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ANTHROPOLOGY, GLOBAL HEALTH, AND RARE DISEASES

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Introduction

When the United Nations (UN) accepted the resolution "Addressing the Challenges of Persons Living with a Rare Disease and Their Families" on December 16, 2021 (United Nations, 2021), the category of rare disease (RD) gained official recognition at the global level. In this resolution, the UN addresses the complex nature of challenges faced by people living with a RD, as well as their families and caregivers, and intrinsically links these to both human rights and the 2030 Agenda for Sustainable Development. Thus, member states, UN agencies, and other stakeholders are urged to design and implement policies and programs to protect and promote the rights of people living with an RD. In February 2022, the International Rare Diseases Research Consortium (IRDiRC), a collaborative initiative with global "aspirations" (Cross and Street, 2022) launched by the European Commission and the US National Institutes of Health in 2011, published its "IRDiRC State of Play" report. This states that the IRDiRC should "facilitate cooperation and collaboration on a global scale" among the various RD stakeholders, and that "global integration and coordination of rare diseases research are necessary to accelerate the understanding, diagnosis, and treatment of rare disorders" (Letinturier-Valencia et al., 2022). If, as many scholars have emphasized, global health (GH) "is about health equity everywhere" (Abimbola, 2018, p. 63; see also, e.g., Farmer et al., 2013; Holst, 2020) and the "burden of illness should be used as a criterion for global-health priority setting" (Koplan et al., 2009, p. 1994), we believe that RDs should be among the most studied areas within GH research and policy. This has certainly not been the case so far.

Drawing from our ethnographic research on RDs in Europe (Rajtar, Knoll) and Asia (Knoll), we critically scrutinize the relationship between the proliferating field of RDs, anthropology, and GH, and we highlight that "the *global* connectedness of diseases and of people and institutions" that characterizes GH (Yates-Doerr and Maes, 2019) is equally important in RDs. Research on RDs is underdeveloped in both anthropology and GH scholarship. Although medical anthropology foregrounds "critical, cross-cultural, people-centered, and transdisciplinary" approaches to health and well-being (Panter-Brick and Eggerman, 2018, p. 234), it has given insufficient attention to RDs. The RD field remains

DOI: 10.4324/9781003284345-31

dominated by biomedical agents in the Global North, despite the UN resolution and bold initiatives such as IRDiRC. Furthermore, even in the resource-rich settings of the Global North, people with RDs rarely experience health equity with patients living with more common conditions.

In what follows, we first characterize RDs and orphan drugs. We address biomedical and anthropological research on RDs, then we present two case studies that illuminate important issues for many people with RDs and aspects of RD policy, such as newborn screening (NBS), migration, and 'therapeutic remoteness': a disadvantageous asymmetry in health care (Knoll, 2021a). Finally, we conclude that more sustained critically reflexive ethnographic inquiry into real-life experiences of RD patients and their families is essential to illuminate both the global expansion of RDs and the prevailing biomedical reductionism of the field. These problems result from the dissonance between the small number of highly diverse RD patients and the GH approach of scaling-up standardized care.

Rare diseases: An emerging global health concern

Also known as 'rare disorders' and 'orphan diseases' (Richter et al., 2015), RDs first appeared on US health policy maps in 1984, followed by those of Japan, South Korea, Singapore, Taiwan, Australia, and the European Union (EU) during the 1990s (Huyard, 2009). Like GH, RDs lack a universal definition (Haendel et al., 2020; Richter et al., 2015; von der Lippe et al., 2017). In the EU and Australia, RDs are defined as conditions that affect no more than one in two thousand people, in the United States those that affect fewer than two hundred thousand people, and in Japan those that affect fewer than fifty thousand. In other countries, such as India, an absence of epidemiological data impedes development of a RD definition (Ministry of Health and Family Welfare, 2021). An estimated 30 million people in the EU and 263 to 446 million people worldwide live with a RD.[1] The number of RDs is currently estimated to exceed ten thousand (Haendel et al., 2020; Rubinstein et al., 2020). Thus, taken as a whole, RDs are not so rare.

Individually scarce but collectively numerous, RDs affect many people. These life-threatening and/or chronically debilitating conditions result in high degrees of morbidity and mortality and thus pose a serious threat to public health across the world (Rubinstein et al., 2020; Council of the European Union, 2009). Some 80% of RDs are of genetic origin; many are inherited and often occur in early childhood. The diagnosis, treatment, and prevention of RDs affect individuals, families, health professionals, public health authorities, population groups, and patient advocacy organizations.

The RD category emerged as a 'by-product' of orphan drug regulations in the United States (Huyard, 2009). Emerging during the 1960s, the term 'orphan drugs' describes pharmaceuticals that are not economically viable for development and marketing, such as those for rare conditions and, therefore, require governmental economic and regulatory incentives (Novas, 2016; Huyard, 2009). The US Orphan Drug Act of 1983 was the first law to address issues relating to rare diseases and conditions and made orphan drugs profitable for the pharmaceutical industry through financial and other incentives (Mikami, 2019). Simultaneously, this act played a pivotal role in fuelling the development of specialist biotechnological companies, thereby increasing therapies for RDs, and it constituted a policy model for countries such as Australia, Japan, Singapore, and the EU (Novas, 2016, p. 183). Rooted in the 'affective economy' (see Aureliano and Gibbon, 2020, p. 249 for RDs) of orphan drugs policies, RDs can be viewed as part of the revitalized magic bullet and phar-

maceutically driven thinking that has marginalized social medicine perspectives (Adams et al., 2019, p. 1385). Despite these targeted political countermeasures, therapies are available for only 5% of RDs.

