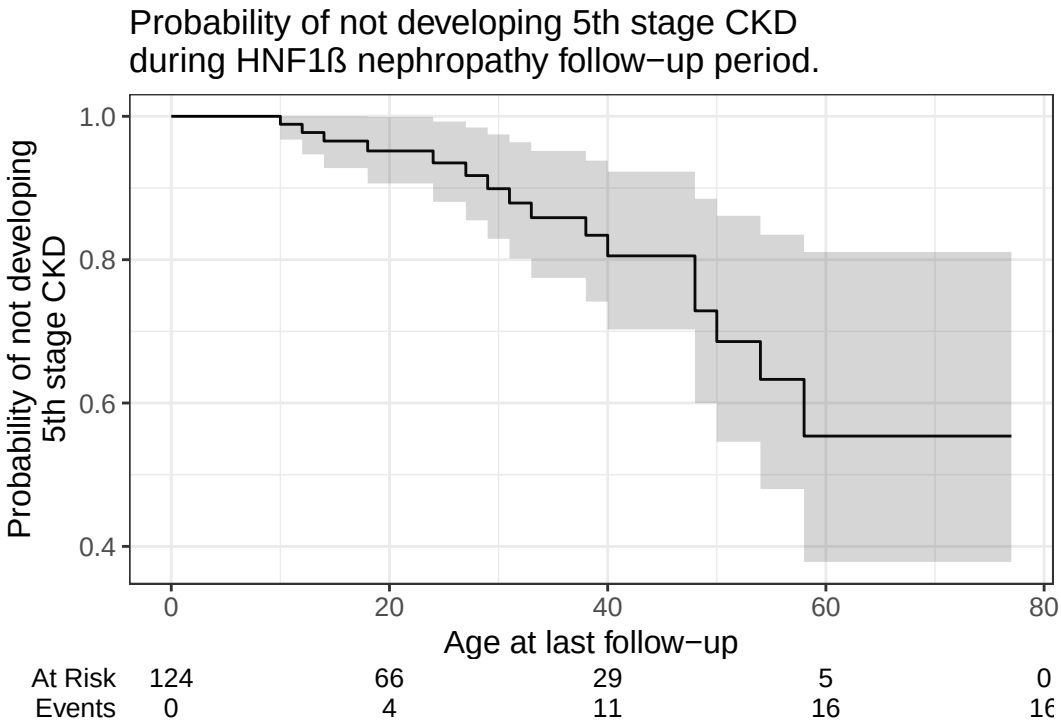
Lauryna_plots

Table of contents

1 Total Incidence

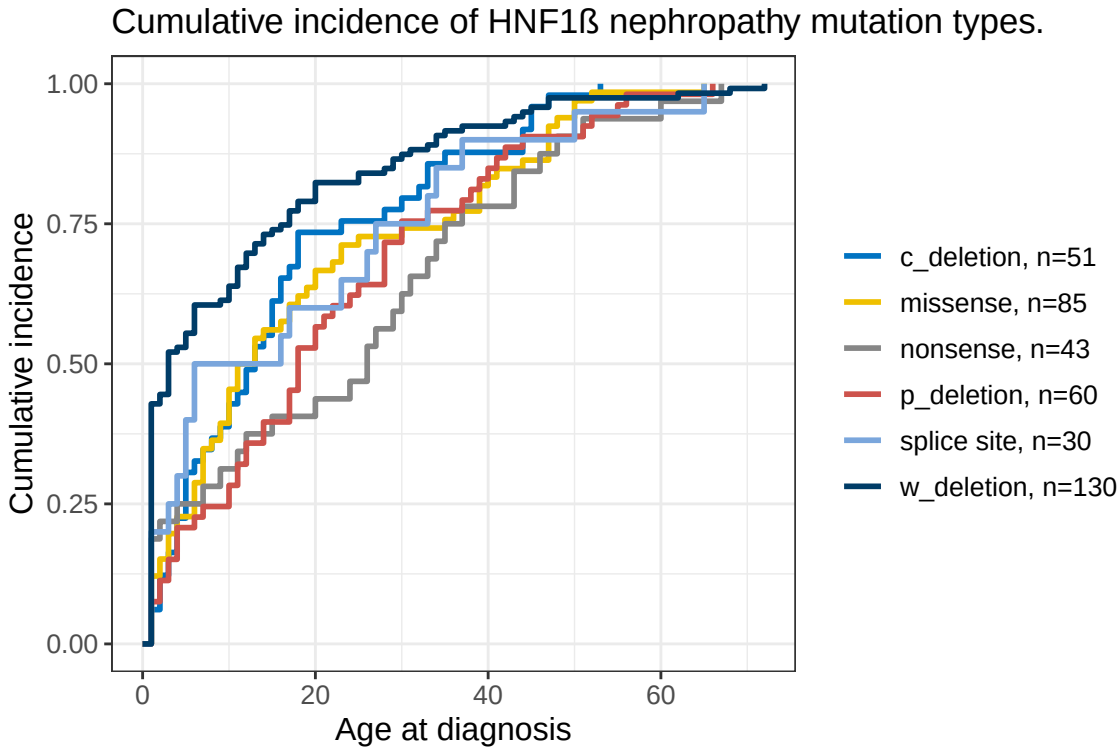
0.5 incidence at 11 years. 50% atvejų nustatoma ties 11 metų.

