```
library(tidyverse)

## -- Attaching packages ------------------------------------- tidyverse 1.3.1 --
## v ggplot2 3.3.6      v purrr   0.3.4
## v tibble  3.1.7      v dplyr   1.0.9
## v tidyr   1.2.0      v stringr 1.4.0
## v readr   2.1.2      v forcats 0.5.1

## -- Conflicts ---------------------------------------- tidyverse_conflicts() --
## x dplyr::filter() masks stats::filter()
## x dplyr::lag()    masks stats::lag()
numberFamilies <- 10000
load(str_interp("../RObjects/summary_tables/summaryTable10000Families_no_censoring.Rdata"))
```
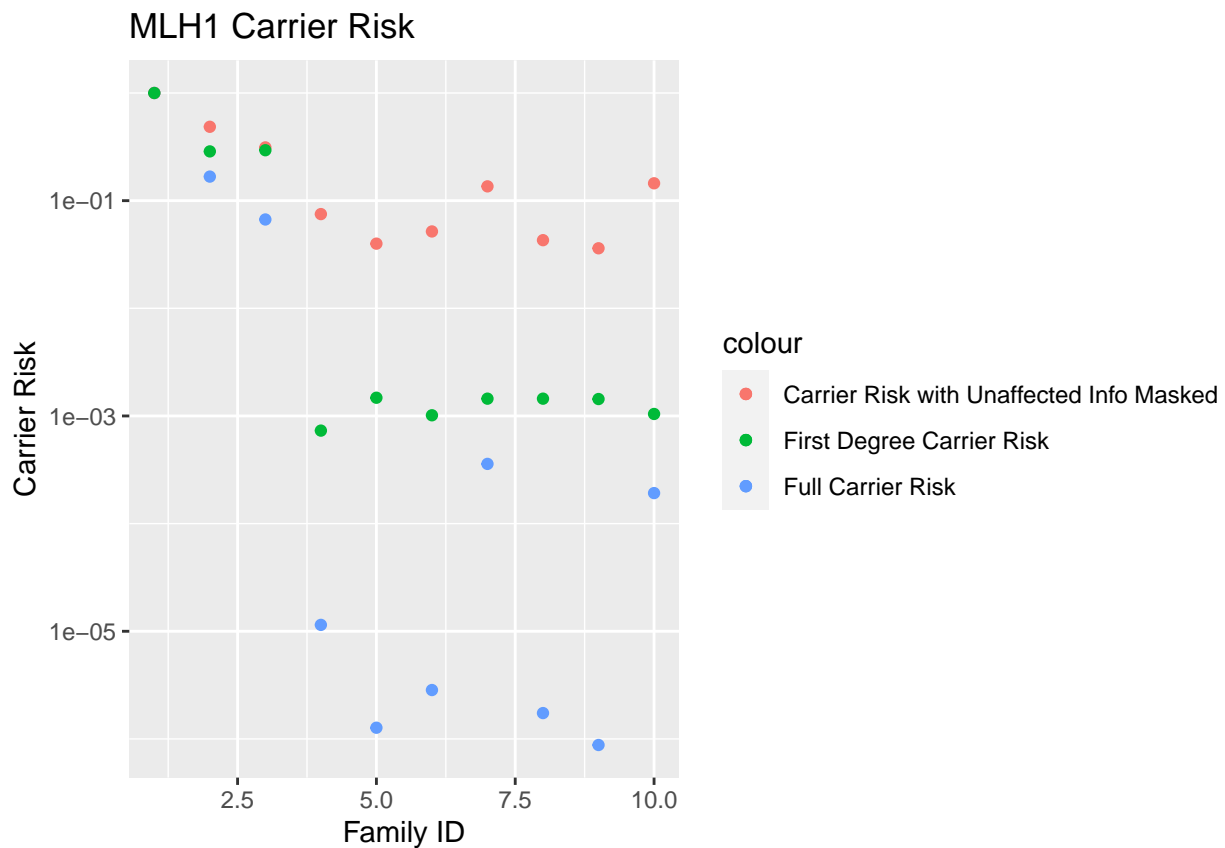
```
summaryTable <- summaryTable[1:10,]
logit <- function(x){
  log(x)-log(1-x)
}
visual = ggplot(data= summaryTable) +  geom_point(aes(x=famID, y=fullCarrierRisk, color="Full Carrier R
  geom_point(aes(x=famID, y=carrierRiskUnaffectedInfoMasked, color = "Carrier Risk with Unaffected Info
  geom_point(aes(x=famID, y= firstDegreeCarrierRisk, color = "First Degree Carrier Risk")) +
  scale_y_log10() +
  labs(title= "MLH1 Carrier Risk", x= "Family ID", y="Carrier Risk")

print(visual)
```

Next we will generate the same plot but only focus on the full family information
and the families with masked unaffected relatives

```
ggplot(data= summaryTable) +  geom_point(aes(x=famID, y=fullCarrierRisk, color="Full Carrier Risk")) +
  geom_point(aes(x=famID, y= carrierRiskUnaffectedInfoMasked, color = "Carrier Risk with Unaffected Info
  scale_y_log10() +
  labs(title= "MLH1 Carrier Risk", x= "Family ID", y="Carrier Risk")
```
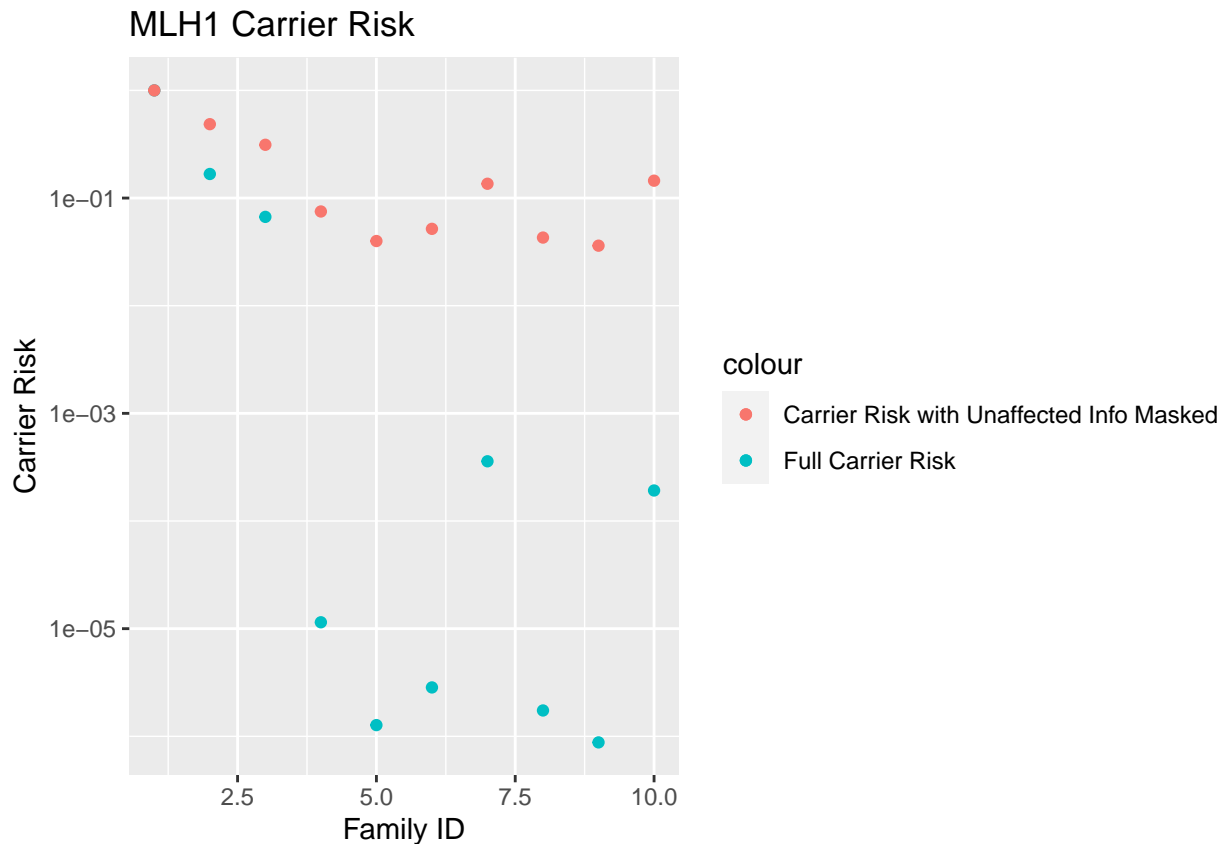
MLH1 Carrier Risk

