

'It's like a bag of pick and mix – you don't know what you are going to get': young people's experience of neurofibromatosis Type 1

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**'It's like a bag of pick and mix - you don't know what you are going to get':
Young peoples' experience of Neurofibromatosis Type 1 (NF1)**

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Conflict of interests

No conflict of interest has been declared by the authors

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Abstract

Aims - To explore the day-to-day experience of young people living with NF1 in the United Kingdom

Background - Neurofibromatosis type 1 (NF1) is a genetic condition which is highly variable and unpredictable. The condition can result in varying degrees of visible difference (disfigurement) which often manifest during puberty; a time during which appearance concerns are often heightened. In addition to the condition's actual affect on appearance, the uncertainty of NF1 may be particularly difficult to manage. However, very little research to date has investigated the psychosocial impact of NF1 during adolescence.

Design - Exploratory qualitative interview study

Methods - Nine people aged 14-24, with a diagnosis of NF1, took part in in-depth semi-structured interviews between March and September 2011. Interview transcripts were thematically analysed.

Findings - Three key themes emerged from the data. (1) 'Different things to different people' reflecting the variability of the condition. (2) 'Relationships and reactions' relating to individuals' social and interpersonal experience, and (3) 'Understanding and misunderstanding' reflecting people's experience with organisations and social structures.

Conclusions - Findings suggest a need for further research to explore young people's adaptation and management of NF1 during adolescence. In particular, raising awareness and understanding of NF1 among professionals and within the general public was an important issue for adolescents. Additionally, access to trustworthy information about NF1 and practical advice to support adjustment to an altered appearance and managing stigma experiences are highlighted as areas to be considered further.

Summary Statement

Why is this research needed?

- Neurofibromatosis type1 significantly impacts on psychological wellbeing and quality of life.
- The psychological impact of NF1 may stem from managing both the unpredictability of the condition and changes to appearance.
- Adolescence is a period of increased appearance salience and a time when health behaviours are consolidated, as such young people with NF1 may be particularly vulnerable to the psychological impact of NF1.

What are the key findings?

- NF1 becomes a more prominent part of many peoples' lives during adolescence
- Particular challenges that young people with NF1 describe include managing other people's reactions to their appearance, living with the uncertainty of the condition and the general lack of awareness of NF1.

How should the findings be used to influence practice and/or policy?

- Evidence based interventions supporting appearance concerns and social skills would be beneficial for young people with NF1.
- Reliable information and social support that is designed specifically for young people in relation to NF1 would be highly valued by young people.
- Raising awareness and understanding of NF1 amongst health professionals may be highly supportive of young peoples' overall day-to-day experience of NF1.

Keywords

Neurofibromatosis type 1, Disfigurement, Appearance, Qualitative, NF1, Nursing, Genetic Counselling, Adolescence, Young Adult, Young People

Introduction

Neurofibromatosis type 1 (NF1) is a highly variable and unpredictable genetic condition with an incidence rate of 1: 2500 - 1:3000 (Ferner et al 2007). Diagnosis is based on clinical assessment and visible signs of the condition include café au lait spots, neurofibromas and plexiform neurofibromas.

Research demonstrates that NF1 can have a significant impact on quality of life and psychological adjustment (Mouridsen & Sorensen 1995; Zoller & Rembeck 1999; Samuelsson & Riccardi, 1989, Graf et al 2006; Krab et al 2009; Noll et al 2007). Adolescence may be a particularly challenging time for people with NF1, since this is often a time during which appearance concerns are heightened (Rumsey & Harcourt, 2012), yet this is when neurofibromas typically start to develop. Furthermore, young people with NF1 may have poor social skills, difficulties processing social information and forming friendships (Benjamin et al, 1993; Huijbregts et al 2010) all skills which have been identified as important in mediating the effects of an altered appearance (Rumsey & Harcourt, 2012). The unpredictability of the appearance changes associated with NF1, and a body of literature that has shown that the extent or severity of a visible difference is not associated with levels of distress, (Moss, 2005, Ong et al 2007) suggest that appearance concerns are as much an issue for people with no, or few, physical signs of NF1 as they are for those with more noticeable changes.