2 5th CKD stage probability



3 General mutation types Incidence

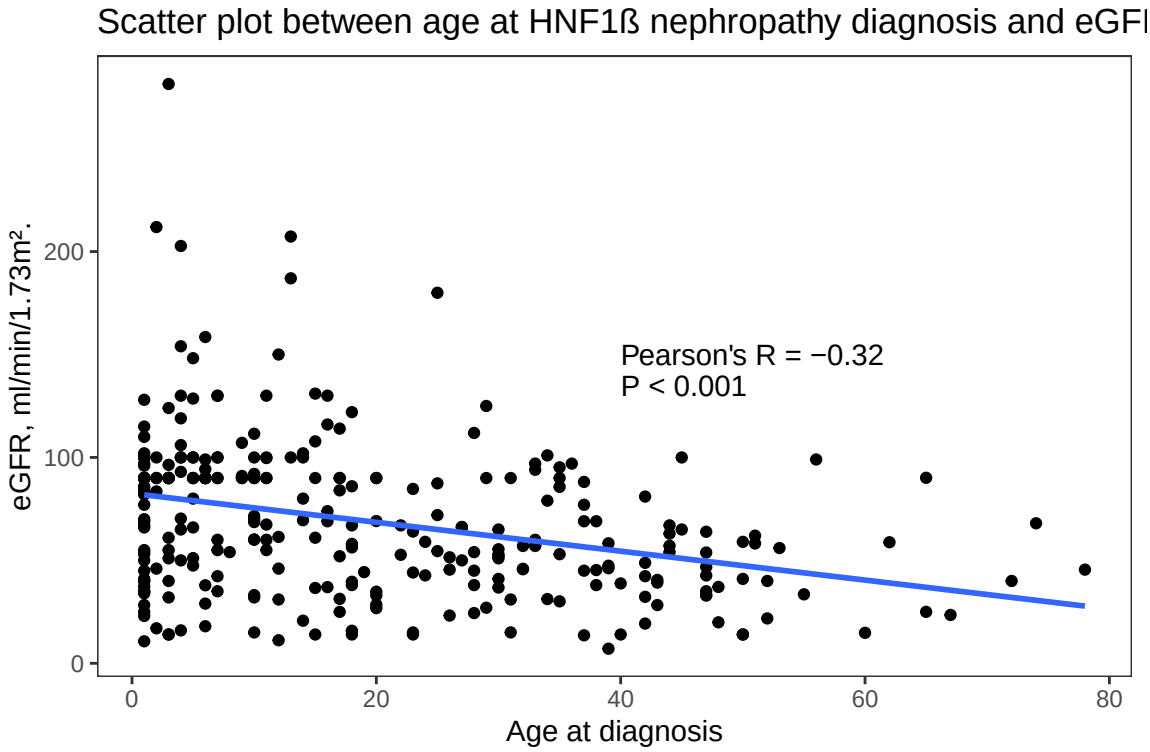
```
# A tibble: 6 x 2
  mutacijos_tipas     n
  <chr>           <int>
1 cdeletion          51
2 missense           85
3 nonsense           43
4 pdeletion          60
5 splice             30
6 wdeletion         130
```



