



Neurofibromatosis type 1 (NF1): Exploring parent's experiences of using a customisable information sharing resource (The NF File) using thematic analysis

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ABSTRACT

Research has revealed the importance of psychoeducational intervention directed at addressing needs regarding NF1. The aim of the current study was to explore the effectiveness of the use of a customisable information sharing resource (The NF File) in parents caring for children diagnosed with NF1 using thematic analysis. It explored: the effectiveness of the NF File in aiding parents and others understanding of NF1; the effectiveness of the NF File in the management of NF1; the ways in which parents used the NF File to support the management of their children's condition; and lastly, the improvements that can be made on the effectiveness of the NF File. Four mothers of children diagnosed with NF1, who had previously received the piloted NF File, took part in the semi-structured phone interviews. Thematic analysis approach was employed. Analysis of the accounts indicated three main themes: (1) uncertainty over the progression of NF1, (2) the importance of support, (3) the need for people to be educated about NF1. Findings revealed the effectiveness of the NF File in the management and aiding the understanding of NF1. Future research is needed to further investigate how the NF File can be advanced to provide families and diagnosed individuals with the care and support needed.

KEY WORDS:	NEUROFIBROMATOSIS TYPE 1	THE NF FILE	QUALITATIVE	THEMATIC ANALYSIS	THE NEED FOR INFORMATION
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Background

What is NF1?

NF1 is a variable genetic condition affecting 1 in 2500 to 1 in 3000 individuals (Ferner et al., 2007), regardless of gender, race, and ethnicity (Hirbe and Gutmann, 2014). The condition leads to observable changes in an individual's appearance (Barke et al., 2016) that are progressive and unpredictable (Saltik and Basgul, 2013). NF1 occurs because of a genetic mutation in the NF1 tumour suppressor gene (Gutmann et al., 2012) that causes the growth of benign and/ or malignant tumours on nerve endings (Barke et al., 2016; Graf, 2010). It is characterised by café-au-lait macules, which are often the first and most common signs of NF1 (Boyd et al., 2009). However, not all café-au-lait macules are NF1 related because of this a genetic test can be used to aid diagnosis (Boyd et al., 2009). Arguably, even though genetic testing is useful for diagnostic confirmation, a positive result cannot predict disease severity (Boyd et al., 2009). Nonetheless, skin-folding freckling, cutaneous neurofibromas (Saltik and Basgul, 2013), and plexiform neurofibromas found in at least 25% of diagnosed individuals (Huson, 1989) are also common features of NF1. Deformities caused by plexiform neurofibromas (Packer and Rosser, 2002) and central nervous system tumours (Gutmann et al., 2002) can also be observed in diagnosed individuals.

Due to the high occurrence of learning and behavioural problems seen in about 30-65% of individuals (Cutting et al., 2004; Draucker et al., 2017), monitoring is important especially in young children (Ferner et al., 2004). However, in some studies, because of the referral of more severely affected cases in a low sample size, the reported incidence of complications may be an overestimation (Huson et al., 1989). Arguably, owing to the unpredictable nature of the condition, NF1 patients are recommended to seek review of any development of unusual symptoms (Ferner et al., 2007). Not seeking medical consultation may lead to an increase in NF1-related complications (Oates et al., 2013).

Impacts of NF1 on Affected Individuals

According to Hummelvoll and Antonsen (2013), appearance concerns have been recognised as a key factor of some patient's experiences of NF1. In some studies, (e.g., Gaff et al., 2007; Krab et al., 2009; Wolkenstein et al., 2009), appearance changes have been found to have a negative impact on quality of life and psychological adjustment. For instance, findings have revealed that some patients find the appearance of café-au-lait macules and skin-folding freckling to be distressing (Ferner et al., 2007). Likewise, disfigurement caused by neurofibromas has been found to result in psychological problems in affected individuals (Ferner et al., 2007). Symptoms of anxiety and depression are also commonly reported (Ferner et al., 1996; Cnossen et al., 1998). However, these symptoms usually respond well to a combination of antidepressant and counselling (Ferner et al., 2007). Nonetheless, lowered social skills (Martin et al., 2012), difficulties in processing social information (Huijbregts et al., 2010), and relationship problems with friends have been recognised in young people diagnosed with NF1 (Barton and North, 2007, 2004; Rumsey and Harcourt, 2012; Noll et al., 2007). Thus, there is a need for the development of interventions to aid the processing, understanding, and recognising of social signals for young people to function competently at a social level (Bechara et al., 2000; Beer et al., 2004).

