

Invisible Challenges of a Visible Disease: A Qualitative Study on Disease-related Impairment, Participation Barriers and Care Experiences in Adults with Ichthyosis

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Ichthyoses are rare genodermatoses characterized by scaling, hyperkeratosis and barrier dysfunction, often accompanied by hypohidrosis and pruritus. Beyond visible symptoms, ichthyosis imposes substantial psychosocial, financial and time-related challenges. This qualitative study explored patient-perceived impairments, disability and experiences with medical and social care. Semi-structured interviews were conducted with 18 adults (13 women, 5 men; aged 18–67 years) with various ichthyosis subtypes, recruited from 2 specialized dermatology clinics in Germany and the patient organization Ichthyosis e.V. Reflexive thematic analysis showed that participants' daily lives were influenced not only by symptoms and time-consuming care routines but also by social and structural factors limiting participation in various life domains. Participants reported inequalities in education and employment, rejection during job seeking and at work. This can lead to social anxiety, depression and negative self-image. Psychological strain also arises from limited treatment options and financial strain. Perceptions of disability vary, though many seek formal recognition. The impact of ichthyosis is often underestimated by healthcare professionals, complicating access to social and medical support. Dermatologists should recognize the crucial role of their diagnosis and documentation and carefully guide patients through the healthcare system to ensure appropriate sociomedical support.

Key words: ichthyosis; health services research; patient narratives; disability; patient experience; qualitative research.

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Inherited ichthyosis is a clinically and genetically heterogeneous disorder of keratinization, caused by mutations in genes crucial for keratinocyte differentiation and epidermal barrier function (1).

The ichthyoses have recently been reclassified as epidermal differentiation disorders (EDDs),

SIGNIFICANCE

Ichthyosis is a rare skin condition that can affect everyday life and emotional well-being. Many people spend hours each day on skin care and still face pain, discomfort, as well as personal, financial and time-related burdens. However, the greatest difficulties often come from social challenges – being stared at, excluded, or struggling to access adequate care. This study provides a novel perspective on the impact of ichthyosis by showing how these experiences affect various life domains. The findings highlight the need for better recognition and support, and form the basis for a national survey to improve care and participation.

comprising non-syndromic forms (nEDDs) (2), such as ichthyosis vulgaris (IV, FLG-nEDD), and syndromic variants (sEDDs) (3), such as Netherton syndrome (SPINK5-sEDD). Keratinopathic ichthyoses, including epidermolytic ichthyosis (EI, KRT1-nEDD), may involve palmoplantar keratoderma. Rare autosomal recessive forms, formerly called autosomal recessive congenital ichthyosis (ARCI), include harlequin ichthyosis (HI), lamellar ichthyosis (LI) and congenital ichthyosiform erythroderma (CIE).

Clinically, ichthyosis is characterized by hyperkeratosis, erythema/erythroderma and blistering; possible symptoms include pruritus, pain, hypohidrosis, ocular complications and external auditory canal problems (1, 4–6). For a more detailed overview of disease subtypes, molecular mechanisms and clinical variability, see recent reviews (1–6).

Treatment

Ichthyoses are incurable; treatment aims to improve skin condition and quality of life. Lifelong topical therapy – including emollients, keratolytics and scale-removal methods – is essential and varies according to individual patient needs and physician experience (1, 7–9). Systemic retinoids may benefit certain subtypes but require careful risk-benefit assessment, particularly regarding teratogenicity. The German S1 (7) guideline and the European Guidelines (8, 9) emphasize assessing psychosocial needs and recommending rehabilitation and emotional support for patients and families.

Ichthyosis in health services research

The extent to which clinical recommendations are implemented in routine care for individuals with ichthyosis remains unclear. Klein et al. (10) report substantial barriers to treatment and specialized care in Germany, yet medical and psychosocial needs are scarcely documented.

While ichthyosis is known to significantly impair quality of life – affected by factors such as gender, pain, pruritus, and scaling (11–15) – 2 qualitative studies have identified influences, including daily life demands, relationships and self-perception (16, 17). Elevated rates of anxiety, depression and suicidality are reported (18–20), though underlying causes remain insufficiently investigated.