Rare diseases have been closely connected to knowledge production and, evolving alongside new genetic discoveries, the emergence of new collective and individual identities and practices that have been termed 'biosocialities' (Rabinow, 1996; see also e.g., Gibbon and Novas, 2008; Mikami, 2020; Navon, 2019). Rare genetic mutations have not only become a new way of classifying human difference, but they have also contributed to research, clinical practice, patient advocacy movements, "new hybrid communities" and biosocial identities (Navon, 2019, p. 5). While patient advocacy organizations have actively engaged in the co-production of RD knowledge and assumed "an authoritative position alongside their research and policymaking partners" (Mikami, 2020, p. 149; see also Rabeharisoa and Callon, 2004), various forms of "genetic citizenship" (Heath, Rapp, and Taussig, 2004) and "biological citizenship" (Rose and Novas, 2005) have also emerged. However, some patients, who do not identify with "groups defined by particular diagnostic or genetic conditions", refrain from participating in patient advocacy organizations (Lesmo, 2022, p. 10).

Enriching biomedical perspectives and counterbalancing biomedical reductionism through ethnography

For patients living with a RD, delayed diagnosis and inadequate care often result from a lack of awareness and/or expertise. Thus, people with RDs are regarded as a vulnerable population in health policy documents (Rajtar, 2020). Yet, RD-related vulnerabilities are highly diverse: they are shaped by distinct national and local settings, including "local biologies" (Lock and Nguyen, 2010), local moral worlds, and cultural forms of social suffering. The 2021 Global Needs Assessment Study report identified 10 common challenges and needs that affect diagnosis, care, and treatment of the global RD population (Lopez Gousset and Bolz-Johnson, 2021). Many of these challenges and needs exceed the scope of biomedicine and life sciences; thus, they may easily escape the attention of prevalent methodologies that utilize quantitative metrics and randomized controlled trials (Adams, 2013, 2016). Actors in GH often hold an "unbroken belief in the unlimited curative power of biomedicine" (Holst, 2020). Research and health policies relating to RDs are primarily shaped by the medical-industrial complex, the impact of biomedical reductionism, and the revitalization of magic bullet and pharmaceutically driven thinking (see Adams et al., 2019). Socio-empirical studies, notably ethnographies, critically examine challenges that RD patients and caregivers face, such as, among others, social acceptance, equality, inclusion, political recognition, geographical and cultural contexts, and empowerment. In resource-poor settings, RDs pose yet another burden over and above existing health issues. Among these, communicable and non-communicable diseases, including neglected tropical diseases, are of particular concern. Thus, medical anthropology is well suited to address RDs as a new frontier in GH and counterbalance biomedical reductionism in this field (Rajtar and Knoll, 2023).

While medical anthropologists and other social scientists have examined the lived experiences of people with RDs, they have seldom explicitly advanced or even operated in the field of RDs. Rather, they focus on sociocultural, medical, and technological phenomena, such as, among others, illness narratives and daily life (Larotonda, 2016; Mattingly, 2014); kinship (Beaudevin, 2013); diagnosis and NBS (Timmermans and Buchbinder, 2013);

genetics and genomics, including in the context of race (Featherstone and Atkinson, 2012; Gibbon, Kilshaw, and Sleeboom-Faulkner, 2018; Wailoo, 2007); the production of knowledge (Lambert and Rose, 1996; Callon and Rabeharisoa, 2008; Rabeharisoa, Moreira, and Akrich, 2014); biosocialities (Navon, 2019); orphan drugs (Huyard, 2009; McGuire, 2020); and the judicialization of health (Aureliano and Gibbon, 2020; Biehl, 2014). These categories often overlap. With some notable exceptions, researchers have tended to conduct their studies in western Europe and North America. Thus, like their counterparts in GH, they have neglected the Global South and the resource-poor peripheries of the Global North. While new biotechnologies may facilitate diagnosis, cutting-edge treatment, and prevention for many RD patients in the Global North, their counterparts elsewhere might, for instance, have to resort to 'judicialization' to access health care, medication, and social services. This right-to-health litigation, especially concerning access to medicines, has been a growing phenomenon in countries such as Brazil (Aureliano and Gibbon, 2020; Biehl, 2014). In Poland, the judiciary has played an increasingly important role in securing access to the rights and resources available to RD people with disability status (Frydrych, 2023). Health policies for RDs – in India, for instance – propose measures such as "voluntary crowd-funding for treatment of patients with RDs" as a solution to "resource constraints" (Ministry of Health and Family Welfare, 2021, p. 23). Finally, in the Global South RDs intersect with other GH concepts and interventions, such as neglected tropical diseases as in the case of African trypanosomiasis (sleeping sickness, see Varanda and Théophile, 2019). In the following, we draw from our ethnographic research on rare metabolic disorders and rare inherited blood disorders to address NBS, migration, and therapeutic remoteness as key issues in RDs and GH.

Newborn screening: Foregrounding 'clear health benefits' and obscuring family experience

Four days after leaving a hospital located in a large city in western Poland with their newborn, Mr. and Mrs. Nowak were informed that NBS suggested their baby was suffering from a rare inherited metabolic disorder (IMD). To not alarm the parents, no reference was made to a specific disorder; instead, they were told to wait for the results of confirmatory tests. The following day, a female doctor called to inform the Nowaks the probability of this IMD was extremely high, and she urged the couple to return to the hospital, where they were given the diagnosis of LCHAD deficiency and briefed on the life-long dietary treatment demanded by this disease. Mrs. Nowak described the week after receiving this diagnosis as "a catastrophe". Her husband added: "Just the fact that you learn that the child has a life-threatening disease, an uncurable disease, and just the name of this uncurable disease makes you think a thousand thoughts; [you worry] what is going to happen to the child. This was hard. It's been hard." Finnish and Polish parents of children with an IMD interviewed by Rajtar and her colleagues often experienced profound distress, disbelief, and doubt (see also Rogalski, 2022, p. 869).[2] For instance, as LCHADD patients cannot consume breast milk due to its fat content, newborns are fed special formulas known as foods for special medical purposes (FSMP).[3] This can lead to mothers feeling devalued, as breastfeeding would endanger their baby's health, and guilty for having breastfed their newborns prior to diagnosis.