This plot shows the difference between the carrier risk with the unaffected info masked and the full
carrier risk

## do box plots by proband status

```
summaryTable <- summaryTable %>%
                  mutate(diffMaskedFull = as.double(carrierRiskUnaffectedInfoMasked) - as.double(full


ggplot(data = summaryTable, aes(x = famNumber, y = diffMaskedFull)) +
  geom_point(aes(x = famID, y = diffMaskedFull, colour= as.factor(numAffectedRelatives), shape = proband
  labs(x = "Family ID", y = "Unaffected Masked Risk - Full Risk", title="Difference for Carrier Risk",
```

Difference for Carrier Risk
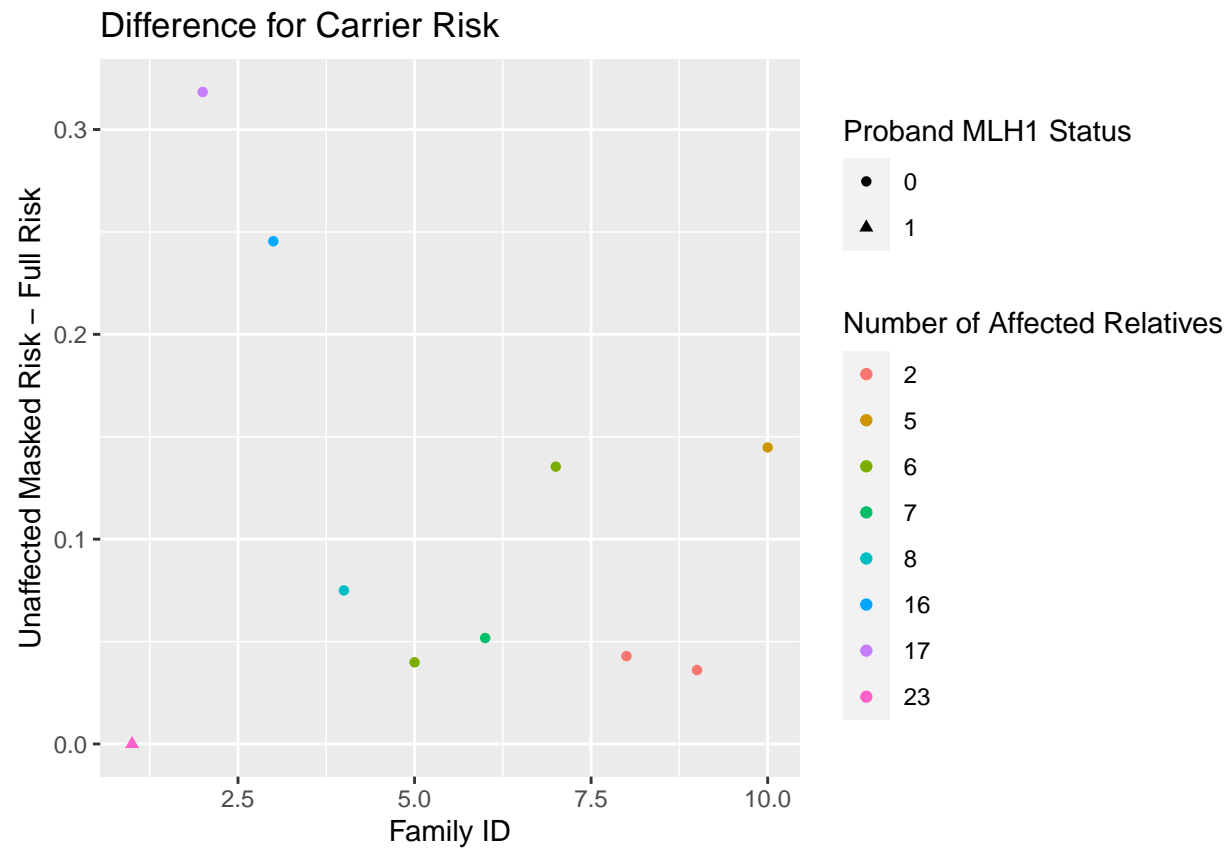
```
ggplot(data = summaryTable, aes(x = famNumber, y = diffMaskedFull)) +
  geom_point(aes(x = famID, y = diffMaskedFull, colour= as.factor(probandAffectionStatus), shape = proba
  labs(x = "Family ID", y = "Unaffected Masked Risk - Full Risk", title="Difference for Carrier Risk",
```