Saltik and Basgul (2013) conducted a systematic review study that aimed to investigate health-related quality of life (HRQoL) in children with NF1. The study made use of the Pediatric Quality of Life Inventory (PedsQoL) Parent Form and PedsQoL scores. These were completed by mothers of children with a diagnosis of NF1 (patient group) and mothers of children with no diagnosis of a chronic disease (control group). Findings revealed that PedsQoL domain scores of the patient group were lower than that of the control group, indicating that HRQoL of the patient group was negatively affected by NF1. These findings also corresponded with earlier studies carried out by Wolkenstein et al. (2009) and Vranceanu et al. (2015) which revealed that NF1 complications, such as plexiform neurofibromas and learning disabilities, affected the quality of life (QoL). In that event, it is recommended for multidisciplinary evaluation and psychiatric management of NF1 to be implemented for the improvement of HRQoL in patients diagnosed with NF1 (Saltik and Basgul, 2013).

HRQoL is a multidimensional construct that explores the way health, illness, and treatment affects physical, mental, emotional, and social functioning (Varni and Limbers, 2009). Accordingly, allowing for the effects NF1 has on patients to be understood (Varni et al., 2005). Parent's level of education, familiarity with NF1 and good family relationships are associated with higher HRQoL (Graf et al., 2016; Oostenbrink et al., 2007). However, it must be noted that there are no valid and reliable QoL scales developed specifically for children with NF1 (Saltik and Basgul, 2013). This may have prevented the study from determining valid and reliable factors that affected HRQoL in the NF1 patients (Saltik and Basgul, 2013). This is important because the development of valid and reliable QoL scales targeted specifically for NF1 patients may enable a better understanding of the correlation between certain factors and their effects on HRQoL. Thus, allowing a more targeted intervention to be established to help increase HRQoL in children diagnosed with NF1. All the same, it has been proposed for the use of HRQoL in clinical practice settings to inform treatments (Varni et al., 2015). Reasons being: physicians have reported its usefulness; it has been shown to improve patient's satisfaction; and lastly, patients have felt the instruments to be helpful (Varni et al., 2005).

The Need for Information

Parents are faced with challenges due to the adversity faced by youth with NF1 (Martin et al., 2014, 2016). Caregivers of chronically ill youth are at greater risk of developing depression, anxiety, and loneliness (Kreutzer et al., 2009; Wood et al., 2008). Perceptions of limited social support have been associated with higher levels of distress in mothers of NF1 patients (Reiter-Purtill et al., 2008). Additionally, the stress of supporting a child with special healthcare needs compromises parents' psychological functioning (Martin et al., 2016). Furthermore, social isolation, withdrawal, and challenges in social roles are commonly reported amongst these parental groups (Klassen et al., 2011; Kratz et al., 2009; Rodrigues and King, 2009; Draucker et al., 2017). It is, therefore, important for parents to be provided with the social support needed for a better management of their child's disorder and improved physical, mental, and emotional health (Janicke et al., 2009; Wang et al., 2013; Draucker et al., 2017; Rearick et al., 2011).

In a qualitative study carried out by Barke et al. (2014) involving nine young people diagnosed with NF1, the lack of awareness and understanding about NF1 in society

was brought to light. Living with the ambiguity of the condition and managing people's reactions to their appearance was also a key issue that arose in the study. Draucker et al. (2017) also discovered that worries about the uncertainty of the progression of the disorder and its effect on children as they age could be a significant issue. Moreover, Barke et al. (2016) conducted a study aimed at examining parents' experiences of caring for a young person with NF1. Parents expressed that the understanding of NF1 is imperative in supporting their children. Parents also felt the understanding of NF1 in professional and non-professional settings to be a compelling challenge. On the contrary, it must be noted that the perception of what it is like to live with NF1 differs between parents and young people. Research has found that Parents take a more pessimistic approach on measures such as social inclusion in comparison to young people (Wolkenstein et al., 2001; Oostenbrink et al., 2007; Garwood et al., 2012; Krab et al., 2009). Thus, showing the difficulties for parents to gain insight into the full effect NF1 has on their children (Ruth et al., 2004).