A 2021 report detailing "common needs" of the global RD community hailed screening as "crucial" for swifter diagnosis and called for an expansion of premarital, prenatal,

and newborn screening "in certain regions to reduce the incidence [of RDs] and eliminate the diagnostic odyssey" (Lopez Gousset and Bolz-Johnson, 2021, p. 4). Since the 1960s, NBS has been introduced in an increasing number of countries to identify infants with one particular pre-symptomatic IMD, phenylketonuria (PKU), a "condition for which early identification and intervention has a significant impact on outcomes for affected children" (White et al., 2021). Thus, PKU is considered "a paradigmatic case for screening" that has transformed public health (Cornel et al., 2021, p. 258). The development of tandem mass spectrometry (MS/MS) in the late 1990s, a technology that can screen for 40 to 50 conditions using a single blood spot, enabled the expansion of NBS (Cornel et al., 2021; Loeber et al., 2021; Raz and Timmermans, 2018), which is now considered "a global pillar of public health" (Raz, Amano, and Timmermans, 2018, p. 98). Nonetheless, even in resource-rich settings, the implementation of expanded NBS depends on national health policies, the availability of human and technological resources and infrastructures, and sociocultural differences. For instance, in Israel, where health policy prioritizes primary prevention, the NBS panel only encompasses 12 diseases; in the United States, where the focus is on secondary prevention, NBS includes over 50 diseases (Raz and Timmermans, 2018). Even within Europe, the NBS panel ranges from one condition in Moldova to over 30 in Italy (Loeber et al., 2021). Despite NBS being considered "the best example of secondary prevention" in India (Ministry of Health and Family Welfare, 2021, p. 19), resource constraints and "a compelling need to prioritize the available resources to get maximum health gains for the community/population" (Ministry of Health and Family Welfare, 2021, p. 23) mean universal NBS is currently unfeasible.

As 70% of RDs start in early childhood, an emphasis on NBS and other testing technologies facilitates "early detection, treatment and care" (World Health Organisation, 2010). The underlying logic of NBS as a public health program focuses on the "clear health benefits" of early intervention and the improvement of health outcomes in children with RDs (Cornel et al., 2021, p. 258). Technological innovations in treatment, fuelled by recent developments in stem cell transplantation, enzyme replacement therapies, and gene therapies, may lead to the inclusion of more disorders in NBS panels. However, as scientific evidence for treatment effectiveness and "health gain" is difficult to establish for such small patient groups, health policymakers face a significant problem (Cornel et al., 2021, p. 260). Moreover, as Rajtar (2023) has argued, small data collected from RD populations are far from stable and objective: culturally, socially, politically, and scientifically embedded, these data play an ambivalent role. While it may engender the production of valuable genetic and epidemiological data, it could also exacerbate stigmatizing practices against groups that scientific research associates with high RD prevalence. Rajtar's study is an example of the critical and people-centred approach that characterizes medical anthropology (Panter-Brick and Eggerman, 2018) and can therefore reveal the unintended effects of biomedically driven GH research in the field of RDs.

However, health policy debates on the global implementation of NBS tend to marginalize qualitative research that presents a more ambivalent picture of NBS and its experiential outcomes for RD patients and their families (White et al., 2021). The experience of the Nowaks recounted at the beginning of this section is far from unique. Ethnographies and meta-ethnographies of NBS (Raz and Timmermans, 2018; Timmermans and Buchbinder, 2013; White et al., 2021) counterbalance an overly optimistic view by revealing that the technology is "poorly understood, and its potential ramifications are not readily considered by parents" (White et al., 2021) and illustrate how "families experience NBS not as a

distinct moment in time, but rather as part of a larger journey spanning decades" (White et al., 2021). Furthermore, the diagnostic uncertainty inherent in MS/MS technology may not be resolved through genetic testing, thus creating "patients-in-waiting": "a population of people who are caught betwixt and between health and pathology" (Timmermans and Buchbinder, 2013, p. 95). In such cases, rather than reducing the time to diagnosis, NBS consigns patients to an extended diagnostic odyssey. Despite appreciating the lifesaving possibilities of the technology, especially when confronted with the daunting experiences of families with children who were diagnosed later in life or even died before the introduction of NBS, parents often express dissatisfaction with physicians' lack of knowledge and/or concrete suggestions for daily care management (Raz, Amano, and Timmermans, 2018, pp. 105–106). This particularly applies to family doctors and/or physicians at emergency clinics who often have little knowledge of a particular RD and may even prioritize their own medical authority over recommendations from specialists. During Rajtar's research in Poland, many families of children with an IMD described admitting physicians endangering their child's life by disregarding the RD specialist's emergency letter detailing necessary treatment. Additionally, dietary treatment for patients with rare metabolic disorders relies heavily on FSMP, which in some countries may be expensive and not readily available (see Raz, Amano, and Timmermans, 2018 on Israel and the United States); in other countries such as Poland, where the costs of FSMP are reimbursed, dealing with bureaucracy can be time-consuming for both families and health professionals. Finally, ethnographic research has revealed unintended consequences of NBS: as demonstrated by the Nowaks, parents not only often feel shame and guilt and even, at times, assign blame, but they also face the ethical and emotional dilemma of whether to disclose existence of the RD to the child, the wider family, and the education system (White et al., 2021).

The "urgency narrative" (Grob, 2019, cited in White et al., 2021) used to facilitate the global expansion of NBS obscures the fact that, even in resource-rich settings, there is no adequate infrastructure for the support of RD patients and their families. This focus on saving lives ignores the lack of care, education, and employment opportunities for RD patients and overshadows issues relating to the transition to adulthood that an increasing number of people with RDs have experienced. Additionally, some national NBS panels do not account for the specific needs of global migrants with inherited blood disorders.

Global migration and rare inherited blood disorders

Given the small number of widely dispersed RD patients lacking easy access to medical expertise, RDs are entangled in the various forms of health-related mobility that have come to characterize GH to an ever-increasing extent (e.g., Sargent and Larchanché, 2011; Bronwyn et al., 2015; Cartwright, Manderson, and Hardon, 2016). Medical experts, patients, and caregivers as well as materialities, such as blood samples, frequently undertake the travel demanded by RD training, diagnosis, and treatment.[4] Moreover, in some cases the 'endemic' versus 'rare' disease distinction is challenged by global migration. Health-care settings in resource-rich Western countries encounter migrating neglected tropical diseases (Gold, 2021) and inherited blood disorders. In this section we focus on the latter.