## Difference for Carrier Risk
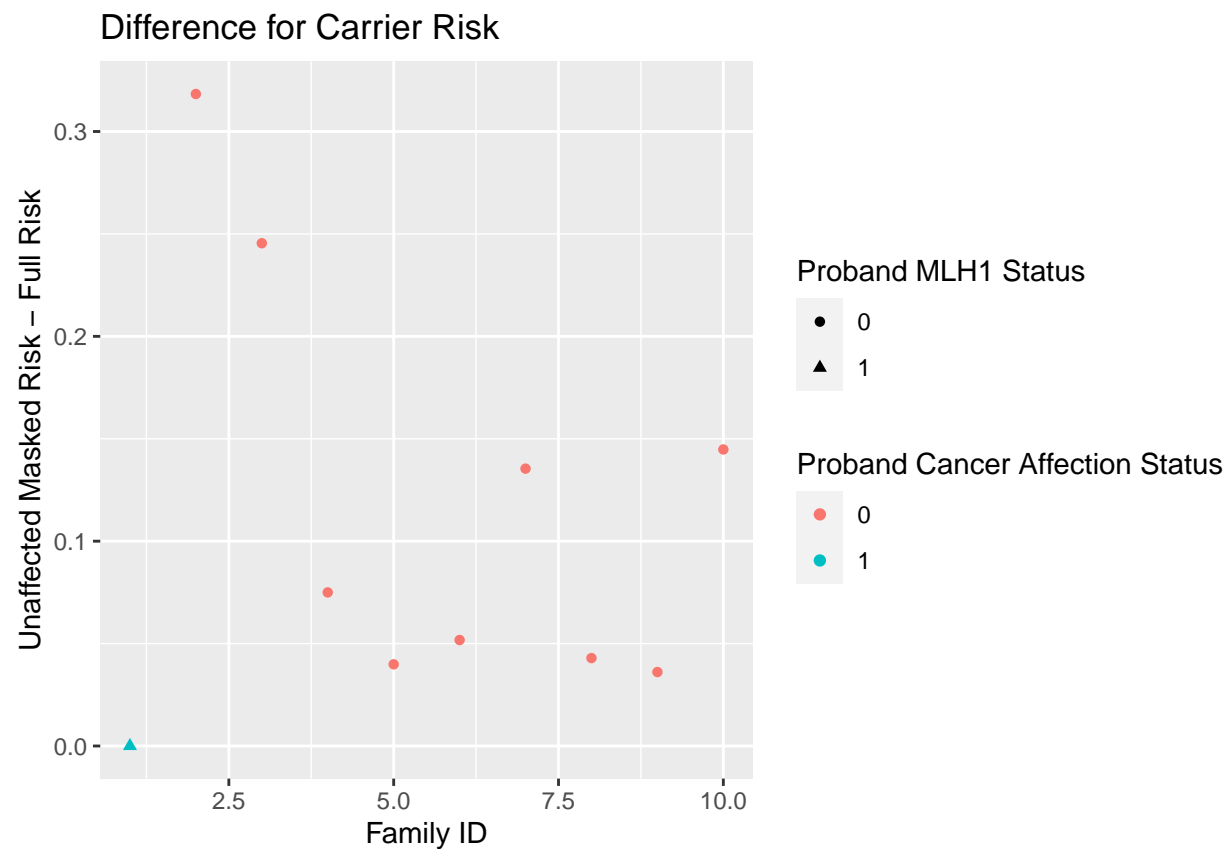

```
ggplot(data = summaryTable, aes(x = famNumber, y = diffMaskedFull)) +
  geom_point(aes(x = famID, y = diffMaskedFull, colour= as.factor(firstDegreeAffectedFamilyMembersBinary
  labs(x = "Family ID", y = "Unaffected Masked Risk - Full Risk", title="Difference for Carrier Risk", c
```
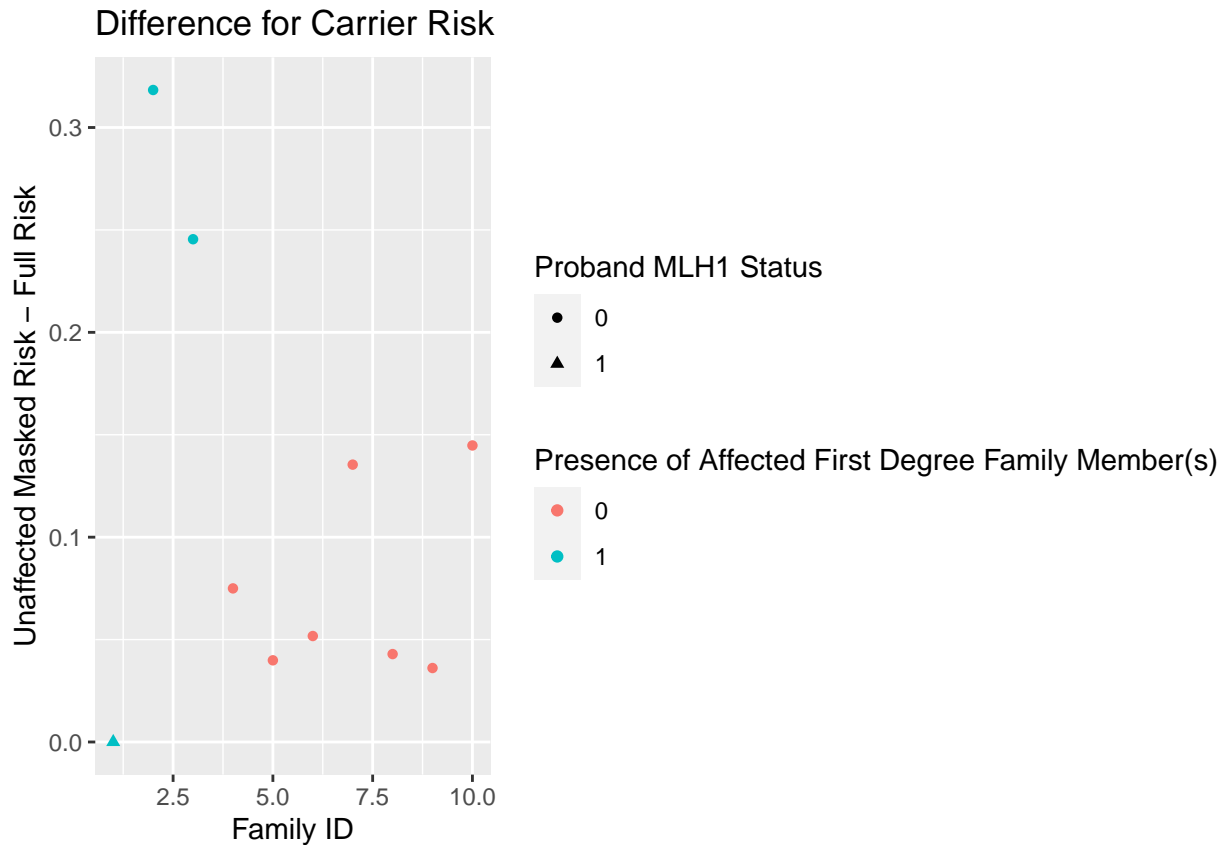
Next we will make a box plot to show by number of affected relatives what the difference in risk is

```
summaryTable$numAffectedRelativesGroup <- cut(summaryTable$numAffectedRelatives,
                breaks=c(-Inf, 0, 1,2, Inf),
                labels=c("0","1","2", "3+"),
                right = TRUE) #the breaks are inclusive on the right i.e. (-inf,0], (0,1]...
ggplot(summaryTable, aes(x=as.factor(numAffectedRelativesGroup), y=diffMaskedFull)) +
  geom_boxplot() +
  labs(x = "Number of Affected Relatives", y = "Unaffected Masked Risk - Full Risk", title="Difference
  stat_summary(
    fun.data = function(x) data.frame(y = max(x), label = paste0("n = ", length(x))),
    geom = "text", hjust = 0.3, vjust = -0.1, size = 4
  )
```
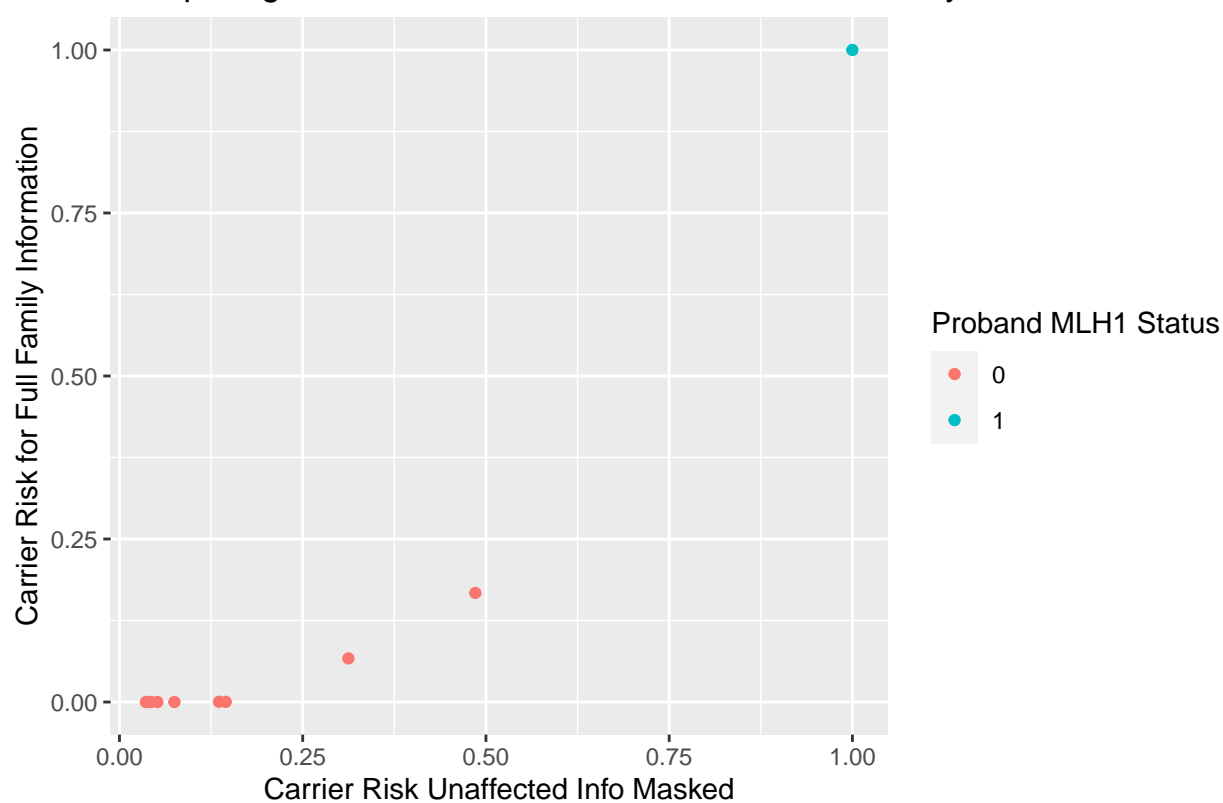
Difference for Carrier Risk by Number of Affected Family Members
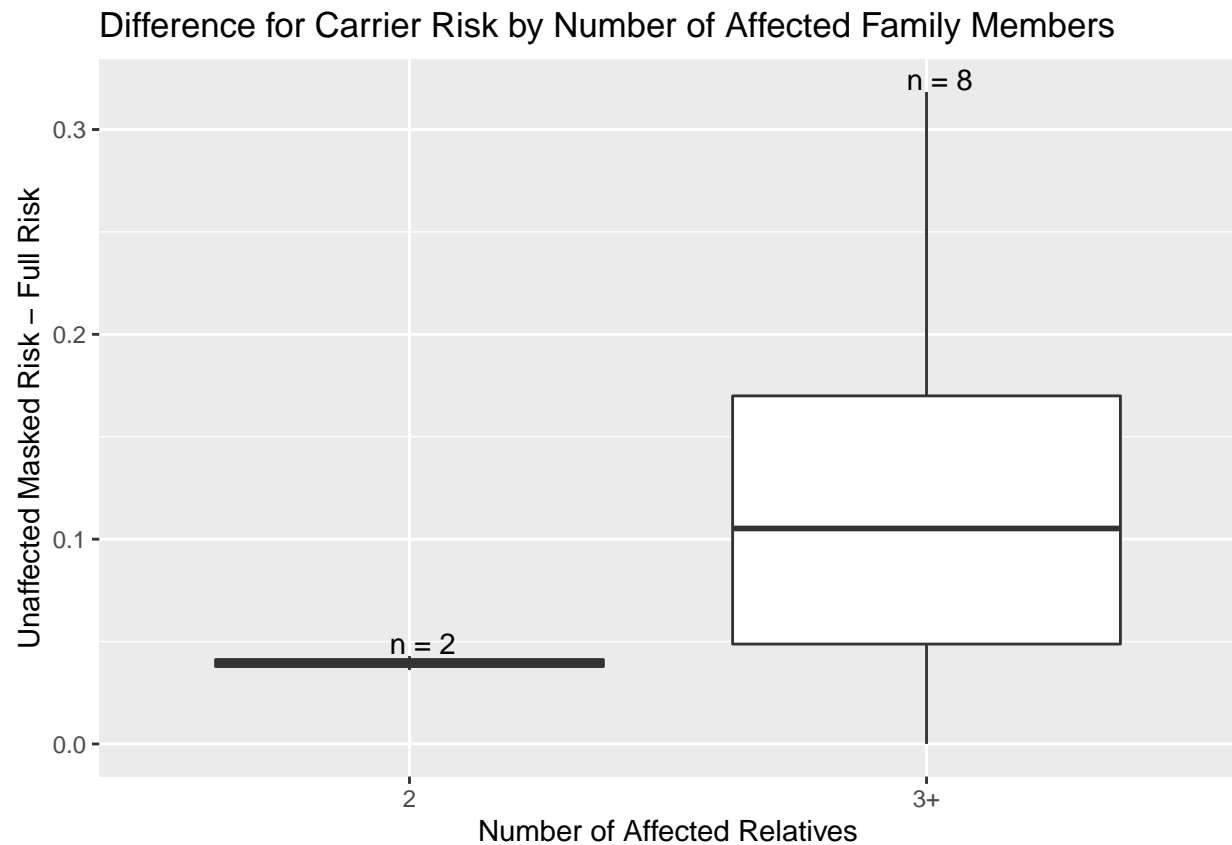
Next we will compare the carrier risk scores for the full families and the masked families
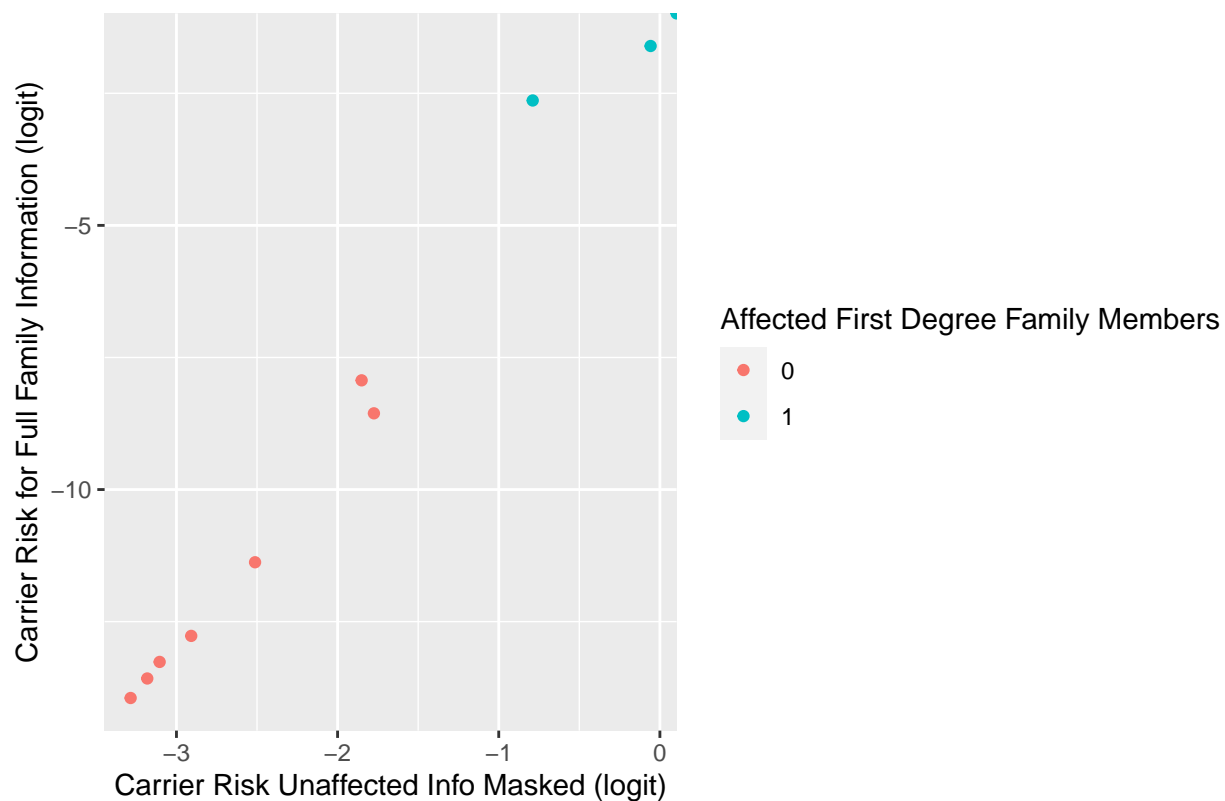
This will serve as a starting point for prediction

```
logit <- function(x){
  log(x)-log(1-x)
}
ggplot(data = summaryTable, aes(x= carrierRiskUnaffectedInfoMasked, y= fullCarrierRisk, color = probandl
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked", y= "Carrier Risk for Full Family Information", title =
```