Despite growing recognition of quality of life and psychological aspects in dermatological healthcare research, important dimensions – particularly the assessment of disease-specific impairments, disability and participation restrictions – have not yet been systematically assessed. Participation – a core concept of the International Classification of Functioning, Disability and Health (ICF) (21) – describes engagement in personally meaningful life areas and is a central concept in both health research and social law in Germany. It encompasses different domains, including social and occupational participation.

The aim of this qualitative pilot study was to explore the experiences, challenges and needs of individuals living with ichthyosis and identify patient-relevant themes to inform a subsequent quantitative survey. Such data can provide a foundation for improving understanding of patient needs, generating hypotheses for future research and informing future evidence-based healthcare practices.

PATIENTS AND METHODS

Study design

This qualitative interview study represents the initial phase of the mixed-methods study. The second part will be a questionnaire study, allowing quantitative assessment of these domains and examination of associations between disease-, patient- and care-related factors and participation in various life domains. The study was developed in advisory exchange with the German Ichthyosis Support Group. This study adopts a contextualist ontology (reality materially real yet socially shaped) and constructivist epistemology (researcher subjectivity generates knowledge), aligned with the ICF framework for understanding disability/health as function in context (WHO, 2001).

Sampling and recruitment

Participants were recruited from 2 university outpatient clinics (University Hospital Münster and Helios Hospital Berlin-Buch) and the German Ichthyosis Support Group e. V. Interested individuals were entered into a database with key characteristics (age, gender, ichthyosis type, region), and participants were then selected in consultation with the study team to ensure diverse representation across these characteristics using maximum variation sampling (22). Recruitment continued iteratively alongside data analysis to target underrepresented groups and ensure sufficient depth and diversity of perspectives, until the dataset provided sufficient richness for theme construction ($N=18$) (23), meaning that no additional codes could be developed. Only adults (≥ 18 years) with confirmed ichthyosis diagnoses – clinically confirmed for clinic patients, self-reported for support group participants – were eligible.

Data collection

Interviews were conducted via telephone or Zoom, according to participant preference, by J.S., an experienced qualitative researcher in sensitive and vulnerable populations. Interviews followed a narrative approach using an open-ended stimulus to elicit participants' experiences with ichthyosis across life stages (24). Immanent prompts deepened emergent topics. Predefined exmanent questions, developed with clinical experts and a patient support group and guided by gaps in the existing literature, were posed postnarrative to elicit contextual information, clarify narrative environments and fill experiential gaps without imposing themes or guiding accounts (**Table I**). Interviews were audio-recorded, transcribed verbatim, checked for accuracy to ensure correct transcription and pseudonymized. Although conducted in German, quotations were accurately translated into English.

Data analysis

Pseudonymized transcripts were familiarized and analysed using reflexive thematic analysis following Braun and Clarke (25). Initial codes were generated inductively from the data in an organic, open, iterative process. MAXQDA facilitated code organization, visualization of coding patterns, and thematic mapping. Semantic codes reflected explicit meanings, while latent codes captured underlying, interpreted meanings. Codes were consolidated into themes when they reflected shared meaning-based patterns across accounts for example, illustrating how symptom-burden interacts with social exclusion to influence participation restrictions.

