation Trust, Wolverhampton, UK

HELEN TATE

EquiPath Analytics Limited, Worcester, UK

Conclusion:

Mental health is an under-recognised yet critical component of ERBD management. Strengthening multidisciplinary models, embedding psychological assessment into routine care, addressing stigma, and expanding research are essential to improving outcomes and ensuring equitable, patient-centred care for individuals and families affected by ERBDs.

PLAIN LANGUAGE SUMMARY

Extremely rare inherited bleeding disorders are lifelong conditions in which the blood does not clot normally. They include shortages of particular clotting proteins and disorders affecting platelets, the blood cells that help stop bleeding. People may experience frequent bruising or nosebleeds, heavy periods, painful joint or muscle bleeds, or serious internal bleeding. Because each condition affects very few people, diagnosis can take years, specialist knowledge may be limited, and someone may never meet another person with the same disorder.

This review was needed because care and research in extremely rare bleeding disorders have mainly focused on controlling bleeding, while the emotional and social effects have received much less attention. The authors brought together published studies, health statistics, guidance and patient-organisation reports. They asked how these disorders affect mental health and everyday life, how delayed diagnosis and stigma add to the burden, what happens when support is missing, and how care could be improved.

The evidence showed that repeated bleeding, pain, uncertainty and limited treatment options can disrupt education, work, relationships and social activities. Anxiety, depression, low self-esteem and isolation were commonly reported, and caring responsibilities also affected families. In one study of Glanzmann thrombasthenia, 30% of participants met criteria for depression and 26% had low self-esteem. More frequent or severe bleeding was linked with poorer quality of life. However, routine mental health checks were uncommon, studies used different measures, and very little research examined these disorders specifically. The available studies were also generally small and could not show how wellbeing changes over time.

These findings matter because good care for extremely rare bleeding disorders should address emotional wellbeing as well as bleeding. Earlier diagnosis, regular mental health screening, access to trained professionals, peer support and better support for caregivers could reduce distress and help people manage treatment and daily life. More long-term, patient-led research is needed so that services reflect the real needs of people living with these rare disorders.

Keywords:

Extremely rare inherited bleeding disorders, Mental health, Psychosocial impact, Stigma, Diagnostic delay, Anxiety, Depression, Caregiver burden, Multidisciplinary care, Rare diseases, Patient-centred care.

BACKGROUND

Rare diseases, though individually uncommon, collectively affect more than 300 million people globally and approximately 3.5 million people in the UK, or roughly 1 in 17 individuals [1]. Mental health challenges are widely reported across rare disease populations, with more than 90% of patients reporting mental health-related difficulties and more than one third reporting suicidal thoughts [2]. Extremely rare bleeding disorders (ERBDs), a subset of inherited bleeding disorders, sit within this wider rare disease context and share many of its diagnostic, clinical, and psychosocial challenges [3].

For the purposes of this review, ERBDs refer predominantly to severe congenital coagulation factor deficiencies and severe autosomal recessive platelet function disorders, each with an estimated prevalence of fewer than one affected individual per million population. These disorders encompass severe deficiencies of fibrinogen and coagulation factors II, V, X, XI, and XIII, as well as severe inherited platelet function disorders, including Glanzmann thrombasthenia and Bernard-Soulier syndrome. Prevalence varies according to disorder subtype, genotype, geographic region, and consanguinity patterns [4]. Some disorders, including Factor VII and Factor XI deficiency, may occur more frequently in milder or heterozygous forms; they are discussed in this review primarily in relation to severe phenotypes associated with substantial bleeding burden and specialist multidisciplinary care requirements.

Together, ERBDs represent a clinically significant population requiring lifelong, multidisciplinary management [4,5,6]. The number of individuals registered with bleeding disorders in the UK has more than doubled in the past two decades, primarily due to enhanced identification of ERBDs and bleeding disorders of unknown cause (BDUC), which now account for up to 40% of all cases [7]. The prevalence of bleeding disorders in the UK is presented in **Table 1**.

TABLE 1. NUMBER OF PEOPLE WITH BLEEDING DISORDERS IN THE UK BY REGISTERED DIAGNOSIS

Data from the UK Haemophilia Centre Doctor's Organisation (UKHCDO) annual report and bleeding disorder statistics (April 2024-March 2025) [7]. Females registered as carriers of haemophilia A or B, or with FVIII or FIX deficiency, are reported by their factor level.

CASE/PARAMETER	NO. OF PEOPLE REGISTERED, BY SEX AS REGISTERED AT BIRTH		
	MALE	FEMALE	TOTAL
Haemophilia A	6,898	0	6,898
Female with <40 IU/dl FVIII	0	943	943
Female with ≥40 IU/dl FVIII	0	2,066	2,066
Haemophilia B	1,372	0	1,372
Female with <40 IU/dl FIX	0	376	376
Female with ≥ IU/dl FIX	0	423	423
Factor IX Leyden deficiency	31	0	31
Factor IX Leyden deficiency carrier	0	19	19
Haemophilia A/B with liver transplant	12	0	12
von Willebrand disease	4,487	7,902	12,389
Probable von Willebrand disease	80	193	273
Factor V deficiency	111	221	332
Factor VII deficiency	1,193	1,416	2,609
Factor X deficiency	166	230	396
Factor XI deficiency	1,850	2,769	4,619
Factor XIII deficiency	60	51	111
Prothrombin (Factor II) deficiency	10	15	25
Dysfibrinogenemia	360	578	938
Hypofibrinogenemia	146	208	354
Hypodysfibrinogenemia	28	30	58
Afibrinogenemia	11	7	18
Bleeding disorder of unknown cause	226	1,655	1,881
Glanzmann thrombasthenia	74	98	172
Bernard-Soulier syndrome	64	75	139
Platelet-type pseudo von Willebrand disease	16	30	46
Other platelet disorder	1,330	2,909	4,239
Combined Factors V and VIII deficiencies	16	20	36
Other disorders*	4	7	11
Acquired haemophilia A	375	356	731
Acquired von Willebrand disease	126	101	227
Other acquired factor deficiency	28	26	54
Miscellaneous/multiple disorders	285	608	893
Total	19,359	23,332	42,691

* Other reported disorders include: Thrombomodulin-associated coagulopathy; Combined factors II, VII, IX and X deficiencies; Alpha 2-antiplasmin deficiency; Reduced one stage FVIII and normal chromogenic FVIII.

