



'We're kind of like genetic nomads': Parents' experiences of biographical disruption and uncertainty following in/conclusive results from newborn cystic fibrosis screening

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ABSTRACT

As whole genome sequencing is being considered as a tool to deliver expanded newborn screening (NBS) globally, the range of equivocal results it could produce are gaining increased attention. For cystic fibrosis (CF) screening, the use of next generation sequencing within existing UK NBS programmes would increase the number of uncertain designations returned within results, including that of Cystic Fibrosis Screen Positive Inconclusive Diagnosis (CFSPID). However, the experiences of families already living with this designation have been under-explored. This study uses in-depth interviews to explore the perspectives of sixteen parents who received positive results from CF NBS, with varying degrees of prognostic un/certainty; parents with a child diagnosed with CF ($n = 6$), CF carrier status ($n = 3$) and those with the CFSPID designation ($n = 7$). The biographically disruptive nature of positive NBS results-regardless of immediate relevance to the child-dominated early experiences of positive results across all groups. For those with CF, biographical reparation involved becoming 'a CF family', underscoring biological kinship bonds and reinforcing familial identity. For those with uncertain results, biographical re-calibration was more complex. Diagnostic and prognostic uncertainty posed a barrier to entry for both the 'CF world' and the 'healthy kid' world, leading parents to attempt to minimise its role, either through rejection, or re-interpretation of their child's result. Other parents, however, experienced biographical reparation more dynamically. The concept of 'genetic nomadism' captures accounts of oscillation between the two worlds; movements that were responsive to evolving health experiences, as well as social, environmental and temporal factors. Through the concept of genetic nomadism, this paper delineates both the productive, as well as divisive, nature of uncertainty for biographical reparation in the aftermath of NBS, as well as the strategies parents use to harness it, in order to successfully navigate the world with a child with an ambiguous genetic future.

1. Introduction

Globally, whole genome sequencing techniques (WGS) are increasingly being incorporated into healthcare systems. Such techniques are capable of quickly and efficiently producing large swathes of data in the form of 'sequences' of DNA base pairs which together constitute that individual's entire genetic code. Whilst initially utilised in the context of clinical care (e.g. The 100,000 Genomes Project), WGS is now also being used in population-level screening programmes e.g. MacKenzie's Mission, Australia (Delatycki et al., 2019) and UMCG, Netherlands (Birnie et al., 2021). The use of WGS for newborn screening (NBS) is currently being piloted in the US (the BabySeq Project), and will also be piloted in the UK (Genomics England, 2021), where it has the capacity to

dramatically increase the number of pathogenic variants identified. This switch to genome-wide screening in newborns would facilitate the pre-symptomatic instigation of therapies and interventions, and has precipitated a renewed interest in exploring and refining the means and ends of NBS, particularly from the perspective of the general public and parents (Hopkins Van Mil, 2021).

Alongside potential benefits of WGS NBS, the number of uncertain genomic results, including variants of unknown significance (VUS) and results that are not of immediate relevance to the newborn (such as propensity to late-onset conditions) would correspondingly also increase (Esquerda et al., 2020). This has given rise to concerns around the screened child's right not to know their genetic status (Moultrie et al., 2020), and their entitlement to an 'open future', uninhibited by

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knowledge of a potential future condition, particularly when its onset is uncertain or inexorable. Fears have also been expressed that healthy children will become over-medicalised (i.e. monitored and prophylactically treated for conditions that may never manifest), families may experience psychosocial burden (e.g. anxiety, stigma) (Howard et al., 2015) and the child's self-identity, and perceptions of their health, may also be negatively impacted (Detmar et al., 2008).

1.1. Liminality and genetic health

The impact, and meaning, of living with an equivocal health status has long been studied within medical sociology. Indeed, the concept of liminality-to demarcate the spaces that emerge between sickness and health-was described by Frank (1991), and later Little et al. (1998), to account for the experiences and status of cancer survivors within 'remission society' (Frank, 1991).

In relation to genetic findings, liminal results have previously been explored in relation to late-onset neurodegenerative conditions (Chillibeck et al., 2011), cancer (Heinsen et al., 2021; Dean, 2016; DiMillo et al., 2012), coronary heart disease (Snell and Hélen, 2019) and developmental syndromes (Whitmarsh et al., 2007). This body of literature has captured the various ways that lives impacted by 'diagnostic uncertainty' are punctuated by practices of medical surveillance and/or intervention, and are governed by a sense of genetic and social responsibility to prevent future disease (Heinsen et al., 2021; Hallowell, 1999). Moreover, the complex- and sometimes ambivalent-relationships that 'at risk' individuals have with their equivocal genetic status has been demonstrated through their range of responses to it; including attempts at its minimisation (Chillibeck et al., 2011), through to its transformation into a site of hope and positivity (Whitmarsh, 2011).

