NF1 Thesis Analysis

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Study Aims

1. To assess whether parental knowledge of NF1 is dependent on the child's mode of inheritance of the condition (familial vs de novo mutations)

2. To determine how exposure to genetic counseling affects knowledge of NF1.

Data Overview & Cleaning

There were a total of 303 responses to the questionare:

- 11 respondents were "Not sure" if they had NF1. These respondents were dropped from the data set since both study aims are contingent upon knowing this information.

- One respondent skipped the question, "Does your child's other parent have NF1?" This respondent was kept since they answered the question, "Do you have NF1?" and their answer to "Does your child's other parent have NF1?" was imputed as "No."

- One respondent skipped the question, "Doe you have NF1?" This respondent was dropped from the data set since both study aims are contingent upon knowing this information.

- 14 respondents do not have NF1 and are "Not sure," if the childs other parent has NF1. These respondents were dropped from the data set since both aims of the thesis are dependent on this information.

After the above adjustments, the final data set contains 277 observations.

Testing Methodology

Although there are only two separate study aims, the first will require a total of 3 pairwise statistical tests, comparing the average test scores of the three groups below to one another using Welch's t-Test.

- **Group A** - Respondents who do not have NF1 and the child's other parent also does not have NF1.

- **Group B** - Respondents who do not have NF1 and the child's other parent does have NF1.

- **Group C** - Respondents who do have NF1 and the child's other parent does not have NF1.

These three tests, combined with the test for the second study aim, create a total of 4 statistical tests. In order to control the family wise error rate, the original significance level ($\alpha = 0.05$) was adjusted using the Bonferroni Correction, reducing the level of statistical significance to $0.05/4 = 0.0125$.

Study Aim 1 - Does the mode of inheritance (familial vs de novo mutations) of the child have an effect on parental knowledge of NF1?

Test 1

- H_0 : Respondents in Group B do not have a different average test score than respondents in Group A.

- H_a : Respondents in Group B have a different average test score than respondents in Group A.

Group A vs. Group B
Mean's of each group displayed as vertical lines: Group A = 0.8277 | Group B = 0.8436
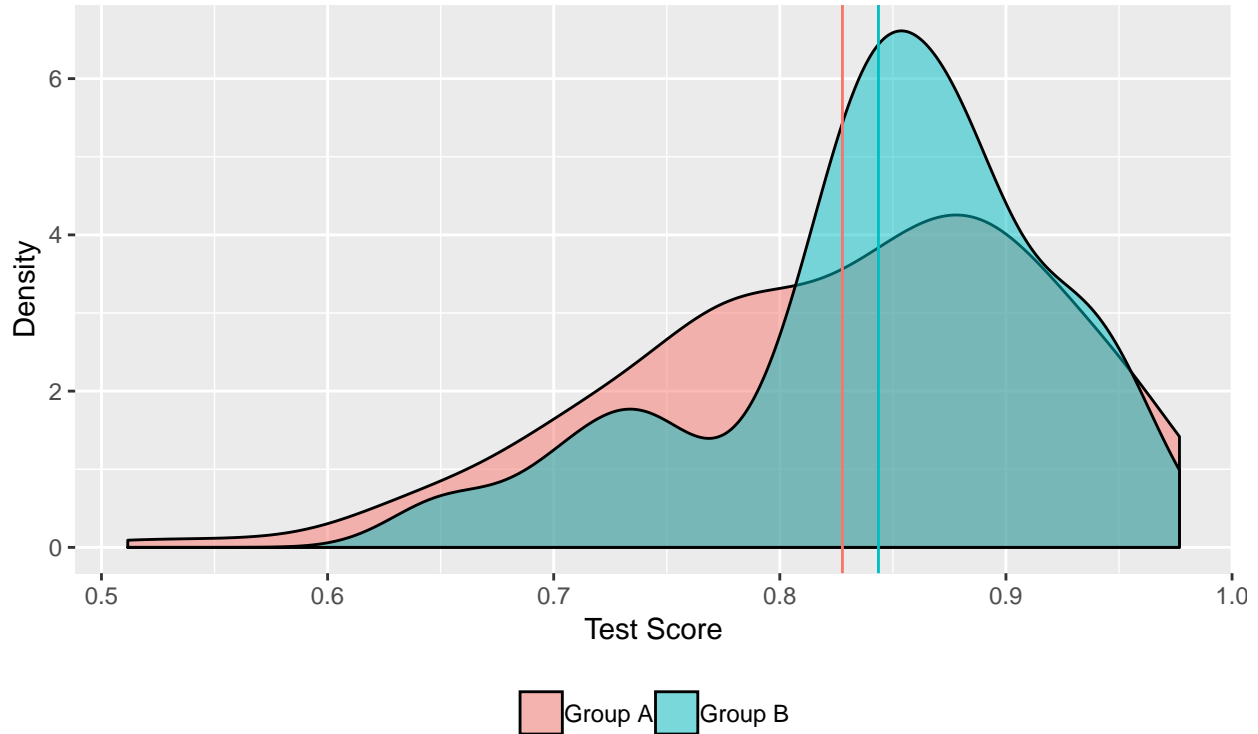

```
## [1] "Test 1 p value = 0.307749444337635"
```
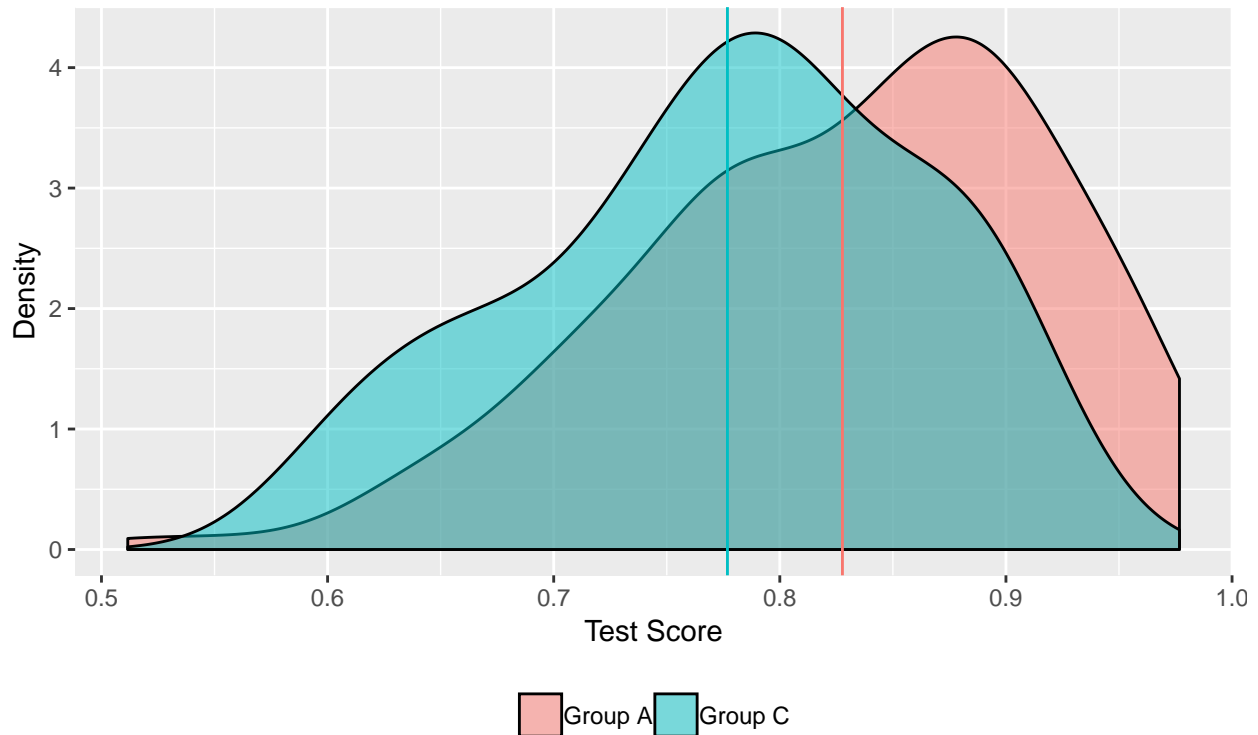
Welch's two-tailed t-Test returns the above p value, with which we fail to reject the null hypothesis.