Individuals with ERBDs frequently endure a prolonged diagnostic journey, characterised by misdiagnosis, uncertainty, and considerable emotional distress [8]. These difficulties are further exacerbated by structural and interpersonal stigma, often leading to isolation, anxiety, and depression. Misdiagnosis is particularly prevalent among women and adolescents, imposing a substantial psychological burden on affected individuals [9]. Families similarly bear emotional, financial, and social burdens, frequently navigating fragmented care systems with insufficient support [10]. Although rare in isolation, these disorders share common challenges, including poor mental health, risk of severe bleeding episodes, and the need for specialised, lifelong care [11]. BDUC adds further complexity, as affected individuals present with abnormal bleeding symptoms despite normal results on standard haemostatic tests [12]. This diagnostic ambiguity can delay appropriate care and further exacerbate psychological distress [13].

People with ERBDs may experience life-threatening symptoms, although presentation varies according to the specific disorder and underlying genetics. Common symptoms include mucocutaneous bleeding, bruising, epistaxis, gum bleeding, and prolonged bleeding following minor trauma or medical procedures, as well as joint bleeds, gastrointestinal haemorrhage, and central nervous system bleeding [8]. Females with ERBDs face heightened health risks, including menorrhagia and obstetric complications such as postpartum haemorrhage, and, in some cases, an increased likelihood of miscarriage and placental abruption [14,15]. Neonates may also be affected if the bleeding disorder is inherited, placing them at risk of haemorrhagic complications during delivery [16]. While rare, intracranial haemorrhage is a life-threatening complication primarily observed in newborns and infants with ERBDs [17,18].

Untreated acute bleeds in ERBDs can result in rapid and potentially irreversible complications. Joint bleeds may cause intense pain, swelling, and permanent joint damage [19,20]; muscle bleeds can lead to compartment syndrome, nerve compression, or tissue necrosis; more critical events such as intracranial or gastrointestinal bleeding can be fatal [21]. Even seemingly minor mucosal bleeds, such as oral or nasal bleeding, may become unmanageable or threaten airway compromise if prolonged [22]. Inadequate treatment of acute bleeds not only increases immediate risk but may also worsen long-term outcomes, including loss of mobility, chronic pain, anaemia, and psychological trauma [23].

The exceptional rarity of these conditions often results in individuals never meeting others with the same diagnosis, thereby fostering profound social isolation and negatively impacting overall wellbeing [2]. This is especially challenging for children and adolescents, who may struggle to comprehend their condition and feel excluded from peer groups [24]. The emotional burden is intensified by the daily realities of managing a chronic, frequently misunderstood condition within healthcare systems that may not be sufficiently equipped to respond [13].

This review aims to synthesise existing evidence on the psychological and psychosocial impacts of ERBDs, identify gaps in mental health provision, and outline implications for clinical practice, service development, and policy.

METHODS

A narrative literature review was undertaken to investigate the mental health needs and psychosocial experiences of individuals living with ERBDs. Narrative reviews are well suited to under-researched areas as they enable the integration of diverse evidence and the generation of new insights within both emerging and established fields of inquiry [25]. The review focused on the psychological and psychosocial consequences of ERBDs, including anxiety, depression, emotional wellbeing, health-related quality of life, stigma, social participation, caregiver burden, and access to psychosocial support services.

Search strategy

Relevant peer-reviewed literature published in English up to March 2026 was identified through comprehensive searches of PubMed, Scopus, CINAHL, and Web of Science. Search terms encompassed concepts related to ERBDs and psychosocial outcomes, including specific disorder names, coagulation factor deficiencies, platelet function disorders, and key mental health constructs such as anxiety, depression, quality of life, stigma, diagnostic delay, and caregiver burden. The screening, selection, and management of records were facilitated using the Covidence systematic review platform.

To provide contextual insight into service provision and broader healthcare challenges within the rare bleeding disorders field, selected grey literature, including clinical guidance documents, organisational reports, conference proceedings, and policy resources, was reviewed. Patient advocacy and informational materials were used exclusively to identify and describe publicly available support services and community initiatives and were not considered primary sources of evidence for scientific or clinical interpretation.

Eligibility criteria

Studies were eligible for inclusion if they examined psychological or psychosocial outcomes relevant to individuals living with ERBDs. Outcomes of interest included anxiety, depression, emotional wellbeing, health-related quality of life, stigma, social isolation, experiences of diagnosis and diagnostic delay, caregiver burden, and access to psychological support services.

Owing to the scarcity of ERBD-specific literature, evidence from broader inherited bleeding disorder populations was also considered where it enhanced understanding of patient experiences and informed clinical practice or service provision. In studies comprising participants with varying levels of disease severity, emphasis was placed on findings relating to severe phenotypes and a significant bleeding burden, reflecting the characteristics most relevant to ERBDs. This inclusive approach was intended to balance specificity with feasibility. Where evidence was drawn from broader inherited bleeding disorder populations, findings were interpreted cautiously and used to illuminate themes likely to be relevant to ERBDs, rather than to imply direct equivalence across all bleeding disorder groups.

Data extraction and synthesis

The included literature was synthesised using an inductive thematic approach. This method supports the identification and organisation of recurring patterns within heterogeneous evidence, facilitating a more comprehensive interpretation of findings across studies [26]. Through iterative analysis, key themes were identified relating to diagnostic experiences, healthcare-related stigma and validation, bleeding-related psychosocial burden, caregiver impacts, and access to mental health support. These themes emerged from the data rather than being specified a priori, and the resulting synthesis was used to inform consideration of clinical implications and highlight areas requiring further research attention.