Commonly reported challenges for people living with a visible difference of any kind, include difficulties forming relationships, negative reactions from others and discrimination (Thompson & Kent 2001, Kent 2005). Despite the potential psychological challenges for young people with NF1, there may also be positive consequences; family relationships may be strong and support adjustment (Graf et al 2006, Krab et al 2009)

Background

Despite the potential impact (both positive and negative) of NF1 on psychosocial adjustment during adolescence, limited research has examined the lived experience of this age group. Much of the available research relies on adults' reports of their own adolescence or parental reports of their child's experiences, yet parental and adolescent concerns and reports have been found to differ, with parents more pessimistic on measures such as social inclusion (Sebold et al 2004, Wolkenstein et al 2008). Additionally, some research has involved younger children, who may be without obvious signs of NF1 since changes to appearance are often not apparent until after puberty (Barton & North, 2007). Much of the research reported as being with adolescents often includes a young sample for instance, whilst the term adolescent is in the study's title, Graf et al (2006) report their participants mean age was 11.6 years, Counterman (1995) reports a mean sample age of 11.8 years, whilst the mean age of participants in Huijbregts et al's (2010) study was 12 years 4 months. Sebold et al (2004) report a mean participant age of 15 years and interestingly they report differences between those under and over 15 years of age suggesting that people's perceptions of their condition change during adolescence. It is also important to note that findings from studies with young adults (Hummelvoll and Antonsen, 2013) have reported differences between the youngest participants aged 18-25 and those aged 26-37 in terms of friendships, depressive difficulties and self-confidence.

The current study specifically investigated young peoples' experiences, concentrating on individuals aged between 14 and 24 and exploring both the challenges of living with NF1 alongside any positive experiences.

The study

Aim

This study aims to obtain a broad, in-depth understanding of young peoples' day to day experience of living with NF1 and aims to use this understanding to identify areas to be investigated further in a primarily quantitative questionnaire.

Design

The research design reflects the researcher's exploratory approach towards this under-researched area and the desire to ensure adolescents were empowered and heard from directly. A qualitative approach was chosen as a way of gaining rich in-depth accounts of young people's different and complex experiences (Rich & Ginsburg (1999).

Participants

Nine young people with NF1 were recruited through the Neuro Foundation newsletter (a UK-based support group for people with Neurofibromatosis), the online forum of Changing Faces (a charity offering support for people living with disfigurement), the Centre for Appearance Research website and advertisements posted on social media sites related to NF1. Participants ranged in age from 14 (the age at which neurofibromas commonly appear) to 24 in line with the World Health Organisation's upper definition of youth in order to obtain a retrospective and current account of experiences during adolescence. See table 1 for participant details.

Table 1: Participants details here

Data collection

People who were interested in taking part contacted the first author and were sent an information pack (including parental information if they were aged under 18). Those who wanted to participate contacted the researcher to arrange an interview. Innovative and visual methods have previously been used in appearance research as a way of engaging people in a potentially sensitive topic (Rumsey & Harcourt 2012, Coad 2007), so participants in this study were offered the opportunity to bring photographs to their interviews or to use timelines during the interview. Using a timeline entailed drawing a line on a piece of paper with their date of birth at one end of the line and the current date at the other, and then filling in significant dates and events in relation to having NF1 between the two. Five participants chose to use timelines or brought photographs to interviews. The photographs and timelines themselves were not analysed but were used as an empowering way for young people to share their experience (Sebold et al 2004, Metcalfe et al 2011).

An interview schedule was developed through a review of literature around engaging young people in research, NF1 and appearance research and with input from people working with young people with NF1. Topics included the role of friends and family, knowledge of NF1, medical treatment, appearance and support. The severity of individuals' NF1 was not recorded as previous research has suggested it is individuals' subjective rather than objective assessment of appearance that predicts wellbeing (Moss 2005, Ong et al 2007), and participants were specifically asked about their subjective experience of their appearance during interviews. Eight interviews were conducted face-to-face and one by telephone (Ros), at the participant's choosing. Interviews were conducted by the first author and took place in participants' homes (n= 6), or another setting of their choice (n= 2). They lasted between 30 and 80 minutes and were audio recorded.

Ethical considerations

Ethics committee approval was obtained from the faculty ethics committee at first author's university. Informed consent was obtained from all participants and parental consent for participants under 18.