Test 2

- H_0 : Respondents in Group A do not have a higher average test score than respondents in Group C.

- H_a : Respondents in Group A have a higher average test score than respondents in Group C.

Group A vs. Group C
Mean's of each group displayed as vertical lines: Group A = 0.8277 | Group C = 0.7768


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## [1] "Test 2 p value = 0.000187363981785434"
```
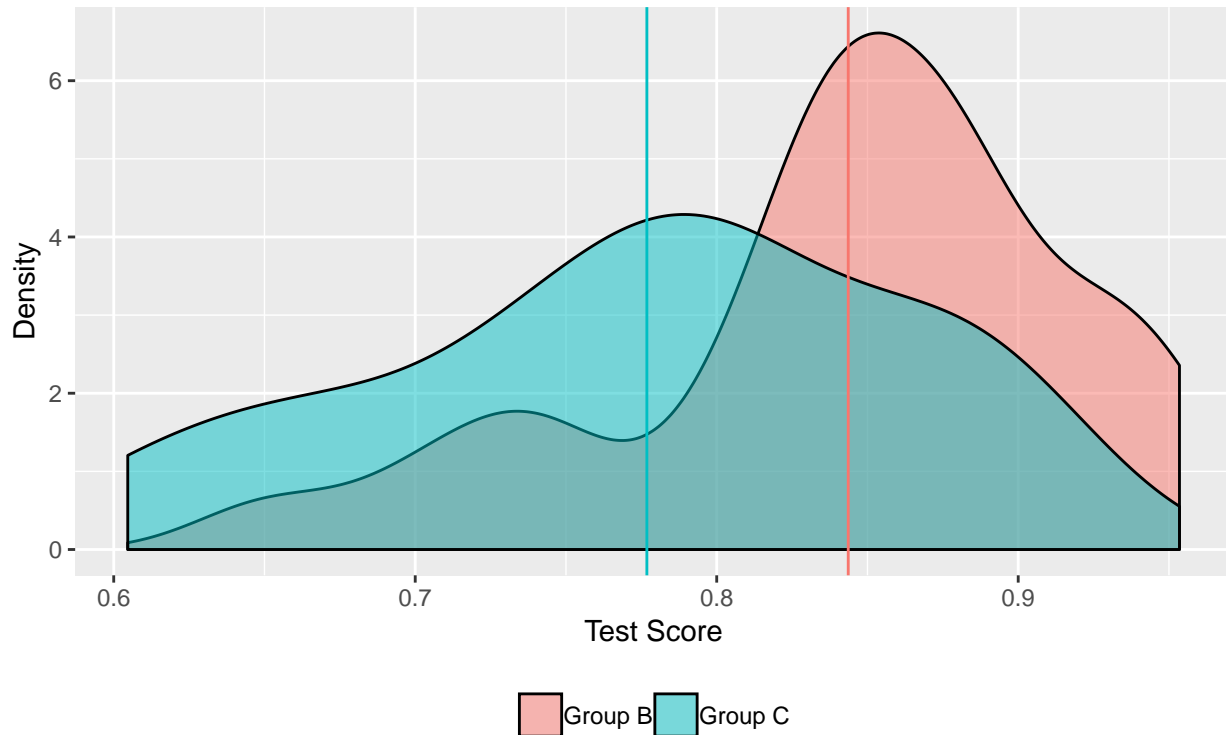
Welch's one-sided t-Test returns the above p value. Being well below the Bonferroni corrected significance level of 0.0125, the null hypothesis is rejected in favor of the alternative.

Test 3

- H_0 : Respondents in Group B do not have a higher average test score than respondents in Group C.

- H_a : Respondents in Group B have a higher average test score than respondents in Group C.

Group B vs. Group C

Mean's of each group displayed as vertical lines: Group B = 0.8436 | Group C = 0.7768
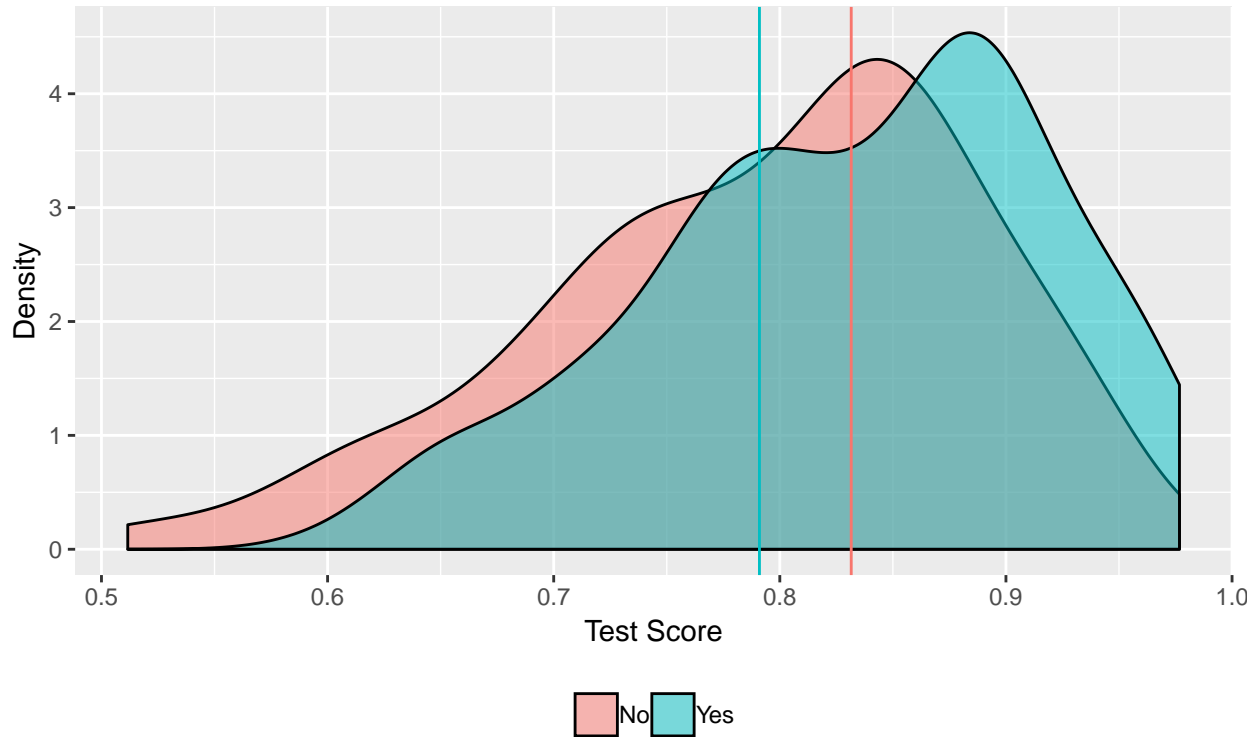