Scope and limitations of the approach

As a narrative review, this study was designed to provide a broad synthesis of the existing literature rather than a systematic assessment of the evidence. Formal quality appraisal and risk-of-bias evaluation were therefore beyond the scope of the review. The available evidence is characterised by substantial heterogeneity and a reliance on predominantly cross-sectional study designs, which constrain the ability to draw robust causal inferences or generalise findings across diverse ERBD populations. The conclusions presented should therefore be viewed as indicative of current knowledge and key evidence gaps, while highlighting areas requiring further empirical investigation.

RESULTS

The evidence was organised into recurring psychosocial themes rather than by disorder subtype, as the included studies varied substantially in design, sample characteristics, and outcome measures. This thematic structure allows common experiences across ERBDs and closely related inherited bleeding disorder populations to be considered together, while recognising the limitations of drawing broad conclusions from a small and heterogeneous evidence base.

The findings are presented below as an integrated narrative synthesis, moving from the scope of the evidence base to the recurring psychosocial themes identified across studies. This structure reflects the limited number of ERBDs specific studies and the need to draw cautiously on closely related inherited bleeding disorder populations.

Seven primary studies exploring psychosocial outcomes, mental health, or health-related quality of life in ERBDs or closely related inherited bleeding disorder populations were identified [27-33]. These included quantitative surveys, qualitative interview studies, and mixed-methods investigations involving Glanzmann thrombasthenia, hereditary Factor X deficiency, Factor VII deficiency, and broader inherited bleeding disorder cohorts. Key study characteristics are summarised in **Table 2**.

Across the included studies, recurring themes included psychological distress, recurrent bleeding-related anxiety, social isolation, educational and occupational disruption, stigma within healthcare encounters, caregiver burden, and unmet psychosocial support needs.

The reviewed studies suggest a substantial psychosocial and mental health burden among individuals with ERBDs, although the evidence base remains limited and heterogeneous. Across both factor deficiencies and platelet function disorders, recurrent bleeding episodes, chronic pain, and delayed diagnosis were frequently associated with psychological distress, including symptoms of anxiety, depression, and reduced self-esteem. In Glanzmann thrombasthenia, qualitative reports described severe disruption to education and social life [28], while the Glanzmann's 360 study reported that 30% of participants screened positive for depressive symptoms and 26% reported low self-esteem, with bleeding severity associated with poorer quality-of-life outcomes [29]. Similar findings were observed among individuals with Factor X deficiency and broader inherited bleeding disorder populations, with surveys reporting reduced wellbeing across educational, occupational, and social domains, alongside moderate caregiver burden [27].

High bleeding frequency emerged as a recurring factor associated with poorer psychosocial outcomes. Evidence from multiple studies suggests that recurrent bleeding episodes can disrupt education, employment, and social participation, potentially contributing to poorer psychosocial wellbeing [27-30]. Participants in two studies reported frequent bleeding episodes, substantial school absenteeism, and impaired daily functioning, highlighting

the broader impact of ERBDs on quality of life [30,31]. Studies involving people with Glanzmann thrombasthenia and congenital Factor VII deficiency also identified educational, occupational and social effects associated with recurrent bleeding. Although both studies reported a substantial psychosocial burden, many respondents expressed satisfaction with their disease management or available personal support [32,33].

TABLE 2. CHARACTERISTICS OF STUDIES INCLUDED IN THE NARRATIVE REVIEW

STUDY	COUNTRY	CONDITION(S)	PARTICIPANTS	METHODOLOGY
Branchford et al. (2024) – Hereditary factor X deficiency in America survey [27]	United States	Hereditary Factor X deficiency	30 adult patients and 38 caregivers, age range 2-65 years	Prospective cross-sectional survey study
Khair et al. (2024) – One day at a time: Life with Glanzmann thrombasthenia [28]	UK / International	Glanzmann thrombasthenia	14 adult patients (aged 22-78 years) and 5 caregivers (children aged 2-7 years)	Mixed-methods study using online quantitative survey and qualitative interviews
Khair et al. (2024) – Bleeding and quality of life in people with Glanzmann thrombasthenia: insights from the Glanzmann's 360 study [29]	International	Glanzmann thrombasthenia	88 adult patients and 29 caregivers of children, age range <2 years to >70 years	Mixed-methods study using online quantitative survey and qualitative interviews
Koekemoer et al. (2022) – Mental health disorders and health-related quality of life in adults with inherited bleeding disorders in central South Africa [30]	South Africa	Inherited bleeding disorders including rare bleeding disorders	38 adult patients (aged 18-65 years) attending a specialist inherited bleeding disorders clinic	Cross-sectional observational study
van Gastel et al. (2025) – A qualitative study on the impact of bleeding disorders on the daily life of Dutch women [31]	Netherlands	Congenital bleeding disorders including rare factor deficiencies and platelet disorders	28 women, aged 18-40 years	Qualitative study
Peltier et al. (2020) – Psychosocial impact and disease management in patients with Factor VII deficiency [32]	International	Congenital Factor VII deficiency	45 respondents: 25 adult patients (aged 18-67 years) and 20 caregivers (children aged 5-17 years)	Cross-sectional survey study
Duncan et al. (2020) – Disease burden in patients with Glanzmann thrombasthenia: Perspectives from the Glanzmann Thrombasthenia Patient/Caregiver Questionnaire [33]	International	Glanzmann thrombasthenia	45 respondents: 24 adult patients (aged 18-61 years) and 21 caregivers (children aged 1-17 years)	Cross-sectional patient/caregiver questionnaire study

