


Received: 14 March 2017 | Revised: 6 February 2018 | Accepted: 22 February 2018

DOI: 10.1002/ajmg.a.38680

## ORIGINAL ARTICLE

WILEY AMERICAN JOURNAL OF  medical genetics

# Worries and needs of adults and parents of adults with neurofibromatosis type 1

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Neurofibromatosis type 1 (NF1) is a neurocutaneous disorder associated with lifelong tumor growth propensity and neurocognitive impairments. Although follow-up of adults with NF1 often focuses on tumor growth, follow-up of cognitive or social problems and other NF1-related comorbidity is often not a part of standardized care. In order to provide optimal care services for these patients, we explored the care needs of adults with NF1. A qualitative study was performed using semi-structured group interviews, exploring worries and care needs in medical, psychological, and socioeconomic domains, also focusing on the transition from pediatric to adult care. Four focus groups were conducted, including young adult patients, patients over age 30, and parents of young adult patients. In total, 30 patients and 12 parents participated. Data were transcribed verbatim and analyzed by computerized thematic analysis. Themes were organized using the World Health Organization International classification of functioning, disability, and health (ICF). Results indicated many and diverse worries and care needs both during the transitional period and in adulthood in medical, mental health, and socioeconomic domains. Worries could be categorized into 13 themes. Parents reported high stress levels and difficulties with their parental role. Participants expressed the need for more information, access to NF1 experts, daily living support, care for mental health and socioeconomic participation, and closer communication between health-care providers. In conclusion, worries and needs of patients and parents underline the importance of multidisciplinary follow-up and continuity of care during and after the transitional period. Additionally, parental stress requires more attention from care providers.

**KEYWORDS**

adults, ICF classification, neurofibromatosis 1, qualitative research, transition

**Abbreviations:** ADHD, attention-deficit/hyperactivity disorder; ASD, autism spectrum disorder; GP, general practitioner; HCP, health-care provider; ICF, International classification of functioning, disability, and health; ISCED, International classification of education; MEC, Medical and Ethical Review Committee; MPNST, malignant peripheral nerve sheath tumor; NF1, neurofibromatosis type 1; NFN, neurofibromatosis patient organization of The Netherlands; WHO, World Health Organization.

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## 1 | INTRODUCTION

Neurofibromatosis type 1 (NF1) is a relatively common autosomal dominant neurocutaneous disorder with an estimated birth incidence of 1/2,700 (Evans et al., 2010). Approximately 40% to 50% of the cases are caused by a de novo mutation in the NF1 gene (Evans et al., 2010; Friedman, 1999). Clinical features of NF1 include multiple café-au-lait spots, axillary and inguinal freckling, multiple cutaneous neurofibromas,

and iris Lisch nodules (NIH, 1987). Serious complications of NF1 include different types of tumor formation, such as central nervous system gliomas and plexiform neurofibromas, with a risk of evolving into malignant peripheral nerve sheath tumors (MPNSTs) (Ferner & Gutmann, 2013) for which guidelines for neuro-oncological follow-up are available. However, although neuropsychiatric and psychosocial problems persist into adulthood, currently these are often not routinely addressed in adults.

Learning disabilities occur in at least 50% of children with NF1, making cognitive problems the most common complication to affect the quality of life in this group (Hyman et al., 2005). Attention-deficit/hyperactivity disorder (ADHD), autism spectrum disorder (ASD), and sleep disturbance are also highly prevalent (Hyman et al., 2006; Leschziner et al., 2013; Morris et al., 2016) and can persist throughout life (Constantino et al., 2015; Descheemaeker et al., 2013; Mautner et al., 2015; van Eeghen AM et al., 2013). Furthermore, patients with NF1 are at risk for psychosocial problems. In adolescence and adulthood, the dermatological and neuronal tumor phenotype can exacerbate in a period in which appearance, acceptance, and social inclusion are of great importance (Van Lierde et al., 2013; Vranceanu et al., 2015). Additionally, the impaired socialization, low self-esteem, and poor interpretation of social cues reported in children with NF1 (Lehtonen et al., 2013) may limit social participation in adulthood. Even when adults with NF1 receive care for the tumor growth phenotype, appropriate care for other NF1-related morbidity is often not part of standardized care (Farre et al., 2016).

For patients with NF1, international health-care guidelines advise multidisciplinary follow-up including evaluation of tumor growth, dermatological manifestations, and neuropsychiatric disorders (Korf, 2015; Rauen et al., 2015). Although pediatric patients often receive and adhere to this multidisciplinary care, studies show that young adults with NF1 may have poor access to health care, limited disease knowledge, and are often lost to follow-up. At the same time, they have a high complication rate and neuropsychiatric and socioeconomic problems persisted or worsened (Oates et al., 2013). Adult patients with NF1 experience decreased quality of life (Vranceanu et al., 2013) and they require developmentally appropriate care (Farre et al., 2016).