```
## [1] "Test 3 p value = 0.000280154187709805"
```

Welch's one-sided t-Test returns the above p value. Being well below the Bonferroni corrected significance level of 0.0125, the null hypothesis is rejected in favor of the alternative.

Test 4

- H_0 : Respondents who have seen a genetic counselor do not have a higher average test score than those that have not seen a genetic counselor.

- H_a : Respondents who have seen a genetic counselor have a higher average test score than those that have not seen a genetic counselor

Have you seen a Genetic Counselor?
Mean's of each group displayed as vertical lines: Yes = 0.8316 | No = 0.791


```
## [1] "Test 4 p value = 0.000280154187709805"
```

Welch's one-sided t-Test returns the above p value. Being well below the Bonferroni corrected significance level of 0.0125, the null hypothesis is rejected in favor of the alternative.
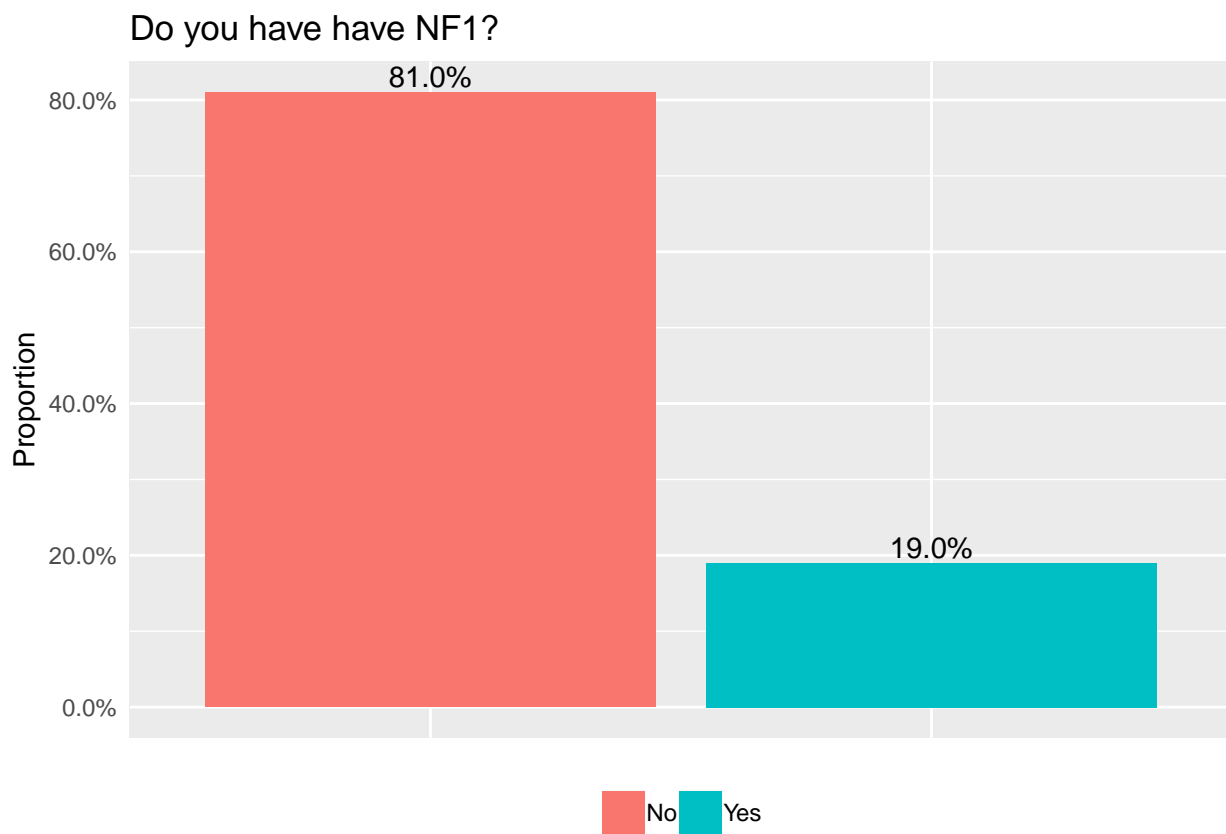
Assumption Testing

Demographic Analysis

Do you have have NF1?

Table 1: Do you have NF1?

Response	Count
No	222
Yes	52