Nevertheless, in line with both Van Lierde et al. (2013) and Ferner et al. (2007) the management of NF1 is dependent on patient's education in self-management and lifelong age-specific monitoring. Both parents and diagnosed individuals need trustworthy information to inform their knowledge about the condition (Barke et al., 2016). Most individuals tend to turn to the internet to construct their understanding of the disorder (Crushing et al., 2015). However, internet searches, especially for NF1 images, commonly reveal pictures that do not accurately represent the disorder for most individuals (Cushing et al., 2015). It has been found that the viewing of online images has caused individuals to feel negatively about the condition, and to express feelings of worry and heartbreak (Crushing et al., 2015). Hence, as supported by previous findings, practical information is highly valued to help individuals manage their conditions, in particular people's reaction to their appearance (Thompson and Broom, 2009; Williamson et al., 2010). Reason being that, misinformation from unreliable internet sources can delay care (Johnson, 2014). This can cause patients to become sceptical of what they learn from their health care professionals (Johnson, 2014). Leading to healthcare providers having difficulties managing parents' fear in relation to the disorder (Van Lierde et al., 2013).

As a result, and as highlighted by Barke et al. (2016, 2014), there is a need for a psychoeducational intervention directed at addressing parents, health professionals, and general publics' information needs regarding the condition. The present study, therefore, aimed to explore the effectiveness of the use of a customisable information sharing resource (The NF File) in parents caring for children diagnosed with NF1 using thematic analysis. It explored: the effectiveness of the NF File in aiding parents and others understanding of NF1; the effectiveness of the NF File in the management of NF1; the ways in which parents used the NF File to support the management of their children's condition; and lastly, the improvements that can be made on the effectiveness of the NF File.

Research Questions

The study tried to answer the following questions:

1. How are parents using the NF File to support the management of their children's condition?

2. What are the experiences of parents using the NF File for their children in regards to its effectiveness in aiding the understanding of NF1?
3. What are the experiences of parents using the NF File for their children in regards to its effectiveness in managing NF1?
4. What are the ways in which the effectiveness of the NF File can be improved for children's use?

Methodology

Design

A qualitative approach was employed to gain rich in-depth accounts of participants' experiences (Rich and Ginsburg, 1999) of using the NF File. It allowed for meanings behind individual's decisions and their lived experiences (Willig and Stainton-Rogers, 2008) to be investigated. It enabled a close researcher-subject relationship permitting the researcher to attain first-hand experience that provided valuable meaningful data (Carr, 1994; Creswell, 2003). The researcher-subject relationship was vital for a genuine understanding of problems faced by parents caring for sick or handicapped children (Baruch, 1981).

A weakness of this, however, is the likelihood of researchers becoming entangled with participants leading to the difficulty of the separation of researcher's own experiences from those of their subjects (Sandelowski, 1986). Resulting in subjectivity (Cormack, 1991) and distortion of research findings (Carr, 1994). Arguably, the complete removal of researcher's influence is impossible (Hammersley and Atkinson, 1995). The present study, as suggested by Bickman and Rog (2009), therefore, aimed to understand the influence of the researcher in the study through a reflexivity account of the researcher's experiences, to gain a broader insight into the study's findings.