Red blood cell disorders and cystic fibrosis rank among the most common hereditary genetic disorders, but nevertheless they belong to the RD family. These recessive inherited single gene blood disorders – most notably thalassaemia and sickle cell disease[5]– are common in some regions but rare in others.[6] Haemoglobinopathies are endemic in the tropical

and sub-tropical malaria-belt and challenge the health-care systems of resource-poor countries with a prevalence or history of severe malaria infection (Knoll, 2020).

Medical anthropologists have documented the ways in which societies have genetically and culturally adapted to malarial pressures and gene anomalies. Research has been carried out in Mediterranean islands such as Sardinia (Brown, 1981); in the Maldives, which has the highest prevalence of beta-thalassaemia in the world (Knoll, 2020); in the Sultanate of Oman, where practices of consanguineous marriage challenge global biomedical discourse on the genetic risks of close marriages (Beaudevin, 2015); and in Senegal, where adaptation techniques "enculturated" the sickle cell gene and mitigated the disorder's severity (Fullwiley, 2011, pp. 20–22). National carrier screening programs in endemic countries, such as India, can racialize sickle cell disease as a tribal disease and reinforce existing health inequalities (Chattoo, 2018, pp. 34–35) and, as in Cyprus, enable local populations to interpret such disorders as a "collective 'ethnic' fate [and] to understand the gene pool as a 'tragic commons' that require[s] collective management" (Beck and Niewöhner, 2009, p. 88). In multi-ethnic immigration societies like the United States, several RDs "have become vehicles for solidifying categories of race and ethnicity" (Lock and Nguyen, 2010, p. 319): Tay-Sachs disease has been described as a "Jewish disease" and sickle cell mutation is associated with African Americans (e.g., Wailoo, 2007). Nevertheless, both RDs have been found in other immigrant populations.

While scholars have critically examined the rigid connection among genotype, territory, and ancestry in global representations of haemoglobinopathies as diseases of origins (Beaudevin, 2013, p. 176), new margins in the GH landscape with intrinsic links to migration deserve attention. By 2010, haemoglobinopathies had become the most common RDs in 10 European countries (Aguilar Martinez et al., 2014, pp. 1–2); the Thalassaemia International Federation estimates that some 855,000 migrants from high prevalence countries arrived in Europe between 2012 and 2019 (Angastiniotis et al., 2021, p. 9803). Scepticism regarding a global health crisis rhetoric, as articulated by Sangeeta Chattoo (2018, pp. 32–33), is certainly justified. Yet health inequities and the social suffering of patients with inherited blood disorders who 'do not fit' into the dominant health-care landscape cannot be ignored. With insufficient data on migrating haemoglobinopathies, they remain 'invisible' within public health. In Austria, as in several other non-endemic central and northern European countries (e.g., Hemminki et al., 2015), inherited haemoglobin disorders are not included in national NBS programs[7] and patients with these disorders have no access to specialised clinics and biosociality groups. Instead, such patients receive treatment in haemato-oncological departments, where they constitute less than 1% of patients. The health-care systems of recipient countries that are geographically and, by consequence, therapeutically 'remote' to the endemic belt and the experience gathered there often lack expertise in clinical management as well as thalassaemia and sickle cell disease prevention. Navigating these unpractised health-care terrains is challenging, particularly for people with diverse migratory, language, and cultural backgrounds. Thus, haemoglobinopathy patients are often unable to benefit from the quality of care their new place of residence can provide (Angastiniotis et al., 2021, p. 9803).

The following ethnographic vignette illustrates a number of these points. In 2019, Ms. Shenmi Huber attended the very first local gathering of patients with transfusion dependent thalassaemia and sickle cell disease in Vienna. The 13 adult patients present were native speakers of 10 different languages. While some attendees, like Shenmi, had arrived as children many years earlier and attained proficiency in the Austrian variety of the German language, others had mastered only the basics of the local vernacular. Shenmi had been

three years old when her parents had left East Asia for political reasons and relocated to Austria. Although the treatment of beta thalassaemia major required her to miss classes twice a month, she had finished school and successfully undertaken an apprenticeship in a paramedical field. Wanting to keep the disease a secret throughout her education, Shenmi had scheduled her blood transfusions according to her off-duty days rather than her body's haemoglobin requirements. Only when offered full-time employment did she feel compelled to reveal her chronic condition in order to arrange the transfusion days every second week that required her presence in the clinic for around eight hours.

At the time of writing, Shenmi is in her early forties and married to an Austrian, with whom she has a four-year-old daughter. Shenmi tearfully revealed that until that initial meeting of haemoglobinopathy patients in a Viennese coffee shop, she had believed she was the only person with the disease: so strong was this conviction that, despite training and working in paramedics, the idea of conducting an internet search on the condition had never occurred to her.

As Shenmi's story reveals, the maladaptation between a health-care setting and a non-endemic disease can cause patient isolation and impede the formation of migrant biosociality. Knoll (2021a) has proposed the notion of "therapeutic remoteness" to illuminate the health inequities and unmet needs that result from an uneven global RD landscape. This concept builds upon ethnographic insights on haemoglobinopathy management in the Maldives (Knoll, 2021b) that is not quite up to international standards due to both the archipelago's location in the Global South and its struggle with health inequities in the more remote outer islands. Drawing on anthropological literature on remoteness, therapeutic remoteness refers to an asymmetric health relationship that is associated with, but *not* identical to, distance, marginality, periphery, and infrastructural weakness. This concept addresses a "remoteness" shaped by global location, genes, and human agency.