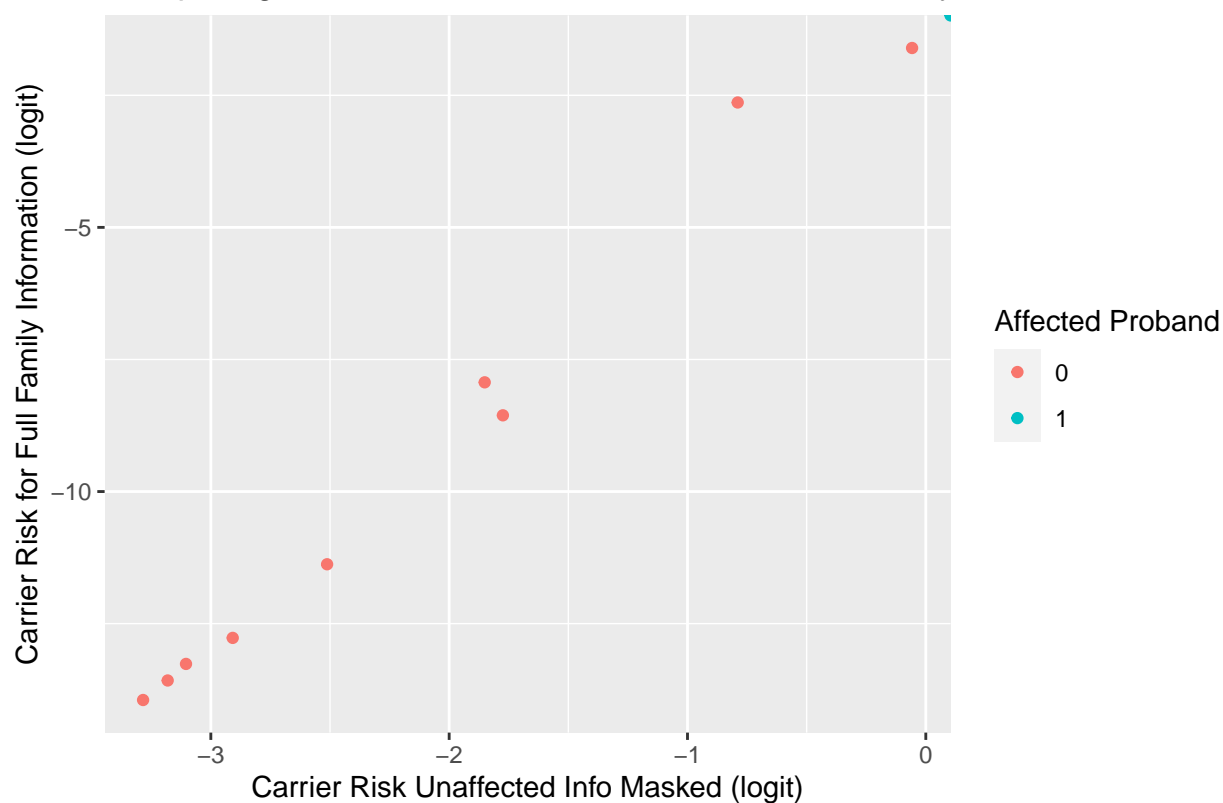## Comparing Carrier Risk for Masked Info and Full Family Information


```
logit <- function(x){
  log(x)-log(1-x)
}
ggplot(data = summaryTable, aes(x= logit(carrierRiskUnaffectedInfoMasked), y= logit(fullCarrierRisk), co
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked (logit)", y= "Carrier Risk for Full Family Information
```