Table I. Interview Guide

Narrative interview guide	
Narrative stimulus/Narration	Methodological notes
-“I would like to ask you to tell me your story living with ichthyosis. This includes experiences with doctors, treatments, as well as your everyday life and the role ichthyosis plays.” “Where does your story begin?”	-The stimulus is designed to be open-ended and to elicit a continuous narrative. -Narration is generally not interrupted. -The participant controls the focus and sequence. -The interviewer notes potential follow-up questions (as relevant to the research question), thematic breaks, gaps, unfinished storylines, or fragments.
Immanent prompts	Methodological notes
“You mentioned that [...], could you describe that in more detail?” “What happened next?” “How did it continue?” “How did you experience the situation?” “What helped you most during this phase?” “What would you have wished for?”	-Open, narrative-generating questions that follow the participant’s ongoing story -Relevant aspects, phases, or biographical stages are explored in depth -Questions are asked in the order in which they appear in the participant’s narrative
Exmanent prompts	Methodological notes
Daily life and self-management	
-“Could you describe your current daily skin-care routine?” -“What does a typical day look like for you? How does skin care fit into your daily life?” -“Are there activities you would like to do but are currently limited in because of ichthyosis?”	
Healthcare and key contacts	
-“Could you tell me about the doctors or clinics you have interacted with regarding ichthyosis?” -“Who do you usually contact regarding your ichthyosis care?” -“How do you get information about your condition or treatment options?”	
Work and education	
-“If you think back to your time in school or education, how did your skin condition play a role?” -“Could you describe how ichthyosis relates to your current work or professional activities?”	
Disability/Legal aspects	
-“Do you have an official degree of disability? If so, could you describe your experience with the application and receipt process?”	
Are asked only after the narrative and immanent follow-up questions Serve to complete the narrative data, not to steer or impose themes -Facilitate contextual elicitation and clarification of narrative environments	
Debrief/Closing phase	
“Is there anything we haven’t discussed that you feel is important?”	

Rigour

Analytical decisions were discussed reflexively within the team, drawing on diverse subjectivities as a resource: J.S. and A.W.’s analysis was informed by an ICF-based understanding of disability and functioning, as well as experience in medical care of vulnerable groups; dermatologists contributed biomedical perspectives and practical clinical experience. In our interdisciplinary team, we intentionally allowed multiple readings of the same text to coexist, enriching interpretation and integrating them into coding and discussion. Clinicians tended to read statements as reflecting symptom-burden, whereas non-medical and ICF-oriented perspectives emphasized contextual factors. Divergent readings were not resolved by consensus but retained as distinct themes or differentiated into subthemes.

RESULTS

Participants

In total, 32 individuals expressed interest in participating and were entered into a database. From this pool, the final sample comprised 9 participants recruited

via support group and 9 via clinics. Between January 2024 and January 2025, 18 interviews were conducted; 12 via telephone and 6 via Zoom (duration: 38–95 min, M=67). Participant characteristics are presented in Table II.

Table II. Patient collective (n=18)

Sex	
Female	13
Male	5
Age	
18–20 years	2
21–30 years	5
31–40 years	1
41–50 years	2
51–60 years	6
>60 years	2
Subtype ^a	
Ichthyosis vulgaris (IV)	2
ARCI, not otherwise specified (ARCI)	3
ARCI, lamellar ichthyosis (LI)	4
ARCI, harlequin ichthyosis (HI)	2
Epidermolytic ichthyosis (EI)	6
Netherton syndrome (NS)	1

^aBased on self-report
ARCI: autosomal recessive congenital ichthyosis.

