Parents' experiences of caring for a young person with neurofibromatosis type 1 (NF1): a qualitative study

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The role of appearance in adolescents' experiences of Neurofibromatosis type 1: A survey of young people and parents

Abstract

Neurofibromatosis type 1 (NF1) is a genetic condition which can result in varying degrees of visible difference (disfigurement). Adolescence is a time when appearance concerns become more salient for many young people and is acknowledged as a particularly challenging time for individuals with NF1. However, there is currently little research into the psychosocial impact of the appearance changes associated with NF1 during this stage of life. In order to address this surveys of young people aged 14-24 years and with a diagnosis of NF1 (n=73), and parents of young people with NF1 (n=55) were developed following interview studies with these groups. The surveys included the Perceived Stigma Questionnaire, Social Comfort Questionnaire, Body esteem (appearance subscale) and the Subjective Happiness Scale. Young people and parents identified appearance as central to young peoples' experience of NF1. . However, no significant difference was found on measures of body esteem, happiness, stigma or social comfort between those young people who reported their NF1 was noticeable to others and those who reported it was not. Parents reported a relationship between the noticeability of their child's NF1 and their interactions with others. Findings highlight the importance of attending to young people's concerns around appearance in general and managing the possibility of future appearance changes, rather than the current noticeability of NF1.

Key words

Neurofibromatosis Type 1, NF1, Young people, Parents, Appearance, Body image, psychosocial,

What's already known about this topic?

- Neurofibromatosis type1 (NF1) can have a significant impact on psychological wellbeing
- The psychological impact of NF1 can stem from managing both the unpredictability of the condition and changes to appearance.
- Adolescence is a period when appearance becomes increasingly important and appearance-related concerns are common.

What does this study add?

- Appearance was identified as an important aspect of NF1 by both parents and young people
- Parents and young people with NF1 differed with regards to the importance they placed on the noticeability of the condition. Whilst parents associated the noticeability of NF1 with their child's social experiences, young people's accounts of social interactions, happiness and appearance did not differ between those who did or did not view their NF1 as noticeable to others.
- Both groups reported that managing uncertainty around changes to appearance is a particular challenge.

Introduction

Neurofibromatosis type 1 (NF1) is a genetic condition which occurs in 1:2500-1:3000 people (Ferner et al 2007). Fifty percent of people with NF1 will have inherited their condition from a parent while the remainder of cases are new to families. The condition can result in varying degrees of visible difference (disfigurement) including cafe au lait spots (coffee coloured birthmarks), neurofibromas (benign tumors on the skin), skin fold freckles, plexiform neurofibromas (diffuse tumors that grow along a nerve) and scoliosis (curvature of the spine).

NF1 is unpredictable and variable both between individuals and over time, making it difficult for those diagnosed with the condition to know how it will affect them over their lifetime. In addition, people with NF1 are at increased risk of varying degrees of learning and behavioural difficulties including Attention Deficit Hyperactivity Disorder (ADHD) and Autistic Spectrum Disorders (ASD) (Barton &

North 2004, Ferner et al 2007, Lehtonen 2012), and have been identified as having lowered social skills and difficulties processing social information (Barton & North 2004, Noll et al 2007, Huijbregts et al 2010). The myriad of challenges that can arise from managing both the uncertainty of the condition and its impact on appearance and social interactions (Ablon 1999; Ferner et al, 2007) mean it can impact on both quality of life and psychological adjustment (Graf et al 2006, Noll et al 2007, Krab et al 2009, Wolkenstein 2009).

Predictability around appearance changes and strong social skills have been identified within the visible difference literature as being important factors in adjusting to an appearance that is in any way different to 'the norm' (Rumsey et al 2010, Rumsey & Harcourt 2012). An existing visible difference may become more challenging during adolescence (Griffiths et al, 2012), although this may also be a particularly difficult time to acquire a disfigurement of any sort (Ben-Tovim & Walker, 1995). Therefore the unpredictable nature of NF1, and its possible impact on social skills can present particular risks to positive adaptation for young people during adolescence.