Have you ever met with a Genetic Counselor?
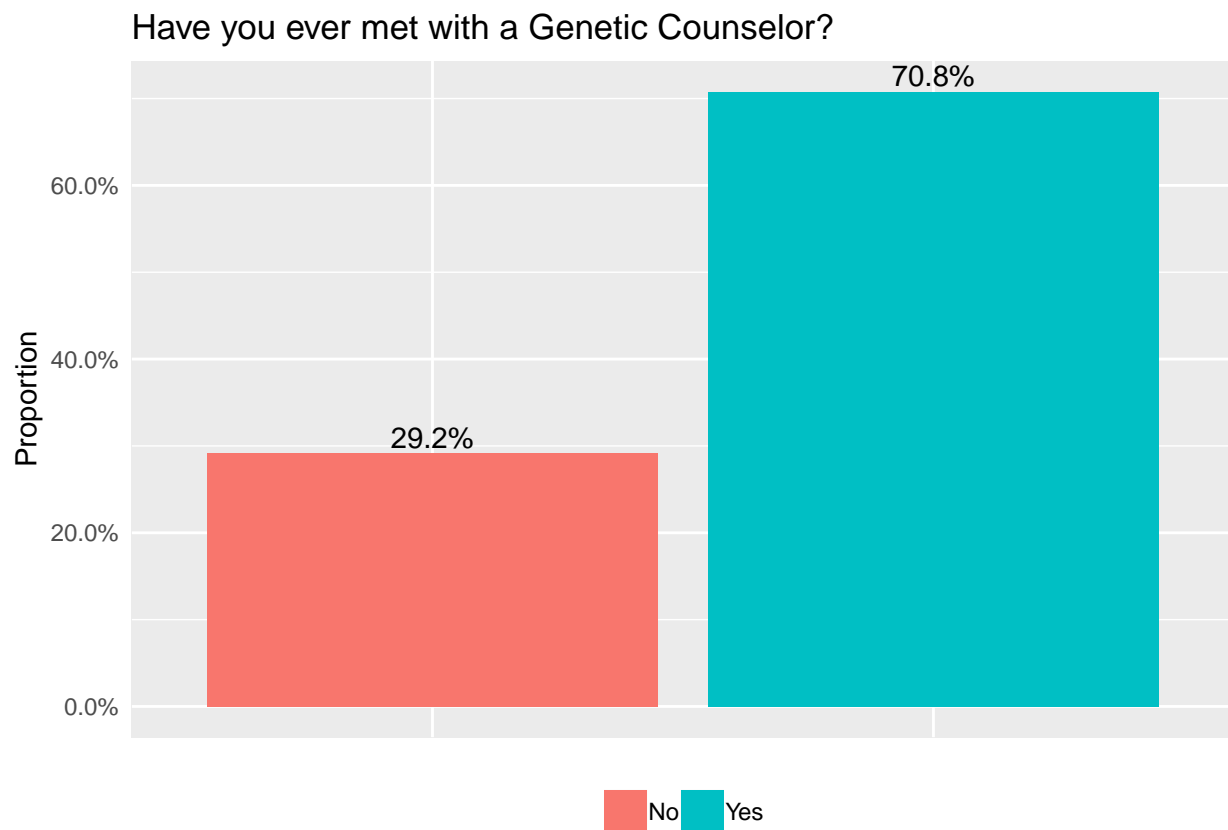
Table 2: Have you ever met with a Genetic Counselor?

Response	Count
No	80
Yes	194
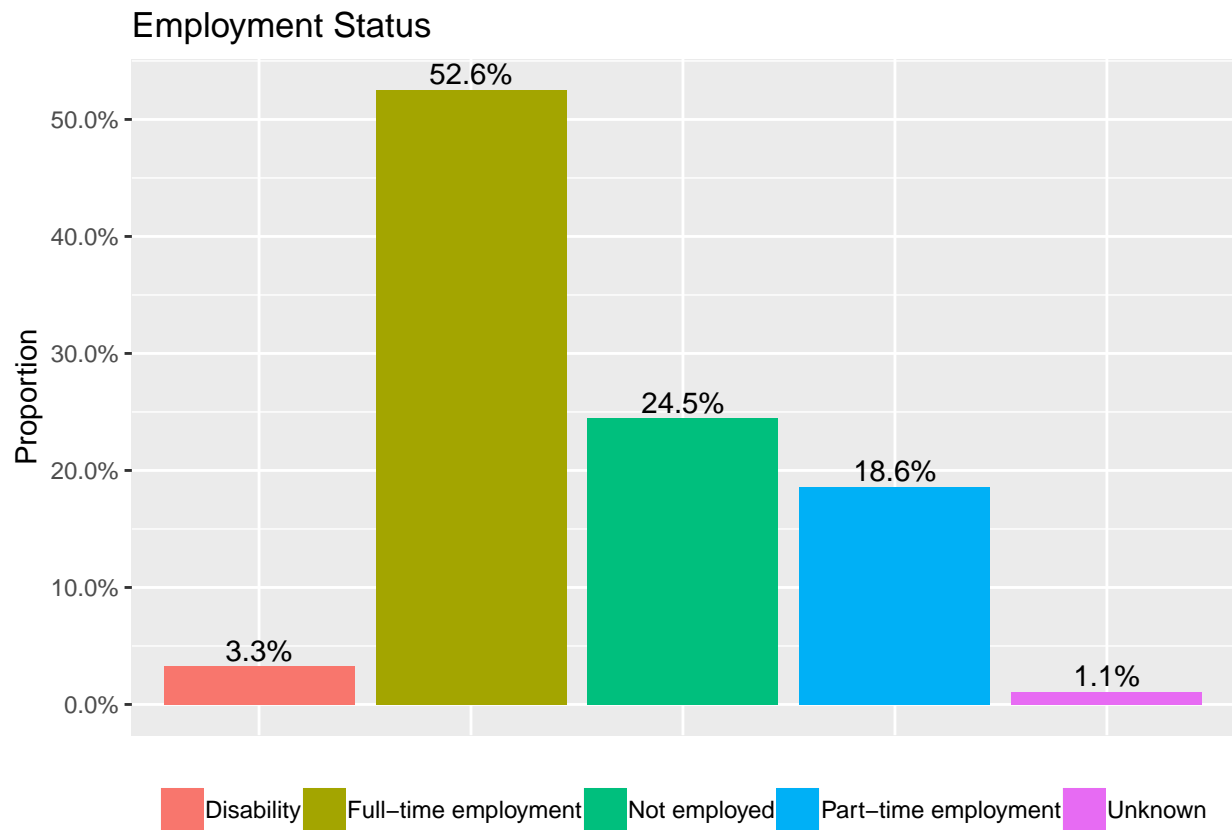
Employment Status

Table 3: What is your employment status?

Response	Count
Disability	9
Full-time employment	144
Not employed	67
Part-time employment	51
Unknown	3
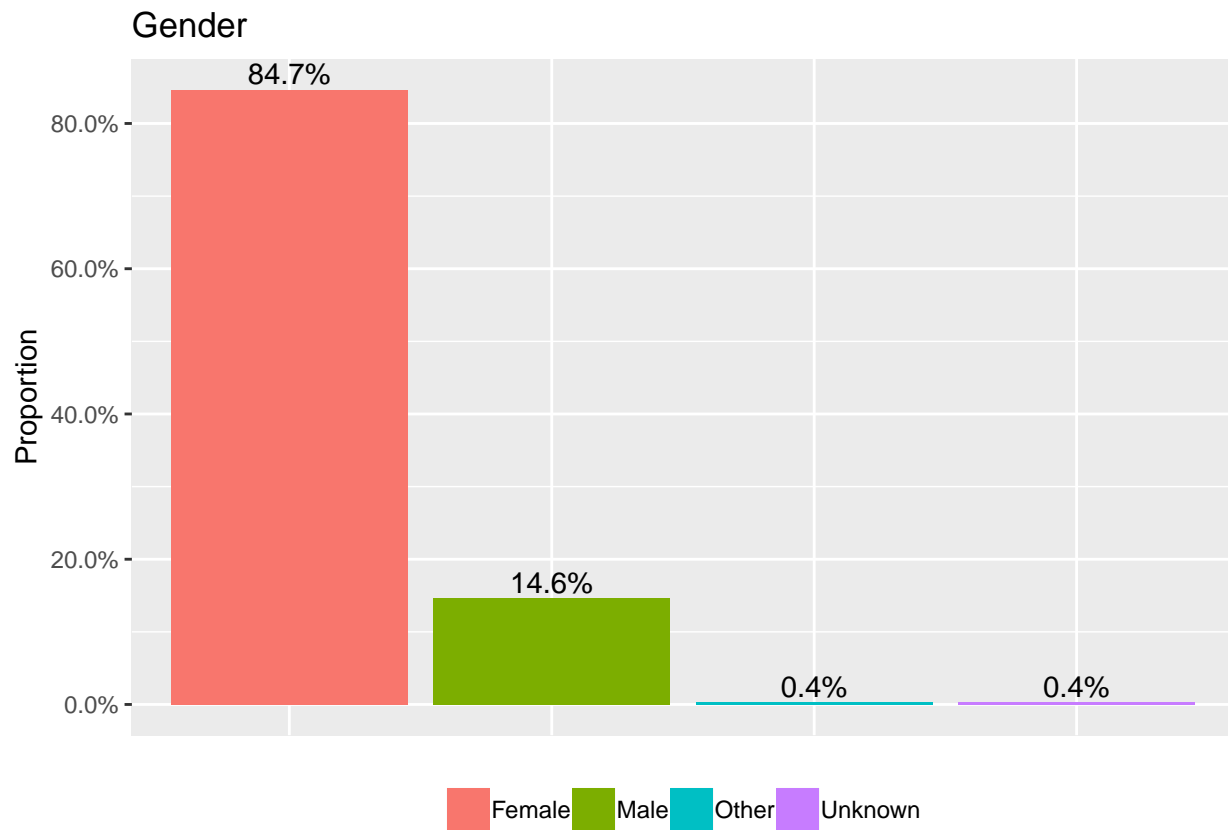
Gender

Table 4: Gender

Response	Count
Female	232
Male	40
Other	1
Unknown	1
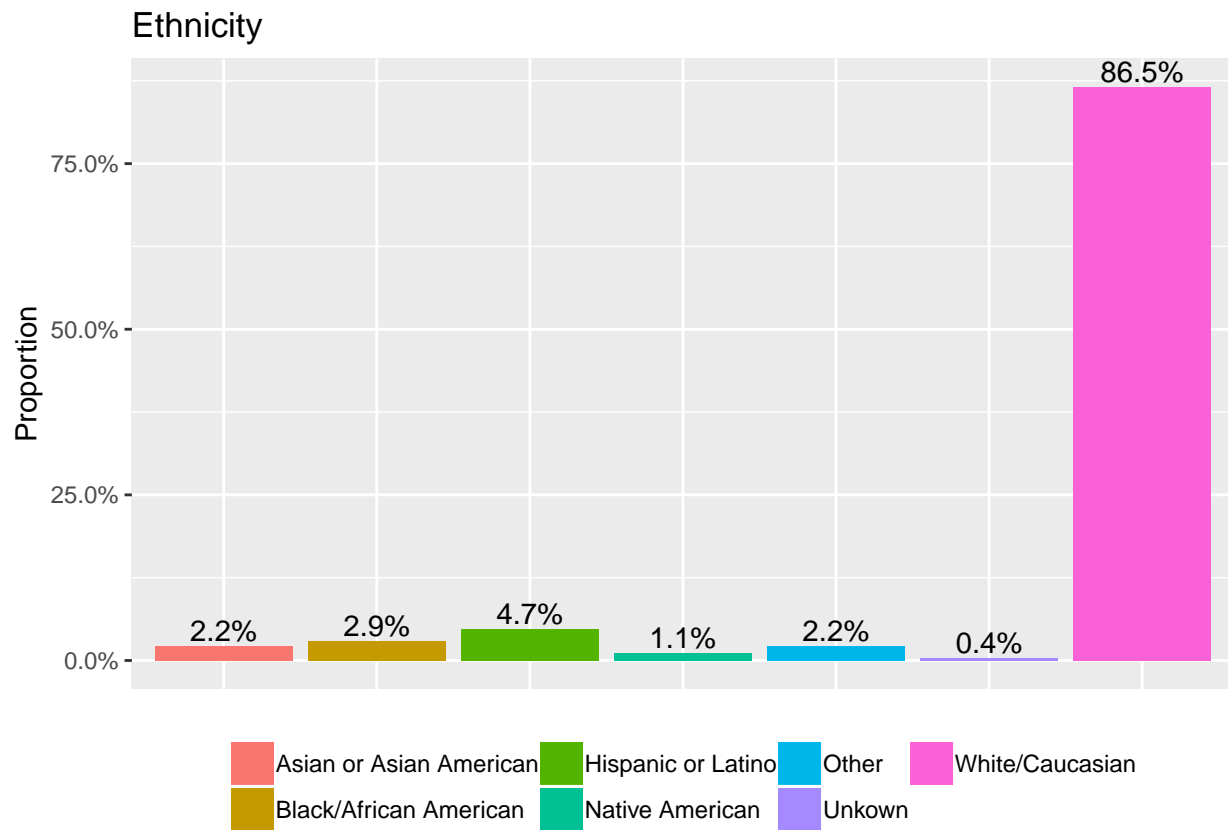
Ethnicity