Thus far, limited data are available on the full scope of worries and health-care needs of young adults and adults with NF1 and their parents. Previous qualitative studies have focused on the impact of NF1 in adulthood, for instance, addressing the effects of plexiform neurofibromas (Lai et al., 2017), health and well-being, quality of life, and transition to adult care (Barke et al., 2014; Crawford et al., 2015; Draucker et al., 2017; Van Lierde et al., 2013). The aim of the current study was to provide more information on the full spectrum of worries and care needs in medical, psychological, social, and economic domains of adults with NF1 and parents of patients with NF1. Using patient-driven data, we formulate recommendations for health-care providers (HCPs) in order to optimize health care for this vulnerable patient group.

## 2 | MATERIALS AND METHODS

A qualitative design with semi-structured group interviews was used to obtain in-depth data on worries and care needs of patients and their

parents. The full spectrum of care needs was explored, including medical, psychological, social and societal domains, and contextual factors. The COREQ (consolidated criteria for reporting qualitative studies) checklist was used as a reporting framework for this qualitative study (Tong et al., 2007).

### 2.1 | Participants

Convenience sampling was performed in close cooperation with the neurofibromatosis patient organization of The Netherlands (NFVN). Young adults (18–30 years) and older adults (30–67 years) with NF1 and parents of patients with NF1 were invited to join focus groups, which were conducted on April 16, 2016, at a conference center in the Netherlands after the annual meeting of the NFVN. This annual meeting is generally visited by 50 to 100 patients, parents of patients, and professionals. The invitation was part of the agenda that was sent to all members of this association. Inclusion criteria for the patients were that they must be 18 years of age or older, and have a diagnosis of NF1 (as stated by the participating members, not necessarily confirmed by DNA testing). The inclusion criterion for parents was having a child with NF1 who is now an adult. Participation of parents was not dependent on their child's participation or vice-versa.

Participants were divided into four focus groups: 1) patients aged 18 to 30 years, 2) parents of patients aged 18 to 30 years, 3 and 4) two groups of patients and parents of patients aged over 30. The cut-off point of 30 years was based on the European Commission definition of "youth and young people" (European-Commission, 2007), evidence that neurodevelopment continues up to the age of 30 years old, and clinical experience that the transitional period can be protracted in patients with neurodevelopmental disorders. Both groups 3 and 4 were mixed groups of parents and patients, who indicated they wanted to join the groups together. In total, 12 parents participated, including four fathers and eight mothers.

### 2.2 | Data collection

Focus groups were 90 min in length and were moderated by a pediatrician (RO), an intellectual disability physician (AE), and two psychologists (AR, JL). Moderators were specifically allocated to ensure they had no treatment relationships with participants. Prior to the focus group, moderators received training to ensure consistency across interviews. Two observers (HH and PB) joined the focus groups.

Semi-structured interviews contained previously drafted questions and probes on worries and needs in medical, psychological, and socioeconomic domains (see Table 1). Participants were explicitly asked to discuss the transitional phase between pediatric and adult care. In the Netherlands, the transition from pediatric to adult health care usually occurs at approximately 18 years of age (van Staa et al., 2011).

### 2.3 | Data analysis

Focus groups were digitally recorded and transcribed verbatim. Transcripts were imported into the qualitative software package ATLAS.ti 6.2 (ATLAS.ti, 2011). Thematic analysis (Braun & Clarke, 2006) was selected for its theoretical freedom and yield of rich and detailed account of data.

TABLE 1 Abbreviated interview guide

Key questions:	Probes
1) What are your worries about...	
2) What are your care needs for...	
Transition from pediatric to adult care?	Transition to adult health care, change in physician, transition to adult life, transition to work, independent living
Medical issues?	Symptoms, NF1-related care, medication, care consultations
Psychological and behavioral issues?	Sleeping problems, ADHD, ASD, depression, anxiety
Social life?	Friendships, romantic relationships, family, family planning, loneliness, sexuality
Work and daily activities?	Work, school, independence, finances, daily living
Paramedical issues?	Language problems, nutrition, motor skills

All transcripts were reviewed and coded by a member of the research team (HH). Initial codes were based on the overall subject of a text fragment. During analysis, initial codes were modified, expanded, or merged as new issues emerged. Decisions about the codes were made based on the most complete representation of the data. Sub-themes were merged where possible. To enhance validity, the coding process and the emerging themes were continually discussed with two co-investigators (PB, AE), and in an additional meeting with other co-investigators themes were discussed until consensus was reached.