Participants were reminded that interviews could stop or pause at any time and that they did not have to answer any questions if they were uncomfortable or uncertain of the answer. If participants were noticeably upset during interviews they were asked if they would like to pause or stop. Participants were reminded of sources of support and were given details of avenues of support verbally and in writing.

The variability of NF1, and the age of participants meant that it was possible that participants may not have thorough knowledge of the condition. As such participants were not asked directly about issues such as future prognosis or heritability/reproductive decision making until the subject was mentioned by the participant themselves. While this may have meant some questions were unasked, and thus unanswered, it was felt this was the most ethical way to ensure the interview process did not introduce areas of concern to young people.

Data analysis

Interviews were transcribed and field notes were kept by the researcher after each interview. Since NF1 is a highly variable condition and participants could be expected to have a multitude of different experiences, thematic analysis was chosen for this study because it maintains the richness of data whilst allowing areas of commonality to be analysed. All transcriptions were coded line by line and codes were developed into subthemes and themes, following the recommendations of Braun and Clarke (2006).

Rigour

To ensure rigor of analysis, themes and interpretations made by the first author were reviewed by the second and third authors and were discussed until there was a consensus.

Findings

The findings presented are based upon interviews with nine participants who ranged in age from 14 to 24 years, six participants were female; three were male. All lived in the United Kingdom.

Central to accounts of their experience of NF1 during adolescence were the variability, unpredictability and visibility of the condition. Its effect on appearance was discussed in terms of adapting to a changing appearance and managing interactions with others. Analysis revealed three key themes: (1) 'Different things to different people' (2) 'Relationships and reactions' and (3) 'Understanding and misunderstanding'

(1) Different things to different people

NF1 can be characterised by its variability (Ablon 1999, Ferner et al 2007). This was particularly evident in how young people described the condition. Some described it as a skin condition, some talked of 'tumors' and 'lumps', some referred to a specific part of their body (eg legs), and others

discussed learning difficulties or ADHD. As Daniel commented: *"It's like a bag of pick and mix you don't know what you're going to get really, it could be anything"*

Accounts also varied regarding the emotional impact of NF1. Some participants felt it held little significance and that there was no real emotional impact at all, whilst others felt it had an extremely profound impact on their life. For instance Robert, commented that NF1 *"Bothers me now and again but it doesn't put me too down"* whereas Ros explained that *".....if I didn't have NF I wouldn't have the problems I have got"*.

While participants had varied experiences and symptoms, many felt their understanding of and feelings about NF1 had changed during adolescence. Katie, the youngest participant, described a growing awareness of having NF1 in recent years *"I don't know if I knew when I was younger or not and I have certainly become more self conscious of it"*. Older participants reflected that this growing awareness could lead to a desire to know more about the condition. For some the desire to learn more stemmed from physical changes:

"I think it was sort of early years of secondary school when I started to notice a few more of lumps and just a bit more curious about it and see how it was affecting me, and I yeah so I got a few more like information packs on it and stuff" (Mark).

Most felt their conceptualisation of the condition altered as they learned more about NF1:

"I think when I was younger it was just the leg stuff that used to bother me, now it is more like the neurofibromas and stuff..... it had always been a leg thing and then to kind of reconceptualise that, it was quite weirdit was just I've got NF that means my legs and my shin bones don't work properly sothere's been a lot of learning since then" (Sarah).

Participants talked of adolescence being a time of change, a time of re-evaluating or learning about NF1 as they came to understand what NF1 meant for them and the impact that the condition might have on their lives.

(2) Relationships and reactions

Participants felt their relationships were affected by NF1 in a myriad of ways. This was discussed with reference to (a) *Family*, (b) *Friends* and (c) *Other peoples' reactions*

Family

NF1 was described as a 'family thing'; a diagnosis impacted on the whole family. Most participants discussed how their families were seen to understand the condition, and were their main source of support and information. Additionally, family members who had NF1 themselves were thought to understand the condition and its impact more than those without it.