Little research has directly explored the role of appearance and NF1 during adolescence. Previous exploratory interviews with young people with NF1 (Barke et al 2014), and parents of young people with the condition (Barke et al, in submission) have identified that thoughts and feelings about appearance, their confidence in managing appearance-related issues and experience of social situations are central to young people's well-being and experiences of NF1. However, the role of noticeability appeared to differ between the two groups. Parents reported that visible NF1 had a significant impact on their child's lives whereas young people themselves reported that how their appearance might or might not change in the future was more of a concern than was the current noticeability of the condition. In the current study we built on this qualitative work to explore the role of appearance and experience of social situations focusing on the impact of subjective noticeability from the viewpoint of young people with NF1 and parents. Specifically we aimed to explore the following:

- How do young people with NF1 feel about their appearance in general and do they report their condition as noticeable?

- How do young people report their social comfort and interactions with others and is this different for those who report their NF1 as noticeable or not?
- How do general feelings about their appearance, subjective noticeability, social comfort and interactions with others impact on young people's wellbeing?
- How do general feelings about their appearance, social comfort and interactions with others relate to one another?
- How do parents describe the role of appearance within their child's experience of NF1 and how noticeable do they feel their child's condition is?
- How do parents report their child's social comfort and interactions with others and does this relate to parents reports of noticeability?

Methods

This study used mixed methods, gathering both qualitative and quantitative data through online surveys completed by young people and parents. The study was approved by the Ethics Committee of the first authors host institution and all necessary NHS ethics and R&D approvals were sought and granted.

Participants

Two questionnaires were developed to further explore and quantify the findings of previous exploratory interview studies (Barke et al 2014; Barke et al, in submission), one for young people and one for parents. The inclusion criteria were (a) young people with a diagnosis of NF1 aged between 14 years (the age at which neurofibromas commonly appear) and 24 years of age (in line with the World Health Organisation's upper definition of youth) or (b) parents of young people who were aged 14-24 years and who had a diagnosis of NF1 (parents were not excluded if they had a diagnosis of NF1 themselves, but it was not an inclusion criteria). Participants were recruited internationally and had to be able to complete a questionnaire in English.

Procedures

Young people were identified through reviewing and searching clinical notes and databases at three NHS clinic sites in England. Information about the study was then sent by their consultant to young

people and their parents. Letters were addressed to young people aged 16 or over, and to the parent/carer if the young person was aged 14 or 15. Those wishing to participate could either complete the questionnaire that was enclosed with the study information letter or complete the survey online which was developed using Qualtrics. Details of the study were also included on web sites, Facebook pages, internet forums and newsletters of relevant support groups, in the UK, USA, Canada, Australia and New Zealand. Informed consent was sought for all participants and young people aged 14 and 15 were asked to provide parental consent. Young people and parents in the same family did not both have to participate; this was made clear in study information.

Instrumentation

Findings from previous qualitative studies defined areas to explore and informed the formation of a series of research questions to guide the development of surveys (as previously detailed). Measures were then chosen to specifically address these questions. The survey contained the following questionnaires:

Young people

Appearance: The Body Esteem (BE) Scale (appearance subscale) (Mendelson et al, 2001) is a 10 item measure of overall feelings about appearance, with potential scores ranging from 0-4 and higher numbers indicating greater body esteem (the self evaluation of one's body or appearance). Young people were also asked whether or not they felt their NF1 was noticeable to others (Yes/No) and completed an open ended statement: *My main concern about NF1 is.....*' to investigate whether appearance was identified as a concern.

Social experience: The Perceived Stigmatization Questionnaire (PSQ) (Lawrence, 2010) is a 21 item measure of how frequently respondents experience various stigmatising social behaviours. Possible scores range from 1 to 5, with a higher score indicating greater perceived stigma. The Social Comfort Questionnaire (SCQ) (Lawrence, 2010) measures social isolation and the violation of privacy (increased staring and questions being asked about the appearance). The scale has 8 items and asks respondents to indicate (on a 5 point scale) how often they feel or think a series of statements. Possible scores range from 1 to 5, with a higher score indicating greater social comfort.