After obtaining a consensus on the open coding, worries and care needs were organized based on the International classification of functioning, disability, and health (ICF) framework. The ICF is a classification of health and related domains, published in 2001 by the World Health Organization (WHO 2001), and describes impairments of body functions and structures, activity limitations, participation restriction, and environmental factors. It is used as a framework to express health and disability using individual and population measures and has been used for holistic evaluation and interpretation of NF1-related disabilities in pediatric patients (Gilboa et al., 2010). A valuable contribution of the ICF classification is the emphasis on the effects of health issues on activities and social and economic (socioeconomic) participation. Socioeconomic participation is defined as a person's involvement in a

life situation and the socioeconomic domain represents the societal perspective of functioning. The theme "parental stress" was classified as an environmental factor for the patient, since parents are part of the environment they live in. "Problems during transition phase" were also considered an environmental factor since the current health-care infrastructure appeared to contribute to these problems.

The education of patients was classified according to the international classification of education (ISCED 2011) (UNESCO Institute for Statistics, 2012).

## 2.4 | Ethical approval

This study was approved by the Medical and Ethical Review Committee (MEC) of the Erasmus Medical Centre (ref. MEC-2016-532). For all participants and recordings, informed consent was obtained and a formal review and waiver of the MEC was given.

## 3 | RESULTS

### 3.1 | Participants

Forty-two participants were included; 30 patients and 12 parents (see Table 2 for patient characteristics). The age of patients ranged from 18

TABLE 2 Description of the study sample

Characteristics	Focus groups			Total
	(1) Patients 18–30	(2) Parents of patients 18–30	(3 and 4) Parents and patients 30+	
Patients (N)	12	–	18	30
Parents (N)	–	7	5	12
Age of patients in years (Median, interquartile range)	24.5 (22.3–29.5)		53.5 (36–59.5)	36.0 (27.3–55.5)
Gender of patients:				
Female N (%)	8 (67)		6 (33)	14 (47)
Male N (%)	4 (33)		12 (67)	16 (53)
Highest level of education of the patient, N (%):				
Primary-lower secondary (ISCED 1–2) <sup>a</sup>	1		3 <sup>b</sup>	4 <sup>b</sup>
Upper-postsecondary (ISCED 3–4)	8		7	15
Bachelor-university degree (ISCED 5–7)	3		7	10

<sup>a</sup>ISCED: International classification of education.

<sup>b</sup>Information on education of one patient missing.

TABLE 3 ICF categories, themes, and corresponding codes

ICF classification	Themes	Codes
Impairment of body functions and structures	Mental health problems	Anxiety, worries in many areas, symptoms of depression, problems with emotion and mood regulation, ADHD symptoms, ASD symptoms, problems coping with having NF1, <sup>a</sup> problems with self-acceptance <sup>a</sup>
	Worries about the future	Worries about future symptoms and prognosis
	Cognitive deficits	Learning disabilities, language, and speech problems
	Physical problems	Sleeping problems <sup>a</sup> , fatigue, headache, pain, growth of neurofibromas, unexplained physical complaints
	Visibility of disability	Problems with visibility of NF1, focus of others on external features <sup>a</sup>
Activity limitations & participation restriction	Limitation of independence	Difficulties adjusting life to complications of NF1 <sup>a</sup> , difficulty reaching independence, patient does not recognize and/or seek help for NF1 symptoms <sup>b</sup> , problems with planning <sup>a</sup> and organizing
	Social deficits limit participation	Lack of self-confidence, <sup>a</sup> trouble with initiating and maintaining social and romantic relationships, loneliness
	Family planning difficulties	Worries about the risk of passing NF1 to offspring, <sup>a</sup> worries about medical procedures to have children, uncertainty about obtaining a relationship
	Limitations with work	Unable to work full time, overestimation, difficulties with finding and keeping work <sup>b</sup>
Environmental factors	Required support in daily life	Support of parents, support of network, support of patient association, involvement of health care professionals in living and working arrangements
	Problems during transition process	Protracted transition, <sup>b</sup> buddy needed in transition time, no consultation between doctors, unsuccessful transition
	Limited access to adequate care in various domains	Knowledge of NF1 absent in doctors, lack of mental health care, <sup>a</sup> difficult to find an NF1 expert. <sup>b</sup> Need for one contact person instead of multiple health-care providers, <sup>a</sup> need for communication between health-care providers, need for multidisciplinary care, <sup>a</sup> need for family planning care, <sup>a</sup> support for parents, <sup>b</sup> need for more social and societal assistance
	Parental stress	No acknowledgment of worries of the parent by care providers, <sup>b</sup> much time spent on assistance and administration, <sup>b</sup> confusion about parental role, <sup>b</sup> decreased quality of parent-child relationship <sup>b</sup>

<sup>a</sup>Mainly discussed by patients.