Table 5: Ethnicity

Response	Count
Asian or Asian American	6
Black/African American	8
Hispanic or Latino	13
Native American	3
Other	6
Unkown	1
White/Caucasian	237

What specialists does your affected child see?

Faceted by mode of inheritance
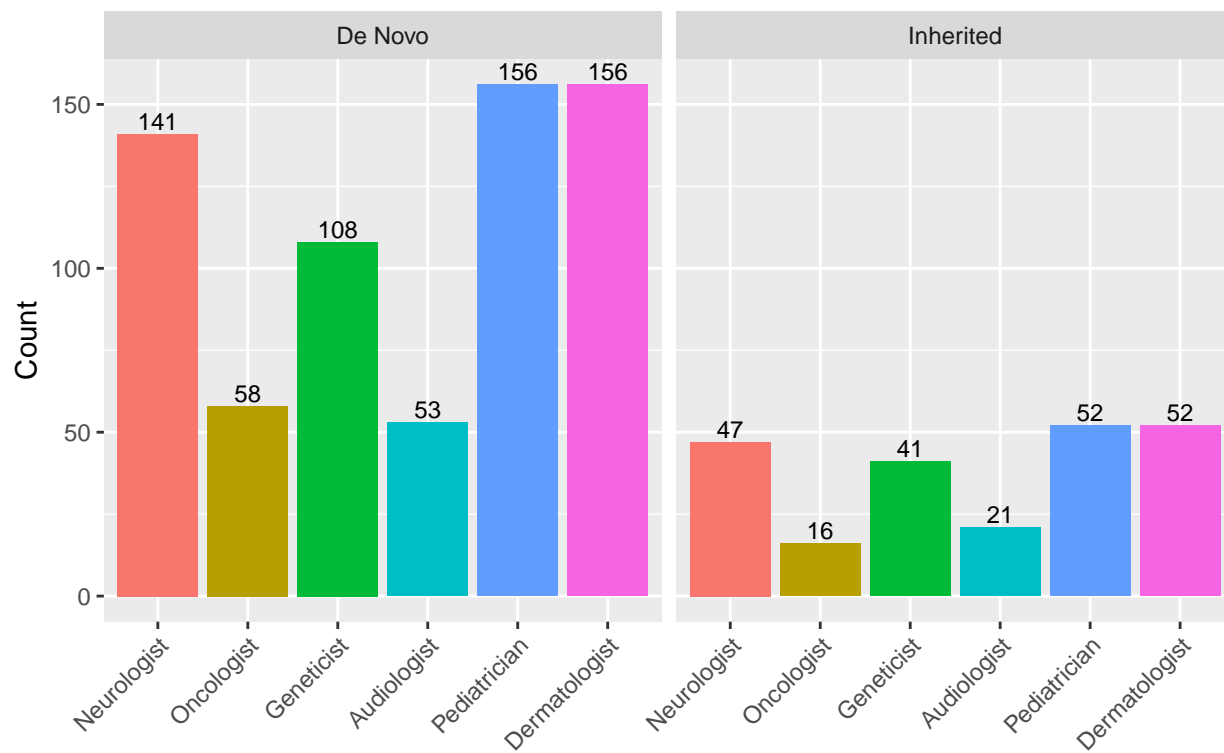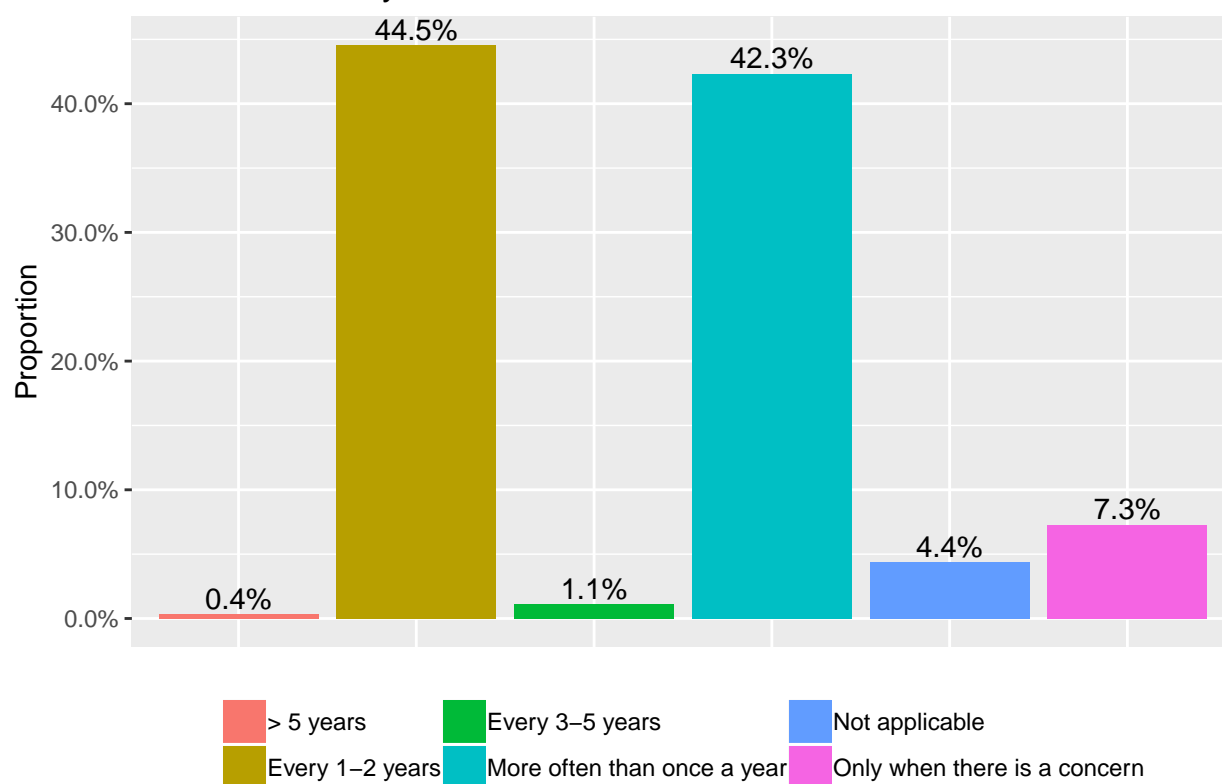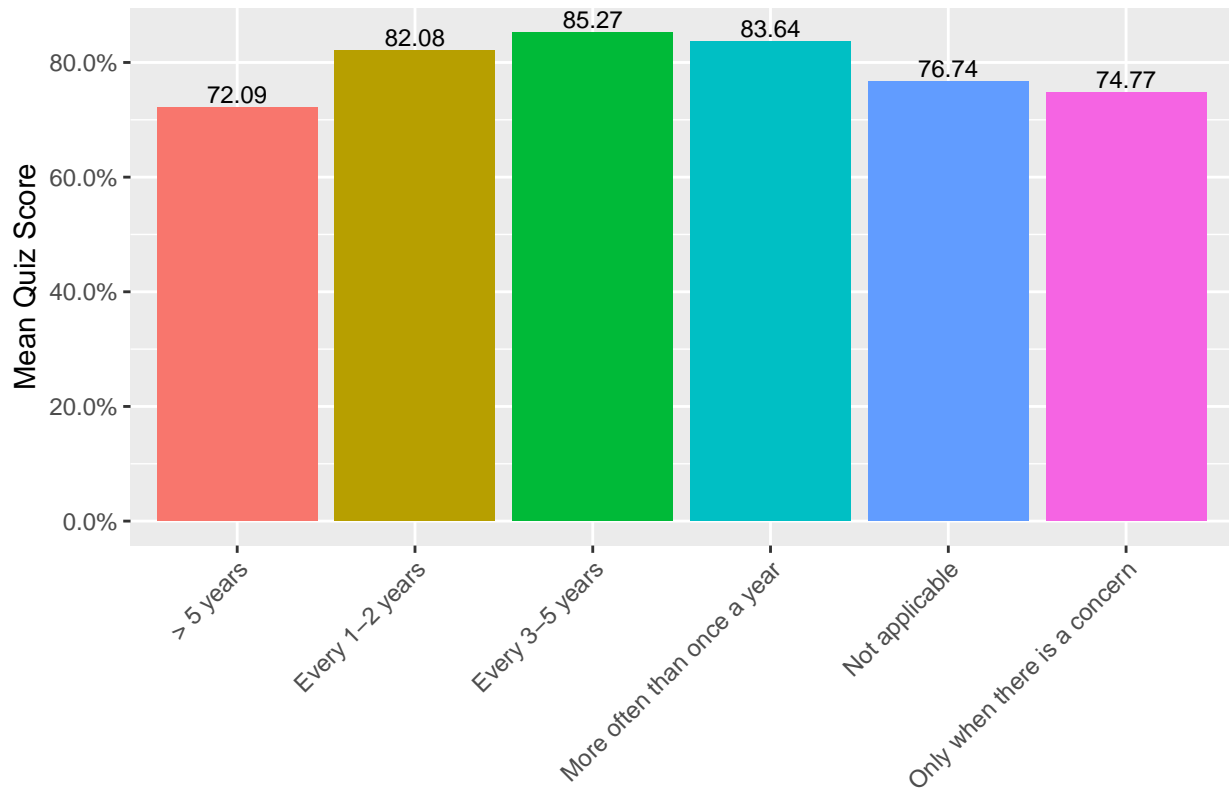
How often does your oldest child see a NF1 doctor?

Table 6: How often does your oldest child see a NF1 doctor?

Response	Count
> 5 years	1
Every 1-2 years	122
