NF1 Thesis Analysis

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Contents

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.

Table 1: Test Score Statistics

	Mean	StandardDeviation
Group A	0.8276901	0.0919368
Group B	0.8436247	0.0750741
Group C	0.7768336	0.0866807

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.

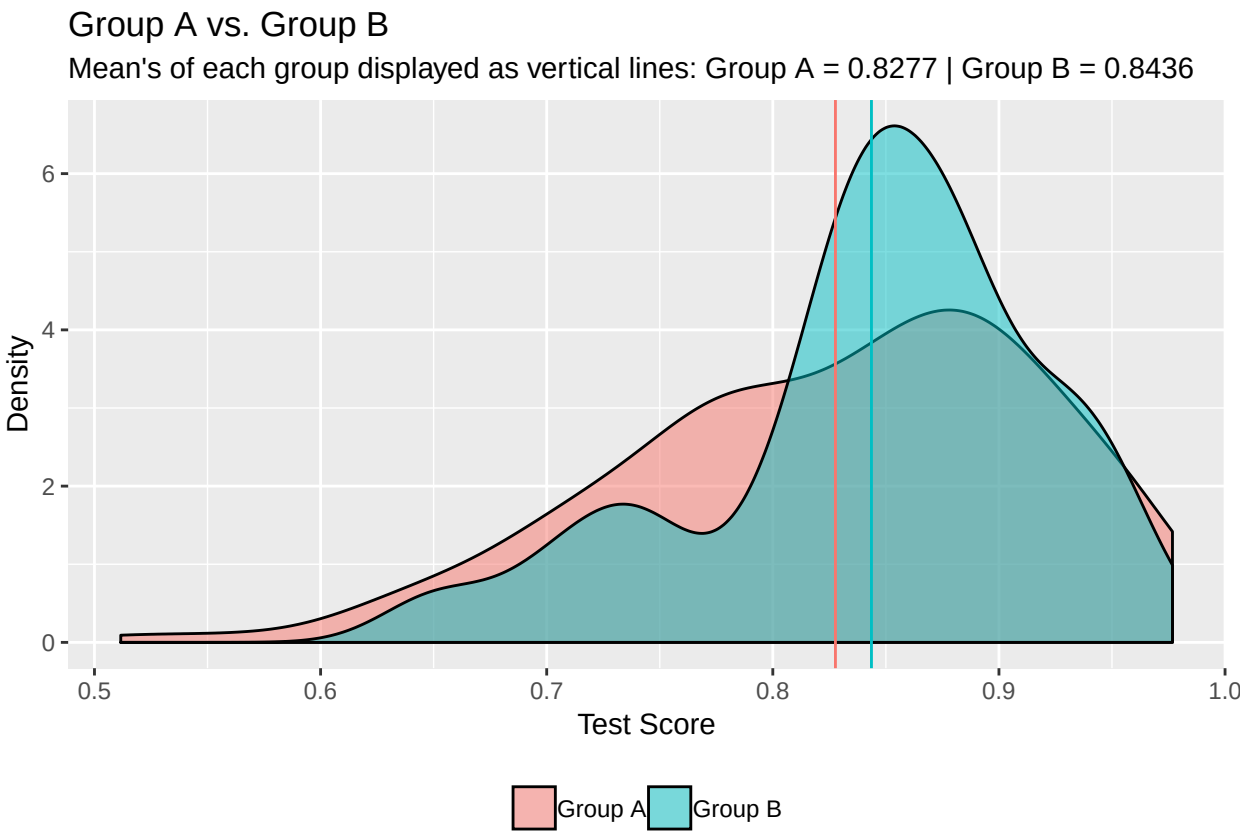
- **Group A** - Respondents who do not have NF1 and the child's other parent also does not have NF1.
- **Group B** - Respondents who do not have NF1 and the child's other parent does have NF1.
- **Group C** - Respondents who do have NF1 and the child's other parent does not have NF1.

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.

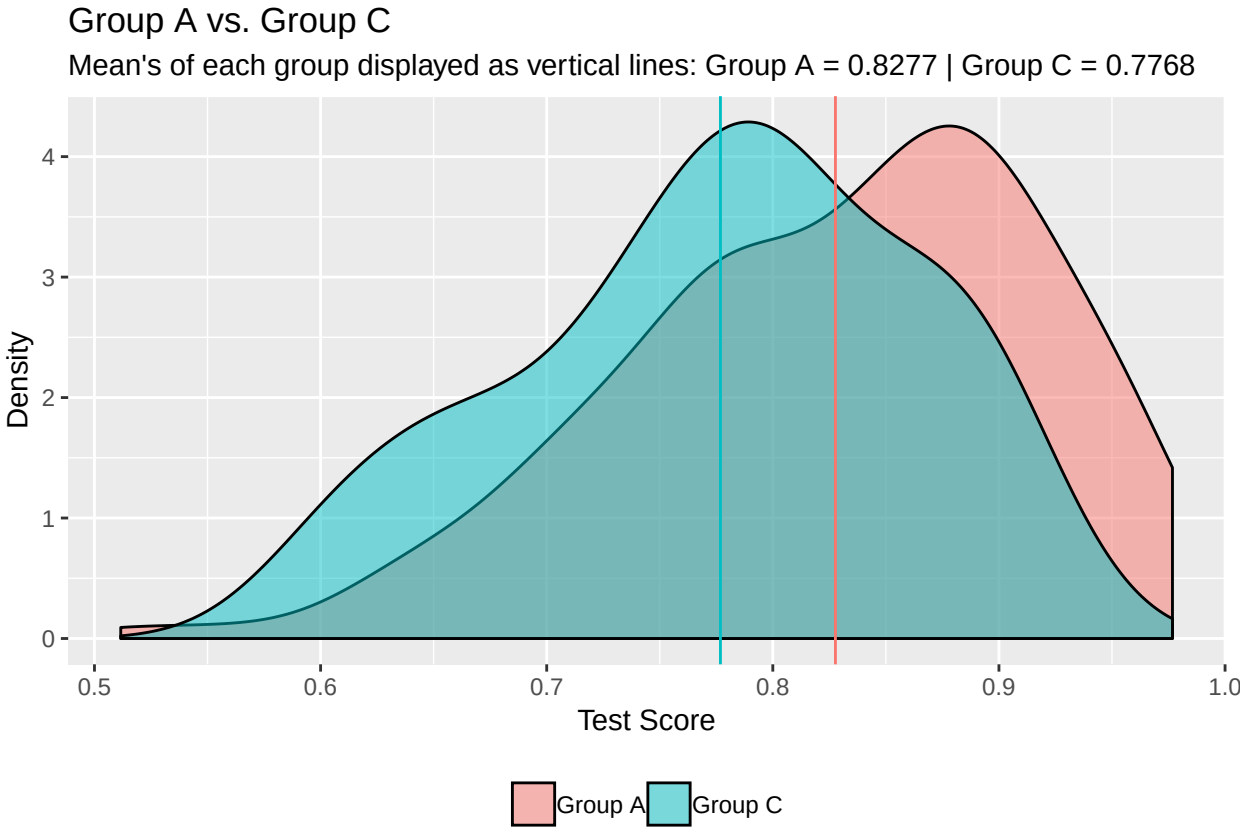



```
## [1] "Test 1 p value = 0.307749444337635"
```

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.

