Social Science & Medicine 388 (2026) 118723

Contents lists available at ScienceDirect

Social Science & Medicine

journal homepage: www.elsevier.com/locate/socscimed

Adults with sickle-cells in a migration-driven rare disease setting: Therapeutic remoteness and graphic medicine in Austria

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ARTICLE INFO

Handling Editor: Susan J. Elliott

Keywords:
Rare disease
Hemoglobinopathy
Sickle-cell disease
Therapeutic remoteness
Remote biosociality
Medical graph
Austria

ABSTRACT

When hemoglobinopathies, notably thalassemias and sickle-cell disease move from countries with a higher prevalence to central and northern Europe these genetic conditions become particularly rare. In such a migration-driven rare disease setting, people with a hemoglobinopathy, their partners and care-givers or carriers of the genetic variation find themselves "remote" from biomedical expertise, socio-cultural disease awareness and biosociality. Yet, rare disease support initiatives and rare disease patient activism in central and northern Europe often lack a diverse ethnic, cultural, linguistic and biosocial presence. Moreover, progress in clinical and self-management has allowed hemoglobinopathies to change from previously fatal pediatric diagnoses into care-intensive chronic conditions. Since rare inherited blood disorders start in early childhood, however, these conditions still are firmly embedded in pediatrics. These new generations of adults living with a hemoglobinopathy thus also find themselves remote to the pediatric expertise with rare diseases.

This article proposes the notion of "therapeutic remoteness" with its spatial and temporal dimensions, to address the understudied rare disease–migration intersections and the often overlooked biosocial challenges adults with a rare disease are struggling with. An Austrian case study demonstrates therapeutic remoteness in its two dimensions. Focusing on a book project by a hemoglobinopathy patient advocacy group in Vienna, this article also scrutinizes the potential of visual story telling as a beneficial medium in the rare disease-migration nexus.

1. Introduction

The rare disease healthcare landscape is globalizing and uneven. Population movements have led to a steady increase of people living with, or carrying a gene variation for inherited blood disorders in Europe. When hemoglobinopathies, notably thalassemias and sickle-cell disease, move from countries with a higher prevalence to central and northern Europe these genetic conditions become particularly rare and the boundaries between endemic and rare disease become blurred (e.g. Aguilar Martinez et al., 2014; Hemminki et al., 2015). Thanks to the development of cross-border European initiatives and targeted National Action Plans, rare disease (RD) patients and care-givers increasingly find care in specialized biomedical reference centers as well as support and advocacy through disease-specific patient organizations. Yet RD support initiatives and patient activism often lack a diverse ethnic, cultural, linguistic and biosocial presence although these variables (and "race" in non-European contexts) habitually play a crucial role in epidemiological data and in clinical trials concerning inheritable RDs. According to the 2021 Census, one fifth of the Austrian population was not born in Austria and 17.5 % of the foreign population has a citizenship of a country from outside Europe (Statistics Austria, 2024, pp. 30–31). This significant share of migrants, however, is not reflected in the institutional composition of offices supporting self-help groups and RD arenas.

Through an Austrian case study, this article explores in how far the concept of "therapeutic remoteness" provides for an improved understanding of strong intersections between RDs and immigrant population groups, if specific visual methods are pursued. When language and institutional barriers characteristic of migration health (see e.g. Onalo and Flaherty-Gupta, 2022) intersect with the rareness of a condition, access to quality care is further complicated. In migration-driven RD settings, I argue, people living with a hemoglobinopathy, caring partners and relatives or carriers of the causative genetic mutation find themselves "remote" from biomedical expertise, socio-cultural awareness and biosociality. This spatial aspect makes up for the first of two dimensions of marginality in the RD landscape that I call "therapeutic remoteness"; the second aspect of marginality addresses time. The notion of

This article is part of a special issue entitled: Rare Diseases, Global Health, and Social Sciences: Counterbalancing Biomedical Reductionism.

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https://doi.org/10.1016/j.socscimed.2025.118723
Received 25 June 2024; Received in revised form 7 October 2025; Accepted 26 October 2025
Available online 31 October 2025

therapeutic remoteness builds upon the anthropological concept of *remoteness*, which draws from Edwin Ardener's seminal text (Ardener, 2012[1987]) and has regained popularity in theorizing within the last decade (Saxer and Andersson, 2019).

The second, time-related dimension of therapeutic remoteness that people with rare hemoglobinopathies face in Austria concerns the expansion of life expectancy into adulthood. At least in affluent countries with robust healthcare systems, progress in clinical and self-management has allowed hemoglobinopathies to change from what had previously been fatal pediatric diagnoses into manageable care-intensive chronic conditions. The first generations of people surviving childhood with such a severe debilitating inherited blood disorder now face additional health challenges as adults. Since about 70 % of RDs, including rare inherited blood disorders, start in early childhood (EURODIS, nd.) and predominately are studied in these early stages of life (Jae, 2018), these conditions still are firmly embedded in the scope of the pediatric branch of medicine. This adds age as a temporal layer to the spatial dimension of therapeutic remoteness: adults living with a hemoglobinopathy in Austria are "remote", both from the disease expertise in endemic countries and from the RD expertise of pediatrics. The dominant biomedical pediatric perspective in RDs, I argue, has a blind spot for the kind of social challenges adults with rare conditions struggle with. For example, the need to explain one's RD to the family and social environment.