We argue that enhancing anthropological knowledge about a highly diverse and expanding group of migrant patients and carriers with rare inherited blood disorders is vital to improve GH. This almost invisible RD group is particularly vulnerable: therapeutic remoteness and migratory health issues add to and amplify the many difficulties faced by RD patients. Haemoglobinopathies are caused by hundreds of different mutations that result in a wide range of clinical manifestations, spanning from almost healthy heterozygotes to the "biomedically mediated lives" (Chatjouli, 2013) of severe cases of homozygotes or compound heterozygotes. As the onset of thalassaemia and sickle cell disease occurs in early childhood, severe case patients rarely survived adolescence until the pharmaceutical breakthroughs of the late 1980s. Therefore, these disorders are still understood as paediatric conditions and targeted care options for adult, procreating, and aging patients are scarce. Thus, patients often describe a difficult transition phase from paediatric to adult care, where they are expected to be self-reliant and assemble their own multi-disciplinary adult care team. In endemic EU countries and traditional immigration countries such as Canada, the United Kingdom, and the United States, thalassaemia and sickle cell mutations have long been "mobilized" (Navon, 2019). In Cyprus, biosociality developed from a local group of concerned parents into the globally operating Thalassemia International Federation. Nonetheless, this consolidated kind of biosociality cannot be easily implemented in central and northern Europe, where haemoglobinopathy-centred collectives must bridge wider ranges of ethnic, linguistic, sociocultural, and religious diversity as well as diverse migratory histories. Including anthropological insights into research questions and the design of GH projects on rare inherited blood disorders would allow the challenges that global migration

brings to the 'endemic' versus 'rare' distinction to be acknowledged, thereby improving both the quality of data and project implementation.

Conclusion

With the 2021 UN resolution and the emergence of the global alliance of RD patient organizations in Rare Disease International, RDs have officially become a GH concern. However, the stated goal of health equity for RD patients will be difficult to achieve unless GH actors go beyond biomedical and metrics-oriented perspectives and allow more space for qualitative methodologies, such as ethnographies, which critically examine the implementation of RD policies across the world and document the real-life experiences of people with RDs and their caregivers. In this chapter, we have traced the creation of the RD category from a Western viewpoint; this classification has been adopted in many countries, alongside biomedically grounded solutions, such as NBS. We argue that the small scale of RDs does not align well with GH, which constantly seeks statistically robust "good data" ready to be appropriated for scaling-up (Adams, 2013, 2016). While the 'urgency narrative' embedded in the global implementation of NBS has certainly saved the lives of many newborns, this technology is still inaccessible for families in resource-poor settings. Furthermore, even in countries where NBS is available, screening for migrating RDs can be non-existent, thus exacerbating 'therapeutic remoteness'. For others, positive or inconclusive NBS results do not guarantee the required follow-up treatment and can initiate an arduous diagnostic odyssey. Sustained medical anthropological investigation is needed to foreground people-centred and critical analyses (Panter-Brick and Eggerman, 2018) of RD policies and practices within GH.

Notes

1 Conditions that affect less than 20 in a million people are classified as ultra-rare.
2 Insights in this chapter are drawn from ethnographic research on IMDs that Rajtar and her colleagues conducted between 2016 and 2023 in Finland and Poland. These countries screen for 21 and 30 conditions, respectively. Results from this research have been published in Rajtar 2020; 2023; Rajtar and Król, 2023, and Rogalski 2022. This research was funded by the National Science Centre in Poland under grants [2015/17/B/HS3/00107 and 2017/26/E/HS3/00291], and the EURIAS Fellowship Programme (Co-funded by MSCA, under the 7th Framework Programme).
3 Used in the dietary treatment of patients with certain medical conditions whose nutritional requirements cannot be fulfilled with 'normal' food, FSMP are intended to be used under medical supervision. In Europe, FSMP are subjected to detailed regulations relating to such aspects as their composition and labelling. The FSMP market is dominated by a handful of global companies, such as Nutricia (a Danone brand) and Vitaflo (acquired by Nestlé in 2010), based in the Netherlands and the United Kingom, respectively.
4 Adult RD patients in China, for example, travelled an average of 562 km and it took an average of four and a half years to receive a definitive diagnosis (Yan, He, and Dong, 2020). In the EU, the mobility of care providers, patients, and data across national borders is facilitated by the Cross-Border Healthcare Directive (Directive 2011/24/EU), which came into force in 2013. An amendment to this directive recognizes the special status of RD patients and guarantees unconditional right of access to health care in other member states prior to authorization.
5 The outdated, misleading, and trivializing term 'sickle cell anaemia' has even found its way into critical anthropological scholarship. Yet, anaemia is merely one of the countless life-threatening problems experienced by patients with this painful disorder.

6 Since 2010, Knoll has conducted ethnographic research into the biosocial impact and history of inherited haemoglobin disorders in the high-prevalence region of South Asia, focusing specifically on the Maldives. In 2019, the scope of Knoll's research expanded to Europe as she tracked the migration of Mendelian blood disorders. Findings from this research have been published in Knoll, 2020 and Angastiniotis et al, 2021.

7 As with the aforementioned rare metabolic disorders, early diagnosis and intervention is equally crucial for the well-being of sickle cell disease patients, who can receive treatment from the age of two months. Currently, only four EU countries (France, Germany, Malta, and Spain) and the United Kingdom include sickle cell disease in their NBS programs (Loeber et al, 2021, pp. 8–9).

References

Abimbola, S. (2018) 'On the meaning of global health and the role of global health journals', *International Health*, 10(2), 63–65. https://doi.org/10.1093/inthealth/ihy010

Adams, V. (2013) 'Evidence-based global public health, subjects, profits, erasures', in Biehl, J. and Petryna, A. (eds.) *When People Come First: Critical Studies in Global Health*, Princeton: Princeton University Press, pp. 54–90.

Adams, V. (2016) 'Introduction', in Adams, V. (ed.) *Metrics: What Counts in Global Health*, Durham: Duke University Press, pp. 1–17.