STUDY	MEASURES USED	KEY MENTAL HEALTH / PSYCHOSOCIAL FINDINGS
Branchford et al. (2024) – Hereditary factor X deficiency in America survey [27]	Hemophilia Well-Being Index, Menorrhagia Impact Questionnaire, QoL and caregiver burden survey items	Participants reported impaired school/work participation, reduced wellbeing, social limitations, treatment burden, and significant caregiver burden. Delayed diagnosis and ongoing bleeding negatively affected psychosocial wellbeing.
Khair et al. (2024) – One day at a time: Life with Glanzmann thrombasthenia [28]	Semi-structured interviews; inductive thematic analysis	Participants described anxiety surrounding bleeding episodes, social isolation, emotional exhaustion, educational/work disruption, invisibility of illness, and unmet psychosocial support needs. Women reported significant menstrual and reproductive burden.
Khair et al. (2024) – Bleeding and quality of life in people with Glanzmann thrombasthenia: insights from the Glanzmann's 360 study [29]	EQ-5D-5L, PHQ-8, Rosenberg Self-Esteem Scale, Self-Efficacy to Manage Chronic Disease Scale, Menstrual Impact Questionnaire	30% screened positive for symptomatic depression and 26% reported low self-esteem. Frequent bleeding was associated with poorer quality of life, fatigue, educational disruption, and reduced social participation.
Koekemoer et al. (2022) – Mental health disorders and health-related quality of life in adults with inherited bleeding disorders in central South Africa [30]	Structured psychiatric assessment and HRQoL measures	High rates of depression, anxiety, and alcohol use disorders were identified alongside impaired health-related quality of life. Mental health difficulties were associated with chronic disease burden and bleeding severity.
van Gastel et al. (2025) – A qualitative study on the impact of bleeding disorders on the daily life of Dutch women [31]	Focus groups and semi-structured interviews; thematic analysis	Women described stigma, healthcare dismissal, shame, insecurity, delayed diagnosis, and disruption to education, employment, relationships, and social activities. Participants highlighted unmet mental health and peer-support needs.
Peltier et al. (2020) – Psychosocial impact and disease management in patients with Factor VII deficiency [32]	Disease management and psychosocial impact questionnaires	Participants reported anxiety regarding future bleeding episodes, treatment burden, emotional distress, and restriction of daily activities. The study highlighted the psychosocial impact of living with an unpredictable rare bleeding disorder.
Duncan et al. (2020) – Disease burden in patients with Glanzmann thrombasthenia: Perspectives from the Glanzmann Thrombasthenia Patient/Caregiver Questionnaire [33]	Disease burden and psychosocial impact questionnaire	Participants reported substantial psychosocial and functional burden associated with recurrent bleeding episodes, including disruption to education, work, social participation, and daily activities. Frequent bleeding resulted in school absenteeism, emotional distress, fatigue, and caregiver burden. Findings highlighted the significant impact of Glanzmann's on quality of life and everyday functioning.

DISCUSSION

The discussion below considers these findings in relation to four interrelated areas:

- The psychological burden of living with ERBDs
- The inconsistent assessment of mental health and wellbeing
- The role of stigma and access barriers
- Implications for multidisciplinary care.

Psychological burden beyond bleeding

The available evidence suggests that psychosocial burden in ERBDs extends beyond the direct physical consequences of bleeding. Across the reviewed studies, individuals reported symptoms of anxiety, depression, reduced self-esteem, social isolation, and impaired quality of life. These outcomes appeared to be associated not only with bleeding frequency but also with the cumulative impact of living with a chronic, unpredictable condition.

A recurring theme across studies was the need for continuous adaptation and vigilance. Participants frequently described modifying educational, occupational, and social activities because of concerns about future bleeding episodes. Many also reported avoiding certain physical activities, restricting travel, planning daily routines around treatment requirements, or limiting participation in social events due to fears of injury or uncontrolled bleeding. Such adaptations may be necessary from a clinical perspective; however, they can also contribute to uncertainty, reduced autonomy, and social exclusion. This suggests that psychological distress in ERBDs may be driven not solely by acute bleeding events but also by the persistent anticipation of future bleeds [34]. Unlike acute episodes of illness, ERBDs often require individuals to continuously assess and manage potential threats in everyday situations [35]. Such findings align with broader theories of chronic illness adaptation, where perceived unpredictability and loss of control are recognised contributors to psychological distress [36].

Consequently, the psychosocial burden in ERBDs should be understood as an integral component of the disease experience, rather than a secondary consequence of physical symptoms alone. Supporting individuals to develop confidence, coping strategies, and self-management skills may help mitigate the psychological impact of uncertainty while promoting participation in education, employment, and social life.

Lack of standardised mental health screening tools

Despite increasing recognition of the psychosocial challenges experienced by individuals with ERBDs, the assessment of mental health and wellbeing remains inconsistent across the literature. Existing research has predominantly focused on clinical manifestations, treatment efficacy, and haemostatic outcomes, with comparatively limited attention given to psychological and psychosocial dimensions of living with these conditions. The measures used to evaluate psychosocial outcomes vary considerably and include condition-specific quality-of-life instruments, qualitative interviews, and a range of validated psychological assessment tools. This methodological heterogeneity limits comparability across studies and makes it difficult to establish the true prevalence, severity, and nature of psychological difficulties within ERBDs populations [37].

The absence of standardised and validated approaches to assessing mental health outcomes presents a significant challenge for both research and clinical practice. Inconsistent measurement impedes comparisons across studies, restricts the accumulation of a coherent evidence base, and hinders the development and evaluation of targeted psychosocial interventions for individuals with ERBDs [37]. This gap may, in part, reflect the historical emphasis within the field on haemostatic outcomes, morbidity, and mortality, which have traditionally been regarded as the primary indicators of disease burden and treatment success [38]. Although these remain critical clinical considerations, an exclusive focus on physical outcomes risks overlooking the broader emotional, psychological, and social impact of ERBDs on everyday life.

Furthermore, the lack of routine psychological assessment within both research and clinical settings may contribute to the under-recognition of psychological distress and unmet psychosocial needs. As aspects of health that are systematically measured are often those most likely to be identified and addressed in clinical care, the limited incorporation of mental health assessment into existing care pathways may inadvertently marginalise important dimensions of patient wellbeing [39].

Greater integration of standardised psychosocial and mental health measures into both research and clinical practice may be necessary to ensure a more holistic understanding of the lived experience of ERBDs, and to support the development of comprehensive, patient-centred care.