These studies all highlight the significant social and cultural implications of uncertain genetic health statuses, and the varying impacts for families and individuals, of living within 'chronicities of risk' (Heinsen et al., 2021). However, the transferability of this literature to NBS contexts is somewhat limited by the familial histories that contextualise participants' interpretations of their genetic status (Snell and Hélen, 2019). Indeed, it has been well documented that lived experience mediates perceptions of genetic risk and responsibility, informing the way that futures are imagined (D'Agincourt-Canning, 2005). In contrast, positive NBS results are typically unanticipated; the majority of families receiving one lack a family history with the condition, fundamentally impacting the way the result is received and responded to (White et al., 2021).

1.2. Biographical disruption, newborn screening and genetic risk

'Biographical disruption' (Bury, 1982) is a concept originally developed as a lens through which to understand the ways that an abrupt health event can fundamentally challenge a person's social and cultural identity, activities, and sense of biographic purpose. Whilst initially employed to explain experiences of a chronic illness, there are parallels with the experiences of those who are thrust into 'narrative reconstruction' (Snell and Hélen, 2019) following an unexpected genetic finding, particularly in the context of NBS. Indeed, the largely 'routinised' nature of NBS (Nicholls, 2012) and the rarity of conditions screened for (Chudleigh et al., 2016) both contribute to the epistemic shock that is experienced by parents when a positive NBS result is returned (Chudleigh and Chinnery, 2020).

For families where NBS results are not only an unanticipated shock, but also imbued with 'diagnostic uncertainty' (Timmermans and Buchbinder, 2010), biographical reparation can be particularly complex. Previous research has explored the impacts of uncertain results from NBS in relation to Pompe Disease (Kwon and Steiner, 2011) type 1 diabetes (Kerruish, 2011), cystic fibrosis (Johnson et al., 2019; Tluczek et al., 2010; Hayeems et al., 2017), Krabbe Disease (Ehmann and Lantos, 2019) and metabolic disorders (Timmermans and Buchbinder, 2010).

This literature variously emphasises the difficulties families experience in reconciling uncertain results within dichotomous schemas of health and illness, and consequently interpreting their social and cultural significance (Johnson et al., 2019).

Despite this area of work, there have been few attempts to make direct comparative analyses between the experiences of families who receive different types of positive NBS results, including those leading to a definitive diagnosis, those that are uncertain, and those not immediately relevant to the newborn (i.e. carrier status). Where this has been attempted, research has found that those with equivocal genetic results experience similar anxiety and uncertainty-if not, more-than those who receive diagnoses (Hayeems et al., 2017). This is particularly striking given the context of burgeoning applications of WGS, and the concomitant increase in ambiguous genomic results it could produce. Given this wider context, the need to better understand the varying impacts on families across the spectrum of un/certain positive results that could be returned through NBS is now critical.

1.2.1. Cystic fibrosis, CFSPID and newborn screening

NBS for cystic fibrosis (CF) is an example of a screening programme that is capable of producing a range of un/certain positive results. CF is an inherited condition, linked to variants in the CF transmembrane conductance regulator (CFTR) gene, affecting one in every 2500 newborns in the UK (Schülter et al., 2020). It causes thick, sticky mucus build-up in various organs, most notably resulting in reduced lung function and susceptibility to lung infections. People living with CF require regular and intensive treatment through physiotherapy, antibiotics and inhaled bronchodilators/corticosteroids (CF Cystic Fibrosis Trust, 2020). Once considered a condition fatal in childhood, new CFTR modulator therapies (e.g. Trikafta, Orkambi) are expected to have a significant impact on life expectancy for people with CF (Balfour-Lynn and King, 2020)- currently 49 years in the UK (CF Cystic Fibrosis Trust, 2020).

NBS programmes for CF are well established internationally, as evidence suggests improved clinical outcomes when CF is detected early (Schlüter et al., 2020). In the UK, dried blood spot cards are used firstly to test for immunoreactive trypsinogen (IRT), with an analysis of the CFTR gene conducted for those infants with high levels (Castellani & Massie, 2014). Infants with changes to the CFTR gene have a sweat test to confirm the diagnosis.

Alongside those with CF, however, current NBS methods also identify infants who have changes in the CFTR gene, but who do not otherwise fulfil the diagnostic criteria for CF (i.e. normal/intermediate sweat test). These newborns are assigned the designation 'Cystic Fibrosis Screen Positive Inconclusive Diagnosis' (CFSPID), otherwise known as 'Cystic Fibrosis Transmembrane Conductance Regulator-related Metabolic Syndrome' (CRMS). The long-term health implications of CFSPID are not clearly understood, and whether or not these infants will go on to develop full CF in the future cannot be accurately predicted (Barben et al., 2020). Recent evidence suggests that around 10% will convert to full CF, with the majority remaining well throughout their lives (Terlizzi et al., 2019). Given this context, relatively conservative management of CFSPID has been recommended (Barben et al., 2020), however there remains wide variation in clinical management (Terlizzi et al., 2021), with many children followed up regularly by CF clinics and/or prophylactically treated (Terlizzi et al., 2019).