Table III. Interview excerpts: Disease-related-burden

Subtheme	Quote
Symptom-related challenges	Q1: "When you get those thick, hardened patches of skin, sometimes you can't fully straighten or bend your knees and elbows, because eventually you just reach a limit. So, this kind of mobility really affects your life. I mean, it meant I couldn't do certain things anymore." (f, 45y, EI)
	Q2: "I have itching, and it really burdens me, especially in the morning and at night. It's so overwhelming that sometimes I have to put on gloves so I don't scratch myself." (f, 54y, EI)
	Q3: "What I really suffered from, even since my childhood, were skin cracks, especially a lot on my neck. They hurt terribly. I didn't like being active because I was always afraid that running or playing would hurt again." (f, 62y, ARCI)
	Q4: "That [Hypohidrosis] is really extreme. My body gets so hot. There's no heat exchange, and that's very distressing. When other people are happy about the nice warm weather and go swimming, none of that happens for me." (f, 67y, IV)
Time-consuming care routines	Q5: "I feel like other people have more hours in the day than I do. Simply because my time is consumed by managing ichthyosis [...] it's just a time factor, where I feel like I could be doing something productive, earning money. I could accomplish other things, but I am limited or restricted." (f, 30y, IV).
	Q6: "Yes, I really have to say, care is very, very difficult, a very challenging topic. So we really suffered a lot because somehow, it was a damn cycle we couldn't get out of." (m, 21y, NS)
	Q7: "I can't just take a five-minute shower. I have to spend an hour in the bath. It's kind of ridiculous. If my day is packed and I need to study or do something else, it becomes tricky." (f, 18y, LI)
	Q8: "I still rely on my mom to help with my head, because I can't manage it completely on my own. That means I depend on help and I already spend at least two hours on it before I can even leave the house." (f, 27y, HI)
Economic challenges	Q9: "I have to live very frugally throughout the year, even keeping the heating on low during the winter months, as minimal as possible. Otherwise, I would end up paying an absurd amount for hot water costs in the utility bills." (m, 55y, LI)
	Q10: "I think that's absolutely unreasonable, because without this cream I couldn't manage my everyday life. And in the amounts I need it, it's really hard to afford." (f, 27y, HI)
	Q11: "But the thing is, every over-the-counter product I've had to pay for myself since I turned 18. [...] and many plant-based products definitely aren't covered. I have to pay for those myself." (m, 29y, HI)
	Q12: "I was already dealt a bad hand with this illness as a young person. And later in life, you just get hit again, because then the money isn't enough to afford proper care without worry, you know? [...] So I really depend on finding a compassionate doctor who will at least prescribe me my ointments." (f, 67y, IV)
Impairments in social participation	Q13: "Traveling and camping, I can't take part in that. Or joining sports classes, that's often not possible either. And of course, it's not a nice feeling when you can't do all these things because of the ichthyosis." (f, 67y, IV)
	Q14: "The worst part is actually the social interactions. Whether I'm taking the bus, in the subway, or shopping, it's always the same. Being stared at, pointed at, people whispering, turning around [...]" (f, 27y, HI)
	Q15: "At times, I just didn't want to go out because I thought, I really don't feel like explaining it now. Or being in the spotlight again." (f, 19y, ARCI)
Impairments in occupational participation	Q16: "But, for example, I think I would have liked to be a teacher, but with the children, that's just really difficult." (f, 27y, HI)
	Q17: "I was always interested in nursing, but they always said, 'you can't do that, for hygienic reasons or something like that.'" (f, 19y, ARCI)
	Q18: "And then it came out that they thought the workplace was too creamy, too scaly, meaning, in their eyes, too dirty [reason for probationary dismissal]." (m, 29y, HI)
Psychosocial consequences	Q19: "What is really deep inside me, are these bad experiences in childhood that I had because of my appearance." (f, 45y, EI).
	Q20: "And that means you're always kind of like an alien, somehow. I think that's probably the hardest thing, what hits me personally, besides all the pain and everything I deal with. [...] it has really affected my personality." (m, 26y, EI)
	Q21: "I experienced exclusion at school already as a child. People didn't want to sit next to me because I always had to apply creams, and sometimes those creams didn't smell very nice — so I did experience what today would probably be called bullying. [...] I definitely had some experiences of exclusion as a child." (f, 54y, EI)
	Q22: "I was basically bullied continuously throughout all of my school years." (f, 54y, EI)
	Q23: "Ichthyosis has shaped my personality a lot. I've always cared about how others feel, which led me to choose a social profession." (m, 26y, EI)
	Q24: "Because of these beliefs that developed from those experiences, I eventually reached a point where I slipped into depression." (f, 31y, IV)
Experiencing disability	Q25: "Because you then experience what life with ichthyosis entails. And it has made me calmer as a person and less open, but unfortunately it has also brought a lot of social anxiety." (f, 32y, LI)
	Q26: "It's a limitation, but not a disability. That was always really important to me." (f, 62y, ARCI)
	Q27: "To me, it's clearly a disability, compared to what's considered 'normal'." (f, 54y, LI)

ARCI: autosomal recessive congenital ichthyosis; EI: epidermolytic ichthyosis; HI: Harlequin ichthyosis; IV: ichthyosis vulgaris; LI: lamellar ichthyosis; NS: Netherton syndrome; y: year(s) at the time of the interview.