4 GFG scatterplot

4.1 eGFR ~Amžius

```
[1] 294  43
```



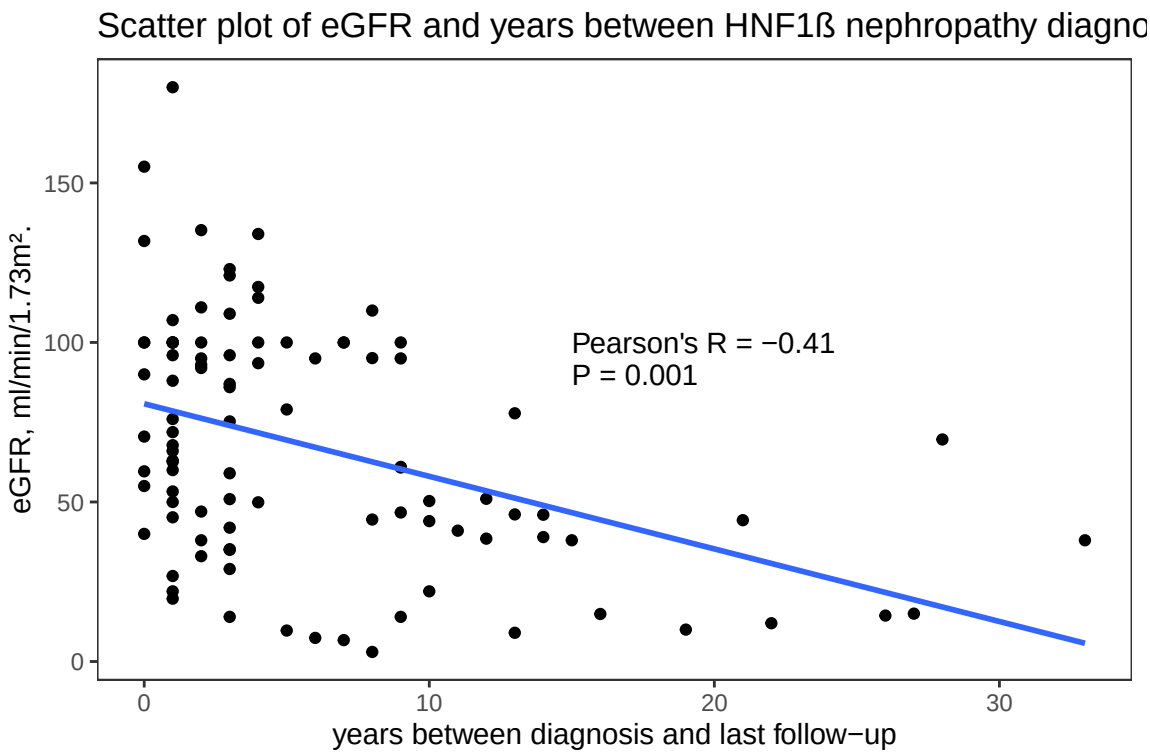

```
Call:
lm(formula = eGFR_pre ~ age_pre, data = HNF1B_simple)

Residuals:
    Min      1Q  Median      3Q     Max
-71.132 -24.195  -1.035  17.921 200.972

Coefficients:
            Estimate Std. Error t value Pr(>|t|)
(Intercept)  82.5336     3.1238  26.421  < 2e-16 ***
age_pre      -0.7018     0.1206  -5.817 1.57e-08 ***
---
Signif. codes:  0 '***' 0.001 '**' 0.01 '*' 0.05 '.' 0.1 ' ' 1

Residual standard error: 35.99 on 292 degrees of freedom
  (233 observations deleted due to missingness)
Multiple R-squared:  0.1038,    Adjusted R-squared:  0.1008
F-statistic: 33.84 on 1 and 292 DF,  p-value: 1.574e-08
```