With an ethnographic focus on the case of developing a book project with a hemoglobinopathy patient advocacy group in Austria, this paper scrutinizes the potential of graphic narratives as a beneficial medium in the RD–migration nexus. Visualizations are indispensable tools in modern medicine, ranging from imaging diagnostics to hasty scribbles used in doctor-patient communication. In the RD field, visual expressions of health, illness and healing such as knowledge graphs, drawings, videos, flowcharts, pedigrees demonstrating inheritance of genetic mutations and medical graphs also serve as practical tools to explain complex content in simple ways. Moreover, in the RD–migration nexus, where inheritable complex multisystem disorders and therapy schemes often have to be communicated across language barriers, visualizations are indispensable. The juxtaposition of text and image effectively conveys information and suggests greater accessibility of messages. Visuals allow for communication, self-reflection or for gaining a different perspective on an RD. Patient experiences communicated in this way may "form a powerful counterbalance to the mystifying medical discourse that can otherwise leave patients feeling overwhelmed, unempowered, and detached from their own bodily experiences" (Hamdy, 2023, p. 225).

This text scrutinizes whether visual storytelling, exemplified by the "Mom has sickle-cells" book project, can serve as a supporting medium in migration-related RD contexts to communicate information, thereby addressing therapeutic remoteness in its spatial and temporal dimensions. Drawing on ethnographic data of engaging in the book project, the article aims to shed light on the understudied RD–migration nexus, to contribute to the expanding and globalizing field of social science research on rare diseases (e.g. Dyson and Atkin, 2011) and to the growing field of medical anthropologists engaging with visual media (recently Hamdy, 2023; AGEM, 2022). In what follows I will shed some light on the underexposed intersection between RDs with contexts and biographies of migration, on crucial constraints and specific forms of vulnerability involved, and on potential fields of agency that may unfold within this intersection. First, I will introduce the theoretical concept of therapeutic remoteness in its spatial and temporal dimensions and present the specific setting of Austria followed by some basic information on sickle-cell disease. Subsequently, I will introduce the comparative ethnographic method I apply to the non/endemic RD fields and the engaged medical anthropology approach I follow in the academic and social struggles against health inequality. Then I will turn to the *Mom has sickle-cells* book project, both to reveal the challenges of biosociality formation in remote contexts and of living with an RD as an adult.

2. Background: two dimensions of therapeutic remoteness and sickle-cell disease in Austria

Hemoglobinopathies are unevenly distributed around the globe. The geographical distribution of cases as well as the rising life expectancy of people with inherited hemoglobin disorders reveals structural vulnerabilities, captured here as therapeutic remoteness in space and time and demonstrated by the case of sickle-cell disease.

2.1. Spatial dimensions of therapeutic remoteness

An estimated 5 %–7 % of the global population carries a gene mutation affecting the production or function of the hemoglobin molecule (Modell and Darlison, 2008). These genetic variations are endemic and thus more common in tropical and sub-tropical regions, where they developed over evolutionary periods in response to the malaria parasite (Piel et al., 2017, pp. 1561-62; Piel et al., 2010). In more temperate regions these genetic traits are rare.

The Republic of Maldives, for example, is an equatorial country with a documented 600-year history of severe "Maldive Fever" (Knoll, 2020). Only the eradication of malaria in the 1970s allowed for the development of the country's signature tourism industry and Maldives' rise to the wealthiest nation in South Asia. In the course of analyzing endemic hemoglobinopathies in the Maldivian archipelago I have introduced the notion of "therapeutic remoteness" (Knoll, 2021a) by discussing margins of care in developing parts of the world and in outer island locations (Knoll, 2021b)(. In its original Maldivian setting the notion addresses health disparities due to both the archipelago's postcolonial location in the Global South – remote from the centers of the Western world that drive international research in hemoglobinopathies – and to its inner socio-spatial struggle with health inequities between the capital Male' and more remote outer islands. The theoretical concept of therapeutic remoteness facilitates to document, analyze and criticize health margins in an uneven global hemoglobinopathy healthscape shaped by geography, genes, and human agency. In the present text, the notion of therapeutic remoteness is applied to and further expanded for the non-endemic case of Austria where hemoglobinopathies are rare.

Building upon the anthropological literature emphasizing that remoteness is not given but made, therapeutic remoteness refers to peripheries "not properly linked to the dominant zone." In an asymmetrical health landscape, "the remote" is relational, "compounded of 'imaginary' as well as 'real' places" (Ardener, 2012[1987], p. 532, 521). Hence remote positions are associated with, but not identical to, infrastructurally weak areas, or to distance or marginality. Therapeutic remoteness is not congruent with rural health, as the case of RD demonstrates, but it addresses equally disadvantageous positions also deriving from "dynamic historical and geographic process[es]" (Saxer and Andersson, 2019, p. 143).