Themes

The themes derived from the analysis are presented within the sections “Disease-Related Challenges” and “Healthcare Experiences,” used to structure the results. Disease-Related Challenges encompass real-life implications from the illness and its environmental interactions, illustrated by excerpts in Table III. Healthcare Experiences capture perceptions of seeking, accessing, and engaging with care, illustrated by excerpts in Table IV. Themes and subthemes are summarized in Table V.

DISEASE-RELATED CHALLENGES

Symptom-related challenges

Participants reported substantial impairment and discomfort due to symptoms such as pruritus, scaling, and pain due to recurrent infections or hyperkeratosis (Table III, Q1–Q3). Hyperkeratosis led to fissures, restricted mobility, and interference with daily activities. Hypohidrosis emerged as core restriction, affecting social participation and requiring careful management with dependence on specific climatic conditions (Table III, Q4). Participants

Table IV. Interview excerpts: Healthcare experiences

Subtheme	Quote
Lack of expertise and acknowledgement	Q1: "Most of the time, I have to explain ichthyosis to the doctors. They don't even know what Neotigason is or what it does." (m, 55y, LI)
	Q2: "I'm now in general dermatology at [German Clinic]. And yes, that brings us to the topic that four out of five have no idea about ichthyosis and how it can be treated." (m, 26y, EI)
	Q3: "This is just how it is in our region. The dermatology clinic, which, I'd say, usually knows about skin conditions, doesn't know about ichthyosis. So, basically, it's very hard—or impossible—to find someone familiar with ichthyosis. I quickly realized that I actually know more about it than the doctors." (f, 45y, EI)
	Q4: "But I also had a doctor who completely messed it up, because he thought I had, uh, eczema, instead of ichthyosis." (m, 29y, HI)
	Q5: "I always feel that there's so much awkwardness even with the doctors. Or maybe a lack of understanding, of what this skin condition actually requires in terms of care and products." (f, 67y, IV)
	Q6: "I would like to see more interest from doctors when it comes to this illness and a bit more empathy with a person who has this illness from head to toe and is often desperate, you know?" (f, 19y, ARCI)
	Q7: "There were no offers for a conversation, or that the doctor ever really talked to me. It was basically just quickly asking how I'm doing, handing me the prescription, and then leaving again." (f, 62y, ARCI)
	Q8: "But very often, it was like, nobody cared. And then suddenly there was this young doctor who was really passionate about it. And that was really, really rewarding for me." (f, 62y, ARCI)
Barriers to accessing essential care products	Q9: "I wish I could get medications and ointments without any issues, without being made to feel like a beggar. It would be nice to receive the medications I need without any problems [Referring to recurring discussions with doctors regarding prescription quantities and eligibility]." (f, 67y, IV)
	Q10: "My dermatologist prescribed a small container—100 grams—and I'm supposed to cover my whole body with it. He says he can't prescribe more, and that's a big problem for me. So I went to my general practitioner, who prescribes 200 grams. I prefer going there, even though he otherwise doesn't know much about my disease." (f, 54y, EI)
	Q11: "[...] because it's really a struggle to get things prescribed at the moment, especially since new regulations keep coming into effect. Sometimes even the doctors themselves don't know them. And then they don't prescribe it, because they want to protect themselves, since in the end the health insurance can come back and say, 'You weren't allowed to do this.'" (f, 31y, LI)
	Q12: "It's really a struggle right now to get things prescribed, especially since the new regulations. For example, I think you're not allowed to have two active ingredients in compounded creams anymore. And then I have to argue every single time about whether I can get my cream or not." (f, 32y, LI)
	Q13: "If I'd only stuck to what my doctors have provided me in the past, I don't think I would have ever gotten as far as I have on my own." (f, 32y, LI)
Social healthcare services	Q14: "I think most people [...], can't really grasp what ichthyosis means. They see the skin and think, well, the skin is red, and there might be some social interaction issues. But that's it. People [even doctors] don't see how much time you spend at home on all the care routines and what it truly means for you." (f, 32y, LI)
	Q15: "Medical assessors said: 'from your face, one would never guess that you're sick.' And that's difficult because you end up being penalized for putting so much effort into taking care of your skin." (f, 32y, LI.)
	Q16: "Legal recognition of my disability had to be hard-fought, because first of all, you can't find any experts in the social service offices. They don't really know what they're doing or understand the problem. Then they just look at you from afar, never actually come by, and only want pictures." (f, 45y, EI)
	Q17: "My legal recognition as severe disabled is temporary. Then the whole process starts all over again. So it's basically an ongoing struggle." (f, 45y, EI)