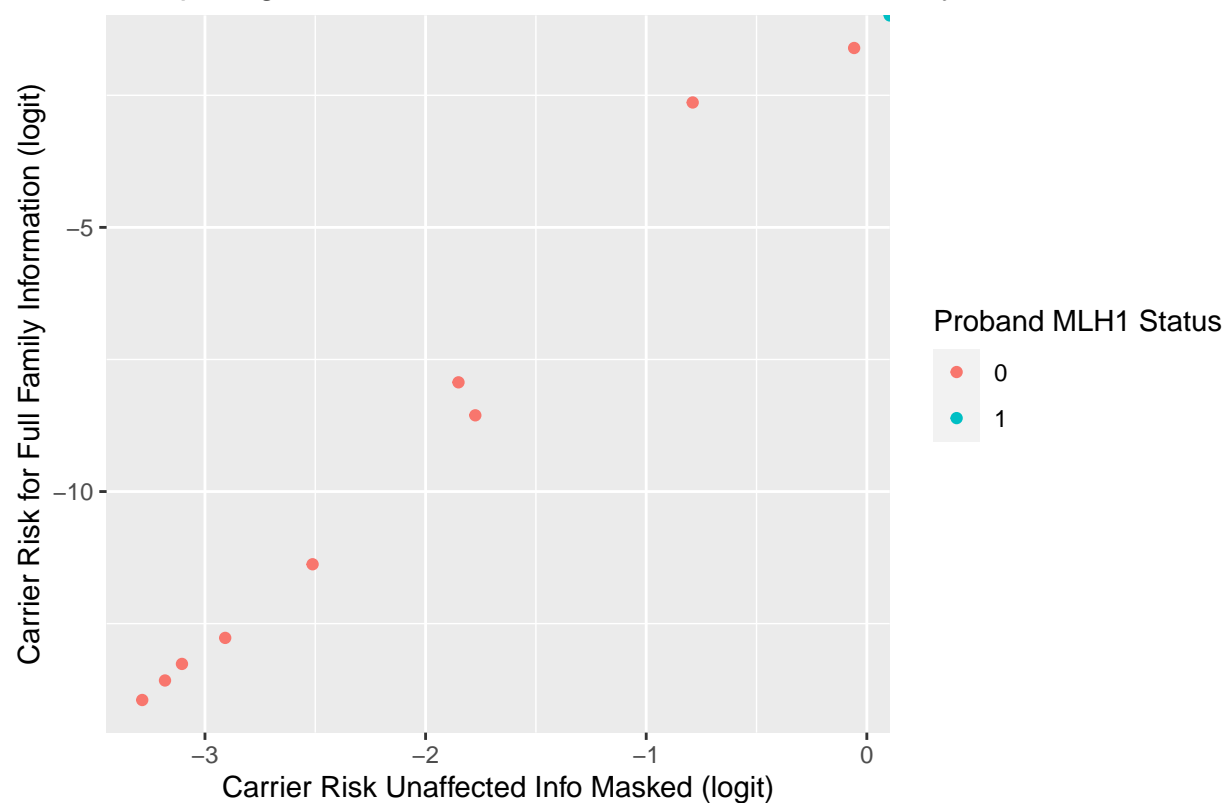# Comparing Carrier Risk for Masked Info and Full Family Information


```
logit <- function(x){
  log(x)-log(1-x)
}
ggplot(data = summaryTable, aes(x= logit(carrierRiskUnaffectedInfoMasked), y= logit(fullCarrierRisk), co
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked (logit)", y= "Carrier Risk for Full Family Information
```

