Social Science & Medicine 372 (2025) 117958



Contents lists available at ScienceDirect

Social Science & Medicine

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



"My full-time unpaid role": Understanding the (extra)ordinary work of founders of rare disease organisations

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ARTICLE INFO

Handling editor: Susan J. Elliott

ABSTRACT

Rare disease organisations can play a crucial role in shaping the medical and scientific landscape. This article draws from interviews with sixteen founders of UK-based, rare disease organisations, all of whom were patients, parents or family members, to understand their experiences and commitment to the organisation and its community. First, we explore the work involved in creating a professional community and addressing the challenge of expert capacity-building for rare diseases. We then utilise the concept of 'translation' to emphasise the efforts of founders at an intermediate stage, for example encouraging health professionals to collaborate and realise that a project is achievable. Third, we consider the personal implications for the founders in their efforts to develop and sustain the organisation. Founders' biographies are intimately entwined with the establishment and development of their organisation, and we highlight how they are fundamentally shaped by the necessity of their hard work, skills and passion. Finally, we recognise that although some of the efforts of founders are undervalued both socially and economically, the founders themselves understand their work and role as crucial to the organisation's long-term success.

1. The political landscape of rare disease

Rare disease has an origin story (Franklin and McNeil, 1993). As an organisational concept, it began gaining traction in Europe more than thirty years ago, drawing resources and political attention by bringing together smaller organisations that were struggling to be heard. Rare disease is broadly defined as affecting less than one in two thousand people (DHSC, 2021). As Bowker and Star (2000) remind us, classifications have consequences. Operating under the logic that strength lies in collective solidarity, 'rareness' has been repurposed as a unifying category. The banners of 'rare is common' and 'together we are stronger' have been successfully used to mainstream rare disease through 'politics in numbers' (Rabeharisoa et al., 2014 p195). Alongside individual rare disease organisations, rare disease now has a collective infrastructure. In the United States, NORD (National Organization for Rare Disorders) was established following the success of patient groups in supporting a change in orphan drug legislation (Heath et al., 2007). EURODIS (Rare Diseases Europe) was then established a decade later by four rare disease support groups to advocate for European Union (EU) laws to support drug development and provision. EURODIS' priority areas include supporting diagnosis, research, access to treatment, and patient care, and accomplishes this by working with policymakers and engaging the community to establish rare disease policies (Courbier et al., 2019). In the United Kingdom, the 2021 Rare Diseases Framework (DHSC, 2021) represents a commitment to improving the experiences of those with rare disease, and given healthcare is devolved within the UK, individual nations have also produced their own rare disease action plans. All of these acknowledge the importance of expediting the diagnostic process, increasing awareness of rare diseases, and supporting access to specialist care and treatment. But what is written down in policy can be difficult to deliver in practice, as Taruscio and Gahl (2024) highlight:

> Rare diseases remain a formidable public health challenge. The key to unlocking breakthroughs in diagnosis and treatment is fostering dynamic international partnerships and streamlined data sharing. The empowerment of patient advocacy groups is essential, as they are pivotal in driving innovative research and elevating health-care standards for these often under-represented conditions. (Taruscio and Gahl 2024, p1)

Explicit within the policy proposals, and as Taruscio and Gahl (2024)

This article is part of a special issue entitled: Rare diseases global published in Social Science & Medicine.

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https://doi.org/10.1016/j.socscimed.2025.117958
Received 30 June 2024; Received in revised form 6 February 2025; Accepted 12 March 2025
Available online 14 March 2025
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emphasise, is the expectation that patients and patient organisations wield the power to shape the landscape of rare disease. Changing expectations about patient experience and expertise have a long history. Epstein (1996) documented activism in the 1980s to explore how communities of patients, families, and allies mobilised their resources to address the urgent needs of the patient population and challenge existing knowledge of AIDS as a newly emerging disease. Many gained 'interactional expertise', mastering the language of a specialist domain (Collins et al., 2023) to legitimate their claims to knowledge.

The impact of patient expertise is particularly prominent in relation to rare disease, facilitated by developments in genetic technology and the identification and delineation of increasing numbers of rare disease. In the mid-90s, Rabinow observed that genetic technologies were beginning to facilitate a new sense of identity and collectivity, which he defined as biosociality. Rabinow (1996) speculated that "it is not hard to imagine groups formed around the chromosome 17, locus 16, 256, site 654, 376 allele variant with a guanine substitution. Such groups will have medical specialists, laboratories, narratives, traditions and a heavy panoply of pastoral keepers to help them experience, share, intervene and 'understand' their fate" (p.102).