In spite of its continued identification through CF NBS, there has been relatively little research into the lived experiences of parents whose child receives a CFSPID result, with a few notable exceptions (Johnson et al., 2019; Tluczek et al., 2010). However, the need for such research is becoming more urgent as sequencing techniques are being considered for use within existing CF NBS programmes to improve their specificity (Doull, 2019). Whilst reducing the number of missed CF cases, this shift would also increase the number of CFSPID cases and carriers identified (Campen et al., 2019; Comeau et al., 2004). Given this context, the need to better understand the experiences of families across the range of

positive CF NBS results possible, is now of critical importance.

This study uses in-depth interviews to comparatively explore the views of parents who received different types of positive results from CF NBS; seven parents of a child assigned the CFSPID designation, six parents who received a conclusive CF diagnosis, and three who received a carrier status result. Through the accounts of these parents, the role of uncertainty in the context of their child's result will be brought into critical relief, highlighting both its restrictive, but also highly productive properties.

2. Methods

The interviews were undertaken as part of a larger mixed methods study of attitudes towards genetic screening amongst families living with CF, haemophilia, fragile X syndrome, thalassaemia and spinal muscular atrophy conducted by the authors (Boardman and Clark, 2022). As part of this mixed methods study, 26 in-depth qualitative interviews were conducted with people living with CF, CF carrier status, or CFSPID in their family to explore their attitudes towards genetic screening (pre-conception, prenatal, newborn). This paper reports on a sub-analysis conducted on the transcripts of 16 participants (parents) who had experience of a positive CF NBS result, extracted from the larger CF dataset.

Participants were recruited to the study through a paediatric NHS CF clinic in the South of England, a call for participants in the Cystic Fibrosis Trust's e-newsletters, the project's social media accounts and through snowball sampling. Parents attending the CF clinics were advised about the study through an information leaflet distributed by CF nurses, and a poster advertising the study was also put up in the waiting room. Those interested in participating were invited to make contact with the lead researcher, and to pass on the study information to others who might fit the recruitment criteria, including partners. A relatively long period of recruitment and data collection (September 2018–June 2019) was used to maximise opportunities to involve people with a range of NBS experiences. Attempts were also made to recruit parents whose child received a false positive CF result, however, these ultimately proved unsuccessful. Ethical approval was acquired through the Health Research Authority (17/WM/0231 01/08/17) and the Biomedical and Scientific Research Ethics Committee, University of Warwick (REGO-2017-190 February 21, 2017).

Fourteen parents made contact, all of whom were eligible for interview. Two invited their partner to participate, resulting in 14 interviews with 16 participants that took place shortly after their first contact. Of the 16, 11 were recruited through the CF clinic (five CFSPID, three CF carrier, three CF) and five through the Cystic Fibrosis Trust advertisements (one CFSPID, four CF) (see Table 1). The children ranged in age from 6 weeks to 11 years (Table 1) and all had received their CF result within the first two months of life. The inclusion of parents at contrasting time-points following their child's positive result proved to be a valuable dimension of analysis, although they were not purposively sampled to reflect this. Rather, the range in time-points was likely a by-product of the recruitment methods employed and the follow-up policies of the CF clinic. All three participants whose child was a carrier were recruited through the clinic, and had all relatively recently received their child's result. This was due to the fact that carriers are not routinely followed up clinically after their initial appointment, and few join the CF Trust. Those recruited through the CF Trust, on the other hand, typically had older children and had been living with CF for some years.

Interviews were conducted around participants' availability and preferences: eight over the telephone, two over skype, and six face-to-face (five in participants' homes and one in a workplace). The interview schedule was developed in conjunction with an expert review panel, made up of two staff members at the CF Trust and two parents of children with CF. The interview schedule used broad, open-ended questions to encourage participants to 'tell their stories' of how CF came into their lives, their reaction to a positive NBS result, the

Table 1
Interview participants.

Participant	Gender	Age	Ethnicity	Screening outcome	Name (gender) and age* of child at interview
Abby	F	36	White British	CF	Harry (m) 10
Simon (married to Abby)	M	40	White British	CF	Harry (m) 10
Tamara	F	42	White British	CF	Max (m) 11
Alex	M	35	White British	CF	Evie (f) 4
Jaqueline	F	29	White British	CF	Matilda (f) 2
Leona	F	40	White European	CF	Bella (f) 6
Olivia	F	33	White British	CF carrier	Noah (m) 7 weeks
Roisin	F	28	White Irish	CF carrier	Benjamin (m) 6
Kayleigh	F	22	White British	CF carrier	Zoe 13 weeks
Emma	F	30	White British	CFSPID	Jaxon (m) 2
Hayley	F	38	White British	CFSPID	Sophie (f) 5
Russell (married to Hayley)	M	41	White British	CFSPID	Sophie (f) 5
Julianna	F	38	White British	CFSPID	William (m) 3
Elena	F	27	White European	CFSPID	Leo (m) 4
Beth	F	39	White British	CFSPID	Freya (f) 6
Rachel	F	34	White British	CFSPID	Thea (f) 2

confirmatory testing process, and the subsequent impacts of this information on their lives. These interviews were designed to generate 'thick description' of the lived experience of a positive CF screening result, and facilitate an exploration of the way these experiences were mobilised in attitudes towards other forms of genetic screening programme (e.g. prenatal, pre-conception).