Visgi eGFR daugiausiai priklauso nuo CKD stadijos

```
Call:
lm(formula = eGFR_pre ~ age_pre + CKD.stage...7, data = HNF1B_simple)

Residuals:
    Min      1Q  Median      3Q     Max
-21.265 -11.474  -4.156   5.402 170.607

Coefficients:
               Estimate Std. Error t value Pr(>|t|)
(Intercept)   140.17659    3.18289  44.041   <2e-16 ***
age_pre        -0.14409    0.07486  -1.925   0.0554 .
CKD.stage...7 -28.95142    1.18163 -24.501   <2e-16 ***
---
Signif. codes:  0 '***' 0.001 '**' 0.01 '*' 0.05 '.' 0.1 ' ' 1

Residual standard error: 20.44 on 252 degrees of freedom
  (272 observations deleted due to missingness)
Multiple R-squared:  0.7299,    Adjusted R-squared:  0.7278
F-statistic: 340.5 on 2 and 252 DF,  p-value: < 2.2e-16



Call:
lm(formula = eGFR_pre ~ CKD.stage...7, data = HNF1B_simple)

Residuals:
    Min      1Q  Median      3Q     Max
-19.934 -11.957  -4.734   5.643 173.043

Coefficients:
              Estimate Std. Error t value Pr(>|t|)
(Intercept)    137.518      2.993   45.94   <2e-16 ***
CKD.stage...7  -29.161      1.100  -26.50   <2e-16 ***
---
Signif. codes:  0 '***' 0.001 '**' 0.01 '*' 0.05 '.' 0.1 ' ' 1

Residual standard error: 20.5 on 267 degrees of freedom
  (258 observations deleted due to missingness)
Multiple R-squared:  0.7246,    Adjusted R-squared:  0.7236
F-statistic: 702.5 on 1 and 267 DF,  p-value: < 2.2e-16
```

4.2 eGFR followup ~ Amžiaus skirtumas

```
[1] 97 44
```



5 HNF1B mutacijos tipo įtaka CKD stadijai

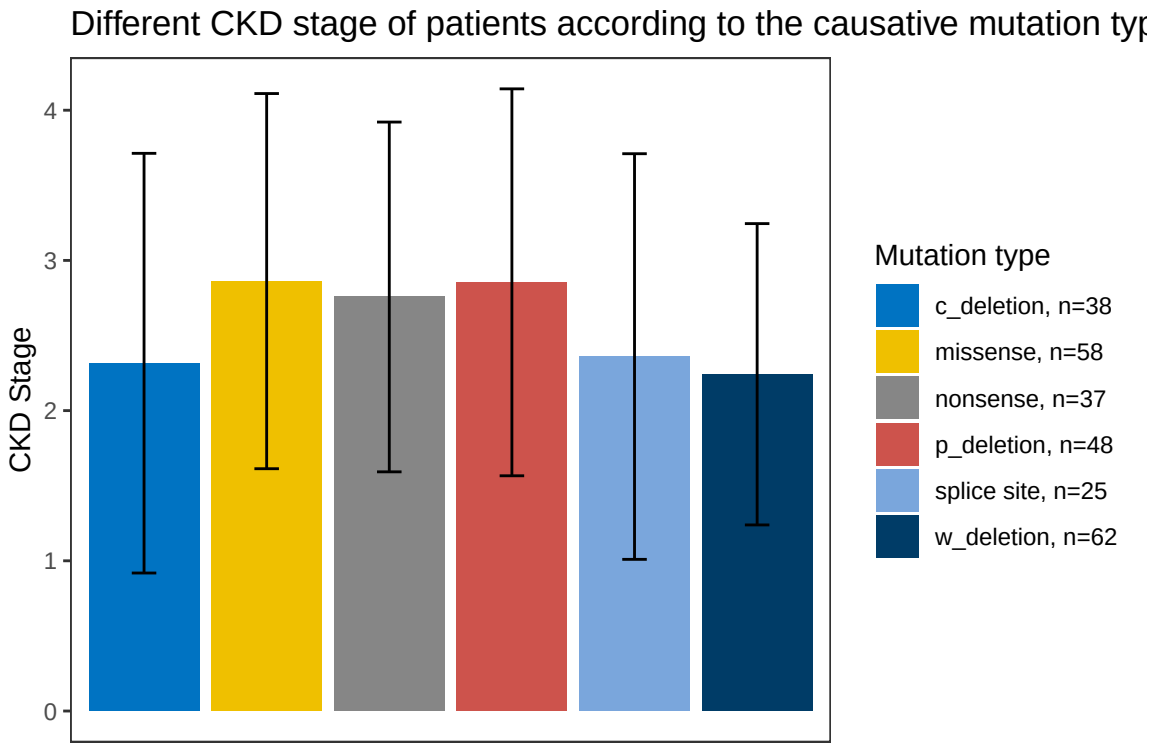
Visos mutacijos

```
# A tibble: 12 x 2
   mutacijos_tipas                      n
   <chr>                            <int>
 1 ?deletion                           78
 2 cdeletion                           51
 3 duplication                          6
 4 frameshift                          10
 5 heterozygous intragenic mutation     5
 6 insertion                            7
 7 missense                            85
```

```
 8 nonsense                          43
 9 pdeletion                         60
10 splice                            30
11 wdeletion                        130
12 <NA>                              22
```