Over the last thirty years, many examples have been documented to explore the emergence of these new patient groupings, working at the intersection of new genetic identities and collective action. Taussig et al. (2003) observed national conferences of patients and professionals, laboratories and medical spaces associated with the Little People of America (LPA), to highlight how stakeholders negotiated the meanings and implications of living with achondroplasia. They found that 'flexible eugenics' was enacted in these settings, establishing what was deemed acceptable in specific situations. They also describe the struggle for parents to be heard, bringing their children to political spaces to force the authorities to acknowledge their experiences. Research documenting the Association Francais contre les Myophathies (AFM) (Callon and Rabeharisoa, 2003, Mayrhofer, 2008) examined the organisation's evolving role in advancing biomedical knowledge of the disease, which could potentially lead to a cure, while also advocating for the recognition of patient's rights. What was remarkable at the time was the AFM established research funding streams and organised their own data banks. Patterson et al. (2023) noted that this is a now more common feature of the rare disease landscape, with about a third of rare disease patient organisations in their study initiating their own registry and/or biobank to facilitate research, with many more playing an active role in recruitment and informed consent for clinical trials and drug development.

Patient organisations can have impact in research by acting as an "organisational platform" linking patients and families with researchers (Mikami, 2020 p152). These connections are vital for accelerating biomedical advances, leveraging patients as cultural capital, and making patient bodies available for "innovative intervention" (Brown and Webster, 2004 p80). We discuss, in this article, how establishing biobanks or patient registries demands significant effort and resources. However, rare disease organisations choose to invest in them due to the significant potential benefits. Novas (2006) described patient investment in new medical and scientific advances as part of the "political economy of hope". According to Novas, 'hope' was not an 'act of the imagination', but was "materialised through a range of social practices" (p290), including for example, patients positioning themselves as expert stakeholders and actively contributing to research. Novas' two case studies, Pseudoxanthoma elasticum (PXE) and Canavan disease highlight interlacing between the production of biomedical knowledge and the generation of wealth, and the risks of commercialisation. PXE international was established by parents of children with PXE, who patented the PXE gene to generate scientific interest and support collaboration across laboratories. By doing so, they ensured that the patient population remained the primary beneficiaries of scientific advances. In contrast, activities around Canavan disease highlighted a misalignment between stakeholders, resulting in a lengthy and costly court case. While patients had donated their biological material and significant amounts of money, scientists claimed ownership over the research, and with it, claimed the right to control access to scientific advances and its wealth.

These examples highlight the high stakes environment in which some rare disease organisations work, characterised by empowerment through community (Rapp et al., 2001) and the possibilities of new collaborations, including patients, clinicians, scientists, researchers, and policymakers (and see, for example, Landzelius, 2006; Rabeharisoa et al., 2014; Gibbon and Novas, 2008). Further, by challenging the status quo, patient organisations have contributed to "novel norms" of medical research, which assign a central role to patients in change-making, informing broader societal expectations about how research could, and should, be conducted (Novas, 2006, p.291).

The movement from patients to knowledgeable experts captures the complexities involved in negotiating illness and biomedical knowledge and the increasing role of citizen scientists in creating knowledge and democratising science (Irwin, 1995). In many areas of medicine, the value of patient and families' knowledge, experience, and expertise has been formally enshrined in patient and public involvement (PPI) initiatives (Locock et al., 2017). However, while PPI might offer some patients and their representatives an opportunity for engagement, this is not the case for all. Rare disease organisations encompass different models of collaboration, depending on the nature of the group, resources available, and the social worlds in which they operate (Huyard, 2009). For example, a patient organisation might be invited to an arena as a 'guest' (Mosse, 2019; Galasso and Geiger, 2021), they might work as an equal partner with scientists or other authorities, or engage in 'uninvited' or 'in the wild' activism (Callon and Rabeharisoa, 2003; Strand and Holen, 2024). Important questions of power, voice and representation continue to be negotiated in practice. And as this article highlights, it is founders who often make these crucial decisions, giving shape and meaning to an organisation and its values.

While sociological research has examined the role and impact of rare disease organisations, the specific role of organisational leaders has garnered less attention. Both Patterson et al. (2023) and Pinto et al. (2018) have highlighted the unique challenges faced by leaders of patient organisations, particularly in securing funding for research and determining the relationship between patients, professionals and research. Pinto et al. (2018) identified how the burden of leading, and representing the needs of a community, can weigh heavily on these individuals. Our work responds to Patterson et al. (2023) call for more qualitative research exploring the experiences, and challenges facing leaders of rare disease organisations.