All interviews were transcribed verbatim with identifiers removed and pseudonyms applied. A modified grounded theory approach to analysis was adopted (Glaser and Strauss, 1967; Ramanadhan et al., 2021) using NVivo 12 to organise and streamline the analytic process. Pragmatic approaches to grounded theory are increasingly being employed as qualitative methodologies that permit analysis to be both 'data driven' (Gibbs, 2007), yet also interpreted within the context of surrounding literature to develop pre-existing theoretical constructs (Ramanadhan et al., 2021). Given the lack of pre-existing literature on experiences with CFSPID and comparisons across the range of positive CF NBS results, the need for a data-driven approach to analysis was chosen, albeit modified pragmatically given the constraints of the larger study for which the data was originally gathered.

Using this combined inductive/deductive approach to analysis (Fereday and Muir-Cochrane, 2006), broad overarching themes were firstly identified iteratively through thematic open coding (Chapman AL and Chapman, 2015), before being refined into conceptual sub-themes (axial coding) to illuminate relationships within and between the overarching themes (Braun and Clarke, 2006). Unlike traditional grounded theory approaches, this second stage coding was subject to interpretive analysis-drawing on concepts developed from the data, as well as those detailed in the surrounding literature. Indeed, it was through this process that the significance of 'biographical disruption' to the experience of newborn screening emerged, which, in turn, framed the remainder of the analysis. Data analysis continued until saturation

occurred - that is, no new themes were being added to the coding framework (Glaser and Strauss, 1967).

The results of this paper have been organised around the core sub-themes developed through the analysis, with references to the literature included where this was used to interpret those subthemes. The quotations included in this paper were selected based on their concise representation of the meaning and content of the subtheme being presented.

3. Results

Fourteen interviews were conducted with sixteen participants (Table 1), the majority of whom were female (81%) and white British. The average age of participants was 34.5 years (range 22–42), reflective of participants of reproductive age. The two overarching themes, 'biographical Disruption' and 'biographical Repair' will be presented in turn, as well as the subthemes within them, to account for differences across participants, and by type of positive result.

4. Screening results as biographical disruption

'Bolt from the blue'

All 16 participants reflected on their CF NBS experiences as highly stressful and deeply emotionally charged. All had only vague recollections of consenting to screening after the birth of their child, with one parent stating that they had not realised it was optional, but something 'you just do when you have a baby' (Abbey, CF), mirroring concerns in the literature regarding the 'proceduralisation' of NBS tests and its implications for informed consent (Nicholls, 2012). Indeed, only three of the parents reported being aware of 'some' (Roisin, carrier) of the conditions that were being screened for, with CF being the one they were most familiar with. Given this context, receiving a positive result was experienced as an unwelcome and unanticipated shock by all. Jaqueline described her daughter's diagnosis of CF in the following way;

It was a complete bolt from the blue- completely. You know, the last thing you'd expect because ... I'd had such a straightforward pregnancy, the birth was ... fine ... [...] ... And when the phone call came through ... I actually missed the calls as ... I'd got all my 'congratulations!' cards ... around the answering machine, I never saw it flashing. Been too busy, enjoying my new baby, you know?..By the time they got hold of me they were already on their way round to the house and a nurse, or ... I forget who she was, turned up, handed me a leaflet on CF on the doorstep and said, 'we need to give you this ... can we come in?'

(Jaqueline, 29, CF)

As has been identified elsewhere, the context of having had a 'straightforward' pregnancy, previous healthy children and/or having 'passed' prenatal screening tests were all mentioned as having heightened parents' sense of shock when an unexpected result was returned (Buchbinder and Timmermans, 2011). Whilst two participants, Alex and Tamara (whose children were diagnosed with CF), had noticed that their children were 'poor feeders' (Alex) or had lost weight, the remaining 14 had no 'trigger warning' (Julianna, CFSPID) that something would appear in their results-either through symptoms, or family history. Julianna, mother to William (aged 3, CFSPID) commented;

He was absolutely perfect, and I'd thought it [NBS] was a bit like rubber-stamping him. They say they're looking for very rare things, so it's not something you're expecting to come back when you've got a baby that's so well ... you're just looking for that seal of approval that your baby is healthy and you're good to go, and I was fully expecting that to be the case.

(Julianna, 38, CFSPID)

Much like pregnancy has come to be regarded as 'tentative' until confirmed as healthy through medical screening (Rothman, 1993), so Julianna's sense of having a 'perfect' baby was subject to validation, or so-called 'rubber stamping' provided by NBS. Indeed, the notion that negative NBS results can be regarded as an implicit assurance of health, a 'seal of approval', serves to only heighten shock when a positive screening result is returned.