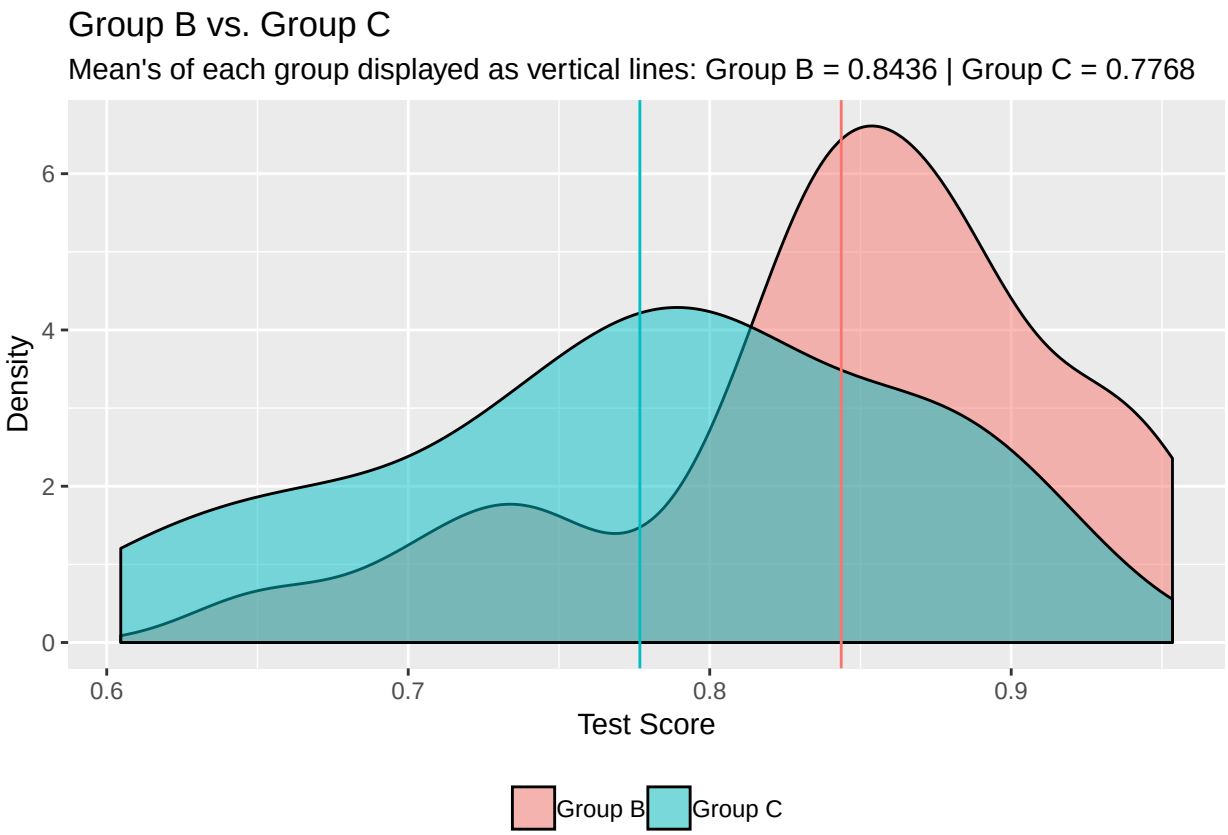



```
## [1] "Test 2 p value = 0.000187363981785434"
```

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.

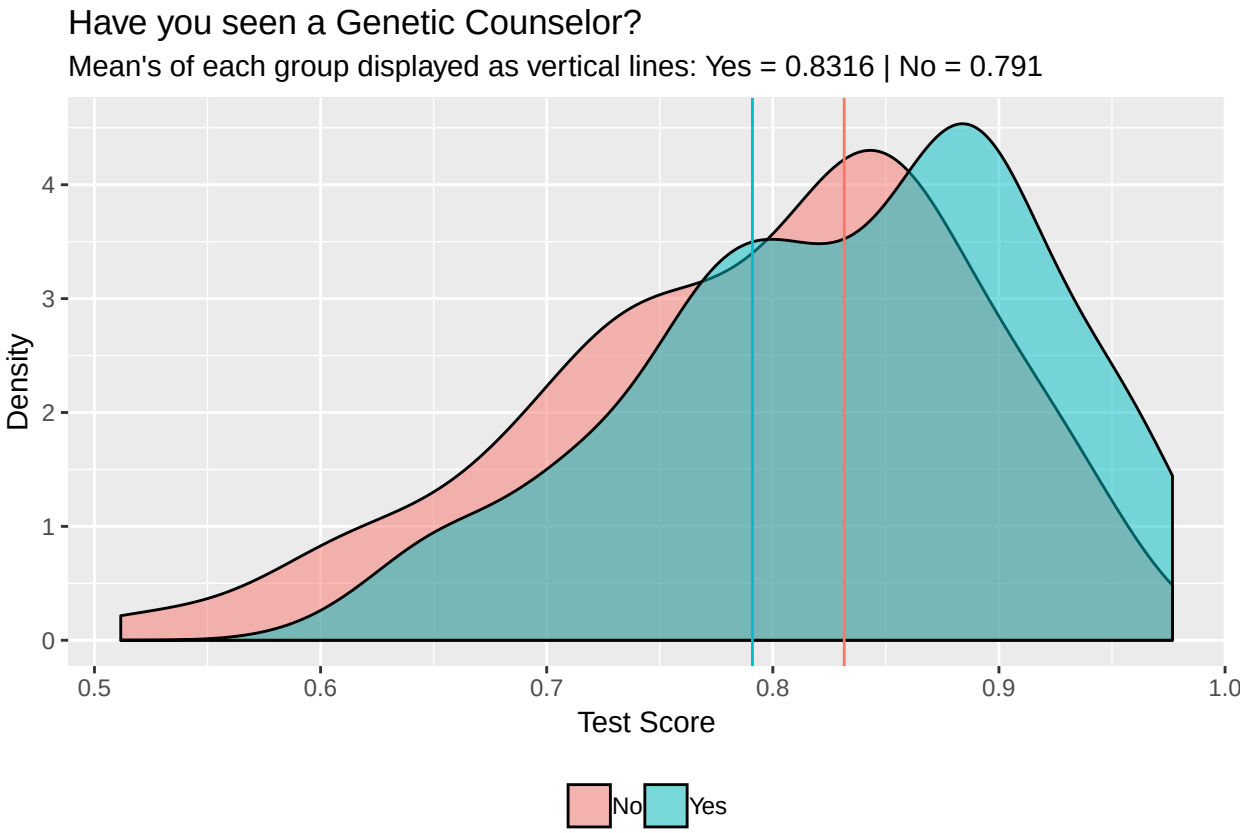



```
## [1] "Test 3 p value = 0.000280154187709805"
```

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