Adams, V. et al. (2019) 'Re-imagining global health through social medicine', *Global Public Health*, 14(10), 1383–1400. https://doi.org/10.1080/17441692.2019.1587639

Aguilar Martinez, P. et al. (2014) 'Haemoglobinopathies in Europe: Health and migration policy perspectives', *Orphanet Journal of Rare Diseases*, 9(97). https://doi.org/10.1186/1750-1172-9-97

Angastiniotis, M. et al. (2021) 'Haemoglobin disorders in Europe: A systematic effort of identifying and addressing unmet needs and challenges by the Thalassaemia International Federation', *Thalassemia Reports*, 11(1). https://doi.org/10.4081/thal.2021.9803

Aureliano, W. and Gibbon, S. (2020) 'Judicialisation and the politics of rare disease in Brazil: Rethinking activism and inequalities', in Gamlin, J. et al. (eds.) *Critical Medical Anthropology: Perspectives in and from Latin America*, London: UCL Press, pp. 248–269.

Beaudevin, C. (2013) 'Old diseases and contemporary crisis: Inherited blood disorders in the Sultanate of Oman', *Anthropology & Medicine*, 20(2), 175–189. https://doi.org/10.1080/13648470.2013.805317

Beaudevin, C. (2015) 'Cousin marriages and inherited blood disorders in the Sultanate of Oman', in Shaw, A. and Raz, A.E. (eds.) *Cousin Marriage: Between Tradition, Globalisation and Genetic Risk*, London and New York: Berghahn, pp. 65–87.

Beck, S. and Niewöhner, J. (2009) 'Translating genetic testing and screening in Cyprus and Germany: Contingencies, continuities, ordering effects and bio-cultural intimacy', in Atkinson, P., Glasner, P. and Lock, M. (eds.) *Handbook of Genetics and Society: Mapping the New Genomic Era*, London: Routledge, pp. 76–93.

Biehl, J. (2014) 'The judicialization of biopolitics: Claiming the right to pharmaceuticals in Brazilian courts', *American Ethnologist*, 40(3), 419–436.

Bronwyn, P. et al. (eds.) (2015) *Bodies Across Borders: The Global Circulation of Body Parts, Medical Tourists and Professionals*, Farnham, Burlington: Ashgate Publishing.

Brown, P.J. (1981) 'Cultural adaptations to endemic malaria in Sardinia', *Medical Anthropology*, 5(3), 311–339. https://doi.org/10.1080/01459740.1981.9986991

Callon, M. and Rabeharisoa, V. (2008) 'The growing engagement of emergent concerned groups in political and economic life: Lessons from the French association of neuromuscular disease patients', *Science, Technology, & Human Values*, 33(2), 230–261. https://doi.org/10.1177/0162243907311264

Cartwright, E., Manderson, L. and Hardon, A. (2016) 'Global quests for care', in Manderson, L., Cartwright, E. and Hardon, A. (eds.) *The Routledge Handbook of Medical Anthropology*, New York: Routledge, pp. 262–281.

Chatjouli, A. (2013) 'Thalassaemic lives as stories of becoming: Mediated biologies and genetic (un)certainties', in Ingold, T. and Palsson, G. (eds.) *Biosocial Becomings: Integrating Social and Biological Anthropology*, Cambridge: Cambridge University Press, pp. 84–105.

Chattoo, S. (2018) 'Inherited blood disorders, genetic risk and global public health: Framing "birth defects" as preventable in India', *Anthropology & Medicine*, 25(1), 30–49. https://doi.org/10.1080/13648470.2017.1381231

Cornel, M.C. et al. (2021) 'Neonatal and carrier screening for rare diseases: How innovation challenges screening criteria worldwide', *Journal of Community Genetics*, 12(2), 257–265. https://doi.org/10.1007/s12687-020-00488-y

Council of the European Union. (2009) 'Council recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02)', *Official Journal of the European Union*, 52(3), 7–10. https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

Cross, J. and Street, A. (2022) 'To fail at scale! Minimalism and maximalism in humanitarian entrepreneurship', *Social Anthropology/Anthropologie Sociale*, 30(2), 101–119.

Farmer, P. et al. (2013) 'Introduction: A biosocial approach', in Farmer, P. et al. (eds.) *Reimagining Global Health: An Introduction*, Berkeley: University of California Press, pp. 1–14.

Featherstone, K. and Atkinson, P. (2012) *Creating Conditions: The Making and Remaking of a Genetic Condition*, New York: Routledge.

Frydrych, J. (2023) 'Disability and rare metabolic disorders', in Rajtar, M. and Król, K.E. (eds.) *Entanglements of rare diseases in the Baltic Sea Region*, Lanham: Lexington Books/Rowman & Littlefield, pp. 167-194.

Fullwiley, D. (2011) *The Enculturated Gene: Sickle Cell Health Politics and Biological Difference in West Africa*, Princeton and Oxford: Princeton University Press.

Gibbon, S., Kilshaw, S. and Sleeboom-Faulkner, S. (2018) 'Genomics and genetic medicine: Pathways to global health?', *Anthropology & Medicine*, 25(1), 1–10. https://doi.org/10.1080/13648470.2017.1398816

Gibbon, S. and Novas, C. (eds.) (2008) *Biosocialities, Genetics, and the Social Sciences: Making Biologies and Identities*, London: Routledge.

Gold, M. (2021) 'Health, migration, and NTDs: An anthropological view', *Medical Anthropology*, 40(6), 491–496. https://doi.org/10.1080/01459740.2021.1953494

Grob, R. (2019) 'Qualitative Research on Expanded Prenatal and Newborn Screening: Robust but Marginalized', *Looking for the Psychosocial Impacts of Genomic Information,* special report, *Hastings Center Report*, 49(3), S72–S81. https://doi: 10.1002/hast.1019

Haendel, M. et al. (2020) 'How many rare diseases are there?', *National Review Drug Discovery*, 19(2), 77–78. https://doi.org/10.1038/d41573-019-00180-y

Heath, D., Rapp, R. and Taussig, K.-S. (2004) 'Genetic citizenship', in Nugent, D. and Vincent, J. (eds.) *A Companion to the Anthropology of Politics*, Malden: Blackwell Publishing, pp. 152–167.