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also described anxiety and worry related to hypohidrosis, which in extreme cases could become life-threatening.

Time-consuming care routines

Interviews consistently highlighted the time-intensive nature of managing ichthyosis. Daily care routines (bathing and applying emollients) often consumed several hours, requiring careful scheduling. This was described as “stolen time”, impacting autonomy, flexibility, and limiting work, education, and leisure (Table III, Q5-Q8).

Extensive care sometimes required physical assistance, creating dependency and emotional strain.

Table V. Themes and subthemes

Theme	Subtheme
Disease-related challenges	Symptom-related challenges
	Time-consuming care routines
	Economic challenges
	Impairments in social participation
	Impairments in occupational participation
	Psychosocial consequences
	Experiencing disability
Healthcare experiences	Lack of expertise and acknowledgement
	Barriers to accessing essential care products
	Social healthcare services

Economic challenges

Ichthyosis imposed substantial financial burdens. Participants reported high costs for essential disease-related products and elevated utility expenses (water, heating, laundry), all integral to daily care (Table III, Q9-Q10).

Some participants experienced financial strain from reduced work hours due to time-intensive care, contributing to long-term insecurity via unemployment gaps, limited pension rights, and early retirement. Early exit further depleted resources, compounding economic challenges (Table III, Q12). Participants described how their medical condition, through financial strain, resulted in structural disadvantages that affected their daily lives.

Impairments in social participation

Social participation involves engagement in meaningful social/community activities (e.g. relationships, family life, leisure, community involvement). Participants' accounts show how ichthyosis symptoms like hypohidrosis and pain restrict such engagement, including peer interactions, sports, and travel (Table III, Q13).

These restrictions can lead to feelings of isolation and loss and may make it challenging to maintain connections with social networks. In addition to

physical challenges, social participation was also perceived as being affected by stigmatization. Symptoms such as skin scaling or redness were described as provoking negative reactions from others, leading to experiences of exclusion and bullying (Table III, Q14). The fear of negative reactions – conceptualized as anticipated stigmatization – led some to avoid social situations and may influence social decision-making (Table III, Q15).

Impairments in occupational participation

Participants reported restrictions in occupational participation, which could be directly related to disease/symptoms (e.g. avoiding high temperatures, influencing career choices). Social factors and external limitations appeared to play an even greater role in shaping opportunities, potentially involving stigmatization and negative reactions (Table III, Q16). External factors, such as discouragement from teachers or parents, also contributed to career limitations in several cases by influencing decisions about career paths – for example, by discouraging choices involving direct interaction with people or clients (Table III, Q17).

Discrimination in job application processes was also reported, with some experiencing appearance-based rejections (Table III, Q18).

Barriers in occupational participation may be mitigated by workplace accommodations and an open, tolerant attitude among employers/colleagues. Participants perceived adjustments like air-conditioned environments, flexible hours or remote work options as valuable for continued occupational engagement.

Psychosocial consequences

Participants described a high psychosocial strain, marked by anxiety, depression, and a persistent feeling of being different. For many, psychological distress stemmed less from physical symptoms than from social and emotional consequences of ichthyosis, including stigmatization and exclusion, particularly during childhood/adolescence (Table III, Q19-Q22).