```
## [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.

Cronbach's Alpha

Using 43 quiz questions returned a Cronbach Alpha value of 0.7010484.

Assumption Testing

Demographic Analysis

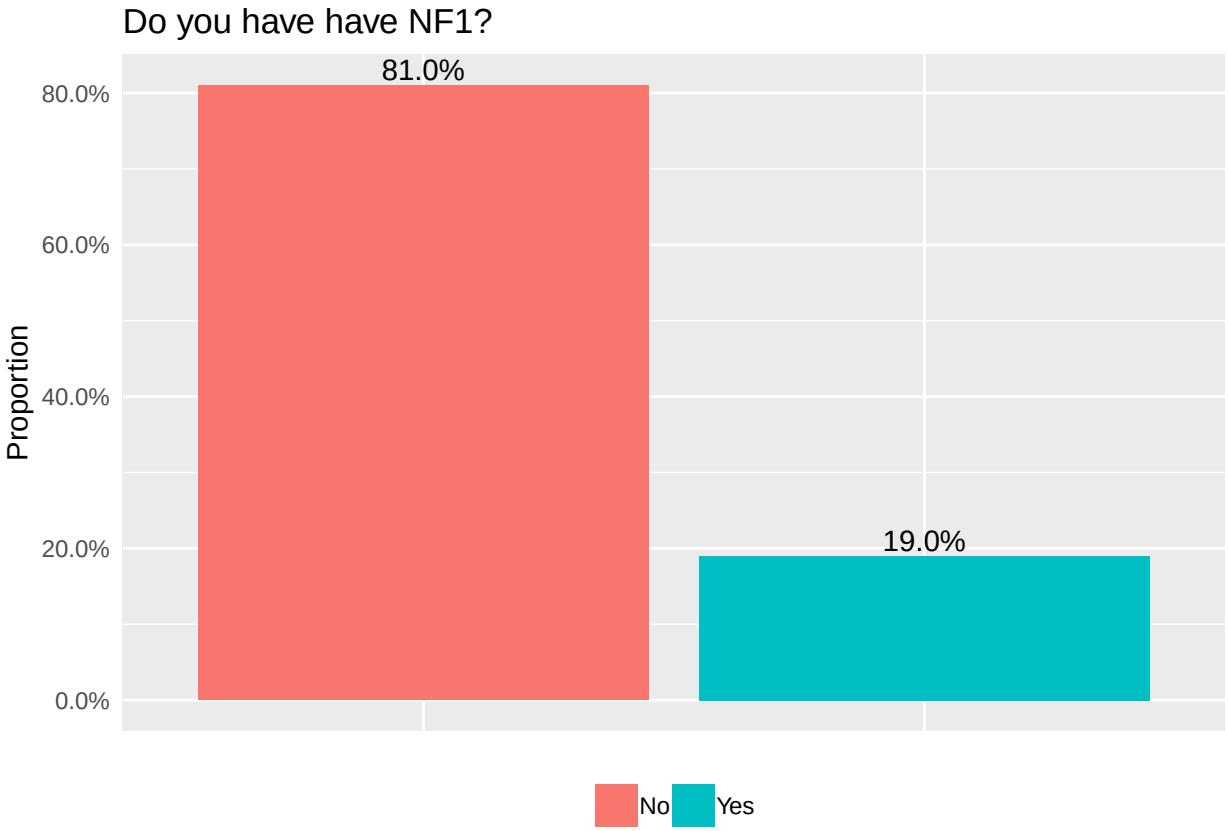


Table 2: Do you have NF1?

Response	Count
No	222
Yes	52

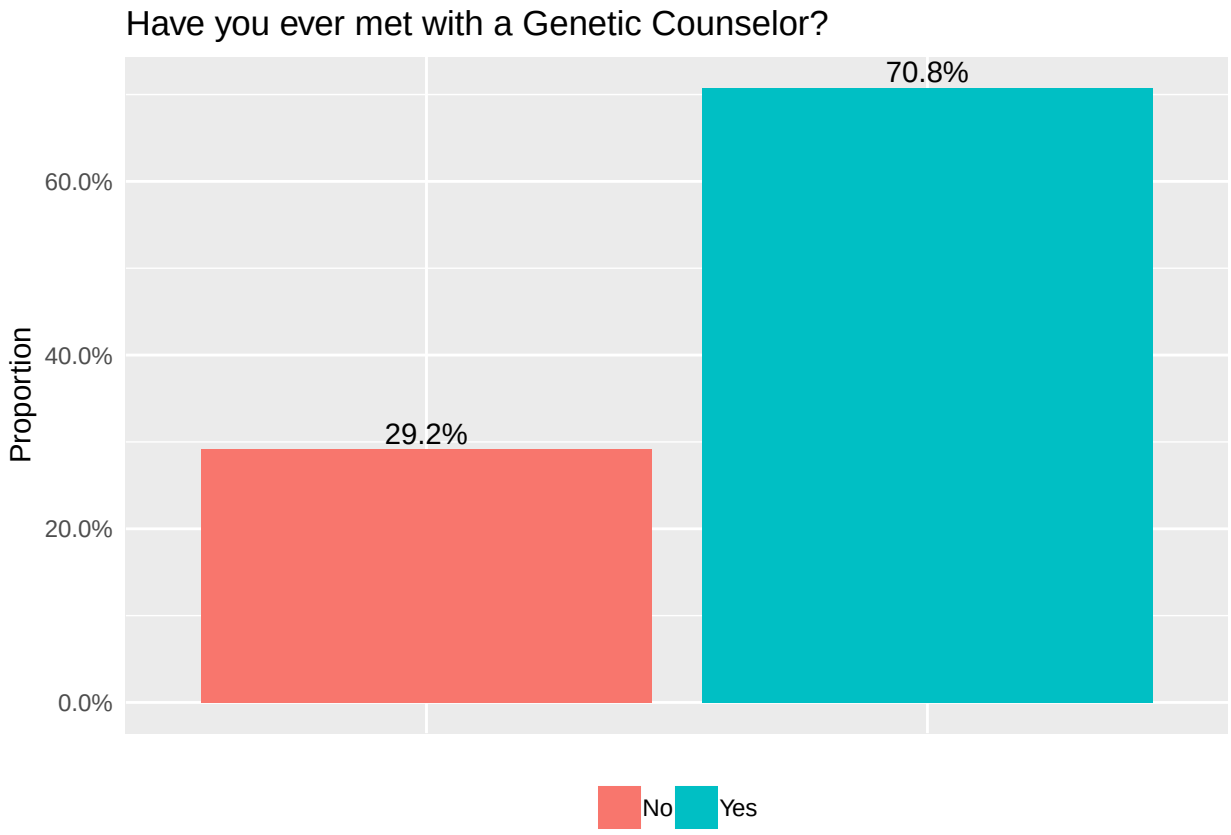


Table 3: Have you ever met with a Genetic Counselor?