Happiness: General wellbeing or happiness was measures using the Subjective Happiness Scale (SHS) (Lyubomirsky & Lepper, 1999). This measure is based upon the evidence that objective circumstances, demographics and dispositional factors are not strongly correlated with happiness or positive wellbeing. People can consider themselves happy in spite of personal circumstances that would seem to predict otherwise. The SHS is a four item scale of global subjective happiness, possible scores range from 4 to 28, with higher scores indicating greater subjective happiness.

Parents

Appearance: Parents were asked how often their child expressed concern about their appearance (generally and NF1 specifically), and how confident they, as a parent, felt managing concerns about appearance. In order to explore if appearance was a concern parents were asked how NF1 affects (a) them and (b) their child and what their concerns were at initial diagnosis and at the time of completing the questionnaire. Parents were also asked how noticeable they thought their child's NF1 was to others on a scale of 0 (not at all) to 10 (highly noticeable).

Child's social experience: The PSQ and SCQ, as described above, were used in order to explore parents' perceptions of their child's social experience. Questions were altered to focus on 'my child' rather than the respondent (parent).

Quantitative data was analysed using SPSS statistical software (version 19). Statistical analyses were undertaken to answer each of the research questions previously outlined. Alongside exploring descriptive data, t-tests, a multiple regression analysis and tests of correlation were employed.

Qualitative data was analysed using content analysis, whereby text is classified into smaller categories that can be quantified; it is systematic and replicable and can deal with large volumes of data (Stemler, 2001). Open ended responses to questions were compiled into a list and were read several times, initial codes were identified and all data was then coded into this list. Data was then quantified by counting the frequency of each code. Codes were verified throughout the research

process; interpretations made by the first author were reviewed by the second and third authors and were discussed until there was a consensus (Morse et al 2002).

Results

Young People

Seventy three young people completed the survey (22 paper copies, 51 online). All confirmed they had a diagnosis of NF1; 34% (n=25) had a family member with NF1, 59% (n=43) had no family history of the condition and 7% (n=5) were unsure. Further details are provided in Table 1. Table 2 summarises results from standardized measures in the young people's questionnaires.

Tables 1 and 2 here

Mean body esteem (appearance subscale) scores in the current study were slightly lower than published means for similarly aged participants in a USA school and college sample (Mendelson et al, 2001). Lawrence et al (2007) suggest scores lower than one indicate very low body esteem. Adopting this cut off, 25% (n=17) of young people in this study scored in this range while 33.9% (n=23) scored three or four indicating positive body esteem. An independent t-test showed no significant difference on any measure (PSQ, SCQ, SHS, BE) between the 33 (47.1%) who reported their NF1 was noticeable to other people and the 37 (52.9%) who did not.

No participants scored total perceived stigma in the 'often' or 'always' categories, 36.2% (n=21) of participants reported perceived stigma in the 'sometimes' range and 63.8% (n=37) scored in the range of 'never' to 'almost never'. The majority of participants (84.6%, n=55) scored social comfort in the 'sometimes' and 'often' range, 13.8% (n=9) reported low levels of social comfort and 1.5% (n=1) felt social comfort 'always'.

Scores on the SHS are within the range (4.63–5.07) reported by Lyubomirsky and Lepper *(*1999*)* for high school and college students (mean age 19 years) in the United States, 61% rated themselves as

slightly to extremely happy, 17.3% were slightly to extremely unhappy while 21.7% scored in the neutral range.

The relationship between the SHS, PSQ, SCQ and BE (appearance) was investigated using Pearson product moment correlation coefficient (see table 3).

Table 3 here

A multiple regression analysis was employed in order to explore whether the variance in happiness scores could be explained by social comfort, perceived stigmatisation and body esteem (appearance). The variance explained by the model as a whole was 49%, $F(4, 50) = 12.077$, p.001. Only the BE (app) scale was statistically significant (beta = .479, $p<.005$) suggesting that the BE appearance subscale explains almost half of the variance in happiness.

An open ended question asked participants what was their main concern about NF1. Sixty four participants responded. Using content analysis, responses were coded into eight categories (see table 4).