Stigma and access to services

Stigma has been described as a social process through which individuals are devalued, marginalised, or excluded on the basis of perceived differences, often resulting in discrimination and social disadvantage [40]. For individuals living with ERBDs, the psychosocial burden associated with the condition may be further compounded by stigma surrounding mental health, which continues to represent a significant barrier to seeking and accessing appropriate psychological support [41]. Several of the studies included in the review identified challenges associated with living with a condition that is both rare and frequently invisible to others. Participants reported experiences of delayed diagnosis, limited awareness and understanding among healthcare professionals, and the need to repeatedly explain or justify their symptoms and healthcare needs. Women described difficulties related to the normalisation, minimisation, or dismissal of heavy menstrual bleeding and reproductive health concerns. Such experiences have been associated with feelings of frustration, self-doubt, and diminished trust in healthcare services [42,43].

The nature of stigma within ERBDs populations may differ from traditional conceptualisations of disease-related stigma, which often focus on overt forms of discrimination. Instead, the psychosocial burden experienced by individuals with ERBDs appears to arise more commonly from experiences of invalidation, misunderstanding, and lack of recognition within healthcare and broader social contexts [38]. These findings suggest that improving awareness and understanding of ERBDs among healthcare professionals may serve not only as a means of facilitating earlier diagnosis and appropriate clinical management but also as an important psychosocial intervention.

Furthermore, the limited availability of comprehensive data on mental health outcomes in ERBDs populations constrains efforts to advocate for service development and inform policy initiatives aimed at addressing psychosocial needs [40]. Earlier recognition of symptoms, validation of patient experiences, and improved access to specialist multidisciplinary support may help alleviate some of the emotional burden associated with navigating healthcare systems and living with a poorly understood condition. Addressing these challenges will be essential to the delivery of more holistic, person-centred care and to improving psychosocial outcomes for individuals affected by ERBDs.

Implications for multidisciplinary care

The delivery of holistic, patient-centred care for individuals with ERBDs requires the integration of psychosocial support within multidisciplinary care models [44]. While psychologists, mental health nurses, and social workers play important roles in addressing psychological wellbeing, psychosocial care is a shared responsibility across the multidisciplinary team. Specialist haemophilia nurses often occupy a central position in comprehensive care, contributing to patient education, emotional support, care coordination, advocacy, and the early identification of psychosocial difficulties [45]. Existing haemophilia nursing competency frameworks developed across the United Kingdom and Europe highlight communication skills, psychosocial assessment, family support, and multidisciplinary collaboration as essential components of high-quality bleeding disorder care [46]. These established models may provide a valuable foundation for enhancing psychosocial support pathways within ERBD services, particularly in settings where access to dedicated psychological services is limited.

Strengthening healthcare professionals' understanding of the psychological and social dimensions of ERBDs is also likely to improve the quality of care provided. Training in trauma-informed approaches and the psychosocial aspects of living with ERBDs may enhance clinical competence, foster more empathetic patient interactions, and enable more effective identification of support needs [47]. Incorporating these principles into clinical guidance and care frameworks may help ensure that psychosocial wellbeing is considered a routine and integral component of ERBDs management rather than an adjunct to physical care [20].

Routine assessment of emotional wellbeing, social participation, family functioning, and caregiver concerns may facilitate the earlier identification of psychosocial difficulties and promote more timely access to appropriate support. Such an approach may also help normalise conversations surrounding mental health within bleeding disorder services, reducing barriers to disclosure and help-seeking. Importantly, the available evidence suggests that psychosocial interventions should not focus solely on affected individuals. Given the substantial emotional, practical, and financial challenges often experienced by families, support services should adopt a family-centred approach that recognises the broader impact of ERBDs on caregivers and family wellbeing. Embedding psychosocial care throughout multidisciplinary services may therefore contribute to improved quality of life, better patient experiences, and more comprehensive long-term management of ERBDs.

Strengths and limitations

This review contributes to a relatively underexplored area of bleeding disorder research by synthesising the emerging evidence relating to psychosocial wellbeing, mental health, and the lived experiences of individuals with ERBDs. Whereas much of the existing literature has focused on diagnosis, bleeding phenotype, and treatment outcomes, this review highlights the broader psychological and psychosocial dimensions of living with these conditions. By integrating evidence from quantitative, qualitative, and mixed-methods studies, it provides a comprehensive overview of the current state of knowledge while identifying important gaps in research, clinical practice, and service provision.

However, several limitations should be acknowledged. Most notably, there is a lack of studies specifically designed to investigate mental health outcomes in ERBD populations. Although psychosocial outcomes are reported in several surveys, observational studies, and reviews, they are frequently examined as secondary outcomes within broader clinical investigations. As a result, conclusions regarding the mental health needs of individuals with ERBDs are necessarily drawn from a limited and heterogeneous body of evidence.

Research in ERBD populations is also constrained by the rarity of these disorders, which inherently limits sample sizes. Most available studies and patient surveys involve fewer than 100 participants. While these studies provide valuable preliminary insights, small sample sizes reduce statistical power and limit the capacity to detect meaningful associations or differences between patient subgroups [48]. Consequently, findings may not be fully representative of the wider ERBD population, particularly individuals with the rarest disorder subtypes, thereby limiting the generalisability of conclusions relating to psychosocial burden and mental health outcomes.

A further limitation of the evidence base is its reliance on predominantly cross-sectional study designs. Such studies provide only a snapshot of patient experiences and are unable to capture changes in psychological wellbeing over time. Furthermore, they do not allow evaluation of the long-term effects of treatment advances, variations in bleeding severity, or psychosocial interventions on mental health outcomes [49]. The lack of prospective longitudinal research therefore limits understanding of how psychosocial challenges evolve across the lifespan and in response to changing clinical circumstances, highlighting an important area for future investigation.

Finally, substantial variability exists in the psychosocial outcome measures used across studies. The absence of standardised and validated assessment tools specifically developed for ERBD populations restricts direct comparison between studies and complicates efforts to establish the prevalence, severity, and nature of psychological difficulties within this patient group. Greater consistency in the assessment and reporting of psychosocial outcomes will be essential for strengthening the evidence base and informing the development of targeted interventions. Collectively, these limitations underscore the need for cautious interpretation of the available evidence while reinforcing the importance of further ERBD specific research focused on mental health and psychosocial wellbeing.

Taken together, these limitations highlight the need for research that is more longitudinal, methodologically consistent, and inclusive of the diverse populations affected by ERBDs. They also point directly to the priorities required to strengthen the evidence base and support more equitable care.

