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KNOWLEDGE AND SELF-ESTEEM IN INDIVIDUALS WITH NEUROFIBROMATOSIS TYPE 1 (NF1)

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A

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KNOWLEDGE AND SELF-ESTEEM IN INDIVIDUALS WITH NEUROFIBROMATOSIS TYPE I (NF1)

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Neurofibromatosis Type 1 (NF1) is a progressive genetic disorder characterized mainly by café-au-lait macules, Lisch nodules, as well as cutaneous and subcutaneous neurofibromas among other traits. Due to the physical manifestations of the condition, it has been observed that individuals with NF1 have lower self-esteem (SE) when compared to the general population. Additionally, a study published over 20 years ago found that overall knowledge of NF1 was poor in individuals affected with the condition. The goal of our study was to reassess knowledge in this population and investigate whether it is related to SE. A survey comprised of knowledge-based questions and the Rosenberg Self-Esteem Scale was distributed to individuals with NF1 through the Texas NF Foundation. Overall, the 49 respondents (13 to 73 years old) had higher than expected knowledge of NF1 (mean score = 77.9% correct answers) across various aspects of the condition. Consistent with previous studies, the SE of our study population was lower compared to general population norms. Although there was no correlation detected between knowledge and SE, SE scores were on average higher if a person reported to have friends with NF1, attended an NF1 support group, attended a NF clinic, or received genetic counseling. Having friendships with people who have the same condition as well as attending support groups may help those affected by NF1 to feel less isolated. Additionally, genetic counseling provides an opportunity for these individuals to potentially identify ways to improve coping through positive coping strategies such as educating peers about NF1.

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Introduction

Neurofibromatosis Type I (NF1) is a progressive genetic disorder caused by a mutation in the tumor suppressor gene, *NF1*. It affects around 1 in 2500 to 3000 individuals. Clinical manifestations of NF1 include multiple café au lait macules, axillary and inguinal freckling, neurofibromas, and Lisch nodules. Additionally, individuals can have plexiform neurofibromas, optic gliomas affecting vision, malignant cancers, skeletal problems, and hypertension (Williams et al., 2009). Children and adolescents can also experience learning and peer problems at school as a result of their diagnosis (Krab et al., 2008). Approximately 35% to 65% of patients with NF1 have learning difficulties, which include deficits in reading, mathematics, language, and memory (Levine, Materek, Abel, O'Donnell, & Cutting, 2006). Approximately 6-7% of people with NF1 have an IQ score under 70 and are considered to have mild intellectual disability (Lehtonen, Howie, Trump, & Huson, 2013). In addition, 50% of people with NF1 have attention-deficit hyperactivity disorder (ADHD) (Levine et al.).

The negative effects of NF1on cognitive abilities and physical appearance have motivated researchers to assess the quality of life (QoL) in affected children and adults. One such study with an adult French cohort investigated QoL through the use of a general health questionnaire (Short Form 36, SF-36) and a skin disease-specific questionnaire (Skindex). They found that the emotional consequences such as depression, fear, embarrassment, and anger about the condition were the most prevalent negative outcomes in affected individuals. Furthermore, there was a greater negative impact on QoL in people with a more visible form of the disorder or in those with more severe medical problems related to NF1 (Wolkenstein, Zeller, Revus, Ecosse, & Leplege, 2001; Chren, 2012). These same measures (SF-36 and Skindex) were also applied in an American cohort with similar results; however, they found that women were more likely to

experience emotional effects of the condition than men (Page et al., 2006). Similarly, a study revealed that children and adolescents with NF1 have impaired QoL, affecting areas of motor, cognitive, and social functioning (Graf, Landolt, Mori, & Boltshauser, 2006). Another study looking at emotional functioning of patients with neurofibromatosis (including individuals with NF1, NF2, or schwannomatosis) found that these individuals were more likely to experience depression and anxiety because of their diagnosis. Additionally, the study population was found to have lower self-esteem on average, when compared to the general population (Wang et al., 2012).

Although there have been numerous studies on QoL of individuals with NF1, Benjamin et al. (1993) is the only study that has investigated level of knowledge of NF1 in those with the condition. They found that there was an overall poor understanding of the condition in individuals with NF1. In fact, knowledge seemed to be reflective of, and even limited to, the patient's personal and/or family's experience with NF1. Factors associated with having a greater level of knowledge of the disorder were: having received genetic counseling, belonging to a higher social class, being diagnosed at a young age, being a member of a support group, having a child with NF1, and when NF1 had influenced reproductive decisions (Benjamin et al., 1993).

