

Checking Error Caused by Using Masked Carrier Risk

```
library(tidyverse)
load("RObjects/summary_tables/summaryTable10000Families.Rdata")
```

Using All Families

```
# calculate average predicted value
avg_full <- mean(summaryTable$fullCarrierRisk) #adjusted means adjusted via prediction model
avg_masked <- mean(summaryTable$carrierRiskUnaffectedInfoMasked)
avg_true <- mean(as.numeric(summaryTable$probandMLH1Status))

sum_full <- sum(summaryTable$fullCarrierRisk) #adjusted means adjusted via prediction model
sum_masked <- sum(summaryTable$carrierRiskUnaffectedInfoMasked)
sum_true <- sum(as.numeric(summaryTable$probandMLH1Status))

# print average predicted value
print(str_interp("Average full carrier risk score: ${avg_full}"))
## [1] "Average full carrier risk score: 0.08222868608331"
print(str_interp("Average masked (unadjusted) carrier risk score: ${avg_masked}"))
## [1] "Average masked (unadjusted) carrier risk score: 0.138178890715236"
print(str_interp("Average number of true carriers of MLH1: ${avg_true}"))
## [1] "Average number of true carriers of MLH1: 0.0978"

mse_full <- mean(as.numeric(summaryTable$probandMLH1Status) - summaryTable$fullCarrierRisk**2)
mse_masked <- mean(as.numeric(summaryTable$probandMLH1Status) - summaryTable$carrierRiskUnaffectedInfoMasked**2)
print(str_interp("Full MSE: ${mse_full}"))
## [1] "Full MSE: 0.0646001237149446"
print(str_interp("Masked MSE: ${mse_masked}"))
## [1] "Masked MSE: 0.0557651202136598"
```

Using Families with Unaffected First Degree Relatives (U1)

```
summaryTableFirstDegUnaff <- summaryTable %>% filter(firstDegreeAffectedFamilyMembersBinary == 0)

# calculate average predicted value
avg_full <- mean(summaryTableFirstDegUnaff$fullCarrierRisk) #adjusted means adjusted via prediction model
avg_masked <- mean(summaryTableFirstDegUnaff$carrierRiskUnaffectedInfoMasked)
avg_true <- mean(as.numeric(summaryTableFirstDegUnaff$probandMLH1Status))

# print average predicted value
print(str_interp("Average full carrier risk score: ${avg_full}"))
## [1] "Average full carrier risk score: 0.0332079222774842"
print(str_interp("Average masked carrier risk score: ${avg_masked}"))
## [1] "Average masked carrier risk score: 0.0952801099097885"
print(str_interp("Average number of true carriers of MLH1: ${avg_true}"))
## [1] "Average number of true carriers of MLH1: 0.046973803071364"
```

```

mse_full <- mean(as.numeric(summaryTableFirstDegUnaff$probandMLH1Status) - summaryTableFirstDegUnaff$fu
mse_masked <- mean(as.numeric(summaryTableFirstDegUnaff$probandMLH1Status) - summaryTableFirstDegUnaff$
print(str_interp("Full MSE: ${mse_full}"))
## [1] "Full MSE: 0.0399033849297642"
print(str_interp("Masked MSE: ${mse_masked}"))
## [1] "Masked MSE: 0.0304030635567485"

```

Using Families with Affected First Degree Relatives (A1)

```

summaryTableFirstDegAff <- summaryTable %>% filter(firstDegreeAffectedFamilyMembersBinary == 1)

# calculate average predicted value
avg_full <- mean(summaryTableFirstDegAff$fullCarrierRisk) #adjusted means adjusted via prediction model
avg_masked <- mean(summaryTableFirstDegAff$carrierRiskUnaffectedInfoMasked)
avg_true <- mean(as.numeric(summaryTableFirstDegAff$probandMLH1Status))
# print average predicted value
print(str_interp("Average full carrier risk score: ${avg_full}"))
## [1] "Average full carrier risk score: 0.461711102398339"
print(str_interp("Average masked carrier risk score: ${avg_masked}"))
## [1] "Average masked carrier risk score: 0.47026945261475"
print(str_interp("Average number of true carriers of MLH1: ${avg_true}"))
## [1] "Average number of true carriers of MLH1: 0.491258741258741"

mse_full <- mean(as.numeric(summaryTableFirstDegAff$probandMLH1Status) - summaryTableFirstDegAff$fullCa
mse_masked <- mean(as.numeric(summaryTableFirstDegAff$probandMLH1Status) - summaryTableFirstDegAff$carr
print(str_interp("Full MSE: ${mse_full}"))
## [1] "Full MSE: 0.255783968716306"
print(str_interp("Masked MSE: ${mse_masked}"))
## [1] "Masked MSE: 0.252099363005274"

```