4.1. Interrupted parent/child trajectory: biographical disruption

Following receipt of a positive result, parents all underwent an intensive period wherein their child was abruptly recalled for diagnostic sweat testing - often the day of, or day after, initial results. Parents described the time between NBS result and diagnostic results as a 'crisis' point in their screening journey, whereby they had to quickly absorb a lot of information and simultaneously contend with overwhelming emotional responses. Leona, mother to Bella (aged six), recalled;

My mind was just racing, and we were scrambling to read as much about CF as we could, even though they had said to us, 'don't google it, just come to see us' ... but of course you do anyway. You're in panic mode. Maybe it was good we didn't have long to do it, we were at the hospital the next day having the sweat test. Up until that point we'd still got a bit of hope that they'd got it wrong, but then they said, 'no, this is what she's got'. And then our world collapsed and I just couldn't take it all in. We went from having a healthy baby to a very sick one in 48 hours. It was like falling from the sky into a whole new world. It was a hard landing that's for sure.

(Leona, 40, CF)

Parents whose child was diagnosed with CF had to respond to an imminent health threat, which shattered their perceptions of their child's identity, and their own as parents. Several reported feeling guilt for having not known, despite the lack of symptoms, impacting their confidence in their ability to parent. All of the CF parents reported that their baby was immediately started on enzyme treatment, and funnelled into regimes of treatment and appointments, requiring them to quickly assimilate into a 'whole new world' of CF. This included re-imagining their own, and their child's life trajectory, as well as confronting their own carrier status, and potentially also those of wider family members.

Simon, father to ten-year-old Harry (CF), reflected;

... I suppose I'd got this idea in my head about what fatherhood would be ... and it sounds ... like a cliché, but I'd imagined him helping his grandad [a carpenter] and me out in the workshop like I did as a boy, carrying on the family business ... and that just isn't possible for someone with CF because of the dust ... so it's having to let go, I suppose, of those idyllic dreams and come to terms with that. It's a bit like grieving ... yeah.

(Simon, 40, CF)

Like Simon, many parents' responses to the biographically disruptive nature of a CF diagnosis was described in terms of grief, as the anticipated trajectory of their parenthood, and their child's life, came to be replaced with one entirely unfamiliar.

This sense of disruption and re-orientation work, however, was not limited to parents who received diagnostic results. Those whose child received a CFSPID designation were similarly faced with a re-imagining of their (and their child's) present and future roles and identities. Russell, father to five-year-old Sophie, described his reaction to CFSPID:

It was a massive shock to us ... [...] ... I mean obviously I'm glad she doesn't have full CF ... but we have to live with the possibility of it every day. You know, how I parent her ... I will always have one eye in that ... direction ... 'if this happens, will she be able to cope with x, y,z?' ... we factor it in to decisions we make in life.. [...] ... like where

we live: are we near a decent CF centre ... ? Is there anyone else CF in her school that could infect her? So we prepare for the worst, but hope for the best.

(Russell, 41, CFSPD)

Whilst the majority of children assigned the CFSPID designation remain well, Russell's account demonstrates that, not only can a diagnosed health condition be 'biographically disruptive' to social roles and identities, but also the *risk* of it (Pranka, 2018). For Russell, this meant preparing for the possibility that Sophie *would* develop CF (and acting in the present to accommodate this outcome) all the while retaining hope that she would not.

4.2. Biographical repair

4.2.1. Making sense of new futures: identities, roles and risk

After receiving unexpected outcomes from CF NBS, participants found themselves needing to re-orientate to new realities and repair biographical ruptures. For those with CF and CFSPID, this meant entering regimes of treatment or medical surveillance. For those whose child was identified as a carrier, however, this meant adjusting to new health information without immediate relevance. Kayleigh, mother to Zoe (aged 13 weeks), explained that biographical repair involved a re-imagining of her daughter's relational and reproductive futures:

She doesn't have CF ... but we do know now that she has this thing, and that when she comes to have her own kids, that's when it comes out. If it's as common as they say [CF carrier status], it is possible she will meet another [carrier] and well ... she's going to have to be more careful than most about unplanned pregnancies! [laughs] ... She might need to tell any future partners because there's going to need to be blood tests done before having kids, or she might choose not to tell anyone at all! But it would probably be sensible, and I'd encourage that.

(Kayleigh, 22, CF carrier)

Kayleigh's account highlights how carrier status can, for some participants, come to be entirely conflated with reproductive risk; a health risk by-proxy. Whilst recent literature has challenged the idea that CF carrier status is an entirely benign health state (Miller et al., 2020), Kayleigh did not regard it as a primary health concern for her daughter. Rather, it was externalised as a health risk to *others*; a dormant risk, that 'comes out', or is catalysed at critical junctures in life, such as partnering/marriage, or reproduction. For Kayleigh, Zoe's status was associated with specific 'genetic responsibilities' (Hallowell, 1999) such as disclosure to future partners, and use pre-conception testing, highlighting how new forms of identity and personhood, and associated obligations, can emerge out of the disruption of unanticipated screening results.