The current study reassessed knowledge of NF1 in patients with the condition and measured their self-esteem using the Rosenberg Self-Esteem Scale (RSES). Correlations between knowledge scores and self-esteem levels were investigated to evaluate associations between having a good understanding of the condition and the potential effect on self-esteem. Additionally, other factors, such as support group involvement, or a family history of NF1 were also explored, to determine if there were any correlations with knowledge and/or self-esteem.

Methodology

Participants

Approval for this study was received by the Committee for the Protection of Human Subjects (CPHS)—The University of Texas Health Science Center at Houston Institutional Review Board (IRB Number: HSC-GEN-14-0596).

The study population consisted of individuals aged 13 years and older with a clinical and/or molecular diagnosis of NF1. The participants were accessed through the Texas NF Foundation, either through their electronic mail (email) database, Facebook group, or at a foundation-sponsored event.

Instrumentation

There was an online and paper version of the questionnaire. The online version was created using REDCap software version 5.9.11. The questionnaire had three main sections: (1) demographic questions, as well as questions focusing on the respondents' experience with NF1, (2) knowledge of NF1, and (3) the Rosenberg Self-Esteem Scale (RSES). The demographic information collected included: gender, current age, age of diagnosis, ethnicity, educational background, and employment status. In the knowledge section, respondents were asked to: 1) identify symptoms of NF1 from those of tuberous sclerosis complex (TSC), 2) answer true and false questions regarding the variability and progressiveness of the condition, and 3) answer multiple choice questions about the genetics, recurrence risk, incidence, and management of people with NF1. Section 3 of the survey consisted of the Rosenberg Self-Esteem Scale (RSES), which is a validated tool that consists of 10 questions that evaluates a person's self-esteem. Each question is graded on a Likert scale with total scores ranging from 0 to 30. Higher scores indicate

better self-esteem, while scores less than 15 are associated with low self-esteem and scores between 15 and 25 with average or normal level self-esteem (Rosenberg, 1989).

Procedures

The Texas NF Foundation sent out an email to all their members (approximately 1600) on November 4, 2014 to invite them to participate in the survey. The email consisted of an invitation letter to participate in the survey as well as a link to the survey. Two reminder emails were sent out in December and January. The survey was available for the participants to complete for approximately three months until February 1, 2015 when data collection ended. The database only included email addresses of adults (over the age of 18 years) with NF1 and the parents of individuals under the age of 18 with NF1. A link to the survey was also posted on the Facebook page of the Texas NF Foundation. Additionally, hardcopy questionnaires were distributed at a foundation-sponsored holiday event, Cookies with Santa, on December 7, 2014.

Parental consent was obtained for participants under the age of 18 years. For the online version of the questionnaire, parents were prompted to read the consent form and decide whether they wanted their child to participate in the study. If the parent agreed, then the child was instructed to read the consent form and indicate whether or not he/she wanted to participate. Adults with NF1 indicated consent by agreeing to opt-in to the study prior to beginning the on-line questionnaire. For the hardcopy version, individuals were asked to read an informed consent document and then provide their signature if they agreed to participate.

Data Analysis

Knowledge scores were calculated by counting each correct answer as one point. The total number of knowledge questions was 35; therefore the highest possible score was a 35/35. The knowledge score was reported as a percentage correct for each participant. Self-esteem

scores were calculated based on the Rosenberg scale methodology (Rosenberg, 1989). Pearson's correlation coefficients were calculated to assess the relationship between the continuous variables for knowledge and self-esteem. A two-sample unpaired *t*-test was used to examine potential difference in knowledge score between those in the "low" self-esteem group and those in the "average or above-average" self-esteem group. Two-sample unpaired *t*-tests were also used to examine potential relationships between one's background or experience with NF1 and that person's level of knowledge or self-esteem. A Mann-Whitney test was used to compare the participant's knowledge score to whether or not they thought they would be able to explain NF1 to a friend. Two-sample tests of proportions were used to compare the knowledge results of the current study to the knowledge results of Benjamin et al. Multivariable linear regression models were fitted to assess the independent effect of various factors on knowledge and self-esteem. All analyses were performed using STATA (v.13.0, College Station, TX). Statistical significance was assumed at a Type I error rate of 0.05.

Results

Study Participants

A total of 115 partial or complete responses were collected through REDCap and 2 complete responses were collected at the Cookies with Santa event. Of the 115 online responses, 48 (42%) respondents had completed all of the relevant sections necessary for inclusion in analysis. One respondent was under the age of 13 and did not meet eligibility criteria and was therefore excluded. The final study population comprised of 49 participants.