By 2010, Europe hosted the largest immigrant population among all world regions (Acevedo-Garcia and Almeida, 2012) and hemoglobinopathies had become the most common RDs in ten European countries (Aguilar Martinez et al., 2014, pp.1-2). Five years later the civil war in Syria significantly increased the influx of refugees and migrants from this endemic region. Estimates speak of some 855,000 migrants from high-prevalence countries arriving in Europe between 2012 and 2019 (Angastiniotis et al., 2021, p. 9803). Sangeeta Chattoo (Chattoo, 2018, p. 32-33), p. 32–33) has rightly warned about a "global health crisis rhetoric" while the Thalassamia International Federation (TIF – Thalassaemia International Federation, nd.), also quite appropriately, emphasizes the challenges some 30,000 new patients in the European Union have to face since they do not fit biosocially into the dominant healthcare landscape: they do not match central and northern Europe's localized biology (Lock and Nguyen, 2018, pp. 319-20). The health realities of people with inherited hemoglobin disorders and migratory roots are "embedded in multiple places" (Acevedo-Gracia and Almeida 2012, p. 2055): genetically connected to an endemic area while

therapeutically and biosocially not yet truly arrived in a non-endemic host country. Only a small number of European countries have, for example, included sickle-cell disease (SCD) in their national newborn screening panels. Austria and Switzerland are not screening for SCD; Germany only since 2021. This article explores forms of therapeutic remoteness shaped by migration and age with a focus on adults living with SCD in Austria.

2.2. Temporal dimensions of therapeutic remoteness: beyond pediatric medicine

Within the last two decades, progress in clinical management and in patient advocacy has brought hemoglobinopathy patients out of the pediatric ward. A previously fatal pediatric diagnosis has changed into a care-intensive chronic condition. At least in affluent countries with robust healthcare systems, first generations of people with inherited hemoglobin disorders such as sickle-cell disease have invested extensive precautionary self-care work to avoid severe crisis and moderate chronic symptoms to be in good enough health to work (Dyson et al., 2021) and start a family. Yet one of the major challenges of living as an adult with an RD is counterbalancing a biomedical pediatric stronghold and finding adult-centered care.

About 70 % of RDs start in early childhood (EURODIS, nd). This is the reason why medical knowledge has been built up, trained for and still remains firmly embedded in the curriculum and scope of pediatric medicine. RD patients find themselves in a challenging phase of transition to adult care (Inusa et al., 2020) that often entails a change of clinics, doctors and modes of care. There are, however, very few truly multidisciplinary centers for adult RD needs, as Toledano-Alhadef et al. (2020) exemplify for neurofibromatosis type 1 syndrome. The challenges of adult living with and aging with an RD are even more understudied than the transition period.

The rareness both of the disease as such, and of people living as adults with this RD adds to the manifold structural factors impacting health outcomes in migrant populations (e.g. Viruell-Fuentes et al., 2021). The range of supportive information brochures for adults with an RD is scanty. The brochure "Sickle-Cell Companion" (Novartis, nd.) is a welcome exception. Yet available information becomes even sparser once it concerns starting a family and being a parent with sickle-cells.

2.3. When remoteness and rareness intersect: the case of Austria

Austria is a small, affluent landlocked Alpine country of nine million people. Some 450,000 of them live with an RD according to Pro Rare, an Austrian umbrella organization for RDs (www.prorare-austria.org) framed by a National Action Plan for RDs from 2015, which is currently being considered for revision. In Europe an RD is defined as a condition that affects fewer than one in two thousand people (Haendel et al., 2020). With no malaria history Austria is not endemic for hemoglobinopathies but nevertheless has – interlinked with migration – some 200 patients with SCD and thalassemias. Yet case numbers remain vague, since in contrast to endemic countries there are no registries for hemoglobinopathies in Austria. With insufficient "small data" (Rajtar, 2023) hemoglobinopathies remain "invisible" within public health. People with hemoglobinopathies, their families and care-givers, I argue, are structurally in a "remote" situation in Austria: remote from the quality of experienced therapeutic care in specialized centers, remote from a disease-aware surrounding population, and remote from the kind of the strong patient voices and vibrant biosociality we see in endemic tropical locations such as the Maldives, or in high prevalence Mediterranean countries such as Cyprus, Italy, and Greece, or in the UK with its large immigrant communities.

2.4. A short note on sickle-cell disease

Sickle-cell disease (SCD) is an autosomal-recessive inherited genetic disorder of the hemoglobin – the iron-containing and oxygen-transporting protein of the red blood cell. In SCD the blood contains sickle-shaped red blood cells in addition to the usual round red cells. The latter, smooth and flexible, easily slip through blood vessels when carrying oxygen to each and every cell in the body. The former, by contrast, are at risk of turning hard and sticky when lacking oxygen. In this case the sickle-cells may intertwine and block the blood flow, resulting in a severe pain crisis and necrosis in the undersupplied tissue. As a complex multi-system disorder, SCD "combines features of acute illness (acute painful crises, life-threatening infections); chronic illness (severe anemia/fatigue and chronic pain) and disability (strokes, recurrent leg ulcers and necrosis of the joints" (Dyson et al., 2021, p. 1). The cumulative effects of these medical complications across the life course have repercussions in limiting options to participation in social life (Swanson et al., 2011).

SCD occurs in approximately 275,000 of 330,000 babies born with a major hemoglobinopathy worldwide. 66 % of the 120 million people living with SCD live in sub-Saharan Africa, where health resources are limited (Inusa et al., 2024; (Integrated African Health Observatory & World Health Organization African Region, 2024). This uneven geographical distribution is driven by two factors: endemic malaria and population movements. The sickle-cell gene HbAS provides protection against severe *Plasmodium falciparum* malaria, which explains its frequency across malarial sub-Saharan Africa and parts of the Mediterranean, the Middle East and India. Population movements, including the slave trade, economic migration, flight and asylum distributed SCD to the Americas and Europe (Kato et al., 2018). Central and northern Europe have no malaria history. In these non-endemic settings in Europe SCD ranks among the RDs, defined as conditions that affect fewer than one in two thousand people (Haendel et al., 2020).