Participants

Four mothers of children diagnosed with NF1 were recruited through the Childhood Tumour Trust (CTT). A small selective sample was utilised due to the in-depth nature of qualitative studies and analysis of the data required (Cornack, 1991). Participants were required to be: 18 years of age and above; a parent of a child/children diagnosed with NF1; living in the UK; and have previously received a copy of the pilot NF File. Three participants with pseudonyms Patricia, Emily, and Jessica, had no family history of NF1 or prior knowledge of the disorder before the diagnosis of their children. The NF File, at the time of the interview, was being used by Emily and the fourth participant Rebecca for two months. Jessica used the NF File for three months. Patricia had the NF File for two months but did not use it. Participants were invited, via email, to take part in the semi-structured phone interview. Interviews took place over the phone for the comfortability of participants during the interview process (Dimond, 2015). This enabled interviewee to fully cooperate in the interview process for the obtainment of a rich account of their experiences.

Research process

An Invitation letter and the study's information sheet was emailed to twenty-five potential participants, four of whom agreed to participate. Potential participants contacted the researcher by email and interview arrangements were made. All

interviews took place at a time favourable for participants to strengthen the quality of data provided (Burke and Miller, 2001). Full informed consent was gained from each participant before the commencement of the interviews. During each interview, participants were informed of the aim of the study and the estimation of the duration of the interview. The estimate was inferred from a pilot study conducted that aided in the identification of question areas that needed clarification for the improvement of data integrity (Burke and Miller, 2001). Open-ended questions were asked to provide rich elaborated data (Burke and Miller, 2001). Each interview lasted approximately thirty minutes and was recorded using an audio recording device. All interviews were transcribed and analysed using thematic analysis.

Data Collection

A semi-structured telephone interview; a verbal interchange where the interviewer attempts to produce information from another person through questions (Longhurst, 2003), was conducted. The semi-structured interview aimed to understand the experiences of participants using the NF File and the meaning participants attached to those experiences (Seidman, 2013). It allowed for participants' responses to govern the information produced about the explored topics and their significance (Green and Thorogood, 2014), leading to the enrichment of the quality of data gathered (Carr, 1994). The telephone interview provided an effective means of data collection (Harvey, 1988) from participants that were locally unavailable and hard to reach on a face-to-face basis due to childcare commitments (Novick, 2008).

The interview utilised an interview schedule that acted as a guide and question prompt during the interview (see Appendix 7: Interview Schedule). The interview schedule was created through a thorough review of literature exploring parent's experiences of caring for young people with NF1, and literature around creating awareness and understanding of NF1. Questions were developed to explore participant's knowledge about NF1, participant's subjective experiences of using the NF File, how participants maintained the use of the file and ways in which the NF File can be developed further. In addition, the questions also focused on understanding the underlying rationality of participant's behaviour, such as their reasoning to either use or not to use the NF File. This allowed the researcher to learn and understand the thought processes of participants from their viewpoint (Green and Thorogood, 2014).

Data Analysis

NF1 is a highly variable condition (Barke et al., 2016). Participants, as well as their children, could be expected to have a diverse experience of the condition (Barke et al., 2016). Hence, thematic analysis (TA), a method for identifying and analysing meanings in a text (Gavin, 2008), and reporting themes in data (Braun and Clarke, 2006) was employed. It captured and maintained the richness of data, and allowed for the analysis of common areas (Barke et al., 2016). TA highlighted the experiences and reality of participants through the generation of themes (Braun and Clarke, 2006). As TA does not require a comprehensive knowledge of approaches, it offered a more accessible form of analysis (Braun and Clarke, 2006).

A realist approach was taken to document the experiences, meanings and realities of participants (Braun and Clarke, 2006) in relations to the NF File. The process involved the transcription of data collected from the interviews to generate initial

codes. Codes were gathered, collated and developed into well-defined themes (DeSantis and Ugarriza, 2000), achieved through the reading and re-reading of data for familiarity (Rice and Ezzy, 1999). The themes were derived both inductively (Hsieh and Shannon, 2005) and deductively (Braun and Clarke, 2006). The inductive approach was applied due to the researcher's analytical interest in understanding the effectiveness of the NF File in managing and aiding the understanding of NF1. Meanwhile, the utilisation of a deductive approach allowed for themes to directly emerged from the data (Patton, 1990). Additionally, themes were identified at a semantic level (Boyatzis, 1998) allowing for the significance of their patterns, meanings and implications to be theorised in relation to previous literature (Patton, 1990). To ensure rigour of analysis, all themes and interpretations were verified. This was achieved through the reviewing and discussion by another researcher as suggested by Barke et al. (2014).