Like Kayleigh, all participants across carrier/CF/CFSPID groups emphasised the familial nature of their child's result. Relevance to related family members created 'ripple effects' (White et al., 2021) across kinship groups, which could both re-affirm biological bonds and collective futures, as well as generate tensions and disassociations. Tamara described the collective nature of her family's response to her son Max's CF diagnosis in the following way:

I think it brought us together. As soon as we got the results, they [family] just rallied around Helen [sister] was found to be a carrier when she was pregnant, so it was all quite tense until Chris [brother-in-law] tested negative. We're a CF family now ... we belong to a lot of groups ... raise money, we all wear the CF awareness ribbons, we've all registered as organ donors it's something that affects the whole family and we became determined to educate people about it and change things, not just for Max, but for future generations.

(Tamara, 42, CF)

As Featherstone et al. (2006) highlight, genetic findings can both illuminate and emphasise ties of biological kinship. For Tamara and her family, responding to CF involved a unification around a genetic identity as a 'CF Family' and a re-calibration of both lateral and vertical genetic responsibilities to existing, and future, family members. For both Tamara and Kayleigh, therefore, biographical repair culminated in the creation of new collective and individual identities, roles, and concomitant responsibilities. Yet, for families living with the CFSPID designation, this process of identity re-calibration in light of NBS results was far more fraught.

4.2.2. Living in spaces of liminality: 'genetic nomadism'

Whilst parents of children with CF, or CF carrier status, perceived their NBS outcomes as unequivocal, parents of children assigned the CFSPID designation occupied a far more ambiguous position in relation to health, illness, and identity. Rachel, mother to Thea (aged 2), reflected that her daughter's CFSPID designation challenged her ideas about the status and role of medicine more broadly:

It [CFSPID] might be something ... or it might be something of nothing. They just can't tell you. ... I thought that medicine was more black and white than that- you either have something, or you don't, and even if it's mild, it's still one thing or the other, whereas now I feel like everything's a bit of a grey area. You can't 100% worry about it, or 100% ignore it, it's just the perpetual in-between.

(Rachel, 34, CFSPID)

Like Rachel, all of the CFSPID parents in the study reflected on the difficulties of living in a state of 'perpetual in-between'; a liminal state between health and illness, which they described as disrupting their very ideas of what illness is. Indeed, the identification of CFSPID as a 'designation' rather than a 'diagnosis' (Barben et al., 2020), also challenged the way in which participants understood medical language and process. They reflected on how this caused difficulties in communicating their child's status to others, especially when their child was being followed-up by a CF clinic, or was undergoing prophylactic treatment. Emma, mother to Jaxon (aged 2, CFSPID), resolved these difficulties by transposing the CFSPID designation onto commonly accepted notions of health and illness:

I find it really hard to explain to people, so we just call it a rare type of CF, which I suppose it is. I explain it doesn't cause Jaxon real difficulties as yet, but it means we have to be extra careful ... with any chest infections, to make sure it doesn't start up, and we have to have ... medication on hand to keep him well for when it does.

(Emma, 30, CFSPID)

By identifying CFSPID as a 'rare form of CF', Emma was able to circumnavigate the uncertainty surrounding Jaxon's health, and situate CFSPID within pre-existing cultural narratives of health and illness as dichotomous and mutually exclusive states. By assigning him a CF identity, she also clarified appropriate roles and behaviours, including keeping medication available to 'keep him well'. This strategy of 'making certainty out of uncertainty' was also found by Johnson et al. (2019), as a means by which to curtail ambiguity, and render CFSPID a tangible health status.

This means of navigating uncertainty, however, was not only evident amongst those who incorporated CFSPID into their child's identity, but also amongst those who rejected it. Beth, mother to Freya (aged 6), described her dismissal of CFSPID in the following way;

It's just a label really isn't it? It's not a diagnosis, she's a healthy child ... We come to the check-ups, but to be honest I'm not sure they're really needed, I think it's more they feel they need to do something. We've probably all got these little quirks in our genes, but in Freya's case, it's just that they've got a name for hers ... I certainly

don't ruminate on it. Perhaps I did worry more when they first told us, when she was tiny, but definitely not now. She's pretty robust!

(Beth, 39, CFSPID)

As previously identified by [Tluczek et al. \(2010\)](#), Beth's account demonstrates the significance of time and ongoing lived experience to her perception of Freya's 'robust' health. This experience was pivotal to her rejection of the CFSPID 'label', and consequently the resolution of the uncertainty and anxiety associated with it. Whilst Beth and Emma, therefore, came to polarised views of their child's status, as 'a healthy child' (Beth) and as having a 'rare form of CF' (Emma), they used similar strategies of uncertainty minimisation, replacing the CFSPID designation with more culturally resonant ideas of health or illness.