When SCD migrates and changes from a pressing public health concern in endemic tropical underserved regions to rareness in Europe "the conundrum of the location of health resources and health need," which an anonymous reviewer of this paper has rightfully highlighted, is by no means solved. Rather it is reversed; comparable to the difficulties migrants face when suffering from neglected tropical diseases (Gold, 2021). SCD is not included in the newborn screening and registries for SCD are not established in Austria. Due to the rareness of hemoglobinopathies in central and northern Europe and due to the even smaller case numbers over 18 years old, adults with sickle-cells, in fact, often are not able to benefit from the quality of care their new place of residence can provide (Angastiniotis et al., 2021, p. 9803). An estimated 120 people live with SCD in Austria; only about 15 % are adults. This article addresses adults living with rare SCD in the non-endemic therapeutic remote context of Austria.

3. Methods

This article draws on a research project focusing on health-equity questions regarding migration-related hemoglobinopathies in Europe and from my involvement with a patient group in Austria. From 2019 to 2021 I contributed to the THALIA – Thalassaemia in Action project of the Thalassemia International Federation (TIF), which was co-funded by the Third Health Program of the European Union (Angastiniotis et al., 2021)(. The formation of patient advocacy groups in selected under-served European countries was an integral part of the project and inspired my conceptual and factual engagement with what I call "remote biosociality".

Since May 2019, I have participated in four delegations to Austria and Germany and three physical and two online workshops as well as in 32 group meetings online and offline in Austria and Germany. I kept an ethnographic field note book for my observations and critical reflections. In addition to participant observations I conducted semi-structured individual and group interviews with clinicians (n = 5), patients (n = 12; five with SCD and seven with thalassemia), partners (n = 3) and care-givers (n = 2) in Austria and Germany. About 40 % of the

research partners agreed to recordings; in the other cases I took notes. The formal interviews were carried out in the offices of the clinicians and in the private homes of the patients, partners and care-givers. All interview partners gave informed verbal or written consent before the interview started. Selected parts of the recordings were transcribed, anonymized and then content analyzed together with my written field notes. I triangulated significant insights in informal talks. Research partners were predominately addressed through established professional contacts and through the activities of the patient organization.

The present article's arguments are further informed by a "controlled comparison" (Gingrich, 2015) between the experiences of living with a hemoglobinopathy in non-endemic Austria and an endemic country such as the Maldives, where I did research for more than a decade. In particular, my ethnographic research about genetic responsibility and remoteness, carried out between 2018 and 2024 in the Maldives, forms the backdrop for this analysis and allows for a more nuanced perspective on the situation in Austria. In response to the travel restrictions during the COVID-19 pandemic years, the project expanded to migration-related aspects of hemoglobinopathies in Austria.

Furthermore, I pick up on Bridget Bradley's (2021) argument for "biosolidarity" as an anthropological method of engagement for my commitment to patient concerns in Austria and the Maldives. Yet my position differs from Bradley's insider/outsider position as a scientist and as someone living herself with a chronic condition and actively engaging in advocacy. Hence as an academic I engage in the dimensions of social critique, collaboration and advocacy according to Low and Merry's (2010, pp. S208-S211) seminal categorization of engaged anthropological work, based on my understanding of a healthcare setting in South Asia, where hemoglobinopathies rank among the most pressing health concerns. I thereby face a kind of "double contention," which Dueholm Rasch, van der Hout and Köhne (2022) have emphasized in engaged anthropology and scholars' activism. In an act of biosolidarity I am involved in social struggles against health inequality while I criticize the structural lack of diversity in RD and support-group settings.

4. Findings

4.1. Group formation across a group of disorders classified as hemoglobinopathies

The first patient advocacy group for inherited rare blood disorders in Austria was formed in 2019. This process has resembled the "formation of new group and individual identities and practices," that Paul Rabinow (Rabinow, 1996[1992], p. 99, 102) describes as emerging around a specific allele variant. The analysis of genetics as "a circulation network of identity terms and restriction loci" is the aspect that is particularly relevant here from Rabinow's seminal work: it highlights the expansive dynamics of the ways that our cultural understandings of "nature" are, and always have been informed by human practices. Coining the much cited term "biosociality" (e.g. Gibbon and Novas, 2008), Rabinow refers to the example of neurofibromatosis groups, "whose members meet to share their experiences, lobby for their disease, educate their children." Rabinow in fact used an RD to explain biosociality, but did not use the term rare disease, which first appeared on US health policy maps in 1984 and was gradually adopted by other states over the following four decades (Rajtar and Knoll, 2024:354). In contrast to neurofibromatosis group formation around gene variants on chromosome 17, and by strategically empowering the small number of patients in a non-endemic location, the Austrian collective is built around a group of disorders classified as "hemoglobinopathies" in Orphanet. Orphanet is an online initiative established in 1997 that provides knowledge of the RD ecosystem and creates and maintains the RD nomenclature for Europe (www.orpha.net). The formation of remote biosociality in Austria across two groups of disorders, thalassemias and sickle-cell disease caused by gene variants on chromosomes 11 and 16 is challenging. Patients living with one of these two groups of diseases have little in common apart from sharing the rarity of a gene anomaly affecting the production of hemoglobin and a chronic and debilitating course of disease progression. Although the majority of them receive treatment in the same hematology departments, hemoglobinopathy patients do not automatically form a collective there. They are rather dispersed as they only account for about 1 % of the predominately cancer patients. It was the collaborative work on a book project that boosted the confluence into a patients' group by heterogeneous and dispersed people.