<sup>b</sup>Mainly discussed by parents.

to 67 years, while the age of parents ranged from 54 to 75 years. The NF1-mutation status of parents was unknown. After transcription and coding, 13 major themes were identified and classified using the ICF (Table 3 provides an overview). The ICF-framework proved to be a useful tool in organizing the diversity of worries and needs ensuring an overview of both functional impairments and their consequences in daily life. Detailed results of the analysis of the transcript are provided below. The addition of new codes diminished greatly during analysis, suggesting data saturation.

## 4 | IMPAIRMENT OF BODY FUNCTIONS AND STRUCTURES

### 4.1 | Mental health problems

High levels of anxiety and stress were broadly discussed, such as fear of failure, social anxiety, and fears and worries about the future. Symptoms of depression were also discussed in all focus groups, as several patients reported needing antidepressants and psychological consultations. Both patients and parents noted problems with emotion and mood regulation, leading to anticipatory stress and frustration by family members.

Anxiety is always present, you know. Fear of new situations, and also fear of meeting new people. I see it in my son, even when he needs to go into a new store.

So every time he goes somewhere new for the first time, he wants me to join him.—Parent 18–30

Difficulties with self-acceptance and coping with the disease were widely present. Especially patients in the 30+ focus groups mentioned that it would have been helpful if psychological care had been offered. Various patients noted that the disorder of NF1 itself limited their coping skills: fatigue, anticipatory stress, and a depressed mood made them feel less resilient.

### 4.2 | Worries about the future

The unpredictable course of NF1 was a major concern of patients. Patients worried about future symptoms and prognosis, future loss of functions, future surgery, future esthetic problems, malignant transformation of neurofibromas in the future, obtaining relationships, and achieving independence.

It is just that you don't know how it will be. Really everything is worrisome.—Patient 18–30

### 4.3 | Cognitive deficits

Language and speech problems were mentioned and some adults received speech therapy. Not many cognitive deficits or learning

problems were presented as worries, although their dependence on others was often mentioned. Some parents had the impression that cognitive development stopped or slowed during the transition period because their children reached independence much later than their peers.

... That's where I have question marks. I wonder if development in those children, in young adults under age 30, if there is still progress.—Parent 18–30

#### 4.4 | Physical problems

There was great diversity in the experienced physical problems. Sleeping problems, fatigue, headache, and limited motor skills often expressed as “clumsiness” had a major impact on daily life. Fatigue and headaches had a large effect on the ability to work full time and to participate in as many activities as peers. Various sleeping problems were reported as a concern; difficulty falling asleep, difficulty staying asleep, difficulty waking up, and need for sleep medication. Furthermore, the growth of neurofibromas was mentioned as a major cause of frustration, because of possible malignant transformation and changing appearance. Symptoms mentioned by a small group of patients were pain, back pain, and scoliosis. Limited motor skills had a more indirect impact on the lives of the patients, for instance being unable to cycle or swim.

#### 4.5 | Visibility of disability

Patients noted being watched, stared at, and/or insulted in public places because of their cosmetic problems. The visibility of the disease caused questions from outsiders which were perceived as annoying, for example, questions about contagiousness.

It was hinted that I had to leave the pool because others had problems with me being in the pool.—Patient 30+

Particularly the younger patients were annoyed about NF1-portrayal in media, for example, only the worst cases being shown on television or the internet, resulting in a false perception of NF1.

There was a documentary also about (person's name), and it's always about getting the worst, very worst, the worst of all ... and showing the most severe [cases]...  
—Patient 18–30

## 5 | ACTIVITY LIMITATIONS AND PARTICIPATION RESTRICTION

### 5.1 | Limitation of independence

Parents and patients mentioned their worries about achieving independence. Parents reported that their children with NF1 lacked initiative. Young adults emphasized that achieving independence was delayed; some patients wanted to complete school before moving out

of their parent's house. Those who lived independently stated that they needed assistance in housekeeping, planning, and organizing. Patients had trouble asking for help, resulting in having to wait for people offering assistance. Additionally, patients encountered problems adjusting their lives to their limitations caused by NF1, for example, planning too many activities while at the same time having limited energy.

I'm having a hard time becoming independent, and it's very scary to separate from my parents and to do it all by myself.—Patient 18–30

Many parents remained in the coordinating role regarding medical affairs and participated in visits with medical specialists, claiming that patients did not recognize and seek help for NF1 symptoms.

### 5.2 | Social deficits

Loneliness and trouble initiating and maintaining social and romantic relationships were the main social problems. Some of the young adults stated to have had social skills training in the past. Furthermore, patients feared a lack of understanding of NF1 in new social contacts. Both patients and parents mentioned that friendships were complicated by limited understanding of the disorder in their environment.