The participants' ages ranged from 13 to 73, with four under the age of 18. The mean age of respondents was 39.2 years (SD: 16.9). While 42 (86%) of the participants indicated that they knew the age at which they were diagnosed with NF1, only 39 of those (93%) provided the age. The median age of diagnosis was 4 years (range <1 year through 34 years).

The majority of the participants identified themselves as non-Hispanic white (61%) followed by Hispanic white at 20%. Approximately two-thirds of the study population had completed graduate school or professional school, college, or some college. Complete demographic information is summarized in **Table 1**.

A large proportion of the participants (n=37, 76%) reported problems with learning in school, with 32% of those individuals requiring special education classes. Additionally, 57% of the study population reported difficulty paying attention and 23% reported an official diagnosis of Attention-Deficit Hyperactivity Disorder (ADHD) (**Table 2**). Participants who reported having learning problems were found to have lower self-esteem (p=0.023) (**Table 3**).

Table 1 Demographics

Mean age (*N*=49)	39 years
Median age of diagnosis (*N*=39)	4 years
Gender (*N*=49)	*n* (%)
Male	17 (35)
Female	32 (65)
Ethnicity (*N*=49)	*n* (%)
Non-Hispanic white	30 (61)
Hispanic white	10 (21)
Hispanic/Pacific Islander	3 (6)
Black or African American	2 (4)
Other	2 (4)
No answer	2 (4)
Education (*N*=49)	*n* (%)
Still in middle or high school	5 (10)
Did not graduate from high school	2 (4)
Graduated high school or GED	7 (14)
In or graduated from a technical school	1 (2)
Some college	14 (31)
Graduated college (undergraduate degree)	14 (29)
Graduate school or professional school	4 (8)
No answer	1 (2)

Table 2 Learning and Attention of Participants (*N* =49)

Problems learning in school	*n* (%)
Yes	37 (76)
No	10 (20)
Don't know/don't remember	2 (4)
Special education classes	*n* (%)
Yes	12 (25)
No	36 (73)
Don't know/don't remember	1 (2)
Attention problems	*n* (%)
Yes	28 (57)
No	21 (43)
Diagnosis of ADHD	*n* (%)
Yes	11 (22)
No	36 (74)
Don't know	2 (4)

Table 3 Factors Influencing Self-Esteem

	Mean Self-Esteem Score (Range 0-30)±SD	*P*-value
Do you or did you have problems learning in school?		
Yes (*n*=37)	18.2±6.7	**0.023**
No (*n*=10)	23.9±5.3	
Don't know/Don't remember (*n*=2)	n/a	n/a
Do you have any friends with NF1?		
Yes (*n*=13)	23.4±5.4	**0.009**
No (*n*=31)	17.7±6.7	
Don't know (*n*=2)	n/a	n/a
Incomplete self-esteem score (*n*=3)	n/a	n/a
Have you ever been to a gathering or support group for NF?		
Yes (*n*=28)	21.3±5.5	**0.006**
No (*n*=18)	15.7±7.3	
Incomplete self-esteem score (*n*=3)	n/a	n/a
Have you ever been to a NF clinic (a clinic that specializes in treating people with NF)?		
Yes (*n*=23)	20.8±6.5	**0.049**
No (*n*=21)	16.8±6.6	
Don't know (*n*=2)	n/a	n/a
Incomplete self-esteem score (*n*=3)	n/a	n/a
Have you ever had genetic counseling/seen a genetic counselor?		
Yes (*n*=16)	22.4±6.5	**0.008**
No (*n*=28)	16.9±6.2	
Don't know (*n*=2)	n/a	n/a
Incomplete self-esteem score (*n*=3)	n/a	n/a

Survey Responses

Self-Perceived Severity

Participants were asked questions regarding self-perceived severity of NF1. The majority (84%) believed that they had a less severe form of the NF1 compared to others with the condition. While a large proportion (71%) reported NF1 affecting their physical appearance, overall, respondents were not embarrassed to go out in public because of their diagnosis (78%).

About half of the study population reported that they had health problems due to their NF1 diagnosis.

Experience with NF1

Participants were asked questions regarding their experience with NF1. Of the 49 respondents, 27% had inherited the condition from a parent (**Table 4**) and overall, 37% of respondents reported having a first degree relative with NF1. Among sources of support and experience with different healthcare professionals, 61% percent reported attending a support group for NF1, 49% had received care at an NF-specific clinic, and 33% had seen a genetic counselor or had genetic counseling (**Table 5**). Respondents were also asked questions regarding peer relationships and the importance of knowledge; approximately two-thirds of the study population did not have trouble making friends because of their diagnosis and almost the entire group (98%) believed that knowing information about NF1 was important (**Table 6)**.