Atrinktos mutacijos

```
# A tibble: 7 x 4
  mutacijos_tipas     n  mean    sd
  <chr>           <int> <dbl> <dbl>
1 ?deletion          54  1.61 0.960
2 cdeletion          38  2.32 1.40
3 missense           58  2.86 1.25
4 nonsense           37  2.76 1.16
5 pdeletion          48  2.85 1.29
6 splice             25  2.36 1.35
7 wdeletion          62  2.24 1.00
```



5.1 ANOVA testas

```
                 Df Sum Sq Mean Sq F value Pr(>F)
mutacijos_tipas   5   20.3   4.065    2.71 0.0208 *
Residuals       262  393.0   1.500
---
Signif. codes:  0 '***' 0.001 '**' 0.01 '*' 0.05 '.' 0.1 ' ' 1
```

5.2 Tukey post-hoc (pairwise comparisons)

```
  Tukey multiple comparisons of means
    95% family-wise confidence level

Fit: aov(formula = CKD.stage...7 ~ mutacijos_tipas, data = anova_data)

$mutacijos_tipas
                            diff        lwr        upr     p adj
missense-cdeletion   0.546279492 -0.1875823 1.28014131 0.2715671
nonsense-cdeletion   0.440967283 -0.3711569 1.25309146 0.6263824
pdeletion-cdeletion  0.538377193 -0.2251443 1.30189871 0.3313253
splice-cdeletion     0.044210526 -0.8612988 0.94971988 0.9999924
wdeletion-cdeletion -0.073853990 -0.7982847 0.65057668 0.9997093
nonsense-missense   -0.105312209 -0.8451413 0.63451690 0.9985262
pdeletion-missense  -0.007902299 -0.6940266 0.67822198 1.0000000
splice-missense     -0.502068966 -1.3433469 0.33920900 0.5241819
wdeletion-missense  -0.620133482 -1.2624739 0.02220694 0.0653085
pdeletion-nonsense   0.097409910 -0.6718489 0.86666867 0.9991628
splice-nonsense     -0.396756757 -1.3071090 0.51359545 0.8109616
wdeletion-nonsense  -0.514821273 -1.2452963 0.21565373 0.3318840
splice-pdeletion    -0.494166667 -1.3614386 0.37310524 0.5755547
wdeletion-pdeletion -0.612231183 -1.2882587 0.06379630 0.1008721
wdeletion-splice    -0.118064516 -0.9511283 0.71499925 0.9985570
```

6 CKD stage

6.1 CKD diagnozės metu duomenys

```
# A tibble: 5 x 2
  CKD.stage...7     n
          <dbl> <int>
```

```
1             1   110
2             2    85
3             3   114
4             4    30
5             5    31
```

6.2 Pacientai su CKD duomenim diagnozes ir follow-up metu

6.2.1 diagnozes

```
# A tibble: 5 x 2
  CKD.stage...7     n
          <dbl> <int>
1             1    23
2             2    17
3             3    19
4             4     8
5             5     1
```