Ethics

The study obtained ethical approval from Manchester Metropolitan University (Appendix 1: Application for Ethics Approval Form) and adhered to the British Psychology Society guidelines (BPS, 2009). Participants received an invitation email (appendix 4: Invitation email), consent form (Appendix 2: Consent form), and an information sheet (Appendix 3: Participant Information Sheet). Participants were briefed and debriefed. They were made aware of the study content and their rights to withdraw from the study at any given point. Before consent was given, participants were told about the recording and transcription of the interview. They were informed that anonymity will be granted through pseudonyms. Data collected was stored on a password-protected computer and was destroyed after the completion of analysis.

Analysis and Discussion

Three themes were identified from the analysis: 'uncertainty over the progression of NF1', 'the importance of support', and 'the need for people to be educated about NF1'. These themes are reflective of all interviews, but for illustration purposes selected quotes are used.

Uncertainty over the progression of NF1

This refers to the difficulties the uncertainty over the progression of NF1 posed to all participants. This was reflected in the concerns participants held about the future of their children. Thus, backing up studies that have also found similar findings (e.g., Barke et al., 2016; Draucker et al., 2017; Barke et al., 2014).

...it's an uncertain future, isn't it? Because you don't know the variable, you don't know how your child is going to be [or] what the future holds for them (Transcript 3, Patricia, Lines 39-41).

just because it's so unpredictable obviously, you've got the concern of what's going to happen in the future (Transcript 2, Rebecca, Lines 35-36).

The importance of the provision of informative resources to help ease feelings of ambiguity over the future of their children was a prominent matter throughout the interviews. Participants found that with the aid of the NF File, they would be able to '...track the progression of NF[1] on [their] child[ren]...' (Transcript 1, Emily, Lines

203-204). Consequently, helping them observe the ways in which the disorder has affected their children over time.

...if she's got the freckling or she's got a lump or something like that, you can take a picture of it and then that would be a reference point then. So then if you want to go back, say come back in six months, you'd already have a picture at that point and you could take another picture and add them up and then see how things change...you'd be able to monitor, you know, things as well... (Transcript 4, Jessica, Lines 203-209).

...I could've documented everything. I think it would've been helpful for me to look back on, it would've been helpful for doctors to have seen erm things and it would've been really helpful to monitor everything... (Transcript 3, Patricia, Lines 87-79).

...it's good to be able to keep records for ourselves and just to see how he's progressing... (Transcript 1, Emily, Lines 160-161).

Through a process of lifelong age-specific monitoring and self-management (Van Lierde et al., 2013; Ferner et al., 2007), the scepticism or concerns felt about the progression of the disorder can be minimised. It was expressed that being able to monitor the progress of the child can act as a reminder of '...what [the disorder] is and how [the child is] dealing with [it]...' (Transcript 1, Emily, Line 181). The NF File, therefore, acted as a '...great essential document...' (Transcript 1, Emily, Line 183). Furthermore, as the monitoring process can permit parents to examine the support system and treatments that are working for their children, with every accomplishment and milestones reached, the feelings of uncertainty can be alleviated.

...cause if it's affecting him now and we go in for an assessment in six months' time and it's no longer affecting him, we can actually remind ourselves that he's actually taken a jump forward there and we've done good in that area (Transcript 1, Emily, Lines 161-164).

These accounts show the way in which parents used the NF File to help ease the concerns they felt about the progression of the disorder. It revealed how the NF File was used to effectively monitor and manage the disorder. The positivity around the file shed light on the value of monitoring resources in supporting families. Nonetheless, despite the benefits of having a monitoring resource to help oversee the progression of NF1, participants also felt that '...it would be more helpful to have [the unpredictability of the disorder] explained...' (Transcript 4, Jessica, Line 44). Moreover, as NF1 is an unpredictable disorder, it must be acknowledged that there is no single best approach to support NF1 patients (Barke et al., 2014) or remove all uncertainty, but the focus should be on supporting and providing the best possible treatments (Rubenstein and Korf, 1990).