Every 3-5 years	3
More often than once a year	116
Not applicable	12
Only when there is a concern	20

How often does your oldest child see a NF1 doctor?

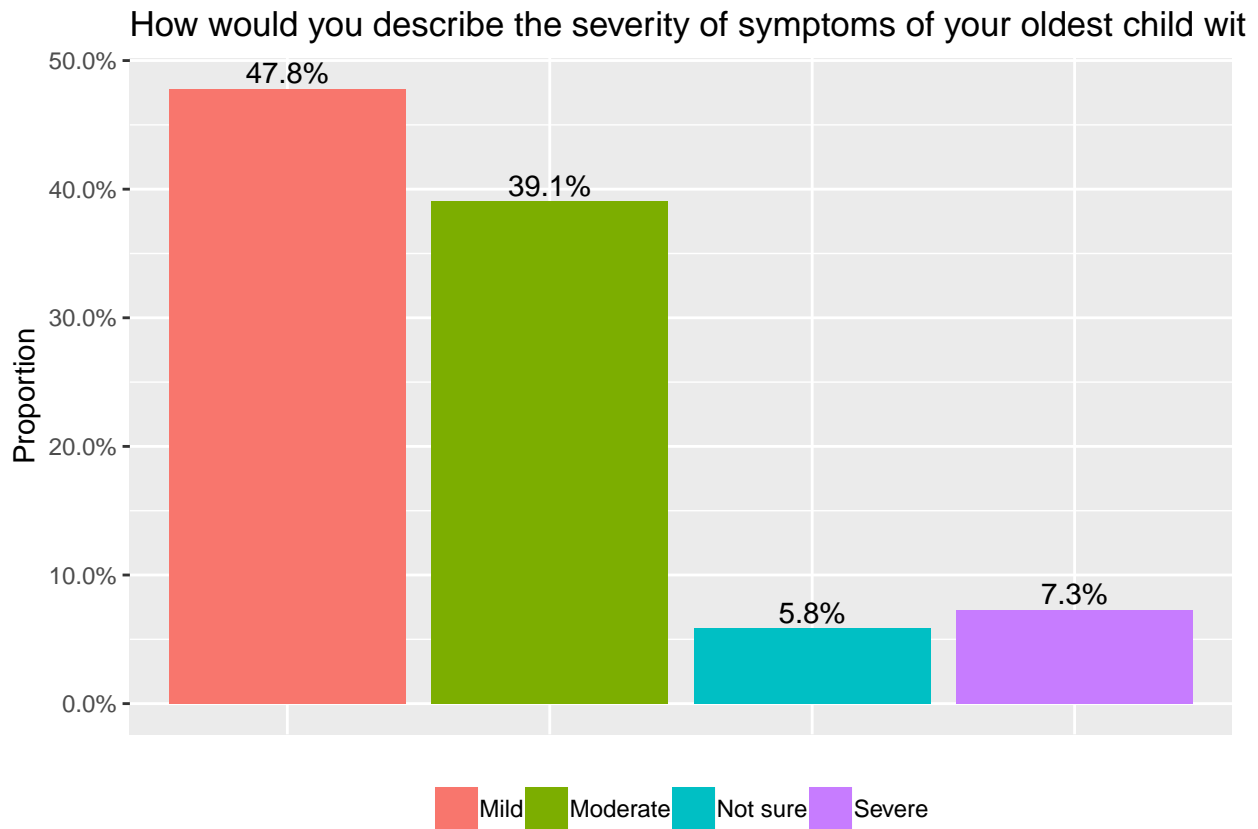## Have you ever met with a Genetic Counselor?
Faceted by mode of inheritance

How would you describe the severity of symptoms of your oldest child wit
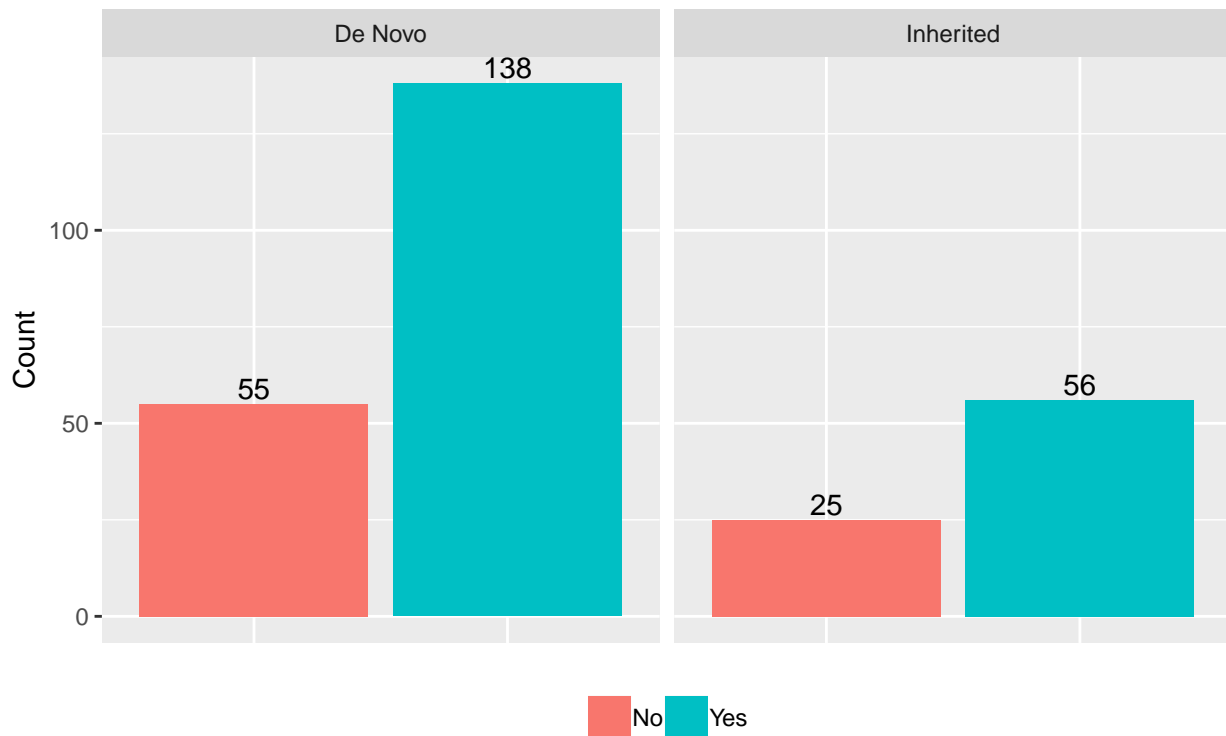
Table 7: How would you describe the severity of symptoms of your oldest child with NF1?

Response	Count
Mild	131
Moderate	107
Not sure	16
Severe	20