4.2. An illustrated book on family life with sickle-cells

What is seen as the most pressing concerns of people living with an RD is predominately defined from a biomedical perspective. Socializing and sharing anecdotes oscillating between treatment experiences and family life, however, the group of adult patients, partners and caregivers in Vienna identifies an urgent need to explain a parent's rare chronic condition to a child. "Speaking with kids about their mom's or dad's disease," has become an issue. An illustrated children's book is envisioned as a helpful tool. It tackles the consequences of SCD labeling parents and their children as socially different, limiting their social participation and requiring certain adjustments in everyday life (Von der Lippe et al., 2017, p. 767). The book is intended to help a child discuss and understand why Mom is different; why she is sometimes tired or in pain, why she's taking medicine, observes some precautions and why she is occasionally in hospital. There might be a particular need for such a book in regions where SCD is rare rather than endemic and where a child most probably is not surrounded by other family members, distant kin, neighbors and schoolmates with SCD. It might help to make more sense of situations where Mom is sticking out with a disease nobody has ever heard of.

The book project provided a space to address and discuss issues entangled with the rare blood disorder though not being discussed within conventional biomedical consultations. The negotiation of different perspectives and attitudes thereby underlines the social component, as the following ethnographic vignette illustrates:

"Never ever you touch my medicine drawer!" she firmly told her kids repeatedly, explains Anália while we stand in front of the drawer in her apartment in the outskirts of Vienna. From early on, since they were toddlers, she taught her two daughters to always stay away from her medicine. Anália lives with sickle-cell disease. When I asked if I could take a picture of her for a lecture I would give in a nursing school she wanted me to show her in front of this drawer, since it plays such a significant role in her life. It contains vitamins, antibiotics, the disease-specific chemotherapy agent Hydroxycarbamid as well as highly effective painkillers including opiate-based analgesics. "The essentials" in managing daily life as a chronic-disease patient; especially in managing looming episodes of pain crisis which are characteristic of Anália's blood disorder (Interview 12 Nov. 2019).

"I always included my kids", counters Shadanaa, also a SCD mother, when months later the three of us meet on zoom to discuss the challenge of having powerful drugs *and* curious children at home. "I always let them be part – of my need to rest, of my efforts to hydrate, of my pain, and, also, of my medication," explains Shadanaa. "Children feel valued," she argues,

> when being included. It takes away some of their fear … and it is important … One day, my son even saved me! He was very small; not even three years old when it hit me in the bathroom. I never had much trouble with pain crisis before, but suddenly, it hit me like a stroke. The pain was so sudden, so intense. I was sitting on this toilet seat, all in tears, I could hardly breathe and I could not move. No way to get up. My little son understood my pressed words "medicine." Off he went and brought the right package.

This lively exchange between two mothers with SCD took place electronically and in English during the COVID-19 pandemic (Notes 20

March 2020). Anália, who came up with the idea of producing a children's book, lives with HbSS; with two genetic alleles of the sickle-cell hemoglobin HbS, associated with the most severe clinical manifestation of SCD. In accordance with the clinical guideline for SCD in Germany (Hoferer et al., 2021) we also avoid the older, trivializing term sickle-cell anemia in Austria. Shadanaa has one son below ten and one teenage daughter. She lives with a compound-heterozygous form of two gene variations causing SCD and beta thalassemia (HbS/βthal); the clinical manifestation, however, resembles SCD. In the end, the online exchange between Anália and Shadanaa (both pseudonyms) culminated in an agreement: since their medicine cabinets make them stick out in a crowd of mothers, "mom's medicine cabinet" definitely has to become a chapter in the prospective children's book.

The book-in-the-making is to serve as a supportive tool in explaining their mothers' SCD to children. It addresses social problems arising from and with this rare chronic condition. Being a mother and a sickle-cell patient means, first, juggling with curious children and powerful, even deadly medicine that should always be kept within easy reach. Secondly, being a mother and a sickle-cell patient in an environment which is decontextualized from family, kin, and neighbors who might also have this disorder requires dealing with the child's questions induced by the situation of therapeutic remoteness.

4.3. Graphic medicine and experiential knowledge

For more than a decade there has been growing interdisciplinary scholarly interest in the therapeutic, communicating and analytical powers of medical visualizations summarized under the term graphic medicine (Czerwiec et al., 2015; Green and Myers, 2010). Recently, these potentials were readily taken up in the COVID-19 pandemic (West et al., 2021). Furthermore, a comic, a picture or a caricature can address what might not be easy to be talked about or captured in spoken language. For instance, in her graphic memoir or pathographic, Sarah Lippett (2019) shares touching moments of her long and eventful journey to the diagnosis of the rare Moyamoya disease. Engaging readers "on a deeper, more sensorial level" the creative genre of visual story telling allows both for "breaking the taboo of silence" or simply "show [ing] things as they are" (Hamdy, 2023, p. 232, 243) and "illustrat[ing] the ordinary" (McMullin, 2016, p. 151). Moreover, graphic narratives provide optimum communication, as McMullin (2016, p. 162) demonstrated for cancer, to narrate lives deeply marked by the difference of experiencing a disease.