There were several factors associated with higher self-esteem scores: having friends with NF1 (p=0.009), attending a support group (p=0.006), attending an NF clinic for care (p = 0.049), and receiving genetic counseling (p=0.008) (**Table 3**). In addition, individuals who did not think that they would be able to explain NF1 to a friend had a lower median knowledge score (p=0.007) and individuals who desired to know more about NF1 also had a lower knowledge score on average (p=0.043) (**Table 7**).

Multivariable linear regression models were fitted to determine the independent effects of having learning problems, having friends with NF1, attending a support group, attending an NF clinic, and receiving genetic counseling after adjusting for all the other factors. Based on the regression models, on average, self-esteem scores increased independently by 4.8, 4.7, 4.0, and 4.1 units in the absence of learning problems (95% CI: 1.4 - 8.3), having friends with NF1 (95%

CI: 0.7-8.7), attending a support group (95% CI: 0.2 - 7.8) or receiving genetic counseling (95% CI: 0.3 - 7.8), respectively. Although attending a multidisciplinary clinic was significantly associated with self-esteem in univariable analyses, this factor was not significantly associated in the multivariable models.

Table 4 Experience with NF1: First Degree Relatives

	Yes *n* (%)	No *n* (%)	Don't know *n* (%)	No answer *n* (%)
Do you have a parent with NF1?	13 (27)	32 (65)	4 (8)	0
Do you have brothers or sisters with NF1?	4 (8)	42 (86)	3 (6)	0
Do you have a child with NF1?	9 (18)	39 (80)	1 (2)	0

Table 5 Experience with NF1: Support and Healthcare

	Yes *n* (%)	No *n* (%)	Don't know *n* (%)	No answer *n* (%)
Do you have any friends with NF1?	14 (29)	33 (67)	2(4)	0
Have you ever been to a gathering/support group for NF?	30 (61)	19 (39)	0	0
Have you ever had counseling/seen a therapist because of having NF1?	8 (16)	40 (82)	1 (2)	0
Have you ever had neuropsychological testing (tests that measure intellectual ability)?	16 (33)	29 (59)	3 (6)	1 (2)
Have you ever been to a NF clinic (a clinic that specializes in treating people with NF)?	24 (49)	23 (47)	2 (4)	0
Have you ever had genetic counseling/seen a genetic counselor?	16 (33)	31 (63)	1 (2)	1 (2)

Table 6 Experience with NF1: Peer Relationships and Knowledge

	Yes *n* (%)	No *n* (%)	Don't know *n* (%)	No answer *n* (%)
Have you ever been bullied because you have NF1?	22 (45)	22 (45)	5 (10)	0
Do you think you have trouble making friends because of having NF1?	12 (25)	34 (69)	3 (6)	0
Do you think that your friends and other people understand what NF1 is?	9 (18)	37 (76)	3 (6)	0
Do you think you would be able to explain NF1 to a friend?	72 (86)	5 (10)	2 (4)	0
Do you wish you knew more about NF1 so that you could explain it better to people?	34 (69)	14 (29)	1 (2)	0
Do you think it's important to know information about NF1?	48 (98)	1 (2)	0	0