Response	Count
No	80
Yes	194

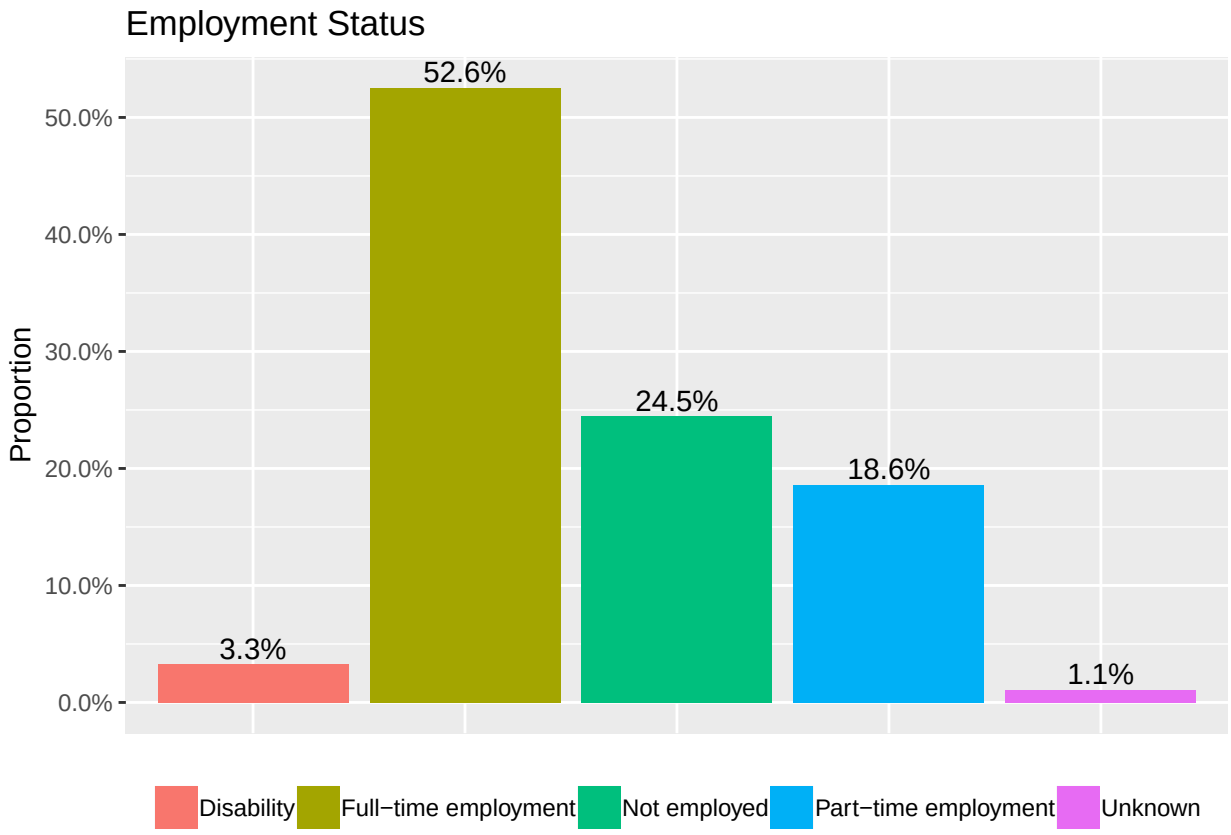


Table 4: What is your employment status?