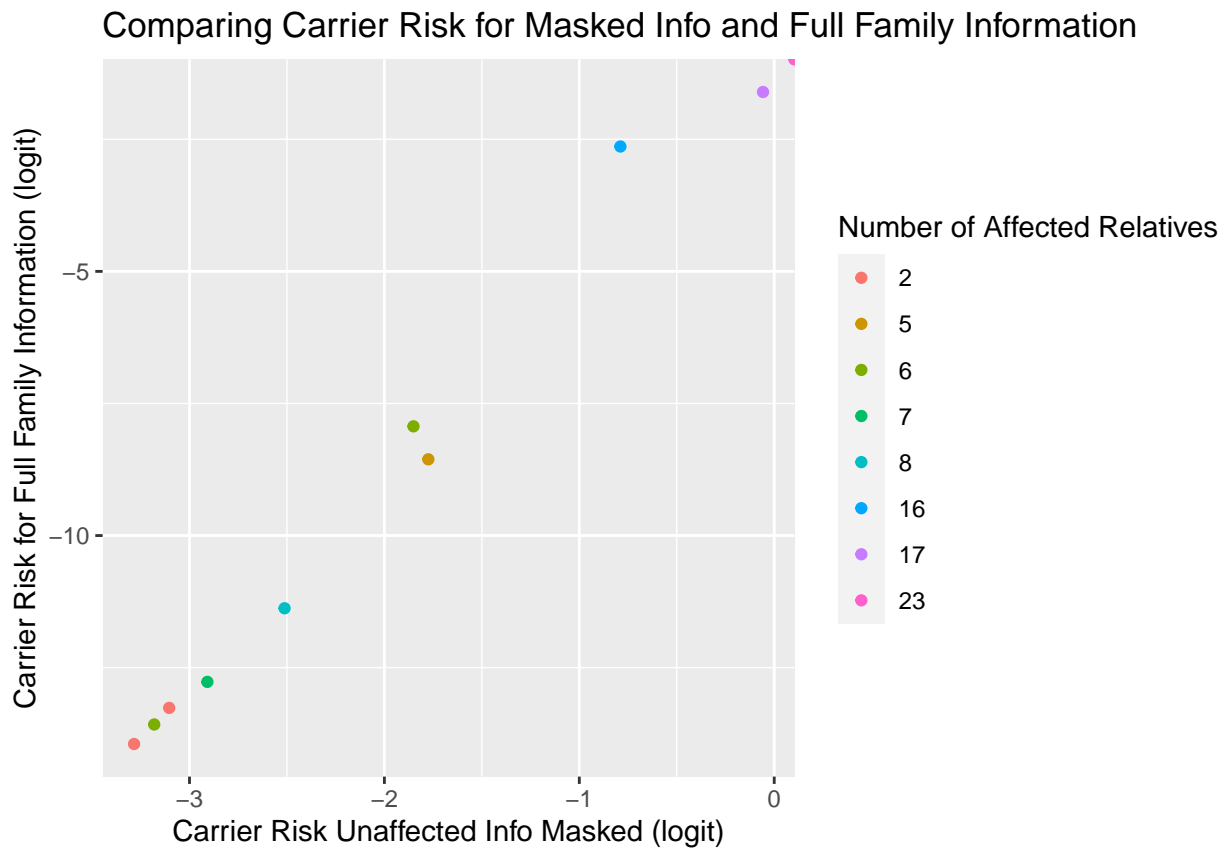# Comparing Carrier Risk for Masked Info and Full Family Information


```
logit <- function(x){
  log(x)-log(1-x)
}
ggplot(data = summaryTable, aes(x= logit(carrierRiskUnaffectedInfoMasked), y= logit(fullCarrierRisk), c
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked (logit)", y= "Carrier Risk for Full Family Information
```