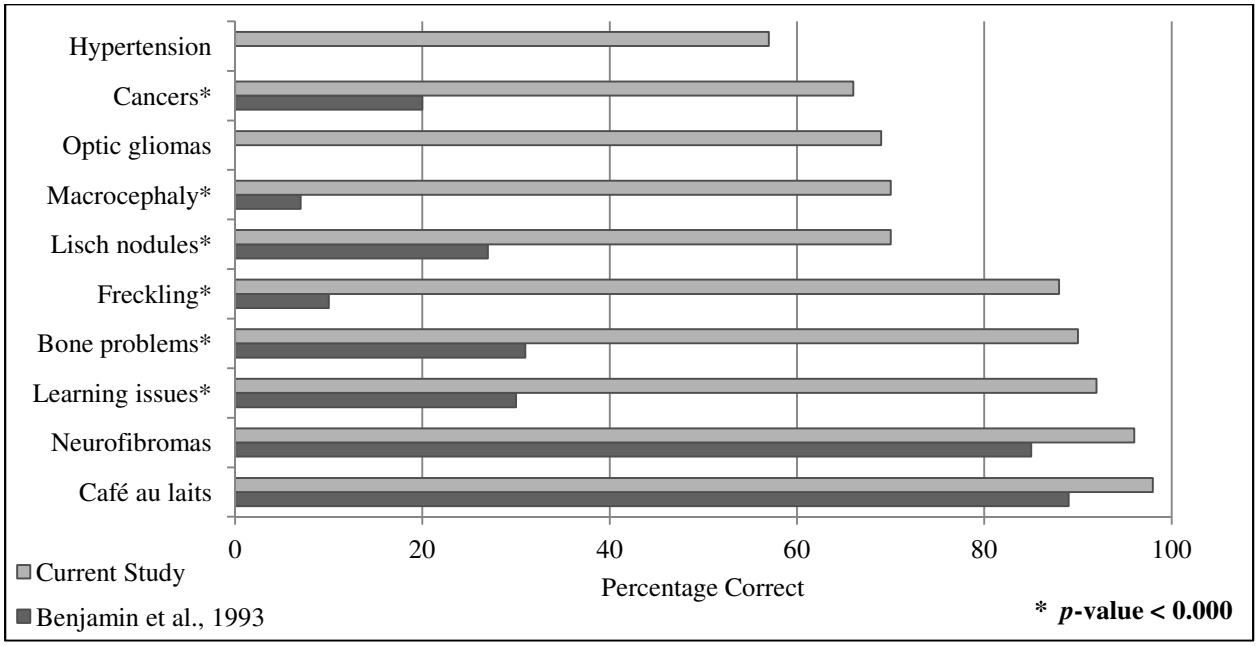
Table 7 Factors Influencing Knowledge Scores

	Median Knowledge Score (%)	*P*-value
Do you think you would be able to explain NF1 to a friend?		
Yes (*n*=42)	80.0	**0.0066**
No (*n*=5)	68.6	
Don't know (*n*=2)	n/a	n/a
	Mean Knowledge Score (%)±SD	
Do you wish you knew more about NF1 so that you could explain it better to people?		
Yes (*n*=34)	76.4±7.6	**0.0428**
No (*n*=14)	82.0±10.7	
Don't know (*n*=1)	n/a	n/a

Knowledge

Participants were asked to identify the features associated with NF1. Overall, correct identification of the features was high with several features correctly identified over 80% of the time, such as, café au lait macules, neurofibromas, learning problems, freckling in axillary and inguinal regions, and bone problems. Compared to Benjamin et al. (1993), we saw a statistically significant improvement in knowledge of the following features: cancers, macrocephaly, Lisch nodules, freckling, bone problems, and learning issues (**Figure 1**).

Figure 1: Features of NF1



Participants were asked a series of true and false questions. Ninety-eight percent of the study population correctly understood that it is not their fault that they have NF1. There was also high knowledge of the variability of the condition, with 93.9% understanding that NF1 could vary within and between families. The more severe outcomes of NF1, such as cancers and blindness, had the lowest correct scores (**Table 8**).

Table 8 True and False Section

Question	*Correct answer*	*Correct responses (%)*
It is a person's fault if they have NF1.	False	98
If multiple family members have NF1, then they will all have the same amount of lumps and brown marks and will have the same health problems.	False	94
Often, people with NF1 from different families will have different symptoms.	True	94
Brown marks (café au laits) are often the first sign that a person has NF1.	True	86
NF1 physical features get better as a person gets older.	False	82
Half of patients with NF1 have a family history of NF1.	True	78
A person with NF1 can develop cancer that causes them to die.	True	67
A person with NF1 can develop a cancer that causes them to be blind.	True	59

There were several other questions covering additional aspects of the condition. Ninety-six percent of individuals knew that NF1 was caused by a mutation in a gene, although only 61.2% knew the function of the *NF1* gene. Additionally, 90% chose the correct recurrence risk and 76% understood that the recurrence risk is the same for each pregnancy. Complete results of the multiple-choice section can be found in **Table 9.** Compared to Benjamin et al. (1993), we observed a significant increase in knowledge of intrafamilial and interfamilial variability and recurrence risk of the condition (**Figure 2**). Overall, we observed good understanding across the majority of the knowledge section, with an average score of 77.9% (SD: 8.8) correct answers.