The importance of support

This refers to the lack of or limited support participants felt they received when their child was first diagnosed with NF1. This backs up the findings of Martin et al. (2016). The perceived lack of support stemmed from the anxiety of being a '...parent with a newly diagnosed child...' (Transcript 3, Patricia, Line 81), which led participants to feel neglected, 'scared', and 'afraid'.

...we were given very limited erm details from [x] genetics... we were just told to go away and not really told anything at all about it... (Transcript 4, Jessica, Lines 29-33).

...quite upset[ing] because you're told that your child's got, you know, a condition that may affect them greatly in their life but may not, but not to an extent or not really where to go for help in between, or what sort of other things can exist with it...so really just quite scared and alone and afraid really (Transcript 4, Jessica, Lines 36-39).

These accounts corresponded with findings revealing that caregivers of chronically ill children are at risk of anxiety and loneliness (Kreutzer et al., 2009; Wood et al., 2008). As a result, it is imperative to provide support to minimise the levels of distress and stress of caring for a child with special health care needs (Reiter-Purtill et al., 2009; Martin et al., 2016). This matters as participants highlighted that they are left to manage their children's disorder.

...There was very little communication between different doctors and hospitals and you end up having to manage all that, and you end up chasing up on your genetic appointment... when your last eye test is [etc]... (Transcript 3, Patricia, Lines 162-164).

Nonetheless, it was believed that the difficulties of parents receiving optimum support could be due to the lack of '...continuity of working with at least one health professional' (Transcript 3, Patricia, Line 52). In addition, medical staff's assumptions of parents being knowledgeable in all aspects of the disorder was also a contributing factor.

...I think sometimes the medical staff forget that you don't necessarily know what all the things are that they're going on about (Transcript 4, Jessica, Lines 249-251).

As young children are not able to fully express the extent to which NF1 affects them, they need to be provided with relevant emotional and physical support (Draucker et al., 2017). This will, in turn, reduce the emotional distress felt amongst families.

...he can't actually tell us and he can't spell how he feels about that, but we can see, you know, it affects him physically, it affects him mentally and hopefully not emotionally... (Transcript 1, Emily, Lines 158-160).

The need for the early provision of support in the management of NF1 was highly valued by participants, as predicted by previous studies (Janicke et al., 2009; Wand et al., 2013; Draucker et al., 2017; Rearick et al., 2011).

...if I had this [the NF File] from the point of diagnosis I would've used it right from the beginning and I think it would've been really helpful... I mentioned before about health professionals, not sort of knowing what one hospital was doing... I think I would've have used it from that point of view (Transcript 3, Patricia, Lines 61-65).

...if you get it [the NF File] at the beginning when your child was starting off or just being diagnosed, I think it would be extremely useful because then you'd

be able to do it as you're going along... (Transcript 4, Jessica, Lines 105-107).

Participants expressed that being able to get the help needed for their children would make them feel '...a bit more empowered...' (Transcript 4, Jessica, Lines 269-270) and self-confident. The NF File acted as a form of '...reassurance that [they were] not overreacting to things...' (Transcript 1, Emily, Line 217). Thus, showing the beneficial purpose of families to receive support for better management of NF1 and improved physical, mental and emotional health (Janicke et al., 2009; Wang et al., 2013).

The need for people to be educated about NF1

This refers to the lack of understanding of NF1 from both healthcare and non-healthcare professionals, and how this brought about hardships. Throughout the interviews, the significance of professionals being educated about NF1 was highly prevalent. The '...lack of knowledge from certain medical professionals and...lack of... [team]work in between hospitals' (Transcript 3, Patricia Lines 43-44) brought about 'challenges'. Participants were unable to recognise symptoms that could potentially be harmful or cause distress to their children. Jessica felt that tumours could be growing internally inside of her daughter and she would be unaware as they '...weren't really told what sort of things to look out for that may cause problems...' (Transcript 4, Lines 47-48).

...the midwife had noticed that [x] had a stridor...which we thought he was just a noisy sleeper at the time, but he had the symptoms of very weak muscle tone which erm was NF and it was at that point that we realised that it was something concerning going on there... (Transcript 1, Emily, Lines 51-55).