Hemminki, K. et al. (2015) 'Thalassaemia and sickle cell anemia in Swedish immigrants: Genetic diseases have become global', *SAGE Open Medicine*, 3, 1–8. https://doi.org/10.1177/2050312115613097

Holst, J. (2020) 'Global health—Emergence, hegemonic trends and biomedical reductionism', *Globalization & Health*, 16(42). https://doi.org/10.1186/s12992-020-00573-4

Huyard, C. (2009) 'How did uncommon disorders become "rare diseases"? History of a boundary object', *Sociology of Health & Illness*, 31(4), 463–477. https://doi.org/10.1111/j.1467-9566.2008.01143.x

Knoll, E.-M. (2020) 'Inherited without history? Maldive fever and its aftermath', in Campbell, G. and Knoll, E.-M. (eds.) *Disease Dispersion and Impact in the Indian Ocean World*, Palgrave Series in Indian Ocean World Studies, Cham: Palgrave Macmillan, pp. 255–284.

Knoll, E.-M. (2021a) 'Therapeutic remoteness: Unmet needs in rare inherited blood disorders', *AAA—American Anthropological Association. Annual Meeting. Session: Anthropology and Knowledge Production in Rare Diseases*, Baltimore, 17–21 November 2021.

Knoll, E.-M. (2021b) 'In motion: Genes, identities and mediated lives in dispersed small-scale contexts', in Gingrich, A. (ed.) *Anthropology in Motion: Encounters with Current Trajectories of Scholarship from Austria*. The RAI Country Series, Volume Four. Canon Pyon: Sean Kingston Publishing, pp. 64–91.

Koplan, J.P. et al. (2009) 'Towards a common definition of global health', *Lancet*, 373(9679), 1993–1995. https://doi.org/10.1016/S0140-6736(09)60332-9

Lambert, H. and Rose, H. (1996) 'Disembodied knowledge? Making sense of medical science', in Irwin, A. and Wynne, B. (eds.) *Misunderstanding Science? The Public Construction of Science and Technology*, Cambridge: Cambridge University Press, pp. 65–83.

Larotonda, A. (2016) 'Rare disease in children in Italy', in Manderson, L., Cartwright, E. and Hardon, A. (eds.) *The Routledge Handbook of Medical Anthropology*, London: Routledge, pp. 26–30.
Lesmo, I.E. (2022) 'Micropolitics of rare diseases in Italy', *Medical Anthropology*, 42(1), 48–61. https://doi.org/10.1080/01459740.2022.2119853
Letinturier-Valencia, M.C. et al. (eds.) (2022) *IRDiRC State of Play: Rare Diseases Research Initiatives 2019–2021*, February 2022 [online]. Available at: https://irdirc.org/resources-2/state-of-play/ (Accessed: 18 July 2022)
Lock, M. and Nguyen, V.-K. (2010) 'Genes as embodied risk', in Lock, M. and Ngyuen, V.-K. (eds.) *An Anthropology of Biomedicine*, Malden: Routledge, pp. 304–329.
Loeber, J.G. et al. (2021) 'Neonatal screening in Europe revisited: An ISNS perspective on the current state and developments since 2010', *International Journal of Neonatal Screening*, 7(5), 15. https://doi.org/10.3390/ijns7010015 (Accessed: 26 July 2022)
Lopez Gousset, V. and Bolz-Johnson, M. (2021) 'Common needs of the rare disease population identified globally', *Rare Disease International*, October 2021 [online]. Available at: https://www.rarediseasesinternational.org/wp-content/uploads/2021/10/final-Common-Needs-of-the-Rare-Disease-Population-Identified-Globally-report-extract.pdf (Accessed: 25 July 2022)
Mattingly, C. (2014) *Moral Laboratories: Family Peril and the Struggle for a Good Life*, Oakland: University of California Press.
McGuire, M. (2020) 'Paces of costly care: Rare disease drug access in Canada', *Medical Anthropology*, 39(4), 319–332. https://doi.org/10.1080/01459740.2019.1573229
Mikami, K. (2019) 'Orphans in the market: The history of orphan drug policy', *Social History of Medicine*, 32(3), 609–630. https://doi.org/10.1093/shm/hkx098
Mikami, K. (2020) 'Citizens under the umbrella: Citizenship projects and the development of genetic umbrella organizations in the USA and the UK', *New Genetics & Society*, 39(2), 148–172. https://doi.org/10.1080/14636778.2019.1693889
Ministry of Health and Family Welfare. (2021) *National policy for rare diseases*. Available at: https://main.mohfw.gov.in/sites/default/files/Finalpercent20NPRDpercent2Cpercent202021.pdf (Accessed: 21 July 2022)
Navon, D. (2019) *Mobilizing Mutations: Human Genetics in the Age of Patient Advocacy*, Chicago: The University of Chicago Press.
Novas, C. (2016) 'Patient activism and biopolitics: Thinking through rare diseases and orphan drugs', in Cisney, V.W. and Morar, N. (eds.) *Biopower: Foucault and Beyond*, Chicago: University of Chicago Press, pp. 183–198.
Panter-Brick, C. and Eggerman, M. (2018) 'The field of medical anthropology', *Social Science & Medicine*, 196, 233–239. https://doi.org/10.1016/j.socscimed.2017.10.033
Rabeharisoa, V. and Callon, M. (2004) 'Patients and scientists in French muscular dystrophy research', in Jasanoff, S. (ed.) *States of Knowledge: The Co-Production of Science and Social Order*, Abingdon: Routledge, pp. 142–160.
Rabeharisoa, V., Moreira, T. and Akrich, M. (2014) 'Evidence-based activism: Patients', users' and activists' groups in knowledge society', *BioSocieties*, 9(2), 111–128.
Rabinow, P. (1996) 'Artificiality and enlightenment: From sociobiology to biosociality', in Rabinow, P. (ed.) *Essays on the Anthropology of Reason*, Princeton: Princeton University Press, pp. 91–112.
Rajtar, M. (2020) 'The concept of vulnerability within research ethics and health policies on rare diseases', *Przegląd Socjologiczny/Sociological Review*, 69(3), 107–127. https://doi.org/10.26485/PS/2020/69.3/5
Rajtar, M. (2023) '"Small" data, isolated populations, and new categories of rare diseases in Finland and Poland', *Anthropology & Medicine*, 31(1), 1–16. https://doi.org/10.1080/13648470.2022.2152633
Rajtar, M. and Knoll, E.M. (2023) 'Rare diseases, global health, and social sciences: Counterbalancing biomedical reductionism', *Brocher Foundation Workshop*. Hermance, Switzerland, 9–11 May 2023.
Rajtar, M. and Król, K.E. (eds.) (2023) *Entanglements of Rare Diseases in the Baltic Sea Region*, Lanham: Lexington Books/Rowman & Littlefield.
Raz, A., Amano, Y. and Timmermans, S. (2018) 'Parents like me: Biosociality and lay expertise in self-help groups of parents of screen-positive newborns', *New Genetics & Society*, 37(2), 97–116. https://doi.org/10.1080/14636778.2018.1451315