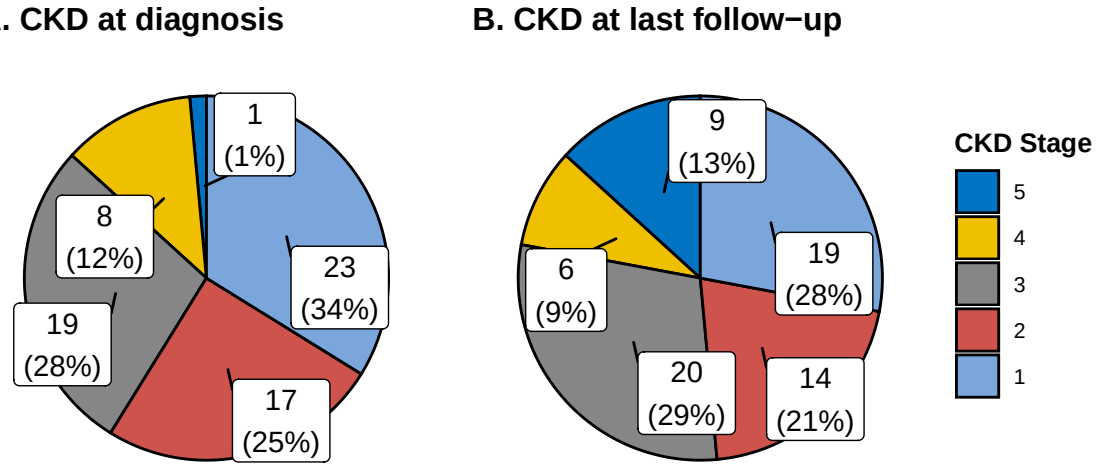
6.2.2 follow-up

```
# A tibble: 5 x 2
  CKD.stage...20     n
           <dbl> <int>
1              1    19
2              2    14
3              3    20
4              4     6
5              5     9
```

6.2.3 Pie charts

```
    Cochran's Q test

data:  CKD_stage and CKD_time and Patient.ID
Q = 15.114, df = 1, p-value = 0.0001012
```

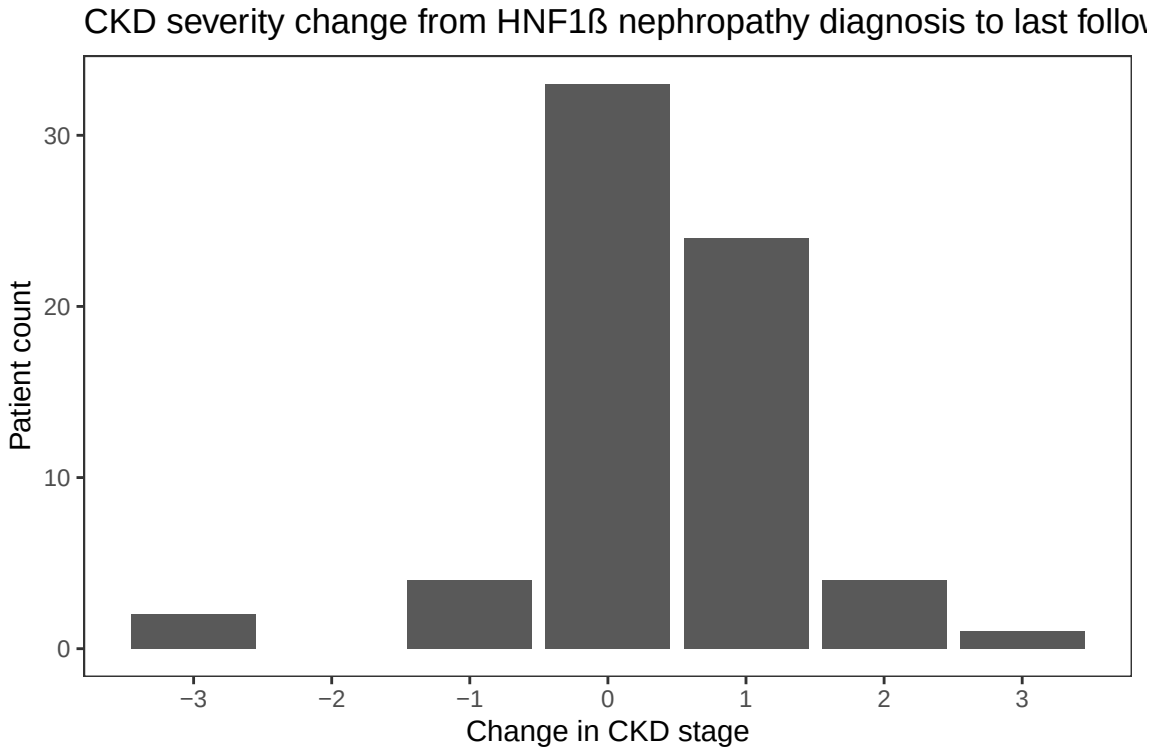
rtions of patients with different CKD stages at HNF1ß nosis and last follow−up (n = 68).