Table 9 Multiple Choice Section

What causes NF1?	*n* (%)
A mutation in a gene	47 (96)
An infection	0
Being in the sun too much	0
Having an allergic reaction to food?	0
Don't know	2 (4)

If a person with NF1 has a child, what is the chance that the child will have NF1?	
0% chance	0
10% chance	1 (2)
50% chance	44 (90)
100% chance	1 (2)
Don't know	3 (6)

If a person with NF1 has a child with NF1, what is the chance that a second child will have the condition?	
The chance is much higher to have another child with NF1	3 (6)
The chance is much lower to have another child with NF1	2 (4)
The chance to have another child with NF1 does not change	37 (76)
Don't know	7 (14)

About how many people have NF1?	
1 in 300	2 (4)
1 in 1,000	0
1 in 3,000	35 (71)
1 in 30,000	4 (8)
Don't know	8 (16)

This is a cure for NF1.	
Surgery	0
Taking certain medicine	0
There is no cure	48 (98)
Don't know	1 (2)

What group of people is most affected by NF?	*n* (%)
Asian	0
Black/African American	0
White/Caucasian	4 (8)
NF1 affects all groups of people equally	40 (82)
Don't know	5 (10)

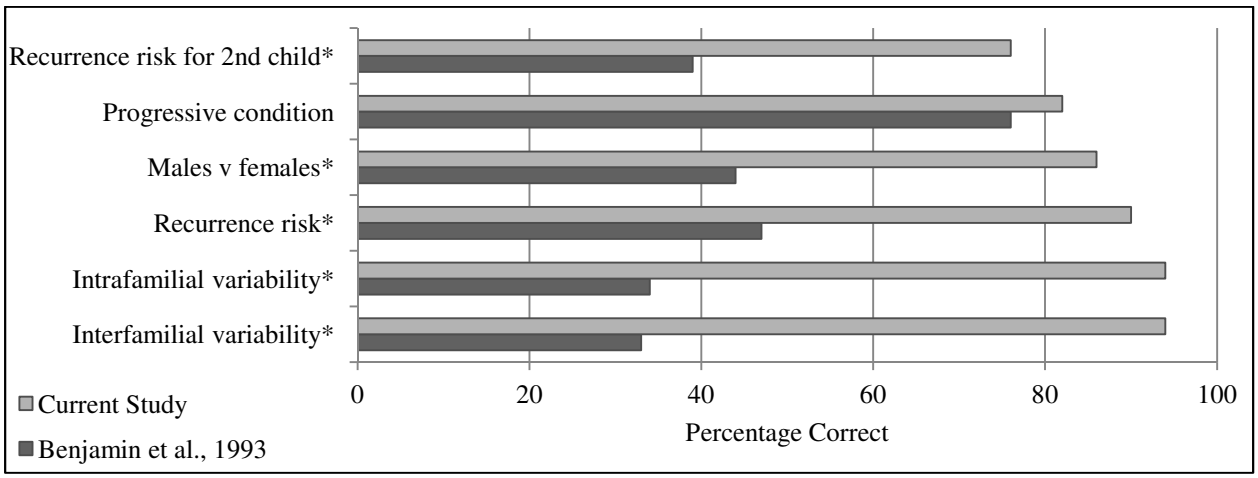
Are men or women more affected by NF?	
Men	1 (2)
Women	1 (2)
NF1 affects both men and women equally	42 (86)
Don't know	5 (10)

A person diagnosed with NF1 usually has this/these test(s) done:	
A skin exam	3 (6)
Brain imaging (MRI)	2 (4)
Eye exam	0
All of the above	44 (90)

What function does the NF1 gene have?	
Determines a person's eye color	0
Determines if a person will be a good athlete	0
Helps the body break down sugar	1 (2)
Keeps tumors from growing in the body	30 (61)
Don't know	18 (37)

How can NF1 be diagnosed?	
If a patient has enough features of NF1 (clinical diagnosis)	9 (18)
With genetic testing	2 (4)
Both a and b	38 (78)
Don't know	0

Figure 2: Knowledge of Recurrence Risk, Variability, and Progressiveness



Rosenberg Self-Esteem Scale

Complete self-esteem scores were collected for 46 (93.9%) of the participants. Of these individuals, 21.7% had low self-esteem scores (<15) with an average score of 9.5, 56.6% had average/normal self-esteem scores (15-25) with an average score of 19.1, and 21.7% had above-average self-esteem scores (>25) with an average score of 28.6. The average self-esteem score of the study population was lower compared to general population norms (p=0.0001) (Sinclair, Blais, Gansler, Sandberg, Bistis, & LoCicero, 2010). The study population was also stratified by gender and compared to the general population norms; both males and females had lower average self-esteem scores compared to the general population (p=0.0432 and p=0.0006, respectively) (**Table 10**).