Stigma was identified as a dominant theme across all interviews, manifesting as external stigma in being stared at, avoided, or treated as contagious, with long-term impacts on self-image and identity in some cases (Table III, Q23-Q25).

The narratives also reveal internalized beliefs (self-stigmatization), sometimes resulting in avoidant behaviour or missed opportunities. Psychological strain did not always correspond to participants' experiences of physical symptoms; several participants reporting comparatively milder symptoms still described significant mental health challenges. Barriers in accessing care and treatment were cited as additional stressors.

Experiencing disability

The perception of ichthyosis as a disability varies among participants. Whether individuals identify their condition as a disability – and themselves as persons with disabilities – largely depends on their associations with the term. For many, it is primarily linked to mobility or intellectual impairments. Some explicitly state that while they experience limitations, they do not define their condition as a disability personally (Table III, Q26). Others, by contrast, refer to their “disability” without hesitation and use the term naturally in describing their situation (Table III, Q27).

Nevertheless, despite these differing personal interpretations, most participants pursue formal legal recognition of their condition as a disability, as this status within the German social welfare system entitles individuals to compensation for disadvantages (e.g. tax relief, additional leave and other support measures).

HEALTHCARE EXPERIENCES

Lack of expertise and acknowledgement

A prevalent issue was the lack of expertise, even among dermatologists. Participants often had to educate doctors about ichthyosis and specify prescriptions they needed (Table IV, Q1-Q4).

Some participants perceived a sense of insecurity among physicians and felt that the challenges of daily life were not recognized, contributing to the experience of not feeling taken seriously or understood (Table IV, Q5). They also reported a lack of interest and engagement from healthcare professionals (Table IV, Q6).

The psychosocial impact of ichthyosis was often not sufficiently addressed in medical consultations. Some participants expressed a need for opportunities to discuss their experiences (Table IV, Q7, Q8).

Participants emphasized the high self-initiative required to navigate the healthcare system – finding knowledgeable specialists, identifying suitable treatment routines or securing support services (Table IV, Q13). Referrals to specialized centers were often delayed or absent. However, when patients were able to consult a specialist familiar with ichthyosis, some described this as highly relieving and psychologically reassuring (Table IV, Q8).

Barriers to accessing essential care products

A central challenge is accessing essential care products/medications due to high costs and restricted prescription eligibility, creating persistent feelings of fighting for everything; this ongoing struggle proved exhausting and psychologically burdensome. Participants reported inconsistent/conflicting information on prescription

eligibility, reimbursement and allowable quantities. A primary concern was finding a doctor willing to prescribe necessary treatments rather than receiving broader medical care. (Table IV, Q9–Q12).

Many participants described diverse and highly individualized topical treatment needs which require trial and error. Preferences in texture, ingredients, packaging and application vary widely. Problems arise when effective products are discontinued or access becomes restricted due to regulatory changes (Table IV, Q11, Q12).

Social healthcare services

A recurrent theme among participants was the burdensome bureaucracy of applying for social healthcare benefits, including legal recognition of ichthyosis as a disability and approval of cost coverage. Participants expressed frustrations with disability assessors who base their decisions on visible skin appearance and fail to acknowledge the hours of daily care needed to achieve what they can see, showing insufficient understanding of the illness and its daily impact on life (Table IV, Q14–Q16). The legal recognition of disability is in some cases granted only for a limited period, requiring that bureaucratic procedures be repeated at regular intervals (Table IV, Q17).

DISCUSSION

Even though disability and participation are not yet systematically addressed in dermatological health services research, several authors highlight their importance, noting that the burden of chronic skin diseases is not fully captured by metrics such as DALYs (26) and calling for more comprehensive assessment of psychological, social and occupational impacts (27). This perspective is reflected in the emerging field of social dermatology, which highlights the role of social and structural influences on health (28).