“... I think myself and my dad can understand each other more, where you know, people without NF can't really” (Ros)

Friends

Consistent with previous research with young people with an altered appearance (Williamson et al, 2010), friends were an important source of support, understanding and trust. Some participants found telling friends about NF1 straightforward:

“it just takes two minutes of conversation and then you don't need to talk about it again” (Mark)

However, others discussed avoiding talking about NF1 with others:

“I wouldn't want to say NF because my fear is they'll go onto Google or Wikipedia and then find this kid has varying disabilities this kid may have a fit and I don't want them to think that” (Daniel)

Other peoples' reactions

Participants discussed managing situations where strangers had stared at them or asked questions about their appearance. Some found this distressing, whilst others had learnt to manage it:

“I am always scared that people are looking at you, are they staring at you?” (Tina)

“I've never really been too uncomfortable because I've grown up with it, so from an early age I've always been used to having to say something.” (Mark)

Most participants reflected that their concerns about other people's reactions to them could make social situations difficult, and in some circumstances cause them to avoid interacting with others:

“...when I meet new people, there is immediately this thing where, oh she walks a bit different, her legs are a bit weird, she's just tripped over her own feet, like you know, and it's like I just feel immediately awkward about all of those things and I think.....just screw it I won't even bother trying to talk to anybody.” (Sarah)

(3) Understanding and misunderstanding

This theme relates to participants' experiences with organisations including healthcare, school, support groups, and the media, and the impact these organisations had on relationships and psychological adjustment.

Healthcare

Whilst it was very important to participants that health professionals were available to give good quality information and advice about NF1, most commented that it was poorly understood within the

medical community. Tina described asking her GP for information and her frustration that he had to look up the condition, *“they looked back on my file and said ‘hang on we have got neurofibromatosis here’.....the doctor was reading like a massive big book in front of me and I was like ‘what are you doing mate, come on’”*

Those who described positive relationships with health professionals commented on their perceived expertise and professionalism. In particular, those who had seen specialist nurses, doctors or Neuro Foundation specialist advisors described being very happy with the advice and care they had received. Experts had a crucial role to play in putting information into context.

Education

A few participants felt NF1 had a positive impact on their education. Katie explained *“with NF I get to go to college and I’ve got experience before people at school”*. Others talked of a ‘battle’ to get the support they needed and to have their NF1 and any associated learning difficulties recognised, and felt this had impacted negatively on their education.

Medical professionals could be particularly important in supporting young people at school, as Ros explains:

“I struggled all the way through primary school up until secondary school. It was only until secondary school where my paediatrician and he stepped in and said this girl needs you know help in her work and he arranged for an educational psychologist and that’s when I got found out that I obviously had dyslexia” (Ros).

Support Groups

The Neuro Foundation was a source of support for some of the young people in this study. Four young people had been to camps or events run by the organisation, which they described as being supportive, particularly because they were designed for their age group, giving them an opportunity to meet others of a similar age and experience to themselves.

Some participants preferred to use online support rather than face-to-face groups, and as shown in the following quote, were sometimes pleased to speak to others without the need to focus on their condition.

“I used the instant messaging thing which was really good because there would be loads of people there and you’d be talking to lots of different people and I’d kind of just go on and wouldn’t always really talk about NF it would just be a shared understanding” (Sarah)

Media

The representation of NF1 in the media (such as the internet, television and in print) was important to many participants. Most felt it is misunderstood or unknown to the general population, and worried about the quality of information available, and the negative and pathologising way it is portrayed.

“...if I see any articles regarding NF, 9 times out of 10 they got it wrong. That really winds me up.” (Daniel).

“.....what if my friends have read that and thought oh that's what {Sarah} has got as well, that Elephant Man disease or whatever. That was, that was really hard because I'm trying to.... I was trying to cope with the things that made me different and trying to make myself as normal as possible and deal with all these things that made me different and then to have those horrible stories like thrown in the mix.” (Sarah)

Discussion

This study aimed to explore young peoples' experiences of NF1 and has generated a rich insight into their views about the condition and its impact on their lives. Findings support previous suggestions (Ablon 1999, Ferner et al 2007) that NF1 may be particularly challenging due to its unpredictable nature and its affect on appearance. Young people face these challenges within a society that holds a robust belief about the importance of appearance and the stereotypical view that “what is beautiful is good” (Dion et al, 1972) and during a life stage when body image concerns are likely to be emphasized (Levine & Smolak 2004). Additionally, findings suggest the lack (or perceived lack) of awareness and understanding of NF1 may make adjustment all the more complicated.