This supports the claim that NF1 is a poorly understood disorder and the lack of understanding proposes difficulties (Barke et al., 2014, 2016). Hence, highlighting the significance of the need for people to be educated about NF1.

Patricia expressed that she found it '...surprising how many consultants you go to who don't know what NF[1] is...' (Transcript 3, Line 84) and that '...a lot of people still in this day and age have not heard of it' (Transcript 3, Lines 173-174). Supporting this, Emily also believed her son is the only child with NF1 seen by their paediatrician and as a result, the paediatrician's knowledge was '...very limited...' (Transcript 1, Line, 89-90). She also voiced out that she was not '...given enough information from the nurse[s]...' (Transcript 1, Line 29) at the time of her son's diagnosis. These accounts demonstrate the need for people to be educated about NF1 in order to avoid the feeling of frustration or scepticism in regards to health professionals not being able to understand the disorder (Barke et al., 2016).

Arguably, one participant relayed that being given a detailed informative resource, such as the NF File, at the beginning of the diagnosis of their children '...would've [been] quite distressing...' (Transcript 3, Patricia, Line117). Despite this, Patricia also indicated that learning difficulties associated with NF1 were not properly understood by schools (Barke et al., 2016) and thus, the NF File would be highly valuable in that respect.

...[x] looks normal, but [the school] didn't quite understand the level of difficulties that he can experience... [the file] would...make them realise the difficulties and the worries that encompass the condition (Transcript 3, Patricia, Lines 120-122).

This account corresponded with the findings of Draucker et al. (2017) indicating that people are unable to fully perceive the seriousness of NF1 and are often unaware of the learning difficulties that can accompany the disorder.

The lack of detailed information from medical professionals led participants to rely on the internet to construct their understanding of the disorder, as found by (Crushing et al., 2015).

Erm, just the usual way, google. Erm, so you get basic limited information given to you and then you just google everything on the internet (Transcript 3, Patricia, Lines 27-28).

This reliance generated more fear instead of educative and supportive information. Internet searches mostly focused on 'worst-case scenarios' making the diagnosis 'daunting' and 'overwhelming'. As proposed by Crushing et al. (2015), internet searches do not accurately represent the disorder and it causes individuals to feel negatively about the disorder.

There were many difficulties faced with the use of the internet to generate information about the disorder. For instance, Rebecca found that internet searches contained '...a lot of jargons [and were] not very understandable...' (Transcript 2, Lines 30-31). Emily, also, expressed that because the diagnosis of her son was rare '...it was very difficult to track down information...' (Transcript 1, Line 47). In addition, the authenticity of the information provided was difficult to established. According to Jessica, '...you don't really know if [the information] are correct...' (Transcript 4, Lines 182-183). Emily demonstrated mix feelings about the reliance of information provided on the internet. She found that information are '...reliable but [at the same time] overwhelming and not always positive' (Transcript 1, Lines 34-35).

Contrarily, not all internet sources were found to be unreliable. Both Patricia and Rebecca found the Neuro Foundation to be 'very helpful'. As found by (Barke et al., 2014), the Neuro Foundation was an important internet source in gathering relevant and reliable information. Nonetheless, there is still a high demand for the provision of educative information to aid the understanding and management of the disorder.

...when you go for medical appointments, you do get told all these medical things that you have... no idea what they are going on about sometimes... instead of just googling it and then coming up with some really random facts, you can just look on there [the NF File] and then just seeing that it's not anything or whatever... so things like that, think would just alleviate some worries... (Transcript 4, Jessica, Lines 229-238).

...for me I (inaudible) google researched all of my information on NF and it can be quite overwhelming, just, you know, having a document that bullet points, you know, key facts, key symptoms erm...it's easier to understand, it's easier erm look at those particular symptoms. If I want to go back to google I

can google those symptoms rather than look, you know, for multiple symptoms (Transcript 1, Emily, Lines 148-153).