Yes, obtaining friendships is difficult because she [my daughter] sometimes behaves a bit awkward.—Parent 18–30

Especially among patients between 18 and 30, feelings of insecurity and low self-esteem were reported. They felt that they were “different” from others and wanted to be “normal.” Many patients were still trying to cope with bullying in their past and some patients even mentioned this prior bullying “still influenced their current social skills.”

### 5.3 | Family planning difficulties

There were many worries regarding family planning. Patients appeared to be informed that they could pass NF1 to their offspring, but discussed their need for timely and adequate information and care on this issue. Some of the patients mentioned the desire to have children but felt uncertain about obtaining a relationship which would allow for that in the future.

I want children, but do not want to inflict upon them the life that I'm leading, [...] so yes, you have to go into that trajectory, but who with, and how, and adopting is an option, but then you can't give birth, and yes, I'm twenty now and I do not have a partner, and I'm getting older...—Patient 18–30

## 5.4 | Limitations with work

For cognitively high-functioning patients (ISCED levels 5–7), fatigue was a large cause of problems at work. They felt unable to work full time, but the reduction of working hours was not always allowed by their employers. Patients mentioned the need for assistance with employment matters, for example: obtaining a suitable job, obtaining a permanent contract, overestimation of abilities, lack of understanding of disabilities, and unequal treatment at work. A job coach or specific organizations were often deployed, but not always successfully. Parents indicated the need for involvement of HCPs in living and working arrangements.

Fatigue, headaches . . . there is zero understanding . . .—  
Parent 18–30

Actually, in my view, a coordinating physician should refer to rehabilitation doctors earlier, or that for the home/work situation a medical advice is provided, about how to proceed with such a child. . .—Parent 18–30

## 6 | ENVIRONMENTAL FACTORS

### 6.1 | Required support in daily life

Parents and patients all mentioned the need for support and recognition, and many of them found this in peer groups of the national patient association. Also, they appreciated people in their direct networks showing interest in NF1, both in real life and in social media networks.

. . . in the peer group, we recognize each other there, fortunately, we are very happy with the patient foundation because we feel supported.—Parent 18–30

### 6.2 | Problems during transition process

Many of the patients declared that the transition to adult health care had been hampered by a lack of communication about the transition to adult care, lack of organization of adequate follow-up, lack of consultation between physicians, and lack of referral to expert care. Often, patients were referred back to their general practitioner (GP) for adult care. Parents noted that contrary to their expectations, care needs increased after becoming 18.

### 6.3 | Limited access to adequate care

A number of patients noted that they appreciated when their GP was closely involved. Patients noted the lack of communication and consultations between physicians. Generally, it was very difficult to find physicians with knowledge and experience with NF1, and often second opinions were required. Patients from outside NF1 expertise centers were not referred to appropriate adult care by their GP's or pediatricians. Psychological support during intensive treatments (e.g., facial

surgery) was not often offered but mentioned as a care need by various patients. Parents felt that their child received inadequate care if parents were not involved.

Some physicians, they didn't know the disorder and they had to sometimes "google" during the consultation, and that I find absolutely unacceptable, personally.—  
Patient 18–30

Patients and parents indicated a need for one easily accessible contact person for NF1-related questions, surrounded by a multidisciplinary team. Also, they were in need of family planning care, social assistance and community support, and support for parents. During the focus groups, the question was raised about whether the expertise center could provide information about NF1 to employers, health insurance companies, and social security offices. These organizations have a lack of knowledge about NF1 and patients would benefit if they would have more information. Interestingly, parents noted that short-term thinking was helpful for their child, because of their slower development and the unpredictable course of NF1. As remarks about possible long-term effects of NF1 were not found to be useful and unnecessarily stressful, parents also advised professionals surrounding the NF1-patient to use "short-term thinking" with short-term recommendations.

### 6.4 | NF1-related stress in parents

Parents were particularly concerned about their children not recognizing NF1-related symptoms such as (growth of) neurofibromas, not seeking health care, not receiving proper transitional care, and losing their child to the complications of NF1. Parents worried about the slow development of independence and the vulnerability of their child (e.g., risking abuse). Many parents reported high stress and fatigue levels due to their persistent coordinating role in the lives of their children—spending much time with the planning, organization, and administration of daily life and medical issues. Some parents even acted as a legal representative. Consequently, by having the coordinating role, the question arose: "Who will take care of our child when we are no longer able to do so in the future?" Parental concerns were generally not acknowledged or addressed by HCPs or people in their environment, leading to increased stress.

. . . Listen to us, really hear us, even when it seems exaggerated.—Parent 18–30

## 7 | DISCUSSION

This study offers patient- and parent-driven data on worries and care needs of adult patients with NF1, with an emphasis on the transitional period. Worries emerged from a broad spectrum of areas, ranging from physical and mental health to areas of social and occupational participation, and transition. This enabled us to formulate recommendations for transitional and adult care.