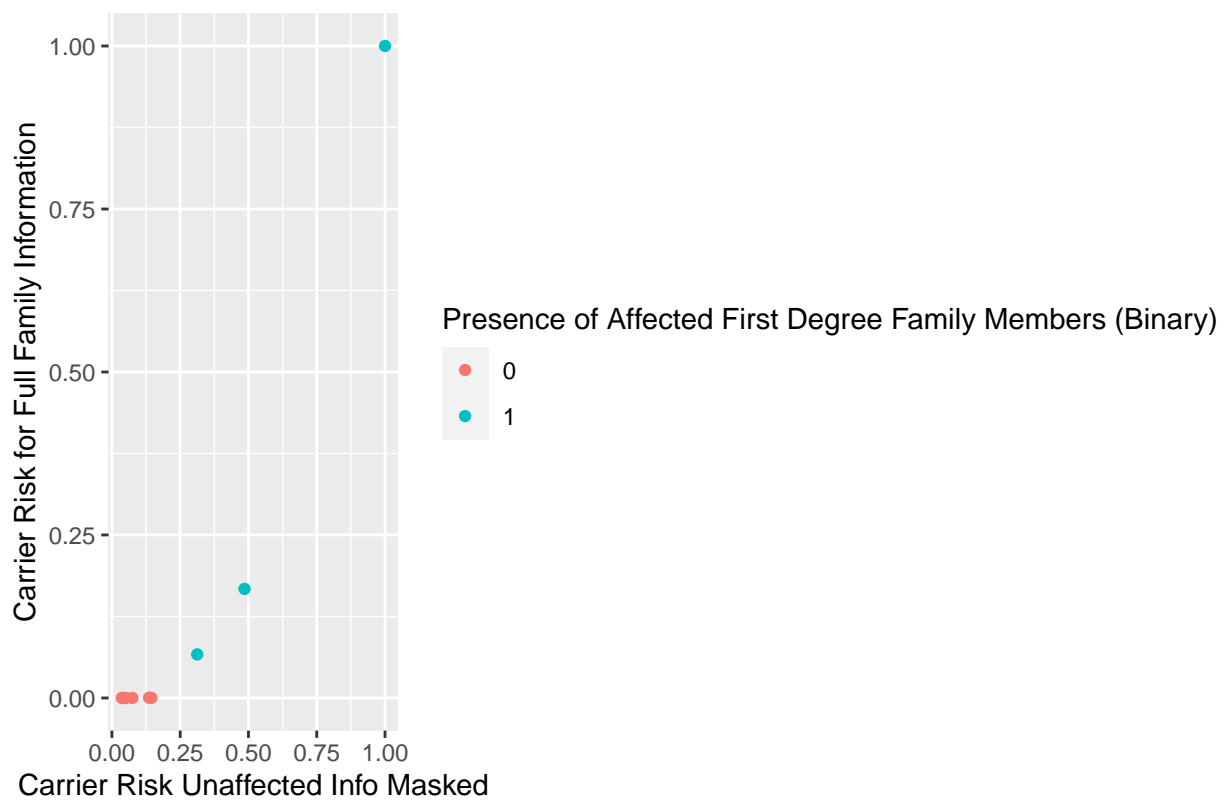## Comparing Carrier Risk for Masked Info and Full Family Information


```
logit <- function(x){
  log(x)-log(1-x)
}
ggplot(data = summaryTable, aes(x= logit(carrierRiskUnaffectedInfoMasked), y= logit(fullCarrierRisk), c
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked (logit)", y= "Carrier Risk for Full Family Information
```

Comparing Carrier Risk for Masked Info and Full Family Information

Here we will color by the number of affected relatives to see the relationship between number of affected relatives and carrier risk scores
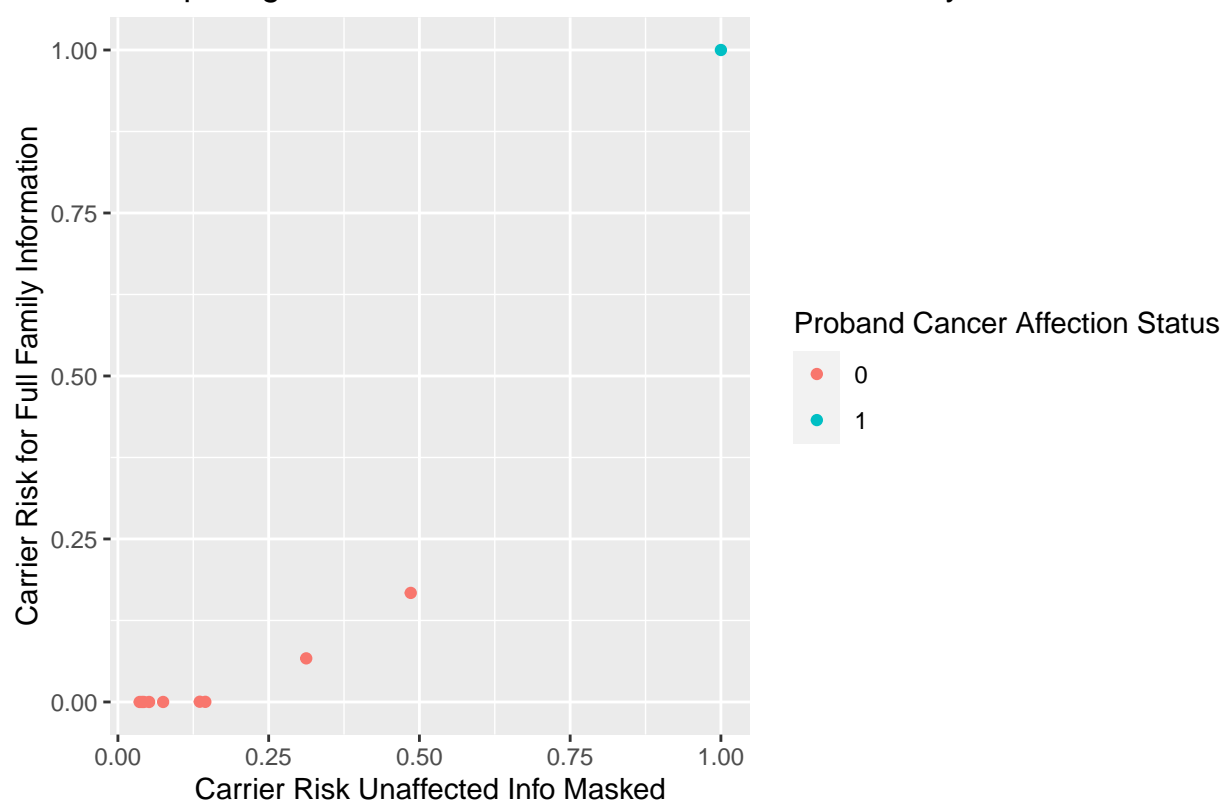
```
ggplot(data = summaryTable, aes(x= carrierRiskUnaffectedInfoMasked, y= fullCarrierRisk, color = as.fact
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked", y= "Carrier Risk for Full Family Information", title =
```

Comparing Carrier Risk for Masked Info and Full Family Information with Li


```
ggplot(data = summaryTable, aes(x= carrierRiskUnaffectedInfoMasked, y= fullCarrierRisk, color = as.fact
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked", y= "Carrier Risk for Full Family Information", title =
```