Table 4 here

Parents

Fifty five parents completed the survey (32 online, 23 paper), 94.5% (n=52) were White British, American or Irish. All respondents indicated that they had a child aged 14-24 with NF1, 45.6% (n=24) of these children were male (1 person did not provided this information). Just over half (56.3%, n=31) of respondents had children aged under 18. Twenty three parents (41.8%) had a diagnosis of NF1 themselves, 43.6% (n=24) reported that their child's NF1 was inherited and 52.7% (n=29) said it was new to the family whilst two respondents were unsure. Further details are provided in Table 5. Table 6 provides details of data from standardized measures in parents' questionnaires.

Tables 5 and 6 here

The majority of parents reported that their child rarely or never expressed concern regarding their appearance in general (79%; n=42) or about appearance-related aspects of NF1 (85%; n=45). Most parents (66%, n=24) were fairly confident (6-10 on a scale of 0-10, with 10 being very confident) in managing their child's appearance concerns. However 34% (n=12) indicated medium to low levels of confidence (0-5 on the scale). The majority of parents (60%, n=32) felt their child's attitude towards their appearance had not really changed at any point. Of those who did feel their child's attitude had changed, many thought this was due to being a teenager (46% n=13).

Around a quarter of parents (28%, n=15) felt their child's NF1 was not at all noticeable to others (scoring 0). The same number felt it was noticeable over the midpoint (between 6-10). The mean noticeability score was 3.57 (SD 3.220). There was a strong positive correlation between noticeability and the PSQ, and a strong negative correlation with the SCQ (see table 7), indicating that greater perceived noticeability related to greater perceived stigma and lower levels of social comfort.

Table 7 here

PSQ scores indicated most parents (n=30, 70%) perceived that their child never or almost never felt they were stigmatized although 9% (n=4) thought that their child often felt stigmatized by others. Scores on the SCQ indicated parents were fairly evenly divided between thinking their children felt socially comfortable almost never (25%, n=12), sometimes (37.5%, n=18) or often (31.3%, n=15). No parents reported that their child was never socially comfortable and three (6.3%) reported 'always'.

Parents' reports of the main ways in which NF1 affected their child and themselves were coded and grouped into categories shown in tables 8 and 9.

Tables 8 and 9 here

In addition, to considering the affect of NF1 on themselves and their child, parents were asked to reflect on their concerns at the time of initial diagnosis and at the time of completing the

questionnaire. The most commonly reported concern at the time of diagnosis related to understanding the condition and the medical prognosis (n=22, 59%). At the time of completing the questionnaire the most common concern related to their child being generally happy and living a normal adult life (n=26, 43%)

Discussion

Previous research has reported negative body image and appearance concerns amongst adults with NF1 (Smith et al, 2013; Granstrom et al, 2012) and a less positive body image amongst young people with a chronic condition than healthy peers (Pinquart, 2013), but there has been a dearth of research exploring body image amongst young people with NF1. The mean body esteem scores for the young people in the current study were similar to those reported in a normative population (Mendelson et al, 2001) and amongst burn survivors and a comparison group (Lawrence et al, 2006). This, in addition to just 15% of those in this survey reporting highly negative body esteem (ie. scoring 0), suggests that while some young people with NF1 have low body esteem and require may benefit from support, many had positive body esteem. It would therefore be premature to assume that NF1 necessarily has a negative impact on body image, although there is still a need for support for those who are negatively affected by the changes to their appearance.

The noticeability of NF1 was important within parents' reports of their child's experience of NF1, but not within young people's own reports. Differences between parents' and young people's perceptions of the impact of severity of NF1, both in terms of appearance and clinical severity, have been reported previously (Sebold et al, 2004; Counterman et al, 1995). Sebold suggests that these differences relate to young people's changing cognitive ability and point out that older adolescents' scores were more closely aligned to their parents' assessments of severity. In the current study, young people were substantially older (survey mean age = 20.4 years) than both Counterman's and Sebold's adolescent groups (mean ages = 11.8 and 15 years, respectively) yet the differing importance of noticeability between parents and young people was still apparent.