## 7.1 | Physical health

Physical symptoms such as fatigue, headaches, sleeping problems, and pain had a large effect on daily life and were the cause of many worries and medical consultations. Most of these generally nonspecific symptoms have been reported in children, adolescents, and adults with NF1 (Afridi et al., 2015; Johnson et al., 2005; Leschziner et al., 2013; Wolters et al., 2015). Although tumor growth must be excluded, further clinical evaluation often indicates that these physical complaints may be associated not only with NF1, but also with mental health, lifestyle, or problems at work. Patients pointed out they wished to know whether these complaints were associated with NF1. Since these complaints have a great impact on quality of life, referral to psychological or occupational care is warranted to cope with these symptoms. Different levels of care may help to answer these questions regarding the role of NF1 in any of these problems. A health-care model with an NF1 expertise center, coordinating NF1-specialists, and several regional treatment centers (a "hub-and-spoke" model, the way complex NF1 services are organized in the United Kingdom) may facilitate close cooperation of GP's with a center that offers the appropriate level of care.

## 7.2 | Mental health

Although many psychological worries were reported, most participants noted a lack of routine assessment of these problems and experienced difficulties finding appropriate evaluation and treatment. Fears and worries about the future occurred very frequently on many different topics and potential growth of neurofibromas was especially worrisome to the participants. Symptoms of depression and anxiety are frequently found in patients with NF1 (Cohen et al., 2014; Pasini et al., 2012) which may be related to the risk of development of malignancies, as is the case in patients at risk for breast and colon cancer (Aktan-Collan et al., 2001; Lindberg & Wellisch, 2001). In NF1, MPNST is the most commonly found malignancy with a lifetime risk of 8–13% in NF1 (Evans et al., 2002). In the current study groups, problems with self-acceptance, low self-esteem, and limited socioeconomic participation also seemed associated with the NF1-related cosmetic burden (Vranceanu et al., 2013) and mental health problems.

Although primary concerns with cognitive deficits were not reported, limited independence and poor social skills were worries of both patients and parents, and symptoms of ADHD and ASD were broadly described. Although more elaborately documented in children with NF1, neurocognitive deficits also result in significant limitations in adults with NF1 (Lehtonen et al., 2013). These findings underline the presence and burden of cognitive deficits and their influence on mental health, activities and participation. Such difficulties can be addressed by an expert psychologist and/or intellectual disability physician (ID physician).

## 7.3 | Activities and participation

Patients expressed frustrations about their inability to function independently, limiting their socioeconomic participation. Difficulties

adjusting their life to the complications of NF1 and asking for help, problems with organizing and planning daily life, and problems with relationships and work were widespread. Poor social skills, reported in children (Barton & North, 2004) and adults with NF1 (Pride et al., 2013), affect the lives of adult patients, manifesting as problems initiating and maintaining social and romantic relationships. Also, negative reactions from others to visible differences in appearance may affect self-esteem and the forming of these social and romantic relationships (Barke et al., 2014). Participants experienced problems with finding and maintaining work, and problems with employers. During the interviews, inadequate socioeconomic participation and loneliness appeared to be associated with psychological complaints—which has been reported before in teenagers and young adults with NF1 (Ejerskov et al., 2015; Hummelvoll & Antonsen, 2013).

Family planning was an important theme, especially for young adults, which is in line with previous research (Crawford et al., 2015). Patients stated that they would want to have children without NF1 and that additional and timely education about genetics and reproduction was needed, although patients could not agree on the age at which this information should be given. This indicates that HCPs should make patient-specific decisions on the appropriate timing and level of detail of this information.

## 7.4 | Environmental factors

Generally, adult patients and parents seemed to be well informed about NF1, but they often noticed a lack of knowledge in people in their direct environment. Both patients and parents experienced limitations in their environment, such as lack of appropriate health care, lack of multidisciplinary care, lack of family care, and inadequate support from family, social networks, and employers.

## 7.5 | Parental stress

Parents of patients have previously participated in research to elucidate the impact of NF1 on their children (Barke et al., 2016), yet the impact on parents themselves has not been described yet. In our study, parents of young adults reported many worries, high stress levels, and little attention from HCPs for their complex parental role. Since the GP is often well informed about family issues, she/he may have a crucial role in referring to the appropriate type of support and mental health care in the community.

Parental stress has been described in other pediatric cohorts with neurodevelopmental syndromes or autism (Briegel et al., 2008; Hartley et al., 2012; Pozo & Sarria, 2015). In children with NF1, mothers reported higher parenting stress than the mothers of typically developing children (Esposito et al., 2014). The uncertainty of tumor progression may increase anxiety in both patients and parents of patients with NF1. In future research, the contributing factors to parental stress should be explored more fully, to identify parents at risk and to provide targets for prevention and treatment. In addition to parental stress, future research should also address the stress of other family members with NF1.