Future research

Addressing the current evidence gaps surrounding mental health and psychosocial wellbeing in ERBDs should be a priority for future research. Longitudinal studies are needed to examine psychological outcomes over time and to evaluate the influence of treatment advances, bleeding burden, and psychosocial interventions on wellbeing. Research adopting an intersectional perspective would further enhance understanding of how demographic and social factors, including gender, age, ethnicity, and socioeconomic status, shape patient experiences and mental health outcomes.

Future studies should also investigate the effectiveness, accessibility, and cultural appropriateness of digital mental health interventions designed for rare disease populations. In addition, greater inclusion of women, adolescents, caregivers, and underrepresented patient groups will be essential to ensuring that future findings accurately reflect the heterogeneity of ERBDs experiences. Strengthening the evidence base in these areas will support the development of targeted psychosocial interventions and help establish mental wellbeing as a core component of comprehensive ERBDs care [50,51,52].

CONCLUSIONS

The available evidence suggests that individuals living with ERBDs may experience a substantial psychosocial burden arising from recurrent bleeding episodes, diagnostic uncertainty, delayed diagnosis, social restrictions, and the wider impact of the condition on caregivers and family members. Emerging research has identified associations between ERBDs and adverse psychological outcomes, including anxiety, depressive symptoms, reduced self-esteem, and impaired quality of life. However, the current evidence base remains limited by small sample sizes, methodological heterogeneity, and a predominance of cross-sectional study designs, meaning that these findings should be interpreted as indicative rather than definitive.

Importantly, the psychosocial impact of ERBDs appears to be influenced not only by diagnostic status but also by factors such as bleeding burden, disease severity, and experiences within healthcare systems. Collectively, the findings of this review indicate that mental wellbeing should be recognised as an integral component of comprehensive ERBDs care, alongside the prevention and management of bleeding complications and other physical manifestations of disease. Multidisciplinary models of care involving specialist nurses, physicians, psychologists, social workers, and other support services may facilitate earlier identification of psychosocial needs and provide more effective support for both patients and caregivers.

Despite increasing recognition of these issues, significant gaps remain in understanding the long-term psychological consequences of living with ERBDs and the effectiveness of interventions designed to address psychosocial needs. Future research should prioritise prospective longitudinal studies, the development and validation of psychosocial assessment tools tailored to ERBD populations, and the evaluation of integrated models of multidisciplinary care. Greater incorporation of patient and caregiver perspectives will also be essential to ensuring that future research, policy development, and service provision reflect the full complexity of living with an extremely rare inherited bleeding disorder. Ultimately, holistic ERBD care will require psychosocial wellbeing to be given the same sustained attention as physical health outcomes within research, policy, and specialist services.

ACKNOWLEDGEMENTS

Author contributions

AOW was a major contributor to writing the manuscript. SM reviewed the article. HT provided input as a lived experience expert. EH reviewed the manuscript as a mental health expert. All authors read and approved the final manuscript.

Conflict of interest

The authors have advised no interests that might be perceived as posing a conflict or bias

Informed consent

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

Data statement

This article is a narrative review based on previously published studies. No new datasets were generated or analysed.

Use of AI

AI-assisted tools were used during the conduct and preparation of this manuscript. Covidence was used to support the organisation and screening of literature during the review process. In addition, AI tools were used to assist with organising and refining the structure and flow of the manuscript. AI was not used for data analysis, interpretation of findings, or generation of the scientific content, and all outputs were reviewed and approved by the authors.

ORCID

Amy Owen-Wyart: <https://orcid.org/0009-0008-7468-4042>

Stacey McGeown: <https://orcid.org/0009-0006-6195-5590>

REFERENCES

1. Department of Health and Social Care. UK rare diseases framework [Internet]. London: Department of Health and Social Care; 2021. Available from: <https://www.gov.uk/government/publications/uk-rare-diseases-framework> (accessed March 2025).
2. Zhang Z. Diagnosing rare diseases and mental well-being: a family's story. *Orphanet J Rare Dis*. 2023; 18: 45. doi: 10.1186/s13023-023-02648-y.
3. The Haemophilia Society. Rare bleeding disorders [Internet]. London: The Haemophilia Society; 2024. Available from <https://haemophilia.org.uk/bleeding-disorders/rare-bleeding-disorders/> (accessed June 2025).