For other parents, however, identification with, or rejection of the CFSPID designation was a far less binary process, instead, it involved a constant movement in and out of health identities. Elena, mother to Leo (aged 4), described this in the following way:

... we don't really fit into either camp- either the CF world, or the healthy kid world. Other parents don't get it, and when I went onto the CF pages [on social media] I got a lot of ... quite rude messages from people with CF and parents too, saying ... things like 'what business have you got posting on here? There's sod all wrong with your kid'. Stuff like that so I was left with nowhere to go. We don't belong fully in either place ... we ... move from one to the other constantly, depending on what's going on we're kind of like nomads really! Genetic nomads! [laughs]

(Elena, 27, CFSPID)

Elena's concept of 'genetic nomadism' succinctly captures the oscillation parents experience as they move between the 'CF world' and the 'healthy kid world' at various points-yet with a fundamental sense of displacement from both. This identity dynamism across contexts was also reflected in the experiences of Hayley, mother to Sophie (aged 5, CFSPID):

You can forget about it most days, but when it comes to a check-up at the clinic, or you know, she gets chesty, then you're worrying about it in the weeks running up to an appointment [at CF clinic] I tune in to it more, start monitoring her for any signs, start seeing CF in everything, but once it's over and they're happy with her, we go back to normal ... until it starts all over again.

(Hayley, 38, CFSPID)

As Hayley identifies, there were cyclical time-points wherein Sophie's 'latent' risk of CF became 'manifest' ([Parsons and Atkinson, 1992](#)), and was brought into conspicuous awareness through clinical appointments, or the onset of illness. At other points, however, it receded into the background, allowing them to return (albeit temporarily) to 'normal'.

Whilst 'genetic nomadism', for these parents, was characterised by a sense of alienation from, and regular movement between both the 'healthy kid world' and the 'CF world', it is important to note that they were not the passive recipients of these shifts over time and context. Indeed, the clinical uncertainty surrounding their child's genetic status created spaces for parents to actively generate hope for the future (Russell) and neutralise the threat of ambiguous futures (Beth). Parents demonstrated that they could actively harness their child's hybrid genetic status strategically to meet particular ends. Juliana, for example, mother to William (aged 3), highlighted how recourse to the CFSPID designation enabled her to accelerate access to healthcare when needed;

I certainly don't make a fuss about it [CFSPID], but ... if he's got a chest infection starting and I'm worried, I will say to the doctors that he needs to be seen as quickly as possible becausehe's a CF

clinic patient, soit's useful to have that to fall back on when I need him to be seen quickly.

(Juliana, 38, CFSPID)

Other parents, both with the CFSPID designation and carrier status, also thought their child's lack of phenotype presentation would provide them with opportunities to 'pass' ([Goffman, 1969](#)), as Emma noted;

I did worry at first that Jaxon might feel ... that other people would view him negatively because of it [CFSPID] like there is something wrong with him ... but it will be up to him ... who he tells- if anyone We tell who we need to, but everything else is up to him, and with any luck it will be a complete irrelevance in his life.

(Emma, 38, CFSPID)

These accounts demonstrate the various ways that parents could employ their child's genetic nomadism strategically, by alternately laying claim to 'the healthy kid world' and/or the 'CF world' across contexts and to achieve particular goals, demonstrating the agentic properties of nomadism.

5. Discussion

By comparing and contrasting the accounts of parents who received a CF diagnosis, a carrier result or a CFSPID designation following NBS, this study charts the universally disruptive nature of positive NBS results. Whilst the notion of 'biographical disruption' has typically been used in the context of chronic illness, its relevance to the experiences of parents facing unexpected health crises in their children, necessitating the re-organisation of identities, roles and imagined futures, has been highlighted. This study demonstrates that, in the context of NBS, biographical disruption occurs both personally (for parents) and by-proxy (in relation to the child's expected life trajectory), and across the range of possible positive results-regardless of their immediate relevance. Through processes of 'biographical repair', positive NBS results triggered the development of new identities, social roles and responsibilities, for parents, the screened child, and the wider family. However, the format these routes of biographical repair took differed across type of positive result.

Parents of children with CF needed to rapidly reorient themselves to a 'new world' (Leona) that typically involved treatment, intervention and an altered sense of their child's future. The concept of different 'worlds', to account for the vast disparities of knowledge and experience that exist between families living with CF and those not, has previously been explored in relation to public attitudes to screening for CF ([McClaren et al., 2007](#)). In the context of diagnosis, this sense of 'crash landing' in a new, and unfamiliar world as parents adjust to having a 'sick child' (Leona), was an inherent part of the 'disruption' of NBS results. Their child's result, however, and sudden belonging to a previously unknown CF world, also brought with it rapid access to resources (both medical and social), and frequently triggered familial support and collectivism ([Heinsen et al., 2021](#)). As identified by Tamara, for many, having a child diagnosed with CF meant becoming a 'CF family', with implications for both immediate and extended biological family members ([Finkler, 2001](#)).