2. Methods

Sixteen semi-structured interviews were undertaken with founders of rare disease patient organisations. Ethical approval was gained through Cardiff University School of Social Sciences. Participants were recruited using the publicly accessible Genetic Alliance UK list of more than two hundred members. Genetic Alliance UK is an umbrella group working to support rare disease organisations, raise awareness of rare disease and influence policy and practice. Although this approach facilitated recruitment, we acknowledge our project only includes organisations who are established enough to seek support, potentially excluding less established groups. The membership list included the name of the organisation with the link to their website, which was then used to assess eligibility. To focus on founders' accounts, criteria for the project included organisations that were UK-based, concerned one rare disease or a group of related rare diseases, and had a website which included the name of the founder. Emails were addressed to the founder of the organisation where contact details were provided, or a generic email address if not. The initial email included a request to interview the organisation's founder, with the information sheet attached. Thirty-two rare disease organisations were contacted. Seventeen replies were

received, and sixteen founders (representing sixteen rare disease organisations) were interviewed. Interviews took place during May 2024 and June 2024. Participants were patients (three male, two female), a parent of a child diagnosed with the rare disease (nine mothers, one father) and a family member (one male). All participants were founders and still involved in the organisation, with most, but not all, continuing to play a key leadership role. Each has been given a pseudonym in this article to protect their personal identity, but with their agreement, organisations have been noted in the acknowledgements.

All interviews were conducted by RD. The interviews followed an interview schedule initially based on the project research aims. These were to identify the barriers and facilitators in establishing the organisation, to understand how they identified and represented the needs of patients and their families and to explore the nature of the relationships formed with health professionals, scientists and/or policy makers. Each interview schedule was then loosely tailored for each founder, based on the detail available on their website. Most of the interviews were undertaken and recorded through zoom, benefitting from increased public familiarity with digital communication and its use in research (see Howlett, 2021), with two conducted by telephone. Most interviews lasted the scheduled time of 1 hour, and only the audio recordings were saved for analysis. Auto-generated transcripts were checked for accuracy against the recordings.

The transcripts were analysed using reflexive thematic analysis (Braun and Clarke, 2021). This is an inductive approach, beginning with reading and re-reading each transcript to enable familiarity with the data. Using Microsoft Word, the transcripts were annotated, with sections of the text highlighted and comments added about topics of interest and links with other transcripts and literature. Initial themes identified through this process were diagnosis, seeking support from other families, creating a family support network, seeking professional expertise, forming a professional network, and looking to the future. The two authors discussed the themes and how these related to literature on the work of rare disease organisations as well as the project's research questions, and it was decided that a focus on professional communities would be productive. The themes generated through further analysis and discussion, and which are explored in this article are (i) establishing a professional community, (ii) translation work and (iii) organisational and personal resilience. We now discuss each theme in turn.

2.1. Theme 1: establishing a professional community

Radu et al. (2021) highlight how rare disease patient organisations are able to harness scientific advances through 'collective intelligence' and through forging strong relationships between patients, scientists and researchers. For many participants, much of this work began in the period immediately following their own diagnosis (or the diagnosis of a family member), as patients and their families embarked on a quest to fill a void of knowledge, expertise and experience. There are benefits and challenges in being a member of, and contributing to, a patient organisation, particularly in terms of sharing knowledge and experiences. But for founders, what distinguished their experiences from many other parents or family members in similar situations, was that they turned this intensely personal quest into one that would have much wider benefits for others. While establishing a national organisation may not have been a founder's formative intention, these grassroots efforts were often transformed into more formalised and structured networks. In many cases they created a community of practice (Lave and Wenger, 1991) based on shared interests and goals coalescing around the specific rare disease.

When establishing the organisation, participants thought carefully about strategies for forming professional relationships, recognizing the value of establishing a scientific or medical advisory board, or a board of trustees. Recruiting for these boards was one of the ways in which someone's interest and involvement could be secured. For some founders, this task was achieved by inviting already known professionals and family members. For others, it was also an opportunity to engage the most recognisable and skilled researchers in their field:

> We knew we needed to set up a medical advisory board, so we knew who we wanted to hit and where we wanted to hit them. So, we went to the top. [Kara]

Participants recognised how the contribution and engagement of prominent experts might provide much needed legitimacy for a patient organisation. In this endeavour, founders reflected the language for acknowledging expertise within a rare disease field (Featherstone and Atkinson, 2013). Going 'to the top' meant working with the clinician who gave their name to the syndrome, or identifying the 'king', 'queen' or to borrow the words of one founder (Ronan), a 'father figure' in that specific rare disease world. For Dev, legitimacy was also secured and strengthened through sponsorship with a well-known company and a relationship with a prominent university, all of which were described as valuable attributes for a newly emerging charity. Solidifying these relationships was seen as a priority. Indeed, founders were explicit in their accounts about professional expertise as a resource. One example was provided by Julie, whose organisation raised funds for research and strategically determined the types of projects to support. Julie emphasised the importance of implementing a robust peer-review system to evaluate grant applications, ensuring that their funding decisions were guided by scientific merit and alignment with the organisation's goals. This system involved assembling a panel of experts to assess the feasibility, innovation, and potential impact of proposed studies:

> Have an absolutely, massively robust peer review system so that even when a project fails, if it has been peer reviewed, if the project has been peer reviewed well, people would be able to build on it […] Secondly, attempt to get the most eminent people you can on your board to look at the applications, because that will, as soon as people see who's on your board it kinda speaks a language to them about the validity of your research. And then, thirdly, you gotta get the best name you can to be the chair of that board, because that's what gets peer reviewers. [Julie]

This structured approach enabled the organisation to maintain transparency and credibility, maximising the likelihood of advancing meaningful research that addressed the specific needs of their patient community. Throughout their accounts, key clinicians or clinician-scientists were noted as influential in helping to shape the organisation's relationship with research. Limited resources (including time, energies and personnel) made it even more essential that an organisation made the right decisions at the right time. Several participants reflected on how much they had achieved over the years. For Dev, organising an international meeting that was attended by the 'Who's Who' in the field, was a notable accomplishment:

> We had an International Board meeting last March and again where we had people in France, US, so that they, in the *tiny* world, the Who's Who in the tiny world of [named rare disease], we brought to London and initially, we sort of funded that ourselves. [Dev]

The phrase "tiny world" stands out here, reflecting a recurring theme in the language of participants. Some described their experiences as working in a "very tiny rare disease organisation" [Julie], a "shoestring organisation" [David], and as Ronan explained in acknowledging interconnections, "as a *small charity* with seven trustees, we could never have done this on our own". The choice of words underscores the close-knit and specialised nature of these organisations, highlighting their unique position within broader medical and research communities. The nature of rareness also means that professional expertise is a scarce resource, with experts often being geographically dispersed and disconnected (Navon, 2019). In this context, participants described their role as not just identifying experts, but also needing to offer encouragement for them to work together. The first stage of this process, for many, was to invite professionals to share the same space. Several

participants emphasised the significance of being 'at the table', a metaphor used to denote meaningful engagement in decision-making processes. Participants elaborated on their role as the host of such meetings, actively creating spaces for knowledge exchange and collaboration to facilitate inclusive and impactful discussions:

> We were there, at the table, and we took the responsibility of being there very seriously. In the early days we sought to bring the treatment centres together. We would host meetings, and bring the doctors, the nurses, the specialist nurses from the centres together for a meeting with us and each other. [David]

Bringing the right people together in the same room reflects a long standing (and since the pandemic, a newly invigorated) debate about the importance of physical events for generating trust and community (Collins et al., 2023). Founders made decisions about how to create the optimum environment to foster meaningful and sustained dialogue. Dev, for example, organised a summit for the 'higher end levels of organisations', those who might be considered the elite, powerhouses, or 'masterminds' in the field:

> And we're all gonna basically go and stay in a large house and basically run through almost like a private sort of mastermind group for lack of a better word. [Dev]

Examples given of outcome-focused projects where such collaboration was considered crucial, included creating multidisciplinary standards of care, exploring the potential and pitfalls of new treatments, and campaigning for access to treatment. Ronan's experience was that strong encouragement was required at times:

> We need to lock them [specialists] together in a room until they agree to do things together, which [is] what we did. We got them together once a year in a conference room. [Ronan]

Although the language of 'locking' people in a room is used here for effect, it illustrates founders' recognition that these professionals were not necessarily used to working together, and had competing demands for their attention. For Ronan, this strategy worked:

> And we got them to talk about the work they're currently doing pre-publication, which most of them admitted it never happened before and they agreed to international collaboration and they adjusted their trials dynamically on the basis of what was the results they were getting in other countries. So, it really pushed forward the gene therapy work trials. [Ronan]

In this example, the professional community was extended by inviting those who were experts in their given field, but not necessarily experts in this specific rare disease. Thus Ronan's efforts overcame the barriers that might normally restrict rare disease research, including working within disciplinary boundaries (Courbier et al., 2019). Successful matchmaking, between individual people, and for particular projects, resulted in people from across the world, sharing their current research and actively changing their practice. We might understand founders' engagement with professionals as a reconfigured form of 'upstream' engagement', a term most commonly used to discuss the involvement of multiple publics in research and development (Pidgeon, 2021). But here it is the founders themselves who are driving the agenda, by facilitating spaces for engagement, inviting those with the potential to make a difference, and directing their engagement. Focusing on founders' accounts reminds us of the difficult landscape of 'rareness' in which they are working, where small organisations can struggle in the battle for resources and influence (Baggott and Jones, 2014), and where building a collaborative community becomes essential for making progress.