The contrast between regular red blood cells and sickle-cells is the prominent iconic visualization of SCD – and the vehicle for explaining the disease as well as for its biomedical reductionism. A limiting perspective on sickle-cells-cause-blockage (Dyson, 2019, p. 20) overlooks the kind of societal contexts and "vital adaptation techniques" Duana Fullwiley (2011, pp. 20-22) describes as preventing or mitigating sickle-cell suffering in West Africa. This "enculturation" of SCD in Senegal combines the consumption of a traditional local medicinal plant with socio-economic care structures and safety nets that draw on idioms of a shared same kind of (sickling) blood and thus on familial solidarity (Fullwiley, 2011, p. 78, pp. 49-52). "Rather than a physical shape (sickle) and a mechanical process (blocking)," argues Dyson (2019, p. 20), "we have a complex series of dynamic biological processes: these processes are complex, dependent on context, interactive and cumulative" – with a significant impact on the aging body.

Since March 2020 three women have formed the core team to develop the general framework and financing of the book; Anália, Shadanaa and myself. We decided on a female perspective and focus of the book and we hired a female creative team comprising an illustrator, a graphic designer and a text writer. Subsequently, the content of the thirteen chapters was developed in six online meetings with group members in Austria and the German-speaking countries.

Mom's sickle-cells are the general topic of the book, though it is mentioned that dads and boys can also have SCD. Fathers are portraied as active and supportive family members, feeding a youger sibling, carrying shopping bags and talking to the doctors in the emergency case of a sickle crisis. In its focus, however, the book draws on the mothering experiences with sickle-cells – on what is identified as "experiential knowledge" in the literature (e.g. Rabeharisoa et al., 2014) in contrast to credentialed (formalized, biomedical) knowledge. Experiential knowledge is gathered by "experts of experience" (Rabeharisoa and Callon, 2004) – and not just by "lay people." "The emotive delivery of the message through visualization [in the book thus] breaks through the opposition between the vulnerable body on the one hand, and the authoritative [biomedical] knower on the other" (Hamdy, 2023, pp. 245, annotation by the author). The social challenges for mothers with sickle-cells cannot be reduced to a limited focus upon diagnosis, treatment, and prevention. It is the experiential knowledge of mothers with sickle-cells that matters.

4.4. Family life with sickle-cells

A biomedical perspective and the core of educational brochures restrict the attention to biochemical relations "within" the body. The "Mom has sickle-cells" book, by contrast, also brings social relations "beyond" into the picture (pun intended). Illustrations and text explain biochemical sickling processes and the biomedical intervention of pain medication within Mom's body and mind though focusing on their impact on family life and on addressing them in a child-friendly manner. Each of the book's thirteen chapters covers a double page. In addition to illustrations, there is a continuous text to read or read aloud and an info box to provide additional information. We envision that parents, family members or care-givers will read the book together with younger children while older ones have the book at hand in their bookshelf.

The book ambitiously aims to serve all age groups, with a plot, a textual and an artistic style "growing along" with the child. In the first chapter a small girl is temporarily staying with her grandparents and visiting mom in hospital. In this chapter, the parents wanted to emphasize that they had been forced to make major compromises in their role as parents. The stories shared in group meetings about their children's early years fueling this chapter included that some had missed the first steps of a child due to hospitalization with a sickle-cell crisis, others had never known what it felt like to carry a child because the family consider this too exhausting.

Proactive prevention of sickling crises and the family support network are addressed in the chapter "Mom's Hydration Day." If things have been too busy, the whole family slows down and spends a quiet day at home. With simple things like drinking a lot and eating watermelon and salads the family aims to positively influenc the body's water balance. Situations in which adequate fluid intake was forgotten and which resulted in a sickle crisis or were barely averted dominated the discussion. The group nevertheless has decided to emphasize agency by highlighting preventive measures.

Chapter 11 is about visiting Mom in a rehabilitation clinic after shoulder replacement surgery. The discussions contributing to this chapter included endless examples of complaints with the musculoskeletal system at a strikingly young age. Equally exhaustively discussed was the difficulty in finding orthopedic specialists who also have some knowledge of SCD and its specific impact on an aging body.

The final two chapters of the book are dedicated to teenagers. A teenage boy gives a presentation on his mom's SCD in school. In the discussions parents with sickle-cells emphasized with admiration how impressive is the scope and quality of sickle-cell knowledge their children have gathered over the years and how supportive they are in their descriptions and arguments. The medical anthropologist Chloe Silverman (2012, pp. 93–196) highlights the emotional connections and affective commitments in parent-child dynamics shaped by chronic disorders such as autism. Empathy and detailed knowledge accumulates through "the common currency of experience" and generates "expert amateurs" among relatives. In this chapter this is a teenage expert

amateur. "How beautiful is that! We are raising a next generation of health advocates," exclaimed Maria Hadjidemetriou, a thalassemia major patient, mother, Executive Board Member at Cooley's Anemia Foundation, USA, and founder of Thalassemia Mommies on FaceBook, when at an international conference she described that her 12-year-old had become so knowledgeable about thalassemia that she gave a talk on her mother's condition at school (Hadjidemetriou, 2021; TIF – Thalassaemia International Federation, 2022, p. 15).