It is unclear exactly why parents reported noticeability as important. However, interviews with parents within our programme of research (see Barke et al, in submission) suggest it may relate to their

vigilance in searching for signs of the condition and concerns over how visible differences could impact on a child's life, both of which Thompson and Kent (2001) have suggested increase the emphasis parents place on appearance. Managing uncertainty has been highlighted as central to the experience of parenting a child with a chronic health condition (Stewart & Mishel, 2000) and vigilance is a coping mechanism that parents use to manage this uncertainty (Jessop & Stein, 1985).

Our finding that young people's reports of noticeability were not significantly associated with happiness, social interactions or body esteem contradicts previous research with adults with NF1 which has linked reported visibility of NF1 and psychological wellbeing (Granstrom et al, 2012; Wolkenstein, 2009; 2001). However, it is important to note that quality of life and body experience (defined as how secure and confident people felt about their bodies) mediated the relationship between visibility and psychological stress in Granstrom et al's study. Similarly, Lawrence et al (2006) found that the importance placed on appearance by burns survivors moderated the relationship between subjectively reported severity and body esteem. This suggests that the importance placed on appearance generally is relevant to people's experiences of living with a visible difference, possibly more so than the noticeability of their visible difference.

Adapting to, and living with, a visibly different appearance is an evolving process (Prior, 2009) and managing a changeable, unpredictable appearance may be particularly challenging (Rumsey et al, 2010). Appearance-related concerns reported by the young people in this survey related to possible changes to appearance in the future, more often their current appearance. Young people with NF1 did not report particularly low levels of happiness, appearance evaluations or negative social interactions. In line with findings with young adults with other genetic conditions, such as Marfan syndrome (Tongerloo & Paepe, 1998), and young people with other visible difference (Rumsey & Harcourt, 2007) many young people with NF1 were happy and felt positive about social interactions and their appearance.

Study Limitations

A limitation of this study is the different ways in which noticeability was measured in the parents' and young people's surveys, since we used questions which reflected how young people and parents discussed the concept in the interviews that informed the development of these surveys. However, this has meant that the findings of the two surveys could not be directly compared. With hindsight, the

surveys could have used the same assessment of noticeability for both the young people and parents. Robust methods for measuring subjective accounts of noticeability that can be used with different population groups are still needed in order to further understand the role of noticeability within people's experiences of a visible difference. Whilst the international reach of the questionnaire has increased the sample size and does not limit our findings to a single service, this could also be considered a limitation when considering the application of findings, since individuals were reporting on experiences in different healthcare system. We attempt to overcome this by discussing the implications of findings broadly rather than particular clinical applications.

Practice Implications

A particular implication of the current study is that whilst some young people clearly require support to manage a visible difference it is important that young people's experiences are not assumed to be negative. Given the highly varied accounts of appearance and NF1, supporting families and young people to be resilient and happy against a backdrop of uncertainty may be particularly beneficial for young people with NF1. However, this is not to suggest that issues around appearance should not be addressed. Parents and professionals working with young people with NF1 should be aware that young people's concerns are not necessarily related to the noticeability of the condition and that any appearance concerns they hold may relate to uncertainty around future changes rather than how they look at a particular point in time.

Health professionals can play a key role in supporting appearance concerns simply by talking about appearance and normalising patients' concerns (Clarke 1999). In light of the findings presented in this study it may be appropriate for health professionals to ask young people directly about appearance,, regardless of the noticeability of the individual's symptoms, and to feel confident in how, when and where to refer on those who may benefit from additional psychosocial support in relation to appearance.

Research recommendations

Further research is needed to explore and understand the relationship between noticeability of a visible difference and psychosocial experience and adaptation. Longitudinal research that explores

this through childhood, adolescence and into adulthood from the perspectives of young people, parents and clinicians would be particularly valuable.

To conclude, this survey highlights the importance of general aspects of appearance and concerns about possible future changes to appearance rather than the noticeability of NF1, and emphasises the importance of parents and health professionals realising that young people's concerns may not necessarily be the same as their own.