2.2. Theme 2: translation work

All the founders of the rare disease organisations outlined the importance of investing in research to generate new knowledge of these rare conditions and advance progress in diagnosis, treatment and care. However, they also acknowledged that making a significant impact in research was extremely challenging. Founders were aware that decisions needed to be made about how best to use the resources they have, and that prioritising research could mean compromising other responsibilities:

> If we talk to the families here in the UK and we ask them, what do you want? They want money to be spent on research and they want to find a treatment and a cure. We're a tiny group of 30 individuals in the UK, who can never raise that kind of money. And we also have to support [patients and families] now, while we're waiting for, you know, potential treatment that could take another 10 years. So, there is always a robust discussion about how a limited resource should be spent. [April]

As April explained, one prominent concern was finding a balance between addressing current patient needs and investing for a better future. Compounding the problem for some founders was their unfamiliarity with the world of medical research. Julie described how she and another parent had met with a clinical expert to explore the best ways for them to support research. The clinician described how findings are modelled on other better resourced and well-known conditions:

> And then he explained to us the concept of translational research, how you take a finding in one condition and you test it and replicate it in another condition. And then we suddenly begin to realise that you could be literally decades behind the more common muscle diseases because the translational work wouldn't have been done. [Julie]

Here, the concept of 'translational research' was proposed as a practical entry point for the organisation to influence the research agenda. Translational research describes a branch of medical research that aims to bridge findings from basic science into practical applications. It is often described as a "bench-to-bedside" or even a "bedside-to-bench" approach (Löwy, 1996), which means taking insights gained from laboratory experiments (the bench) and developing them into clinical applications (the bedside), though this metaphor has been criticised for assuming linearity and concealing its complexity (Lewis et al., 2014). There are considerable barriers to progress in rare disease research, particularly in relation to drug development, where such bridging work is essential (Lumsden and Urv, 2023). Small patient populations make clinical trials difficult, and regulatory hurdles slow the translation process, ultimately making such research even less attractive to stakeholders (Martin, 2022). By funding translational research, the founder was encouraged to see how the rare disease organisation might 'fit' within a research structure, where initial funding could act as a catalyst for further research:

> [The clinician] said what you need to do is fund the 100,000 proof of principle projects. So, you're in the realm of funding something that will get a theory over the line, a scientist who has a hypothesis to do with your condition, they need to test it and if it gets over the line and you prove it, then in behind you will come bigger funders, because it becomes more likely for them. And then behind them, if it really becomes likely, would come the biotechs. [Julie]

While the clinician in Julie's account explained how translational research could work for this rare disease organisation, we recognise that a broader concept of 'translation' is highly relevant to the work and experiences of founders of rare disease organisations. We thus use 'translation work' to describe the efforts of founders in working out how to bridge the knowledge and expertise gap which characterises the rare disease world. Further, we emphasise the role of translation in founders' accounts, essential work performed as part of a larger process, for example, forming relationships with professionals, encouraging them to engage and work together, required a considerable amount of effort,

both emotional and physical. And of course, this was not the desired endpoint, but simply a first stage in creating the optimum conditions for experts to collaborate and forge a community of action. We use translation instead of concepts such as articulation to highlight the fundamental nature of this work to the role and experience of a founder or leader. Translation work is an essential element in the success of a venture (Trupia et al., 2021), it is neither temporal or contingent, and therefore cuts across the visible/invisible dichotomy which has been used to define and value different kinds of work (Star and Strauss, 1999; Lydahl, 2017).

Translation work in its broad sense can be seen in founders attempts to make something understandable, desirable or relevant to others. For example, founders identified how work was required in negotiating 'rareness'. Strategies included making the disease 'appealing and motivating' and potentially appearing 'less' rare, in order to encourage engagement:

> You have to try and for want of a better word, make it look sexy and, you know, appealing and motivating … we all have this saying, 'we're not too rare to care'. Unfortunately, some of the conditions are too rare for people to care about. [Anja]

When describing the grant review process, Julie recognised what she needed to do for experts to engage, "Tell us what you think, we'll write the notes. We'll forward things. We'll copy things, we'll do everything, but we make your life as easy as we can and so they stay with us" [Julie]. Another example of this essential, but often less visible work was provided by April, who described how she motivated and encouraged others during the establishment of specialist services. April had attended a rare disease conference and heard a presentation by a representative from the NHS specialist commissioning services. On her return she related the information to the clinical team, actively encouraging them to take the opportunity seriously, despite their misgivings that they did not have the experience or capacity:

> They [the clinical team] went oh, we can't do that, we've never done that before, that seems like really far too much work, it's gonna be really difficult. But slowly, I think, they came around to like, actually, maybe it could work. It was the first time they'd ever applied for that kind of funding, you know, it took a bit of time as well while we worked together on that, but it was successful and I think that was great. And then you know, from there we managed to run the first clinical trial in the world. [April]

Here, the eventual success of the endeavour was noted by the founder. The project led to the first clinical trial relating to that rare disease, and the founder was able to acknowledge their role, stating that "we managed to make it sustainable" [April]. The nature of such efforts was that founders occasionally found it challenging to substantiate their role in achieving a successful outcome, particularly when their contributions were less tangible. Examples included forging relationships over time, or speaking to key stakeholders in opportune meetings. This was the case for Sean, who discussed the inclusion of the rare disease in newborn blood spot screening. Although he described attending meetings, and forging connections with some of those involved in the decision-making process, he felt unable to claim the successful outcome as his own, "I would love to have said that we were involved".