Cochran's Q test: $X^2(1)=15.114$, $p < 0.00$

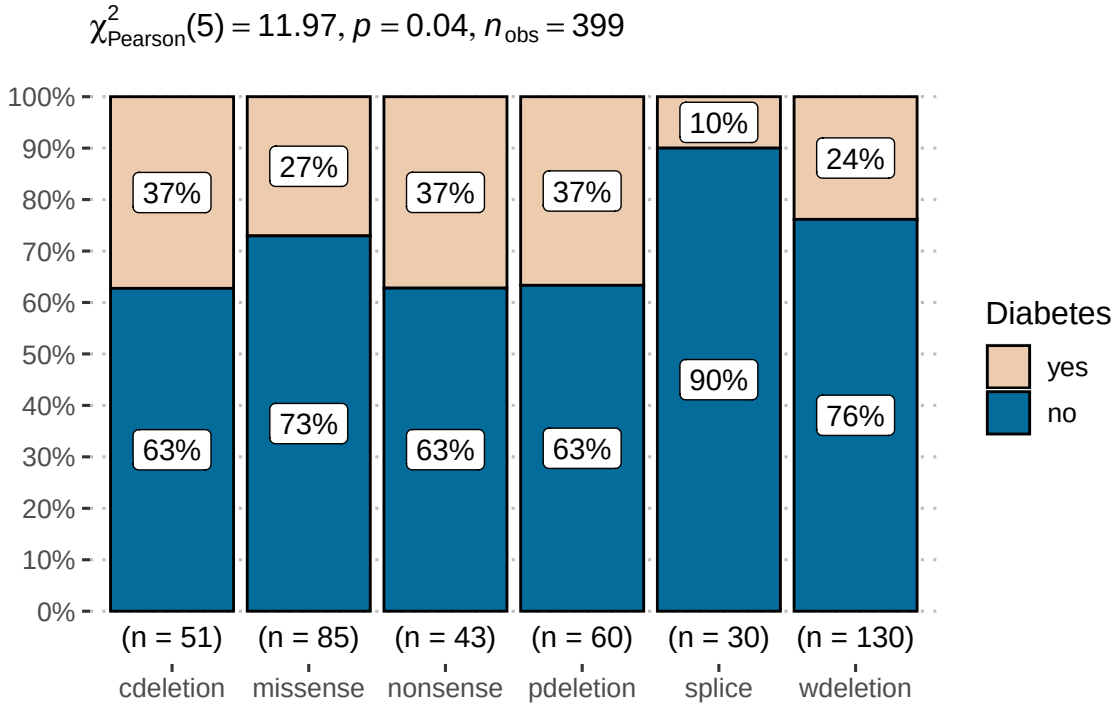
6.2.4 Histogram

Žiūrime kaip keičiasi CKD lygis nuo diagnosis iki follow-up

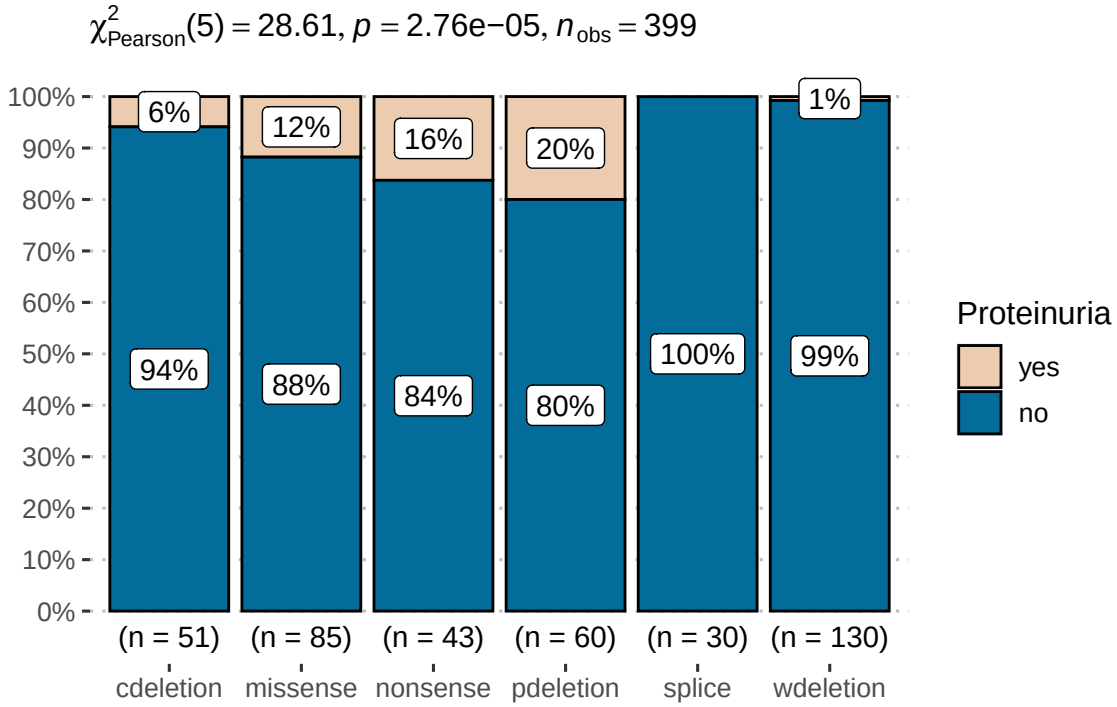