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

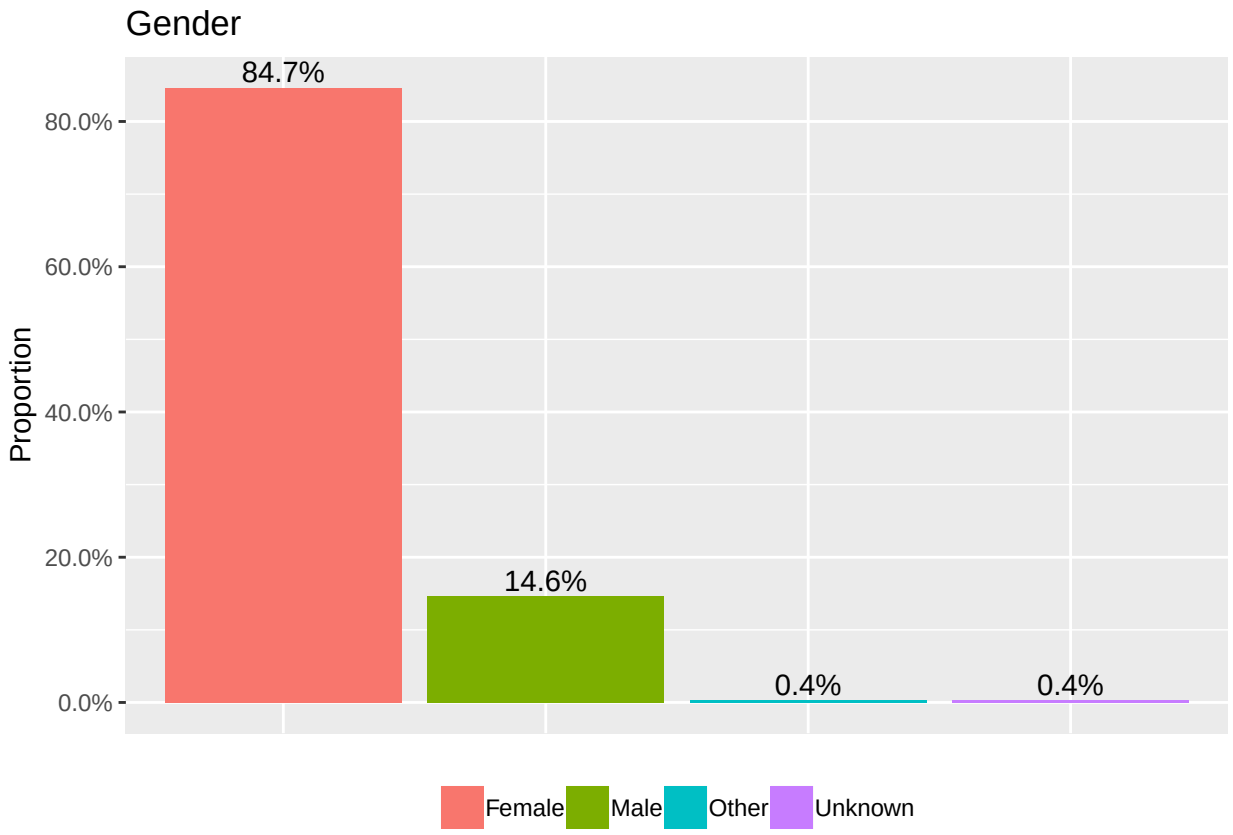


Table 5: Gender

Response	Count
Female	232
Male	40
Other	1
Unknown	1

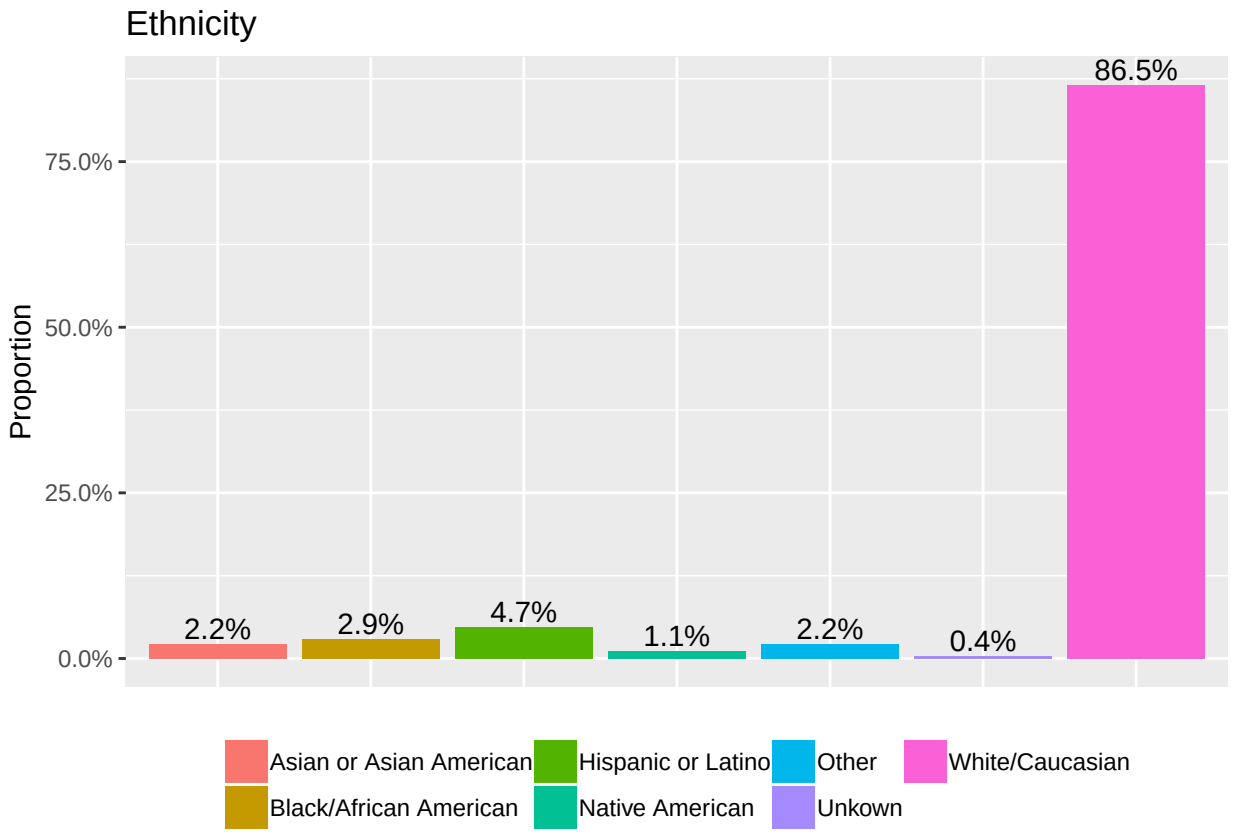
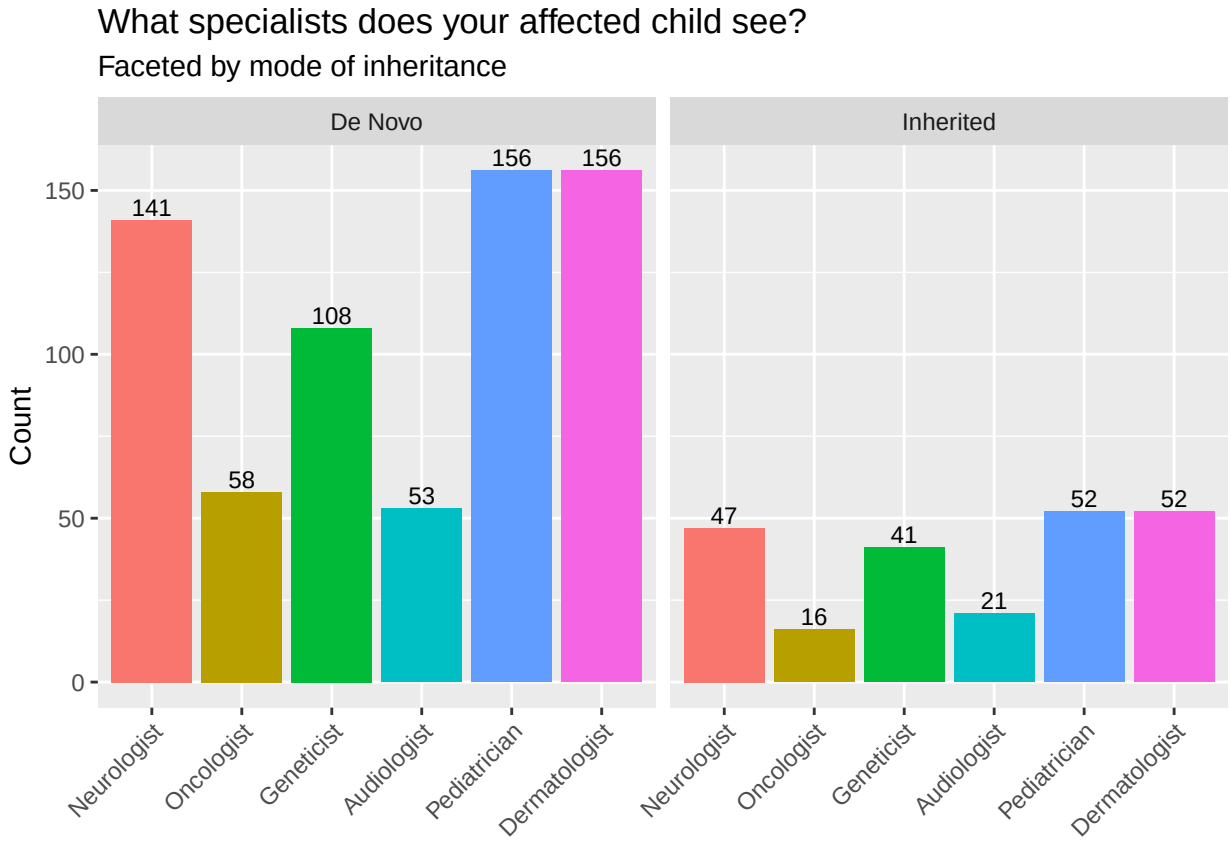


Table 6: Ethnicity

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