The last chapter of the book is "What have mom's sickle-cells to do with me?" The fact that their children inevitably inherited one copy of the sickle-cell gene is of concern. The Mendelian genetics repeatedly pop up in the discussions, according to which a union between two (heterozygous) carriers has a 25 % chance of conceiving a (homozygous) child with SCD in each pregnancy. Endemic countries like the Maldives run governmental screening programs in collaboration with schools to identify hemoglobinopathy carriers in the population already in their teenage years. Teenagers in non-endemic Austria most probably learn about the pea plant experiments by the Augustinian cleric Gregor Mendel (1822–1884) in the Austrian-Habsburg realm and his discovery of the fundamental laws of genetic inheritance. Yet Austrian teenagers certainly do not discuss the probability of being a so-called healthy carrier of an RD trait in school and do not take the carrier test collectively. Parents with sickle-cells and their partners therefore feel compelled to talk about the family's genetic heritage. The chapter pictures a girl on a first date with a boy. The text reveals that she feels this might turn into a serious relationship. Of course, as a 16-year-old she is not yet thinking about getting married and having children, but she is wondering when would be the right time to address her mother's sickle-cells and the fact that she is a carrier of the causative gene.

The general idea of the book is to provide an accompanying and supporting tool. With the goal of teaching and empowering in mind, the envisioned book should help, a) to explain mother's disease; b) to teach children how to respond to their mother's condition; c) to share knowledge about the condition with the family to overcome stigma and fear; and d) to change the perception of SCD of the child, the family, health personnel and the wider public. The book is thus meant to be positive by presenting the rare condition and resulting challenges and risks along with care options within and beyond the biomedical realm. Mothers with SCD should not be portrayed as fundamentally ill, tired, weak and fragile. Quite to the contrary, people who are active in patient advocacy often call themselves "warriors" (see e.g. Knoll, 2020, p. 256; Dyson, 2019, p. 9; Leah, 2016, p. 1) to emphasize a fight on many frontlines: they fight a debilitating disease, but also insufficient healthcare, lack of public awareness and various forms of stigma, neglect and discrimination. Chapter nine emphasizes agency by highlighting mom's warrior activities. The *Mom has sickle-cells* book will be a supportive tool in these fights and demonstrate that there are many children with SCD parents around the world.

5. Discussion

The members of the patient advocacy group in Austria identified an urgent need for a book illustrating family life with sickle-cells. On the international stage ample educational materials depict, explain and discuss SCD in graphics and text. Produced by pharmaceutical companies, medical experts or patient organizations such brochures, picture books and comic books address patients, both pediatric and adult, as well as parents and care-givers in various languages (recently e.g. Simon and Kühler, 2024). Some SCD booklets are written from the patients' perspective. The "My Friend Jen" children's book series, for example, is written in rhymes by Jenica Leah (2016, 2020), a so-called "SCD warrior" herself. Yet parenting as a significant aspect of adulthood – being a sickle-cell patient *and* a parent – is lacking as a topic in the field of RD educational reading.

It was thereby particularly important for Anália and Shadaana, "to make it a *real* book; a beautiful book with professional drawings, with an attractive design and of haptic qualitative material." It should "not just (be) another one of these clinical brochures or self-knitted patient group booklets." Instead, SCD should become equal to all the other topics dealt with in appealing books on a child's bookshelf. It should be "a book you like to take out time and again to read with your child on a cozy Sunday afternoon" (Notes 9 Dec. 2022). "Julie and the sea shells" is an example of a hardcover book for children. Although not an RD, people living with lichen sclerosus often realize that this is "a skin condition even doctors are often not familiar with" (Lichen Sclerosus Association Switzerland, 2016, p. 3). With pre-school protagonist Julie the booklet addresses other children with lichen sclerosus. The "Mom has sickle-cells" book, by contrast, has the potential to step out of both the pediatric and biomedical realm by addressing what concerns adults with SCD in Austria. Why this twofold stepping out is not an easy task for the Austrian group becomes clear when we compare their situation in a non-endemic healthcare setting with the endemic Maldives.

SCD has an endemic stronghold in the African continent while in many other parts of the world SCD is linked to historically enforced movements such as the slave trade and to contemporary forms of migration. In the biomedical literature SCD is captured as "an increasing global health problem" based on the argument that the estimated number of approximately 300,000 infants born every year with SCD could rise to 400,000 by 2050 (Piel et al., 2017, p. 1561).

In both endemic as well as migration-inflicted settings, people with sickle-cells often experience health-related stigma – "an attribute that is deeply discrediting" in Goffman's (1986, p. 4) classic definition. SCD is infamous as "the black disease" in the United States, in contrast to two other rare diseases, cystic fibrosis and Tay Sachs, associated with Caucasian and Jewish populations (Wailoo, 2007, p. 236). In India, as a contrasting example, SCD is constructed as a disorder of subaltern segments of the society, in particular of the indigenous Adivasi communities (Das, 2023). In Austria and other parts of Europe such as Germany (Kunz et al., 2020) or Sweden (Kjellander et al., 2023, p. 2; Hemminki et al., 2015) – all of these obviously being locations remote from the endemic belt – SCD is linked to recently intensified migration flows from endemic countries and therefore ranks among the less known of the RD category.