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Conflict of interests

The authors declare that they have no conflict of interests

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Table 1: Demographic details of respondents to the young people's survey (n=73)

		N (%)
Gender	Female	52 (71.2%)
	Male	20 (27.4%)
	Information not provided	1 (1.4%)
Age	Mean = 20.4 (sd 2.91)	
Ethnicity	White	59 (80.8%)
	Mixed	6 (8.2%)
	Asian	5 (6.8%)
	Black	2 (2.7%)
	Information not provided	1 (1.4%)
Country of residence	England	39 (54%)
	Scotland, Wales, N Ireland and Ireland	9 (12%)
	North America	16 (22%)
	Other (Europe, New Zealand, Australia, Philippines, South America & China)	8 (11%)
	Information not provided	1 (1.4%)

Table 2: Descriptive statistics for standardised measures included in the young people's survey

Scale	**N**	**Min**	**Max**	**Mean**	**Std. Deviation**
Subjective Happiness Scale (SHS) (possible range 4-28; higher score indicates greater happiness)	69	4	27	18.19	5.465
Perceived Stigma Questionnaire (PSQ) (possible range 1-5; higher scores indicates higher levels of perceived stigma)	58	1	3	2.19	.585
Social Comfort Questionnaire (SCQ) (possible range 1-5; higher scores indicate higher levels of social comfort)	65	1	5	3.10	.778
Body Esteem (appearance subscale) (BE) (possible range 0-4; higher scores indicate greater body esteem)	68	0	4	2.01	1.126

Table 3: Correlations between standardised measures in young people's survey

Scale	SHS	PSQ	SCQ	BE (app.)
SHS	-	-0.485**	0.529**	0.667**
PSQ		-	-0.673**	-0.559**
SCQ			-	0.535**

**. Correlation is significant at the 0.01 level (2-tailed).

*. Correlation is significant at the 0.05 level (2-tailed).

Table 4: Young people's self-reported concerns about NF1

Main concern	**N (%)**
Specific medical concern	18 (28%)
Appearance changes in the future	16 (25%),
Passing NF1 on to future children	15 (23%),
Current appearance concern	5 (8%),
Learning difficulties and educational issues	4 (6%),
Social concerns	4 (6%),
Others not knowing about NF1	1 (2%)
No concerns	1 (2%)

Table 5: Demographic characteristics of respondents to the parents' survey (n=55)

		N (%)
Parent's gender	Female	47 (85.5%)
	Male	8 (14.5%)
Country of residence	England	32(58.2%)
	Scotland	2 (3.6%)
	Wales	2 (3.6%)
	USA and Canada	17 (31%)
	Other (New Zealand & Mexico)	2 (3.6%)

Table 6: Descriptive statistics for standardised measures included in the parent survey

Scale	**N**	**Min**	**Max**	**Mean**	**Std. Deviation**
Perceived Stigma Questionnaire (PSQ) (possible range 1-5; higher scores indicates higher levels of perceived stigma)	43	1.00	4.00	2.0875	.71481
Social Comfort Questionnaire (SCQ) (possible range 1-5; higher scores indicate higher levels of social comfort)	48	2	5	3.11	.848

Table 7: Survey of parents: Pearson's correlations between noticeability and the PSQ, SCQ

	R
TOTAL PSQ	.729**
TOTAL SCQ	-.590**

**. Correlation is significant at the 0.01 level (2-tailed).

Table 8: Content analysis of parents' reports of the main affect of NF1 on their child

Main affect on child	**N (%)**
Educational	14 (23%)
Medical	13 (22%)
Social	10 (17%)
Appearance	10 (17%)
Employment and career	1 (2%)
Uncertainty of the condition	4 (7%).
No affect on their child.	3 (5%)

Table 9: Content analysis of parents' reports of the main affect of NF1 on themselves

Main affect on self	**N (%)**
A general sense of worry and monitoring their child's symptoms	21 (41%).
Managing learning and behavioural difficulties,	13 (26%)
The impact on career and work schedule	5 (10%),
Guilt	4 (8%)
Specific medical concerns	3 (6%).
Child's NF1 had no affect on them	5 (10%)