```
##  [1] "respondent_id"
##  [2] "age"
##  [3] "gender"
##  [4] "ethnicity"
##  [5] "educational_background"
##  [6] "employment_status"
##  [7] "do_you_have_nf1."
##  [8] "does_your_childs_other_parent_have_nf1."
##  [9] "have_you_ever_met_with_a_genetic_counselor."
## [10] "does_your_oldest_child_see_a_neurologist_for_nf_care."
## [11] "does_your_oldest_child_see_an_oncologist_for_nf_care."
## [12] "does_your_oldest_child_see_a_geneticist_for_nf_care."
## [13] "does_your_oldest_child_see_an_ent.audiologist_for_nf_care."
## [14] "does_your_oldest_child_see_a_pediatrician_for_nf_care."
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## [15]  "does_your_oldest_child_see_a_dermatologist_for_nf_care."
## [16]  "does_your_oldest_child_see_a_different_doctor.specialist_for_nf_care._.please_specify."
## [17]  "how_often_does_your_.oldest._child_see_an_nf_doctor."
## [18]  "how_would_you_describe_the_severity_of_symptoms_of_your_.oldest._child_with_nf1."
## [19]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_your_doctor."
## [20]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_family_members_with_nf1."
## [21]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_online_searches."
## [22]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_an_nf_organization_website."
## [23]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_social_media_sites_.such_as_f
## [24]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_other_families_you_know_that_
## [25]  "do_you_not_have_any_questions_regarding_nf1."
## [26]  "if_you_have_questions_regarding_nf1_do_you_not_obtain_additional_information."
## [27]  "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_other_sources._.please_specify
## [28]  "score"
## [29]  "group_id"
## [30]  "seen_gc"
## [31]  "mode_of_inheritance"
```