Whilst participants described the challenges they faced as a result of having NF1 (in particular other people's reactions to visible signs on their skin, their concern over the uncertainty of dermatological changes and the general lack of awareness of the condition) , it is important not to pathologise the experiences of people living with an unusual or altered appearance (Rumsey & Harcourt 2004, Egan et al 2011) and to highlight that the participants in this study displayed considerable resilience and were living positively with NF1 despite facing numerous challenges. For example, as suggested in previous research (Ablon 1999, Ferner et al 2007) adolescence was, for many, a time during which the condition became a more prominent part of their life, partly due to physical changes (condition specific or pubertal), but also as they reconceptualised their understanding of the condition. This reconceptualisation, alongside the reported desire for information as symptoms change, suggests adolescence may be a time during which availability of good quality information and access to specialists is crucial.

Young people in this study called for a greater awareness of NF1 by health and education professionals and the general public. As has been suggested previously, participants felt this would be helpful (Dheensa & Williams 2009) and that distressing mistruths such as the persistent (inaccurate) link to ‘the elephant man’ should be challenged (Ablon, 1999).

In line with the findings of studies with young people with other appearance-altering conditions, participants valued social support from friends, family and support groups, as well as practical information (Thompson & Kent 2001, Thompson & Broom 2009, Williamson et al 2010) to help them manage other peoples' reactions to their appearance. An important first step may be for health professionals to recognise the possibility of appearance concerns thus legitimizing the young person's concerns (Clarke 1999).

Additionally social skills training has been shown as beneficial for many people with a visible difference (Rumsey & Harcourt, 2007, Clarke 1999) and adolescence has been found to be a transitional point with regards to young peoples' social skills in studies with other conditions (Pitt, 2009). Given that adolescents with NF1 may have poorer social skills than peers (Barton & North, 2007); this would seem to be a potentially very supportive and beneficial intervention.

An important concern for young people in this study related to managing other people's questions and reactions and explaining NF1 to others. Some young people described a reluctance to discuss NF1 due to concerns that others would look up the condition and see the worst case scenarios and classify them in this way. The variability of the condition can make it difficult for young people to explain. This finding supports other studies that have focused on young people (Hummelvoll & Antonson 2013 and Sebold et al 2004) suggesting that finding ways to talk about NF1 in a simple and positive manner may be particularly significant during adolescence.

Limitations

There are some limitations to this study. Considering the variability of NF1, including the possibility of associated behavioural and learning difficulties, it is not possible to generalise the findings from this study to all young people with NF1. However, this exploratory study has informed the development of a survey to establish the extent to which the results might be applicable to a wider sample of young people with NF1. Our research has also included a survey of parents of young people with NF1 and health professionals (including dermatologists) following interview studies with these groups. A strength of this programme of research overall is that it includes input from these 3 key groups - young people themselves, health professionals and parents – who are best placed to comment on the experience of living with NF1 during adolescence and to inform the provision of appropriate care in the future.

Conclusions

In conclusion, this study has identified 4 key areas warranting further examination and which may be amenable to intervention on different levels. First, a need for increased awareness and understanding about NF1 amongst the general public and professionals such as teachers and doctors is highlighted. Second, ensuring adolescents have easy access to trusted information about NF1, including health professionals with appropriate expertise. Third, availability of social support that is age appropriate

both in person and through online forums and, finally, a range of interventions and advice to support the development of social skills and adjustment to an altered appearance.

Until now very little research has specifically examined the experiences of young people living with NF1. Whilst this study starts to explore this overlooked area, more research is needed in order to support clinicians looking to provide the best possible care for people with NF1 during adolescence in order to positively support the development of resilience.

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Participant pseudonym	Gender	Age	Parent present during interview.	NF1; Inherited or new mutation	Highest educational level to data
Tina	Female	21	No	Unsure	University
Sarah	Female	23	No	New	Postgraduate
Joanna	Female	17	Yes, both parents	New	College
Robert	Male	20	Yes, Dad	New	College
Ros	Female	24	No	Inherited	University
Daniel	Male	24	No	Inherited	Postgraduate
Katie	Female	14	Yes, Mum	Inherited	School
Lucy	Female	16	Yes, Mum	Inherited	School
Mark	Male	21	No	New	University

Table 1: Participant details