## 7.6 | Implications for transitional care

The transitional period from pediatric to adult care is known to be a challenging phase for patients with NF1 and their parents (Oates et al., 2013; Van Lierde et al., 2013), which was confirmed by our participants. For many patients, this transition did not proceed well. Although expert adult care for the tumor phenotype was often accessible, finding NF1 experts in other domains was difficult. The neuropsychological deficits that frequently occur in patients with NF1 may decrease their self-care and organizational capabilities, which puts them at risk for problems during the transition to adult care (Van Lierde et al., 2013). At this age, as is also the case in our sample, differences between parent- and patient-reported worries underline that both parents and young adults should be involved in the transitional process.

Previous studies on the transitional stage in patients with special needs, such as intellectual disabilities or chronic disorders, have shown that many adolescents experience significant problems with the continuity of care during the transition to adult health care (Bindels-de Heus et al., 2013; Fegran et al., 2014; Reiss et al., 2005). Often, after years of periodic checkups by the pediatrician before adolescence, patients discontinued clinical evaluations or did not have contact with any health-care professionals (Lotstein et al., 2009; Oates et al., 2013). In our study, adult patients expressed a need for well-informed, accessible, and multidisciplinary NF1 expertise in close communication with the patient's local network and care providers. Continuous "chronic" care before, during, and after transition (NICE, 2016) could be facilitated if these specialists could also be "generalists" in adult care for people with neurogenetic disorders in the way pediatricians are in pediatric care (Schor, 2015).

## 7.7 | Implications for adult care

From the worries mentioned above, conclusions can be drawn regarding the organization and the content of care provisions. These findings expand on current recommendations for the management of NF1 (Ferner et al., 2007). Suggestions for clinical practice are listed in Table 4: "Addressing health care needs during NF1 consultation."

In all groups, in addition to a local network of health-care professionals including a GP, a desire for multidisciplinary expertise on adults with NF1 was expressed, to obtain treatment and information on all domains. As our results suggest, this expertise team should communicate closely with local HCPs and would be responsible for diagnosis, timely follow-up, second opinions, education, and assistance for the patients, parents, and even the socioeconomic network. Screening and follow-up for mental health problems should be included in routine follow-up, as this patient group is at risk for psychiatric morbidity (Cohen et al., 2014; Descheemaeker et al., 2013; Mautner et al., 2015; Pasini et al., 2012; Pride et al., 2013). In practice, this implicates that individuals with NF1 should be offered structured care, including an accessible leading NF1-expert team who routinely screens for physical and mental health and for limitations in participation in social, occupational, or other meaningful daily activities. Prevention and early intervention of mental health problems can be efficient and cost-effective (WHO, 2016) and will ultimately improve socioeconomic participation and quality of life.

Since this study offered us a clear view of worries and care needs within patients with NF1 and parents of patients with NF1, comparisons across different genetic neurodevelopmental syndromes might help to find common factors and to contrast these findings to neurocognitive, neurobehavioral, and environmental factors.

The organization of multidisciplinary and multilevel care requires a method for severity assessment that includes the full impact of NF1 on all ICF domains, including limitations in activities and participation. Although the Riccardi scale for disease severity (Riccardi, 1992) has frequently been used to assess impairments (Upadhyaya & David, 2012; van der Vaart et al., 2013; Wolkenstein et al., 2001), many worries and health-care needs of our respondents are not addressed by this scale, since it mainly focuses on disease characteristics at a somatic level. This suggests there is a need for assessing the full impact of NF1 on all ICF domains, including limitations in activities and participation. In Tuberous Sclerosis Complex, another neurocognitive and tumor predisposition disorder, an attempt has been made to capture the burden of the disease by introducing the TAND checklist, encompassing "Tuberous sclerosis-associated neuropsychiatric disorders." This could serve as an inspiration for such initiatives in the field of NF1 (de Vries et al., 2015).

## 7.8 | Strengths and limitations

In a qualitative observational study, the subjects that are to be discussed can be standardized, but conversations themselves cannot be standardized. Given the sample size and the nature of this study, these differences cannot be explored in a quantitative way. Future studies with larger sample sizes and a quantitative may focus on systematic differences between age groups, genders, or between parents and patients.

Using focus groups as a means to explore worries and needs has the possible drawback of participants feeling limited in their ability to express feelings or concerns in the presence of other patients or parent. In our focus groups, the atmosphere was quite open and stimulating, and almost all participants actively joined the conversation. However, problems with more intimate matters such as sexuality, addiction, suicidal thoughts, and finance were hardly reported.