4. Davidson S, Gomez K. Laboratory and molecular diagnosis of Factor XI deficiency. *Semin Thromb Hemost.* 2025; 51(2): 145-154. doi: 10.1055/s-0044-1792033.
5. Yoon HS, Han Y, Kim YJ, et al. Epidemiology of congenital bleeding disorders: a nationwide population-based Korean study. *J Korean Med Sci.* 2020; 35(39): e350. doi: 10.3346/jkms.2020.35.e350.
6. Grainger JD, Thachil J, Will AM. How we treat the platelet glycoprotein defects; Glanzmann thrombasthenia and Bernard Soulier syndrome in children and adults. *Br J Haematol.* 2018; 182(5): 621-32. doi: 10.1111/bjh.15409.
7. UK Haemophilia Centre Doctors' Organisation (UKHCDO). UKHCDO annual report & bleeding disorder statistics for April 2024 to March 2025 [Internet]. 2025. Salford: UKHCDO. Available from <https://www.ukhcd.org/home-2/annual-reports/> (accessed September 2026).
8. McGeown S, Owen-Wyrd A. Understanding the diagnosis, symptoms and treatment of rare bleeding disorders. *Nurs Stand.* 2025; 40(11): 47-52. doi:10.7748/ns.2025.e12465.
9. Parker M, Hannah M, Zia A. "If I wasn't a girl": Experiences of adolescent girls with heavy menstrual bleeding and inherited bleeding disorders. *Res Pract Thromb Haemost.* 2022; 6(4): e12727. doi: 10.1002/rth2.12727.
10. Faye F, Crocione C, Anido de Peña R, et al. Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey. *Eur J Hum Genet.* 2024; 32: 1116-26. doi: 10.1038/s41431-024-01604-z.
11. Raheja P, Abdul-Kadir R, Knudsen MJ, Burgess C, Schols SE; Very Rare Coagulation Factor Deficiency Working Group and Women and Girls with Bleeding Disorders Working Group of European Association for Haemophilia and Allied Disorders (EAHAD). Dealing with heavy menstrual bleeding in very rare bleeding disorders: From a patient's perspective. *Res Pract Thromb Haemost.* 2026; 10(1): 103368. doi: 10.1016/j.rpth.2026.103368.
12. Ackbarali S, Ackbarali T, Kowalski K, et al. Addressing clinical management challenges in the treatment of rare, non-malignant hematologic disorders through educational interventions. *Blood.* 2022; 140(Suppl 1): 157219. doi: 10.1182/blood-2022-157219.
13. Avgeropoulos MN, James PD. (2025). Diagnosis of bleeding disorder of unknown cause: how many tests are enough to diagnose BDUC? *Hematology Am Soc Hematol Educ Prog.* 2025; 2025(1): 116-119. doi: 10.1182/hematology.2025000695.
14. Solgun HA. Diagnosis, treatment, surgical practices and review of the literature in rare coagulation factor deficiencies. *Ital J Pediatr.* 2025; 51(1): 3. doi: 10.1186/s13052-024-01806-7.
15. James PD. Women and bleeding disorders: diagnostic challenges. *Hematology Am Soc Hematol Educ Program.* 2020;2020(1):547–552. doi:10.1182/hematology.2020000140.
16. National Bleeding Disorder Foundation (NBDF) Medical and Scientific Advisory Council (MASAC). MASAC Guidelines for pregnancy and perinatal management of women with inherited bleeding disorders and carriers of hemophilia A or B. MASAC Document #265 [Internet]. Washington (DC): NBDF; 2023. Available from <https://www.bleeding.org/healthcare-professionals/guidelines-on-care/masac-documents> (accessed September 2025).
17. Suresh N, Kurian B, Jeladharan R, Sao A, Sidharthan N, Abraham RM. Rare coagulation factor deficiencies: a five-year experience from a single tertiary care center in South India. *Blood Coagul Fibrinolysis.* 2025; 36(2): 37-43. doi: 10.1097/MBC.0000000000001339.
18. Kluge MC, Vachharajani A, Mazuru D, Gruner B, Severance TS. Intracranial bleeding in a neonate. *NeoReviews* 2024; 25(3): e159-e162. doi: 10.1542/neo.25-3-e159.
19. Srivastava A, Santgostino E, Dougall A, et al. WFH Guidelines for the Management of Hemophilia, 3rd edition. *Haemophilia* 2020; 26 Suppl 6: 1-158. doi: 10.1111/hae.14046.
20. Peyvandi F, Menegatti M, Palla R. Rare bleeding disorders: worldwide efforts for classification, diagnosis, and management. *Semin Thromb Hemost.* 2019; 45(6): 581-9. doi: 10.1055/s-0033-1349221.
21. Lim CC, Ling SW, Vijayan KV, Lee MJ, Devesayaham PR. Acquired hemophilia A: rare cause of upper airway hematoma and a literature review. *Indian J Otolaryngol Head Neck Surg.* 2023; 75: 3886-8. doi: 10.1007/s12070-023-03943-7.
22. National Bleeding Disorders Foundation (NBDF) Medical and Scientific Advisory Council (MASAC). MASAC Recommendations regarding physical therapy management for the care of persons with bleeding disorders. MASAC Document 275 [Internet].

Washington (DC): NBDF; 2023. Available from: <https://www.bleeding.org/healthcare-professionals/guidelines-on-care/masac-documents> (accessed August 2025).