Table 10 Comparison of Self-Esteem Scores to General Population Norms

	Study Population		**General Population***		*t* value	*P*- value
	n	Mean ±SD	*n*	Mean ±SD		
Overall	46	19.09 ±6.77	503	22.62 ±5.80	3.89	**0.0001**
Gender						
Male	17	19.29 ±5.31	242	22.43 ±6.21	2.03	**0.0432**
Female	29	18.97 ±7.58	261	22.79 ±5.41	3.45	**0.0006**

*General population norms taken from Sinclair, Blais, Gansler, Sandberg, Bistis, & LoCicero, 2010

Discussion

Knowledge of NF1

This study aimed to assess knowledge about NF1 and self-esteem in individuals with NF1. We identified changing trends in the knowledge of individuals with NF1 and identified other factors associated with knowledge and self-esteem in these individuals.

Compared to the Benjamin study done in 1993, knowledge of NF1 seems to have improved in individuals with the condition. Correct identification of features was high in the current study and showed improvements from the previous study. For example, 90% of participants correctly identified freckling in the inguinal and axillary areas as a feature of NF1 compared to only 10% in the previous study (Benjamin et al., 1993). Additionally, knowledge of recurrence risk was higher with 88% of our study population identifying the correct recurrence risk of 50% in a multiple choice question. We used true and false questions to ask about the variability of the condition and found a stark difference between the two studies, with 94% of the respondents in the present study understanding that NF1 could vary within and between families compared to the previous study which showed that only 33% knew it could vary between families and 34% knew it could vary within a family (Benjamin et al., 1993). Of note, the previous study used an interview-based questionnaire in which the majority of the questions were open-ended.

There are several possible reasons for this apparent increase in knowledge of NF1. First, there are more patient-friendly educational materials available today compared to 20 years ago. For example, in 2005 the Texas NF Foundation (TNFF) published an educational storybook, *14 Stories*, for newly diagnosed patients and their families (http://texasnf.org/). Second, there has been improved availability of the internet over the last couple of decades, making access to

information about NF1 easier. Third, there is likely greater patient awareness of the condition through membership in support group organizations like the TNFF. The TNFF mails out packets of information if requested and directs members to reliable online resources including their web page. The organization's educational goal is to get accurate information about NF1 into the hands of their members (C. Hahn, personal communication, April 3, 2015). The TNFF has ample resources for families affected by NF available on their website and membership in the organization is not required to access this information (http://texasnf.org/). In addition, in the phenomenon of "patient activation," or patients playing a more active role in their healthcare, which requires knowledge of one's condition, could be in effect. This may be influenced in part by the patient's relationship with his or her physician. Traditionally, the physician had a more paternalistic role, while the patient was more passive about their healthcare. However, there has been a shift to a more collaborative relationship between the patient and the physician (Alexander, Hearld, Mittler, & Harvey, 2012).

Lower knowledge scores were seen in participants who reported they would have difficulty explaining NF1 to a friend and in those who desired to gain more knowledge about NF1 so that they could explain it better to others. These findings were not surprising as we would expect those who are having trouble explaining NF1 to others to desire to know more about the condition. The majority (98.0%) of the study population expressed that it was important to learn information about NF. This stresses the importance of continually striving to make educational resources readily available to the NF community.

We hypothesized that individuals with greater knowledge of NF1 would have higher self-esteem. However, we did not find any association between the two factors. Perhaps knowledge itself is not enough to boost self-esteem, but instead it is the use of knowledge in interactions

with others that can help to reduce stigma, which could potentially have the positive impact of improved self-esteem. Knowledge of one's condition may be beneficial in being better able to educate others about NF1 and combat stigma associated with a diagnosis of NF1. Education, or teaching peers about a disorder, has been identified in previous studies as a coping strategy to combat stigma, in addition to the coping strategies of secrecy (hiding one's condition) and withdrawal (avoiding social or professional situations) (Peters et al., 2005). In a study on individuals with Marfan syndrome coping with stigma, it was demonstrated that 51% of individuals used educating others as a coping strategy. It was also observed that those who used "withdrawal" or "secrecy" as coping strategies tended to have lower self-esteem and experience more symptoms of depression (Peters et al., 2005). Furthermore, several studies demonstrate that the use of "preventive disclosure" of a chronic diagnosis to peers is helpful in minimizing negative peer views (Troster, 1997; Berlin, Sass, Davies, Jandrisevits, & Hains, 2005; Marcks, Berlin, Woods, & Davies, 2007). These studies do not take into consideration the individuals' level of knowledge of the particular condition, which could influence the tendency to self-disclose. Although we did not investigate what coping methods were most frequently used by the study population, further studies on stigmatization of the NF population are necessary in order to elucidate the most helpful coping strategies for these individuals.