De Novo
Inherited
Count
150
100
50
0
141
58
108
53
156
156
47
16
41
21
52
52
Neurologist
Oncologist
Geneticist
Audiologist
Pediatrician
Dermatologist

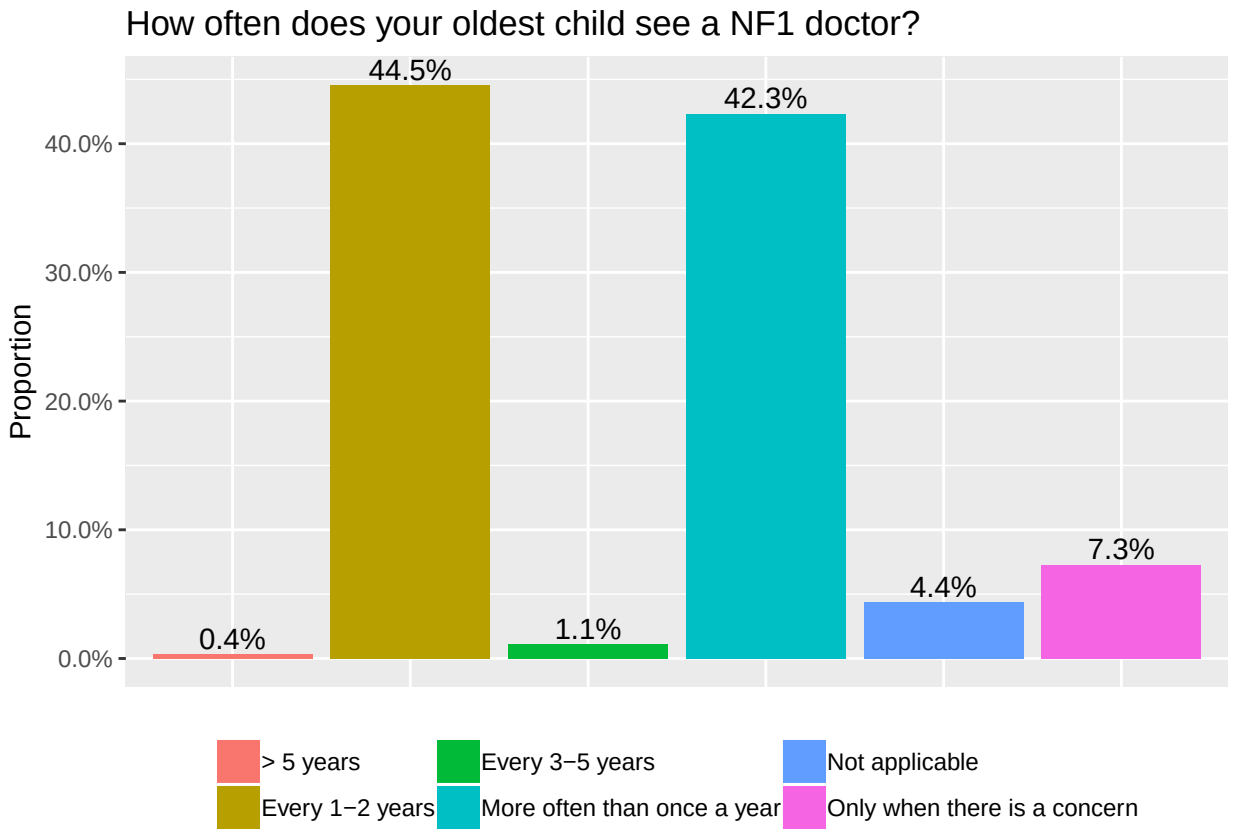
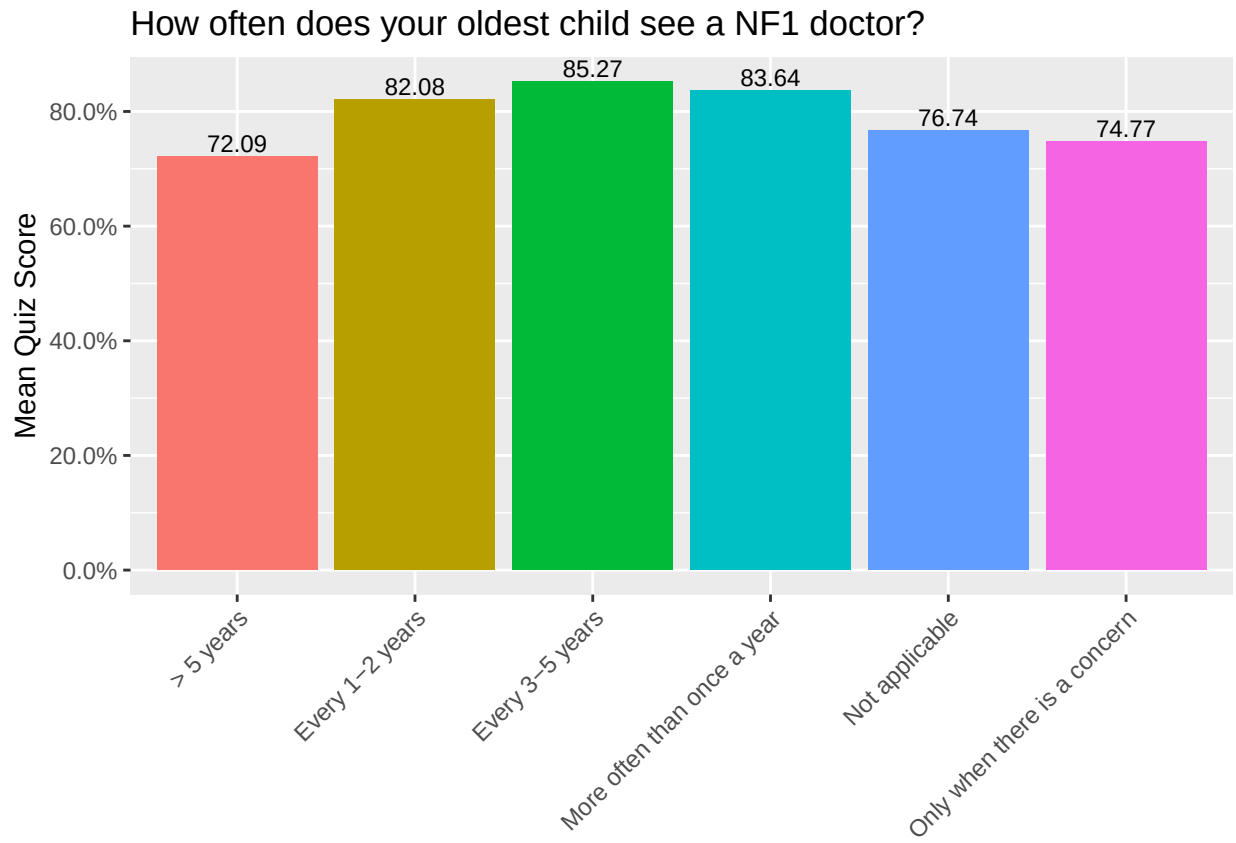
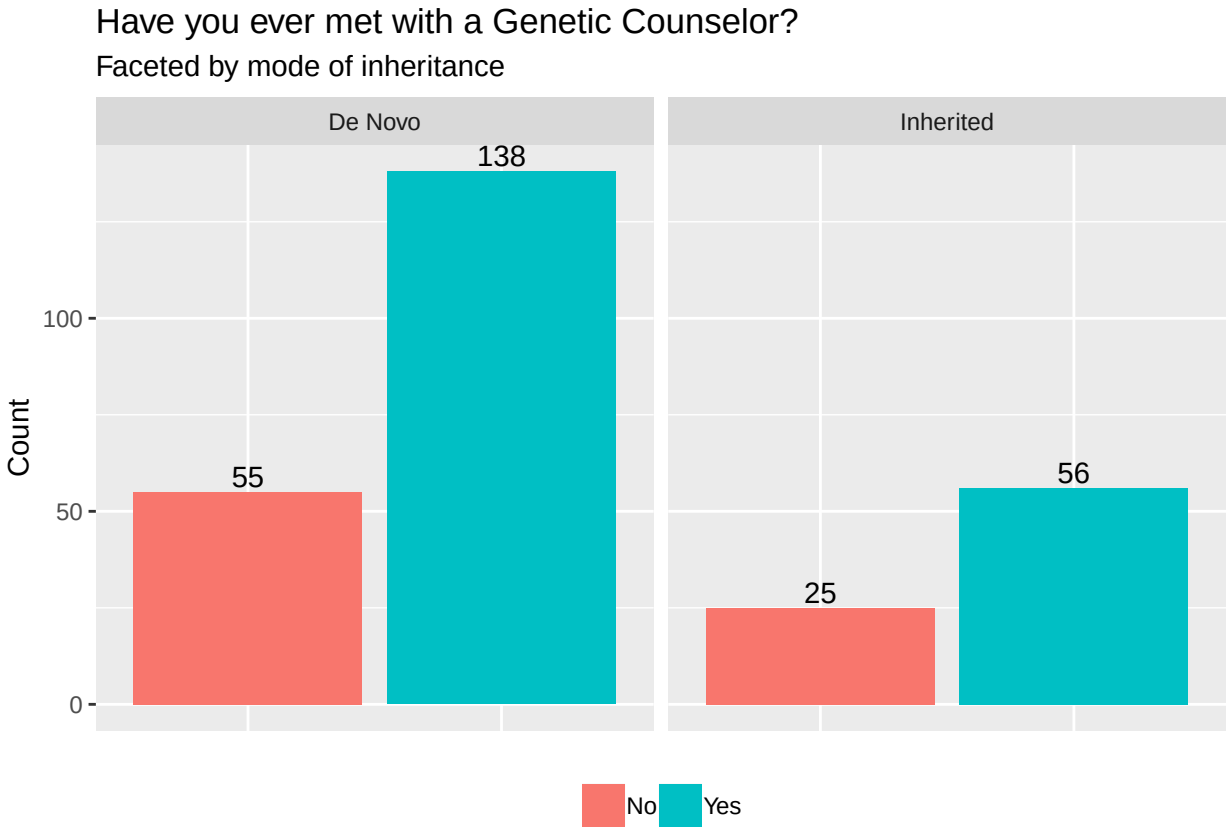


Table 7: How often does your oldest child see a NF1 doctor?

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?
Mean Quiz Score
80.0%
60.0%
40.0%
20.0%
0.0%
72.09
82.08
85.27
83.64
76.74
74.77
> 5 years
Every 1–2 years
Every 3–5 years
More often than once a year
Not applicable
Only when there is a concern


Have you ever met with a Genetic Counselor?