All patients who participated in this study were members of the Dutch NF1 patient organization (NFVN). For this reason, our sample may be biased toward relatively well-informed patients with a higher level of education, and parents and patients may also have a more severe phenotype. The Netherlands is a high-income country in Western Europe. This should be taken into consideration when extrapolating our results and recommendations to other societies where limitations in care infrastructure, expertise, and finances may be more prevalent.

Since a large part of the participants were adults over 30 years old, their reporting about the transitional period could have suffered from "recall bias." For some of them, this period was quite some time ago. Health-care services probably may have changed in the past years. However, we do feel that the recommendations in this article directly result from the information of both younger and older participants.

The apparent data saturation during analysis suggests sufficient sample size and data quality. Other strengths of the study were the



TABLE 4 Improving clinical practice: Addressing health-care needs during NF1 consultation

Care needs	Screen patient for	Provide
Follow-up by NF1-experts	Complexity and comorbidity. Local network of health-care providers.	Easily accessible NF1 expert. Multidisciplinary NF1 expertise center for periodic screening of children, adolescents, and adults. Close communication between GP, care professionals, and NF1-experts.
Information and education on tumor phenotype and prognosis	Tumor phenotype according to current guidelines, including tumor growth, pain, loss of function. Knowledge of tumor-related symptoms, prognosis.	Regular monitoring of tumor-related symptoms. Patient information and education. Referral to patient association, websites, and brochures.
Other physical symptoms	Fatigue, sleeping problems, headache, medication side effects. Somatic comorbidity.	Exclude tumor growth; identify appropriate care in collaboration with GP. Inform about NF1-related complaints. Psychomotor therapies. (Neuro)psychological evaluation and treatment.
Mental health problems	Worries, social problems, isolation, feelings of loneliness, depressive symptoms including suicidal thoughts, ADHD, ASD.	Referral to community support by GP. (Neuro-) psychological and psychiatric evaluation and treatment, peer groups, social worker. <sup>a</sup>
Social participation Economic participation Daily life	Involvement of friends and family, social activities, romantic relationships, support network. Suitable work or daytime occupation. Independence, needs for assistance, financial space. Planning and organizing daily life, self-care, chores.	Evaluation of social and occupational skills. Social worker. <sup>a</sup> Information and advice for employers, insurance companies, and social services. Occupational medicine, occupational therapy.
Family planning	Knowledge of birth control methods, inheritance, family planning options, preconception consultation.	Inform GP, refer to clinical geneticist, gynecologist, psychologist.
Need for information, education, and support for patients, parents, siblings, partners, GP's, employers, and network	Knowledge of symptoms, care infrastructure, prognosis.	Accessibility for consultation. Patient education. Care guidelines for GP. Information brochures for friends/family, teachers, employers. Communication during transitional phase. Information or guidelines for care providers, periodic letters with advice.
Reduction parental stress, especially in parents of young adults	Screen both parent/caregiver and patient, if necessary in separate consultations.	Address parental concerns. Increase support for patient in daily life to alleviate parental responsibilities. Support for parent through GP, social services or psychologist.
Continuity of care during transitional period	Screen for all care domains starting at age 14.	Close communication between NF1 HCP, GP, and social worker until appropriate work, living, and medical care arrangements are in place.

<sup>a</sup>Social worker is used here, but may be substituted or supported by ambulatory help, informed volunteers or other local care where available.

large age range, the range in level of education, and reports from both patients and parents. The ICF turned out to be an appropriate method to describe patient-related problems in almost all areas of life. The universal nature of the ICF and the apparent shortcomings of Dutch health care make our findings applicable to other countries where multidisciplinary care is not routinely offered.

## 8 | CONCLUSION

This qualitative study offers a unique perspective on the worries and health-care needs of adult patients with NF1 and their parents. The care needs of NF1-patients and parents (during and after transitional age) are diverse and need a multidisciplinary approach with systematic evaluation of tumor growth, neuropsychiatric symptoms, and

socioeconomic limitations. Care infrastructure for adults should include local health-care providers with experience in NF1 supported by an accessible multidisciplinary NF1-expertise center. These different levels of care could also address the need for information about the association of NF1 with physical and mental problems, which could greatly decrease the uncertainty both patients and parents need to live with. Addressing physical health, mental health, and socioeconomic participation during regular consultations may improve health care and consequently the quality of life of people with neurodevelopmental disorders.

## ACKNOWLEDGMENTS

We would like to thank all the participants, their family members, and the Dutch patient's organization (Neurofibromatosis Vereniging

Nederland) for their participation and support in this study. We are grateful to Alanna Jacobs, who helped to edit the final version of the manuscript.

## CONFLICT OF INTEREST

None.

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**How to cite this article:** Rietman AB, van Helden H, Both PH, et al. Worries and needs of adults and parents of adults with neurofibromatosis type 1. *Am J Med Genet Part A*. 2018;176A:1150–1160. <https://doi.org/10.1002/ajmg.a.38680>