5.1. Remote biosociality

Biosocial remoteness has a tremendous impact on the "Mom has sickle-cells" book project and informs its perspectives on options for agency. First of all, because biosocialities do not simply exist; illnesses themselves do not cause communities to form (Silverman, 2012, p. 193). Sociable relationships between scientists have to be actively generated to promote research in a field where the rarity of a disease "produces the basic condition of marginalization" in research (Panofsky, 2011, p. 32). Also the formation of a patient collective in marginalized RD conditions requires social investment. Condition-centered communities must be found and formed and their sustainability requires ongoing work (Bradley, 2021, pp. 1–3). Yet the coming together of patients, parents, carriers and care-givers in a marginalized remote context, I argue, differs significantly from the kind of gravity that biosociality can build upon in an endemic context. In endemic Cyprus, for instance, the high prevalence of the inherited blood disorder thalassemia is interpreted as a "collective 'ethnic' fate" and "the gene pool as a 'tragic commons' that require[s] collective management" (Beck and Niewöhner, 2009, p. 88). Also in non-endemic yet immigration-shaped countries like Canada, the US or the UK people with hemoglobinopathies find established patient-support groups within the large immigrant communities (see e.g. Dyson, 1998, p. 126). The highly heterogeneous and diverse remote biosociality in Austria, by contrast, has no comparable ethnic, religious, linguistic or genetic gravity or "bio-cultural intimacy" (Beck and Niewöhner, 2009, pp. 87–88) to build upon – beyond a rather recent immigration background and a shared RD experience. At the first meeting of the emerging patient group in Austria I counted 13 patients

with five different hemoglobinopathies, speaking ten different mother tongues; they further had diverse migration trajectories, different legal status in Austria and they mastered the Austrian variety of German as a lingua franca in highly diverse ways (Notes 17 May 2019). The *Mom has sickle-cells* book therefore also has the socio- and health-political mission to draw attention to the small, marginalized and highly heterogeneous group of hemoglobinopathy patients in Austria as well as to propel and empower this remote form of biosociality.

6. Conclusions

This article has proposed the notion of "therapeutic remoteness" to analyze migration-driven healthcare settings where people with rare inherited blood disorders often face inexperienced healthcare providers and an ignorant host population. The Austrian case example has shown how the concept of therapeutic remoteness provides for an improved understanding of the migration-RD nexus, particularly when specific visual methods are elaborated and implemented. The spatial dimension of therapeutically remote RD patients in countries where inherited blood disorders are not endemic intersects with the temporal aspect of aging. RDs and groups of adults with hemoglobinopathies and migratory backgrounds both operate somewhat at the margins.

In such therapeutically remote settings, I argue, graphic medicine helps in voicing social dimension of living with a rare blood disorder and thus in counterbalancing the dominant biomedical discourses. The greater accessibility of illustrated text can be particularly favorable both for RD groups with highly diverse migrant – read language backgrounds – in non-endemic settings and for RDs as complex though marginalized disorders. The collaborative work and group discussions about the content of the *Mom has sickle-cells* book strengthen the formation of RD identities and communities in the non-endemic and thus remote context of Austria. The introduced notion of a "remote biosociality" shall highlight the hardship of assembling and sustaining a supportive patients' group across internal diversity.

Ethics approval/statement

This article draws on research on health-equity questions regarding hemoglobinopathies in Europe and Asia carried out within the project "Genetic responsibility & remoteness" in the Maldives (2018–2024) which received ethical approval by the National Health Research Committee of the Ministry of Health, Republic of Maldives on July 25, 2018; project "THALassaemia In Action: The THALIA project" by the Thalassaemia International Federation (TIF) (2019–2021) co-funded by the Third Health Programme of the European Union under Specific Grant Agreement (SGA) No.101015571, in accordance with the ethical principles of EU-funds and programs; and project "Transition in rare diseases" (since Sept. 2024) funded by the Austrian Academy of Sciences' International Relations Department which received ethical approval by the Commission for Science Ethics of the Austrian Academy of Sciences on 19 Nov. 2024.

Acknowledgements

I would like to thank my colleague Dr. Malgorzata Rajtar (Polish Academy of Sciences) for the fruitful and pleasant cooperation in drafting and organizing the Brocher Foundation workshop on "Rare Diseases, Global Health, and Social Sciences: Counterbalancing Biomedical Reductionism" (May 9–11, 2023, Hermance, Switzerland), which laid the foundation for this SI. I am grateful to the workshop participants for inspiring contributions and comments as well as to my research partners in Austria, Germany and Asia for sharing their experiences with, and thoughts about living and mothering with a rare disease. Substantial procedural and financial support by the Brocher Foundation and by the Austrian Academy of Sciences for the workshop and its academic outcome is duly acknowledged. An earlier verbal version of this paper entitled "Raising Sickle-Cell Advocates – Learning from a medical booklet in the making" was presented and discussed at the "Visual Expressions of Health, Illness and Healing", 34th Annual Conference of the Association for Anthropology and Medicine (AGEM) in Cooperation with the Austrian Ethnomedical Society and the Weltmuseum Wien, June 2–4, 2022, Vienna (Austria). My special thanks go to Andre Gingrich, Malgorzata Rajtar, David Westacott and two anonymous reviewers, who contributed valuable suggestions to the quality of this article.

Data availability

The data that has been used is confidential.

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