Raz, A.E. and Timmermans, S. (2018) 'Divergent evolution of newborn screening: Israel and the US as the gene worlds', *BioSocieties*, 13(5), 580–600. https://doi.org/10.1057/s41292-017-0110-z

Richter, T. et al. (2015) 'Rare disease terminology and definitions—A systematic global review: Report of the ISPOR rare disease special interest group', *Value in Health*, 18(6), 906–914. https://doi.org/10.1016/j.jval.2015.05.008

Rogalski, F. (2022) 'Epistemologies of living with and treating rare metabolic disorders', *Medical Anthropology*, 41(8), 866–878. https://doi.org/10.1080/01459740.2022.2141630

Rose, N. and Novas, C. (2005) 'Biological citizenship', in Ong, A. and Collier, S.J. (eds.) *Global Assemblages: Technology, Politics, and Ethics as Anthropological Problems*, Malden: Blackwell Publishing, pp. 439–464.

Rubinstein, Y.R. et al. (2020) 'The case for open science: Rare diseases', *JAMIA Open*, 3(3), 472–486. https://doi.org/10.1093/jamiaopen/ooaa030

Sargent, C. and Larchanché, S. (2011) 'Transnational migration and global health: The production and management of risk, illness, and access to care', *Annual Review of Anthropology*, 40(1), 45–61. https://doi.org/10.1146/annurev-anthro-081309-145811

Timmermans, S. and Buchbinder, M. (2013) *Saving Babies? The Consequences of Newborn Screening*, Chicago: University of Chicago Press.

Tumiene, B. et al. (2021) 'Rare diseases: Past achievements and future prospects', *Journal of Community Genetics*, 12(2), 205–206. https://doi.org/10.1007/s12687-021-00529-0

United Nations. (2021) *Resolution Adopted by the General Assembly on 16 December 2021*. Available at: https://www.rarediseasesinternational.org/wp-content/uploads/2022/01/Final-UN-Text-UN-Resolution-on-Persons-Living-with-a-Rare-Disease-and-their-Families.pdf (Accessed: 23 January 2022)

Varanda, J. and Théophile, J. (2019) 'Putting anthropology into global health: A century of anti-human African trypanosomiasis campaigns in Angola', *Anthropology in Action*, 26(1), 31–41. https://doi.org/10.3167/aia.2019.260104

Von der Lippe, C., Diesen, P.S. and Feragen, K.B. (2017) 'Living with a rare disorder: A systematic review of the qualitative literature', *Molecular Genetics & Genomic Medicine*, 5(6), 758–773. https://doi.org/10.1002/mgg3.315

Wailoo, K. (2007) 'Inventing the heterozygote: Molecular biology, racial identity, and the narratives of sickle-cell disease, Tay-Sachs and cystic fibrosis', in Lock, M. and Farquar, J. (eds.) *Beyond the Body Proper: Reading the Anthropology of Material Life*, Durham: Duke University Press, pp. 658–671.

World Health Organisation. (2010) *Birth Defects—Report of the Secretariat, Sixty-Third Third World Assembly*, Geneva: WHO [online]. Available at: https://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf (Accessed: 25 July 2022)

White, A.L. et al. (2021) 'Absorbing it all: A meta-ethnography of parents' unfolding experiences of newborn screening', *Social Science & Medicine*, 287, 114367. https://doi.org/10.1016/j.socscimed.2021.114367 (Accessed: 20 July 2022)

Yan, X., He, S. and Dong, D. (2020) 'Determining how far an adult rare disease patient needs to travel for a definitive diagnosis: A cross-sectional examination of the 2018 National Rare Disease Survey in China', *International Journal of Environmental Research & Public Health*, 17(1757). https://doi.org/10.3390/ijerph17051757 (Accessed: 30 July 2022)

Yates-Doerr, E. and Maes, K. (2019) 'Global health', in Stein, F. (ed.) *The Cambridge Encyclopedia of Anthropology* [online]. Available at: http://doi.org/10.29164/19ghealth (Accessed: 6 November 2021)

THE ROUTLEDGE HANDBOOK OF ANTHROPOLOGY AND GLOBAL HEALTH

Edited by Tsitsi B. Masvawure and Ellen E. Foley

Routledge
Taylor & Francis Group
LONDON AND NEW YORK

Designed cover image: altmodern, Getty Images

First published 2024
by Routledge
605 Third Avenue, New York, NY 10158

and by Routledge
4 Park Square, Milton Park, Abingdon, Oxon, OX14 4RN

Routledge is an imprint of the Taylor & Francis Group, an informa business

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ISBN: 9781032256375 (hbk)
ISBN: 9781032256405 (pbk)
ISBN: 9781003284345 (ebk)

DOI: 10.4324/9781003284345

Typeset in Sabon
by Deanta Global Publishing Services, Chennai, India