The establishment of a patient registry serves as our final example for examining the translation work undertaken by the founders. Several participants recognised that, given the rarity of the disease, a well-constructed patient registry would be an invaluable resource. It not only facilitates the collection and analysis of critical data but also fosters collaboration among researchers, supports clinical trials, and provides patients with a platform for engagement (Hageman et al., 2023). This was the case for Beth, who explained how their international register was "stimulating research":

> We set up our own patient register, which is […] really good because that's now global. We've now got pharma companies potentially to help use that to help, to recruit people for trials. [Beth]

Establishing a patient registry comes with administrative challenges, or as June described, "the hoops you have to jump through". These point to the large amount of effort and resources required to collect and store data, and managing ethical and legal issues associated with the Charity Commission and General Data Protection Regulation (GDPR). An additional hurdle that some founders highlighted was the resistance they met when attempting to encourage professionals to invest in a registry. Ronan exemplified the situation, acknowledging that if professionals were unwilling to commit, the rare disease organisation would have no choice but to take a more proactive role in its establishment:

> We tried for years to get clinicians to do it, but, um, it was never going to work basically. [Ronan]

Ultimately in Ronan's case, the rare disease organisation was able to employ an individual to collect the information for it then to become a useful resource. Difficulties with encouraging professionals to take the initiative was also Julie's experience, "patient registries are notoriously hard to get funded, and you layer in the fact that it's a rare disease, it's just impossible". Once again, in the absence of clinical and research professionals willing to do this work, Julie made the decision that the rare disease organisation would develop the registry themselves. And as in Ronan's case, the registry became a more appealing proposition to others after it had secured some coherence. On these occasions, investment was required by the founders and rare disease organisation to make registries doable for the professionals. In the latter example, a university hospital took over the administration of the registry and it has continued to mature, with potentially far-reaching benefits for the patient population. The extensive work involved was not lost on the founder and it was not without personal cost:

> So, we started it, maybe I suppose 10 years ago. We lost a lot of hair and had a lot of wrinkles for a couple of years as we ran that thing as well as everything else. At one point that was like a 7 day a week job, that was excruciating. [Julie]

Julie metaphorically and literally described the implications of her investment. Though not manual labour, this was work that clearly impacted the body, leaving visible markers as an outward sign of embodied and emotional labour (Hochschild, 1979). Yet despite its significance in terms of the outcomes for the organisation and the personal experiences of the founders, such translation work can be harder to recognise or celebrate as an achievement. Thus, much of the essential work of founders in establishing, strengthening and sustaining a community, risks being un-documented, under-resourced, and ultimately remaining 'back-stage' (Goffman, 1959).

2.3. Theme 3: organisational and personal resilience

The range of tasks performed by the founders of rare disease organisations are all closely aligned with a desire to improve the experiences for current and future patients. Although the founders were candid about their experiences, they acknowledged personal benefits of establishing the organisation, and took pride in their work. Sandra highlighted how this work was "cathartic", reflecting on how far she had come from the origins of "a little computer in my kitchen at home", to "look at us now, I can't really believe it". This is not to say that founders would be doing this work given the choice, as Dev said, "it's exciting as much as it is rubbish to *have* to do it". But most prominent in their accounts was acknowledgment of the intensity, and effort involved in establishing and sustaining the organisation. Often the founders felt overwhelmed and under-supported. Anja described how she was "walking the thinnest tightrope". This was work that was all consuming and draining of their resources, often performed alongside paid

employment, all the while when many had care needs at home precisely because of the disease. The founders were aware of the physical and emotional toll on themselves and their personal lives. In this context they discussed the importance of introducing resilience into the organisation, to support its momentum. Many positioned themselves as currently indispensable for the organisation's sustained existence:

> If I got run over by a bus, the whole charity would suffer [Ronan]

> If I get knocked down by a bus tomorrow would everything carry on. But I think it probably would [Sandra]

> If something happens to one of us [a small number of parents who established and run the organisation], you know, it could all start to then fall apart [Kara]