Factors Influencing Self-Esteem

The results of this study are consistent with previous reports of lower self-esteem in individuals with NF compared to the general population (Wang et al., 2012). Having a history of learning problems was correlated with lower self-esteem. This indicates that perhaps the visible/cosmetic effects of NF1 are not the sole cause of lower self-esteem in this population, but that the cognitive effects of the condition also play a major role. The results of Martin et al.

(2012) suggested that impaired cognitive abilities may negatively impact emotional well-being in individuals with NF1. There are no known studies that have looked specifically at the impact of learning disabilities on self-esteem. Moreover, several factors were observed to have a positive influence on self-esteem: having friends with NF1, attending a support group, attending a NF clinic, or receiving genetic counseling. We suspect that having friendships with others who have the same diagnosis can help reduce feelings of isolation and lead to an increase in self-esteem. Similarly, involvement in support groups help improve confidence of members by providing long-term support and making individuals affected with a genetic condition or families of affected individuals feel less isolated ("Psychological & Social Implications," 2010). Most support groups also aim to provide accurate information to their members about the specific condition and the associated genetic risk. Therefore, parental participation in support groups can have the positive effect of making a family be more open to talking about the genetic condition, which helps with family coping overall (Plumridge, Metcalfe, Coad, & Gill, 2011).

Improved coping techniques in people who have seen a genetic counselor may partially explain the higher self-esteem scores seen in our study. Genetic counselors are taught to be aware of stigma associated with genetic conditions so as to avoid adding to stigmatization unintentionally (McCarthy Veach, LeRoy, & Bartels, 2003). Furthermore, they are proficient in helping patients with genetic conditions "understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease," (Resta et al., 2006). The genetic counseling process can provide an avenue for a person with NF1 to explore what coping mechanisms they have developed as a result of their diagnosis or perhaps identify ways to improve coping (Gaff & Clarke, 2007). The specialized skills that genetic counselors have may play an important role in helping people with NF1 learn positive ways to cope with their

diagnosis, as well as give the person room to express their concerns about NF1 and how it affects their relationships with others.

Studies have found that medical personnel can be a source of stigma for individuals with chronic conditions (Ablon, 2002). Perhaps receiving care at a specialized NF clinic, where the doctors and staff are familiar with the condition, can help to reduce the chance of being judged or misunderstood by medical professionals. Interestingly, we observed that attending an NF clinic was correlated with receiving genetic counseling (p=0.001) and attending a support group (p=0.033). The multivariable models reflected that attending an NF clinic was not independently associated with an improvement in self-esteem, whereas receiving genetic counseling/seeing a genetic counselor or attending support groups did positively impact self-esteem. This underscores the need for genetic counseling and other sources of support for these patients within and without the multidisciplinary model of care. However, this is not to discount the multidisciplinary approach for treating NF1 as these clinics provide many benefits to patients and can be a source of genetic counseling.

Study Limitations

One limitation of our study is that the knowledge portion of the survey has not been validated. In addition, participants were contacted through a support group organization and were given the option to participate in the study, creating a selection bias and/or self-selection bias. However, this could not have been avoided as it is difficult to find cohorts with NF1 using other recruitment methods. This population may be different from individuals with NF1 who are not members of a support group, in that they could be more information-seeking or have greater knowledge of the disease, therefore altering our results. This factor along with our small sample size limits the ability for our results to be generalized to the NF population as a whole.

Furthermore, responses to questions about NF1 were based on the "honor system" as it was impossible to control whether or not a participant looked up answers to questions on the Internet. In order to discourage the use of the Internet to search for answers to questions and minimize guessing we provided, "Don't know" as an option for each question.

Research Recommendations

It would be interesting to investigate the coping strategies used by individuals with NF1 to combat stigmatization and to assess whether increased knowledge of the condition leads to better coping. While the scope of this study did not include gathering medical information regarding the severity of the participant's disease from their physician, collecting this information would be useful in gaining insight to how severity correlates with self-esteem and other factors, such as, disclosure of the diagnosis.

Practice Implications

- Healthcare professionals should encourage patients with NF1 to participate in support groups like the Texas NF Foundation, in order for them to meet and learn from others with the condition and to have access to reliable information.
- Greater effort should be made in the development of multidisciplinary NF clinics throughout the United States.
- Referral to a genetic counselor is recommended for individuals and families affected by NF1.

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Vita

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