For participants with positive NBS results that had less certain, direct, or immediately relevant impacts on the child's life (CFSPID/carriers), biographical repair and biographical recalibration were more complex, and parents reacted in different ways to their child's altered future. Some sought to re-interpret their child's result as being more certain than it was, or outright rejected it. These responses can all be understood as biographically reconstructive activities, enabling parents to minimise uncertainty, and reconcile their child's result within pre-existing cultural frameworks that posit health and illness as mutually exclusive categories ([Johnson et al., 2019](#); [Chilibeck et al., 2011](#)).

Whilst the minimisation of uncertainty was an important aspect of

biographical repair, other participants acknowledged the significance of context in determining responses, and consequently the fluidity and ever-evolving process of biographical disruption and repair (Trusson et al., 2016; Locock et al., 2009). The notion of ‘genetic nomadism’, as a participant-derived concept, captures the various ways that responses to positive, but uncertain, NBS results are not fixed one-off events, but continually shift over time and place. Participants moved in, and out of, the ‘CF world’ and ‘the healthy kid world’ at key moments and sometimes at their own volition. This demonstrates how the world of health, and health identities, can still be accessible to those with equivocal NBS results, despite a preponderance of ‘proto-illness identities’ reported in the literature exploring liminal health statuses (Jauho, 2019).

6. Conclusions

This study demonstrates the utility of the concepts of ‘biographical disruption’, and subsequently ‘biographical reparation’, in understanding parental responses to positive NBS results—regardless of the result’s immediate relevance to the child’s health. It also demonstrates the key role that uncertainty plays in disrupting schemas of health and illness, which, in turn, impact the means and resources available for parents to conduct ‘biographical reparation’. Biographical reparation emerged from this study as a fluid, incomplete process, and one continually subject to revision in response to the many ‘ripple effects’ associated with positive NBS results over time (White et al., 2021).

Whilst the concept of liminality has been developed to account for the uncertain spaces between health and illness generated by equivocal health statuses, this research suggests that parents of children with uncertain NBS results did not necessarily dwell for extended periods in these tentative spaces as ‘patients-in-waiting’ (Timmermans and Buchbinder, 2010), ‘partial patients’ (Greaves, 2000) or the ‘pre-symptomatically ill’ (Heinsen et al., 2021). Rather, they frequently demonstrated dynamic and active reactions to uncertainty, whilst retaining socially sanctioned conceptual distinctions between the worlds of health and illness. The concept of ‘genetic nomadism’ is offered as a means by which to capture these experiences of oscillation and displacement, as well as to highlight its generative potentiality. Whilst movement between worlds could be triggered by external events or contexts, parents could also initiate transition. As such, genetic nomadism may be understood as a consequence of medical uncertainty and liminality, but as also providing the mechanism through which that uncertainty could be harnessed. By belonging to neither world, yet occupying both across time, participants were able to conduct biographical reparation in ways that retained socially validated distinctions between ‘health’ and ‘illness’ when needed—as well as enabling them to better navigate the daily realities of life with a child with an unclear genetic status.

6.1. Limitations and opportunities for future research

A relatively small sample was recruited, as is typical in qualitative studies, however, all participants were from white backgrounds (Table 1) meaning that ethnic minority groups were under-represented. This lack of ethnic diversity may be explained by the relatively high incidence of CF within white populations vis-à-vis other ethnic backgrounds (Rohlf s et al., 2011). The study would, however, have been strengthened by greater representation of the ethnic diversity of the UK.

Whilst attempts were made to recruit participants with a false positive result, these were ultimately unsuccessful. The lack of clinical follow up of these families, together with their typical non-engagement with CF support groups, rendered recruitment particularly challenging. Whilst studies have documented a degree of ‘residual risk’ amongst families receiving this type of result (Schmidt et al., 2012), others suggest that false positive results may not have the same long term sequelae as other types of positive result (Vernooij-van Langen et al., 2014). Nevertheless, future research might usefully explore the experiences of

these families and their negotiation of ‘disruption’ and uncertainty.

Recruitment also proved challenging for participants with children identified as carriers ($n = 3$). As carriers are not routinely followed up, and their reporting is minimised within CF NBS in the UK, carriers were under-represented in the study, and limited to those who very recently received their child’s result. Further research may usefully focus on the experiences of parents and carriers, as they negotiate the implications of NBS results over time.

Finally, as participants in this study were at various stages post-NBS result (4 weeks to eleven years (Table 1)), they had highly contrasting levels of experience of adjusting to, and living with, their child’s result. Whilst this might have introduced recall bias, it may also have strengthened the analysis by allowing a comparison across time, and in the context of ongoing experiential knowledge. The need for longitudinal data exploring adjustment to equivocal NBS results has been acknowledged, and research in this area still emerging (Kerruish, 2016). Research exploring the disclosure, communication and retention of equivocal results within families would also usefully contribute to an understanding of the reach and endurance of genetic nomadism within wider family networks over time.

Credit author statement

FB designed the study, secured funding, collected data, analysed data and prepared the manuscript. CC analysed the data and prepared the manuscript.

Data statement

Due to the ongoing nature of this project, the data are not yet deposited in a repository. Data available from lead author upon reasonable request.

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