The "bus" analogy vividly captures the sense of vulnerability founders experience for them and their organisation. Some participants described how they responded by reorganising or reprioritising their resources to adapt to this challenge. For example, Ronan described his own "resilience plan". This involved working out a new structure for the organisation, and for his role to change, enabling him "to become a true chairman of the trustees and not the day-to-day coach and then chief cook and bottle washer as well". Here, the founder summed up his own experience of establishing and running an organisation, particularly the need for him to multi-task, which meant not necessarily using his skills or abilities in the most efficient way. Securing adequate payment for roles was identified as a way of ensuring sustainability, representing a move away from amateur *ad hoc* relations towards a more scaffolded and resilient approach. On this, as Dev explained, the economic implications were clear:

> I work in my [commercial] company half a day to a day, a week. This [work with the rare disease organisation] is very much now my full time, unpaid role. One day I think it will have to be paid in the sense of like, I can't take the wage cut that I take forever. [Dev]

Many founders were in paid employment elsewhere, describing this as essential to support their home and family. But where possible, this work was deprioritised to focus on developing the rare disease organisation, reflecting the shift in priorities for parents or patients following a diagnosis (Pinto et al., 2018). Participants strongly expressed how leading a rare disease organisation was a full-time job and that its under-resourcing was a problem. April explained how her organisation had been reorganised to enable her to transition from a "full time, unpaid job" to a paid role, a change that was vital because "I can't carry on not bringing in a salary". This was similar for Beth, who explained the value of being paid for her work. She described the 'difficult and challenging' position of juggling her previous employment alongside her investment in the rare disease organisation:

> So then I spoke to the trustee board […] I think what one of my trustees said was, 'this is where the risk is, because let's not worry about [funding] research, let's get you on the salary' […] I dropped my career to make everything work at home. It's tough. Like really, really, really, really, really tough. I can't, it's just like, I know it's really tough. Anyway, um, and then that's probably one of the best decisions we've made because I've just got another grant for the work. I've been able to work for grants, and we've got enough money to employ someone else, so we're going to be employing someone else to help me. [Beth]

Here, Beth was retelling the words and actions of a trustee, who had recognised her critical role in the organisation. The trustee identified that the fundamental 'risk' was not a failure in securing future grants, but it was losing the founder who had contributed, and continued to contribute so much. And as we know from her account, Beth had already experienced personal cost, in terms of giving up her previous career. The result of this discussion was that colleagues were willing and able to move resources and shift priorities, in order to sustain her involvement. Participants communicated and made tangible the burden of their work in the organisation. In this case, the misalignment between 'volunteering' as an under-resourced passion and paid employment was recognised and addressed (Taylor, 2005; Dean, 2022), but of course, this would not be an opportunity afforded to all.

The need to introduce paid roles to sustain a rare disease organisation potentially challenges the foundational values on which it was built. Beth for example, acknowledged that a paid role was possible, "even though we were very much volunteer-led". A similar tension was raised by Kara when talking about recruiting new trustees who might not be parents, and the implied move away from the values of being 'parent-led':

> It's something that we are struggling with as a team, because we very much feel it's our driving force, our commitment and I suppose, is focused on our children and why we're doing this is because of that. [Kara]

Sam also found it troubling to bring others on board, particularly if this was part of a strategic business approach. He highlighted how patients needed more than just "9-to-5, 3–5 days a week", making an implicit contrast between a business approach to resource management and the unlimited passion and support that might be offered by patient or parent-led organisations. April's account highlighted the personal burden of planning for the future, which ultimately manifested as a need to become professional:

> It's hard to keep the momentum going. I think it wears you down. That battle for money and resources, you know, to get to the next level because you start off small. But then you quickly realise that you've got to become more and more professional, to achieve your aims. And all of those things do take time and money and you know, expertise. [April]

Professionalising a rare disease organisation requires compromise for its founders. It comes with a cost, in potentially losing some control and ownership and diluting the passion that underlies their *raison d'etre*. At the same time, responding to change and becoming agile was recognised as essential for enabling rare disease organisations to continue their work. One founder, Julie, described the challenge of being prepared to navigate future obstacles, "we don't know what rocks are coming our way". She continued:

> We've had a few occasions where we've stopped and halted and thought about things. And we refocus what the research targets would be and change the kind of wording of what our, of our grants round slightly and then on, you know, structure and decisions in the charity and where we spend time, we would talk to the trustee board about and kind of observe, observe how things change as we go along. [Julie]

Rare disease organisations are not static, but live, dynamic organisations. The growing workload of a successful organisation, coupled with the emotional toll it takes on its founders, presents a significant risk of burnout, potentially leading them to step away from their roles. This not only affects the founders' well-being but also has far-reaching implications for the organisation, its members, and the broader community it serves. Throughout their accounts, the founders highlighted their awareness of the complex needs of a patient community, and the difficulties of addressing these in a changing, resource limited landscape. It was clear that founders played a crucial role in sustaining the organisation. Responding to these changes demands a form of professionalisation which while providing resilience, risks moving the organisation away from its grass-roots foundations.