23. Königs C, Motwani J, Jiménez-Yuste V, Blatný J, on behalf of the Factor Think Tank. Teenagers and adolescents with hemophilia—need for a specific approach. *J Clin Med*. 2024;13(17):5121. doi:10.3390/jcm13175121.
24. Belzer LT, Wright SM, Goodwin EJ, Singh MN, Carter BS. Psychosocial considerations for the child with rare disease: A review with recommendations and calls to action. *Children (Basel)*. 2022; 9(7): 933. doi: 10.3390/children9070933.
25. Sukhera J. Narrative reviews: Flexible, rigorous, and practical. *J Grad Med Educ*. 2022; 14(4): 414-417. doi: 10.4300/JGME-D-22-00480.1.
26. Braun V, Clarke V. Using thematic analysis in psychology. *Qual Res Psychol*. 2006; 3(2): 77-101. doi: 10.1191/1478088706qp063oa.
27. Branchford B, Clark K, Stanford RH, Garner DA, Huang SP, Wolford E. Hereditary factor X deficiency in America survey: impact on quality of life and burden of disease in patients and caregivers. *Blood Coagul Fibrinolysis*. 2024; 35(3): 73-81. doi: 10.1097/MBC.0000000000001275.
28. Khair K, Fletcher S, Jenner K, Holland M. One day at a time: Life with Glanzmann thrombasthenia—Qualitative results from the GT 360 study. *Haemophilia*. 2024; 30(6): 1373-82. doi: 10.1111/hae.15126.
29. Khair K, Fletcher S, Boyton M, Holland M. Bleeding and quality of life in people with Glanzmann thrombasthenia – insights from the Glanzmann's 360 study. *Res Pract Thromb Haemost*. 2024; 8(3): 100142. doi: 10.1016/j.rpth.2024.102586.
30. Koekemoer HT, Joubert J, Nichol RJ, Joubert G, Coetzee MJ. Mental health disorders and health-related quality of life in adults with inherited bleeding disorders in central South Africa. *Haemophilia*. 2022; 28(2): e64-e67. doi: 10.1111/hae.14486.
31. van Gastel TC, Teela L, Degenaar-Dujardin ME, et al. A qualitative study on the impact of bleeding disorders on the daily life of Dutch women. *Womens Health (Lond)*. 2025; 21: 17455057251362582. doi: 10.1177/17455057251362582.
32. Peltier S, Kellum A, Brewer J, Duncan A, Cooper DL, Saad H. Psychosocial impact and disease management in patients with congenital factor VII deficiency. *J Blood Med*. 2020; 11: 297-303. doi: 10.2147/JBM.S259909.
33. Duncan A, Kellum A, Jain S, et al. Disease burden in patients with Glanzmann thrombasthenia: perspectives from the Glanzmann Thrombasthenia Patient/Caregiver Questionnaire. *J Blood Med* 2020; 11:289-295. doi: 10.2147/JBM.S259904.
34. Arya S, Wilton P, Page D, et al. "Everything was blood when it comes to me": Understanding the lived experiences of women with inherited bleeding disorders. *J Thromb Haemost* 2020; 18(12); 3211-3221. doi: 10.1111/jth.15102.
35. Doherty TM, Kelley A. Bleeding disorders. 2023 Apr 3. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2026 Jan–. PMID 31082094.
36. Tareke M, Bayeh AB, Birhanu M, Belete A. Psychological distress among people living with chronic medical illness and the general population, Northwest Ethiopia: A comparative cross-sectional study. *PLoS One*. 2022; 17(12): e0278235. doi: 10.1371/journal.pone.0278235.
37. Newson JJ, Hunter D, Thiagarajan TC. The heterogeneity of mental health assessment. *Front Psychiatry*. 2020; 11: 76. doi: 10.3389/fpsyt.2020.00076.
38. Mahlangu J. Enhancing the evidence for care in underserved bleeding disorders communities. *Haemophilia*. 2026 Jan 26. doi: 10.1111/hae.70214. Online ahead of print.
39. Scott PJ, Brown AW, Adediji T, et al. A review of measurement practice in studies of clinical decision support systems 1998-2017. *J Am Med Inform Assoc*. 2019; 26(10): 1120-1128. doi: 10.1093/jamia/ocz035.
40. Ahmedani BK. Mental health stigma: Society, individuals, and the profession. *J Soc Work Values Ethics*. 2011; 8(2): 4-1–4-16.
41. Habeb M, Ciobanu AM, Al-Ani M, Mottershead R. Stigma in mental health: The status and future direction. *Cureus*. 2025; 17(6): e85398. doi: 10.7759/cureus.85398.
42. Dutton B, Kai J. Women's experiences of heavy menstrual bleeding and medical treatment: a qualitative study in primary care. *Br J Gen Pract* 2023; 73(729): e294-e301. doi: 10.3399/BJGP.2022.0460.
43. Radhakrishnan R, Thomas J, George J, Varadarajan R. A comparative analysis of the bleeding profile and quality of life among women with hemophilia genotype compared to other bleeding disorders. *Cureus*. 2024; 16(1): e52837. doi: 10.7759/cureus.68636.

44. Durand F, Fleury MJ. A multilevel study of patient-centered care perceptions in mental health teams. *BMC Health Serv Res*. 2021; 21(1), 44. doi: 10.1186/s12913-020-06054-z.
45. Mulders G, Uitslager N, Kavanagh M, et al.; European Association for Haemophilia and Allied Disorders (EAHAD) Nurses Committee. The role of the specialist nurse in comprehensive care for bleeding disorders in Europe: An integrative review. *Haemophilia*. 2024; 30(3): 598-608. doi: 10.1111/hae.14974.
46. Pollard D, Harrison C, Dodgson S, Holland M, Khair, K. The UK haemophilia specialist nurse: Competencies fit for practice in the 21st century. *Haemophilia*. 2020; 26(4): 622-630. doi: 10.1111/hae.14002.
47. Proctor EK, Landsverk J, Aarons G, Chambers D, Glisson C, Mittman B. Implementation research in mental health services: an emerging science with conceptual, methodological, and training challenges. *Adm Policy Ment Health*. 2009; 36(1): 24-34. doi:10.1007/s10488-008-0197-4.
48. Fok CC, Henry D, Allen J. Maybe small is too small a term: Introduction to advancing small sample prevention science. *Prev Sci*. 2015; 16(7), 943-949. doi: 10.1007/s11121-015-0584-5
49. Wanat M, Boylan AM, Borek AJ. (2021). Value, challenges and practical considerations when designing, conducting and analysing a longitudinal qualitative study in family medicine *Fam Med Community Health*. 2021; 9(Suppl 1): e001283. doi: 10.1136/fmch-2021-001283
50. Hemophilia Federation of America (HFA). Mental health matters: you're not alone in the bleeding disorders community. 14 May 2025 [Internet]. Washington (DC): HFA. Available from <https://www.hemophiliafed.org> (accessed June 2025).
51. Ahad AA, Sanchez-Gonzalez M, Junquera P. Understanding and addressing mental health stigma across cultures for improving psychiatric care: a narrative review. *Cureus*. 2023; 15(5): e39549. doi: 10.7759/cureus.39549.
52. National Bleeding Disorders Foundation (NBDF) Medical and Scientific Advisory Council (MASAC). Recommendations on increasing access to mental health and substance use disorder facilities and programs for persons with bleeding disorders. MASAC Document 289 [Internet]. Washington (DC): NBDF; 2023. Available from <https://www.bleeding.org/healthcare-professionals/guidelines-on-care/masac-documents> (accessed June 2025).

OPEN ACCESS

This is an Open Access article distributed under the terms of the Creative Commons Attribution-NonCommercial-NoDerivs 4.0 International License (<https://creativecommons.org/licenses/by-nc-nd/4.0/deed.en>) which permits use and distribution in any medium, provided the original work is properly cited, the use is non-commercial, and no modifications or adaptations are made. Copyright is retained by the authors.

HOW TO CITE THIS ARTICLE:

Owen-Wyrd A, McGeown S, Hallard E, Tate H. Identifying mental health needs in extremely rare inherited bleeding disorders (ERBD): A narrative review. *Bleeding Dis Care Pract* 2026; 13(1): 24-37. <https://doi.org/10.17225/bdcp-2026-0004>

Authors ©2026:

Amy Owen-Wyrd, Stacey McGeown, Emma Hallard, Helen Tate

Published by Haemnet Ltd

eISSN: 2978-4204

Journal contact: publishing@haemnet.com