Comparing Carrier Risk for Masked Info and Full Family Information with Li


```r
ggplot(data = summaryTable, aes(x= carrierRiskUnaffectedInfoMasked, y= fullCarrierRisk, color = as.facto
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked", y= "Carrier Risk for Full Family Information", title =
```
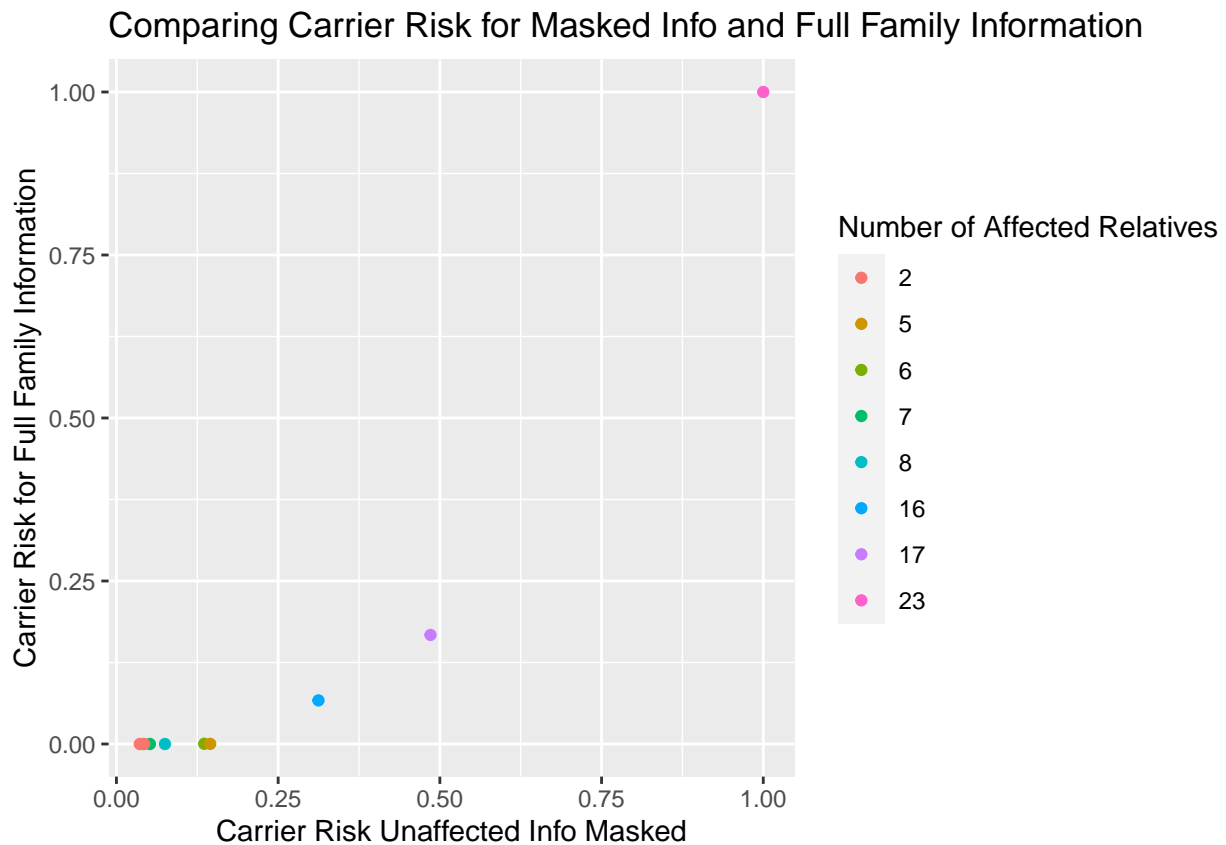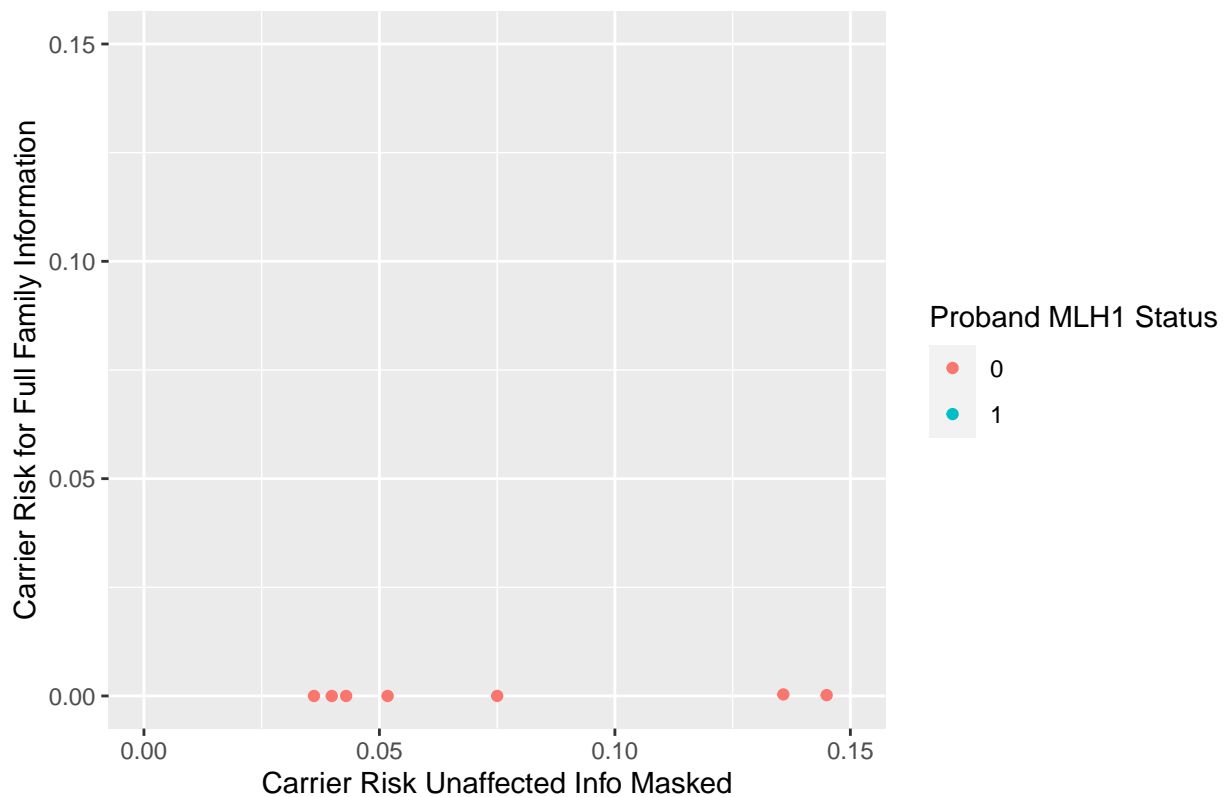
Comparing Carrier Risk for Masked Info and Full Family Information

Zoom in to the bottom left corner to see if there are any trends there

```
ggplot(data = summaryTable, aes(x= carrierRiskUnaffectedInfoMasked, y= fullCarrierRisk, color = probandl
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked", y= "Carrier Risk for Full Family Information", title =
  xlim(0, 0.15) + ylim(0, 0.15)
```

```
## Warning: Removed 3 rows containing missing values (geom_point).
```

Comparing Carrier Risk for Masked Info and Full Family Information Zoome

use more distinct colors for the color scale

an alternative to use 0, 1, 2, 3+ as the categories of the families numaffectedrels

```
ggplot(data = summaryTable, aes(x= carrierRiskUnaffectedInfoMasked, y= fullCarrierRisk, color = as.fact
  geom_point() +
  labs(x = "Carrier Risk Unaffected Info Masked", y= "Carrier Risk for Full Family Information", title =
  xlim(0, 0.15) + ylim(0, 0.15)
```

```
## Warning: Removed 3 rows containing missing values (geom_point).
```

Comparing Carrier Risk for Masked Info and Full Family Information Zoome