3. Conclusion

This article contributes a deeper knowledge of the personal and professional challenges facing rare disease organisations. It responds to

Patterson et al.'s (2023) call for qualitative research to explore leaders' experiences. Our focus on the accounts of the *founders* of rare disease organisations, rather than solely its current leaders, is crucial. Founders play an essential role in organisational identity. Intimately entwined with their own biographies, founders provide unparalleled insight into the organisation's origin story—the motivations, challenges, and values that drive and shape the organisation's creation and continued existence. Understanding how and why the organisation was established reveals the foundational principles and context that shaped its trajectory, offering a deeper comprehension of its purpose and evolution. As Mikami (2020) illustrates, differences in the constitution of rare disease groups can, in part, be explained through the visions and personalities of their founders. Founders' narratives often capture the initial vision and its challenges, which may no longer be as visible in the day-to-day operations led by current leaders. Hearing directly from those present at the beginning provides an authentic perspective on the organisation's ethos, highlighting the passion and determination required to bring it into existence. This historical lens helps to appreciate the transformative role of the organisation in its field and provides a baseline for assessing its growth and adaptation over time. Our research highlights that founders of rare disease organisations are the driving force and passion behind its formation; they are innovators, entrepreneurs and decision makers. Founders' stories thus serve as a vital piece of institutional memory (Linde, 2008) with research interviews being an essential tool in recording this social history.

Prior studies have documented the extraordinary and powerful work of rare disease organisations in generating knowledge and expertise, thus transforming their social and biological worlds (Strand and Holen, 2024). We have highlighted the less discussed, but equally important aspects of this work, where founders address fundamental challenges of building expert capacity in rare disease research, policy and practice. This article demonstrates that when nurturing a professional community, founders make efforts not just to identify experts but also do the work of bringing them together and encouraging collaboration. Success, however it is measured, is often dependent on the continued resources, energy and passion of charismatic and self-motivated individuals. Founders tended to draw on their own resources as a catalyst, to make projects attractive, and 'doable' for professional investment. They expressed a strong desire to sustain the organization, to enable it to adapt to change whilst supporting its community, but voiced concerns about their ability to maintain such a high level of input. Throughout this article, we recognised and valued the founders' experiences and efforts as 'work'. Labelling and recognizing the activities, thoughts and motivations of founders as work, enabled us to examine what it entails, as well as its costs and rewards, helping us to address its undervalue (Hatton, 2017).

Drawing on Star and Strauss (1999), and writing in relation to the Russian Multiple Sclerosis Society, Endaltseva (2024) highlighted the overlooked role of invisible work in rare disease organisations. Our article builds on this by exploring founders' hidden yet valued efforts in leadership and decision-making. We term this 'translation work,' emphasizing that the founders were cognizant of the value of this work and its role in sustaining organisations and communities. All of the contributions of the founders, at least in the beginning, were unpaid, strongly reflecting elements of the 'invisible', and socially and economically undervalued nature of voluntary work (Daniels, 1987). One of the reasons why the labour of founders fits so readily into the category of essential yet devalued, is its close entanglement with the personal. We interviewed founders precisely because of their personal biographies. Their experiences were the reason for establishing the organisation, and remain the motivating force for their continued involvement. Their work is also closely entangled with the home. Founders made constant references to the home as the origins of the organisation, some explicitly related their kitchen table to a workspace, or described how plans were made from an initial meeting in their kitchen. And of course, what for many was a "full time unpaid job" had personal implications, blurring any distinctions between work and home, and between public and private. It also blurs distinctions between the formal and informal, or everyday practice, often used to make sense of visible and invisible labour in the workplace (Grant et al., 2016).

We agree with Patterson et al. (2023) and Pinto (2018) that there are steep learning curves for patients and families in establishing and sustaining a rare disease organisation, and we have highlighted that founders are at risk of emotional exhaustion (Allen and Augustin, 2021). We encourage future researchers to consider the challenges for rare disease organisations, and make visible and value the ordinary and extraordinary work of their founders.

CRediT authorship contribution statement

Rebecca Dimond: Writing – review & editing, Writing – original draft, Project administration, Methodology, Formal analysis, Conceptualization. **Jamie Lewis:** Writing – review & editing, Writing – original draft, Conceptualization.

Acknowledgements

With their agreement, we would like to thank the rare disease organisations who took part in the project: Alex, the Leukodystrophy Charity; Arthur's Quest/ SLC6A1 Connect UK; the British Porphyria Association; Barth Syndrome UK; Cavernoma Alliance UK; Fanconi Hope; Fragile X Society; Gauchers Association; Glut1 Deficiency UK; Keep Me Breathing; Myotubular Trust; PXE Support Group; Rett UK; SCN8A UK & Ireland; Stargardt's Connected; XLH UK.

Data availability

The data that has been used is confidential.

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