Faceted by mode of inheritance
De Novo
Inherited
Count
100
50
0
55
138
25
56
No
Yes

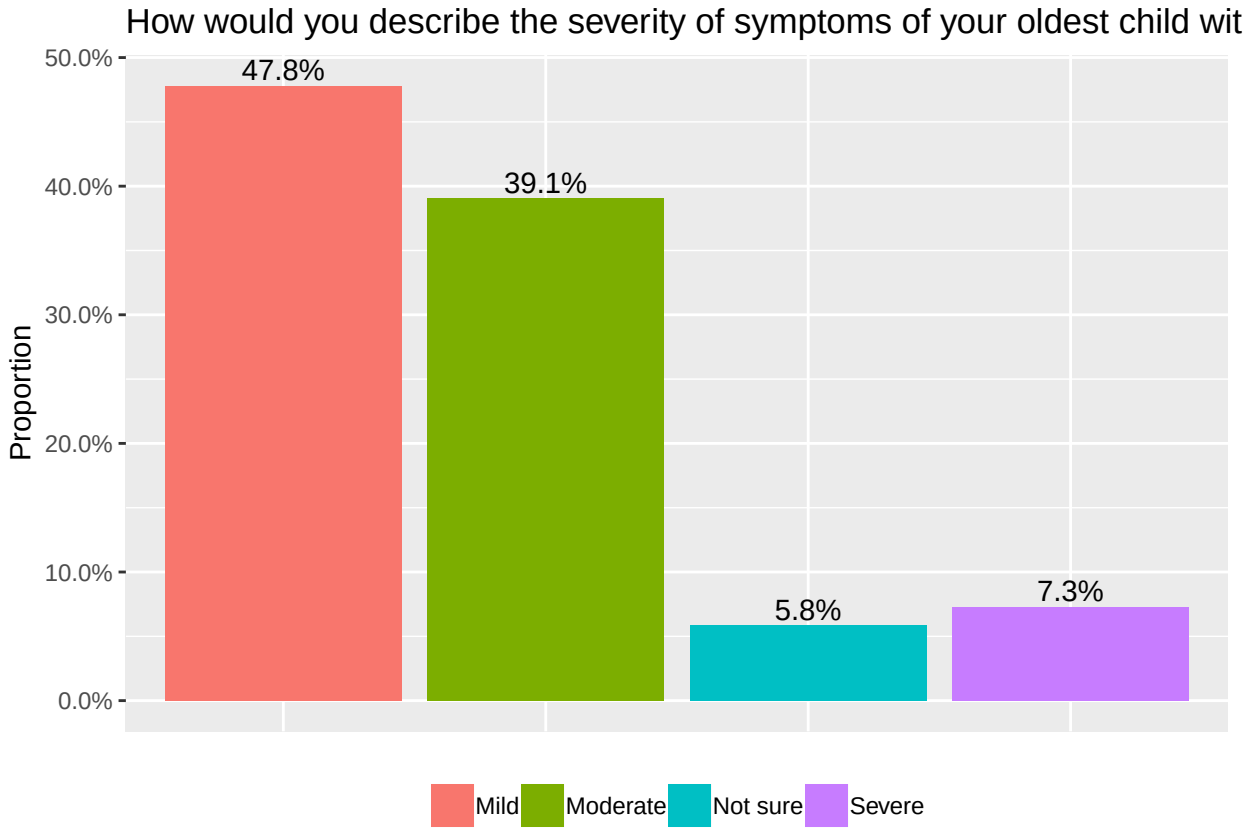


Table 8: How would you describe the severity of symptoms of your oldest child with NF1?

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_fa
## [24] "if_you_have_questions_regarding_nf1_do_you_obtain_knowledge_from_other_families_you_know_that_l
## [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] "one_half_or_50._of_genetic_information_is_passed_down_from_mother_to_child"
## [30] "there_is_more_than_one_gene_that_causes_nf1"
## [31] "a_father_can_pass_down_an_nf1_gene_mutation_to_his_daughters"
## [32] "all_people_who_have_an_nf1_gene_mutation_will_develop_cancer"
## [33] "nf1_symptoms_vary_from_one_person_to_another"
## [34] "a_person_is_born_with_nf1"
## [35] "nf1_can_skip_generations"
## [36] "if_a_woman_with_nf1_has_one_child_with_nf1_her_second_child_will_definitely_not_have_nf1"
## [37] "people_with_nf1_are_generally_more_likely_to_develop_cancer_at_a_younger_age"
## [38] "if_a_woman_with_nf1_has_scoliosis_then_her_child_will_also_develop_scoliosis"
## [39] "tumors_in_nf1_can_appear_anywhere_in_the_body"
## [40] "people_with_nf1_from_different_families_will_always_have_different_symptoms"
## [41] "cafe.au.lait_spots_.brown_marks_on_the_skin._are_often_the_first_sign_that_a_person_has_nf1"
## [42] "a_baby_with_nf1_may_be_born_with_a_tumor"
## [43] "all_cases_of_nf1_can_be_detected_in_a_womans_pregnancy_by_ultrasound"
## [44] "half_of_people_with_nf1_have_a_family_history_of_nf1"
## [45] "a_person_with_nf1_can_develop_tumors_that_may_lead_to_vision_loss_or_blindness"
## [46] "women_with_nf1_are_at_an_increased_risk_for_breast_cancer"
## [47] "is_scoliosis_associated_with_nf1."
## [48] "is_attention_deficit_hyperactivity_disorder_associated_with_nf1."
## [49] "are_clubbed_feet_associated_with_nf1."
## [50] "are_congenital_heart_defects_associated_with_nf1."
## [51] "are_seizures_associated_with_nf1."
## [52] "are_bumps_on_the_skin_associated_with_nf1."
## [53] "are_allergies_associated_with_nf1."
## [54] "is_high_blood_pressure_associated_with_nf1."
## [55] "are_learning_disabilities_associated_with_nf1."
## [56] "are_optic_gliomas_associated_with_nf1."
## [57] "is_infertility_associated_with_nf1."
## [58] "are_lisch_nodules_.dark_spots_on_the_iris._the_colored_part_of_the_eye._associated_with_nf1."
## [59] "is_small_head_size_associated_with_nf1."
## [60] "are_cataracts_associated_with_nf1."
## [61] "how_is_nf1_diagnosed."
## [62] "what_types_of_exams_may_be_helpful_when_trying_to_make_a_diagnosis_of_nf1."
## [63] "what_causes_nf1."
## [64] "what_is_the_function_of_the_nf1_gene_in_the_body."
## [65] "are_men_or_women_more_likely_to_be_affected_by_nf1."
## [66] "how_many_copies_of_a_non.working_nf1_gene_must_one_have_to_be_affected_with_nf1."
## [67] "if_a_person_with_nf1_has_a_child_what_is_the_chance_that_the_child_will_have_nf1."
## [68] "what_is_the_increased_risk_for_a_person_with_nf1_to_develop_cancer_in_their_lifetime_compared_
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## [69] "during_childhood_what_kind_of_screening_may_be_done_for_optic_gliomas_.eye_tumors.."
## [70] "how_do_the_symptoms_of_nf1_change_as_a_person_gets_older."
## [71] "what_is_the_most_concerning_symptom_of_a_tumor_to_suggest_it_has_become_malignant_.cancerous.."
## [72] "group_id"
## [73] "seen_gc"
## [74] "mode_of_inheritance"
```