Participant appreciated that the NF File was validated by an NF specialist, it gave the file an ‘...authenticity feel...’ (Emily, Line179) which encouraged them to customise it. This evidenced for the value of trustworthy information in informing knowledge, as claimed by (Barke et al., 2016). Participants were able to use the NF File to inform and educate professionals and non-professionals about the disorder and its effect on their child.

...we can put what things they’re good at and what things that they struggle with. And then, when we go into like schools, and they say “oh what things are they struggling with?”...you’ve got it all there already... (Transcript 4, Jessica, Lines 94-97).

...It was good to have something written down to tell other people about it (Transcript 2, Rebecca, Line 99).

...when you’re speaking to, you know your community paediatrician or your surgeon’s consultant, it’s very good that they can actually see a plan, the symptoms or NF so we can tackle erm exactly what’s wrong (?) with [x]... (Transcript 1, Emily, Lines 133-135).

The accounts given by the participants provided a detailed reason as to why it is imperative to educate families, professionals, and non-professionals about NF1. It showed how the NF File can effectively be used to educate others about the various symptoms of the disorder and the impact it can have on diagnosed children. This supports the importance of practical information to help individuals manage their condition (Thompson and Broom, 2009; Williamson et al., 2010).

Analysis of transcripts revealed how parents used the NF File to support the management of their children’s condition and how the NF File was used to aid the understanding of NF1. It captured the importance of families, health professionals and non-health professionals to be aware and educated about NF1 in order to provide the support needed by the diagnosed children. It shows how the perceived lack of understanding of NF1 can bring about challenges in accessing social support, as found by Barke et al. (2016). Being given a customisable file that provides detailed information about NF1 that can be used to educate others, especially health professionals, about the disorder, was highly desired by all participants. Participants suggested having the NF File on a more centralised system, as opposed to having a paper copy, can be a more effective approach to managing the disorder. It was also believed that support and educative information should be given once a diagnosis has been made for children to receive the best possible care.

The current research focused on a small sample of participants which provided in-depth knowledge on the experiences and the way participants used the NF File to manage and educate others about NF1. Participants talked positively about the file and the way in which it had enabled them to keep organised and support their children. However, it must be taken into consideration that the current study’s research findings were not reflective of individuals who did not: customise or use the NF File, find it effective in managing or aiding their understanding of NF1, and use the file to inform others or increase awareness of NF1. Moreover, as the NF File is a

new development, data collected represented the usage of the NF File for a short-term period, mostly two months. Therefore, the usage of the file on a longer-term outcome has not yet been established. For this reason, additional research is needed into the long-term benefits of the NF File to validate the overall effectiveness of the file and to assure the consistency of the file in aiding and managing NF1.

Future research is needed to further investigate how the NF File can be advanced to provide families and diagnosed individuals with the care and support needed. Ways in which the NF File can be incorporated into healthcare setting to aid the education of healthcare professionals about NF1 needs to also be explored. Future research would benefit from a larger sample size and more varied sample involving participants that: did not use the file, stopped using the file or did not find the NF file an effective method of managing the disorder. This is to explore and gain an understanding into the reasons why the NF File was either beneficial or not beneficial to people, and how it can then be further improved to reach out to a wide range of individuals.

Reflexive analysis

As an intern research assistant at Manchester Metropolitan University, I worked, alongside my research supervisor, with individuals diagnosed with NF1 and their families. I explored how the NF File can be used to provide support and information to them. This prompted me to investigate the effectiveness of the NF File in managing and aiding the understanding of NF1. I wanted to learn more about the experiences of caring for individuals with a chronic disorder and the difficulties lack of resourceful information posed in the management of the disorder.

I was expecting positive feedbacks in regards to the file and its effectiveness. This was mainly due to my awareness of the need for a psychoeducational intervention amongst these parents. These expectations were met from the outcome of the research. Nevertheless, I was quite surprised about the number of participants that took part in the study. I originally planned to interview six participants but only managed to interview four. The process of recruiting participants turned out to be more difficult than I imagined. If I had interviewed more than four participants I would have been able to gather a varied number of insights. Despite this, I was still able to get in-depth insights into the experiences of these families, how the lack of information and limited support affected them, and the benefit of the NF File.

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