Our findings support and extend this perspective by demonstrating that the challenges associated with ichthyosis arise not only from physical symptoms, but from their interaction with contextual factors, including social responses, medical structures and access to support systems. Interpreted through the lens of the ICF, these findings illustrate how such contextual factors shape participation in social and occupational life. Participants' accounts describe how visible symptoms, as bodily manifestations, become socially interpreted within specific environments, leading to stigmatizing reactions and exclusion. These experiences are internalized and contribute to psychological distress and withdrawal, which in turn restrict participation in various life domains.

These findings are further reflected in the nature of the psychological strain associated with ichthyosis: while previous studies have documented psychological comorbidities, they have not examined the underlying mechanisms (18–20). In our study, psychological burden was influenced by social factors, unequal opportunities for participation and negative experiences with healthcare services. The variation in psychological burden across participants, despite similar physical manifestations, underscores the importance of contextual and individual factors in shaping lived experience.

Future research should therefore focus on identifying factors that promote successful adaptation and effective disease management. Based on our findings, such factors may include early psychosocial support within the family during childhood and adolescence – a strategy whose positive influence on symptom severity, quality of life, parental stress and disease control is well documented for other chronic skin conditions (29). Additional key elements may encompass assistance in developing individualized care routines, facilitation of access to social-medical benefits and, importantly, recognition and validation by healthcare professionals.

A recent qualitative study from the Netherlands (16) analysed lived experiences through a biopsychosocial quality-of-life framework, reporting similar themes like psychological burden, managing treatment and dermatological care and social and occupational challenges.

Our study explicitly examines the dimensions of participation and disability. These findings are particularly relevant in the context of the German social security system, which is embedded in social law (SGB IX) and provides the legal basis for compensatory measures for individuals who meet criteria for disability. Depending on the degree of disability certification, patients may be eligible for various forms of support, (e.g.) free companions at social events enhance security and reduce stigma-related strain; financial compensations like tax relief offset condition-related costs; legally mandated extra leave aids time-intensive care and appointments. Our results suggest that participation restrictions and contextual barriers, as described by participants, are highly relevant for these assessments and should be more systematically considered in clinical documentation and decision-making processes.

Considering ichthyosis from an ICF-based perspective – rather than focusing solely on HRQoL – is meaningful for several reasons, particularly from a social medicine viewpoint. While HRQoL captures subjective health, well-being, and daily functioning, it neglects societal, legal and structural factors like discrimination, limited accessibility, insufficient support or lack of recognition in social systems. A

participation- and disability-oriented framework ensures patient-centred care aligned with WHO's people-centred healthcare (28, 30).

Patients' narratives align with Narrative Medicine approaches (31), demonstrating how illness stories can inform clinical training and improve care delivery for rare and complex diseases. Integrating such lived experiences into dermatological education, alongside biomedical knowledge, could enhance clinicians' recognition of care inequities (32).

Dermatologists should be aware of their role not only in medical treatment but also in supporting patients in navigating social support systems. This includes the assessment and documentation of functional impairments and participation restrictions, as well as the provision of medical documentation required for access to social and financial benefits.

Limitations

Limitations include underrepresentation of individuals with a migration background and potential selection bias toward patients with existing support structures, possibly underestimating the overall impact. The sample also shows an uneven distribution across gender and age groups; this does not compromise the qualitative insights, as the aim is to capture a broad range of experiences rather than population-level representativeness. The subsequent questionnaire study will target a broader and more representative sample. The presentation of results only partially reflects the interpretative depth of the analysis, as priority was given to capturing the breadth of thematic domains, resulting in a more semantic presentation.

Conclusion

This study identified patient-centred dimensions of lived experiences with ichthyosis, illustrating how the condition affects multiple life domains and shapes patients' social and occupational biographies. Some of the greatest challenges according to the patient narratives in this study were less about the symptoms of the disease itself, and more about the social barriers faced on a daily basis. These findings are now used to inform the development of a patient-centred questionnaire, which may serve as a basis for the future improvement of patient care.

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from the corresponding author and may be provided upon reasonable request, subject to ethical and data protection requirements.

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