CKD severity change from HNF1ß nephropathy diagnosis to last follo
Patient count
30
20
10
0
−3
−2
−1
0
1
2
3
Change in CKD stage

7 Categorical Data

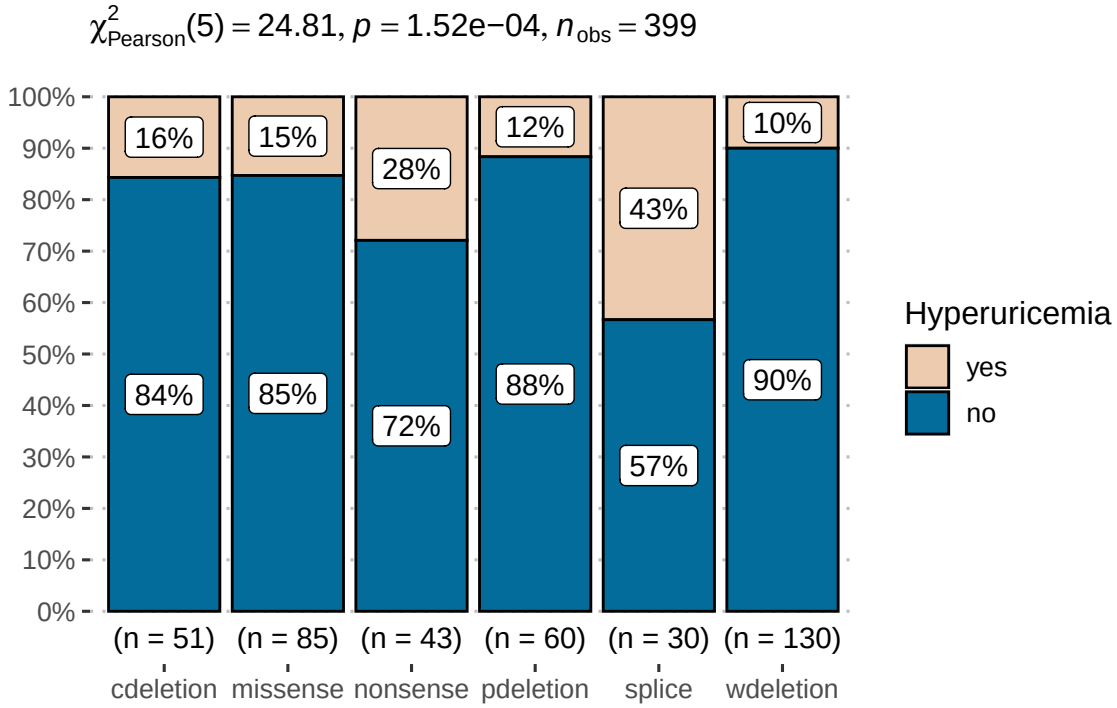
7.1 Diabetes



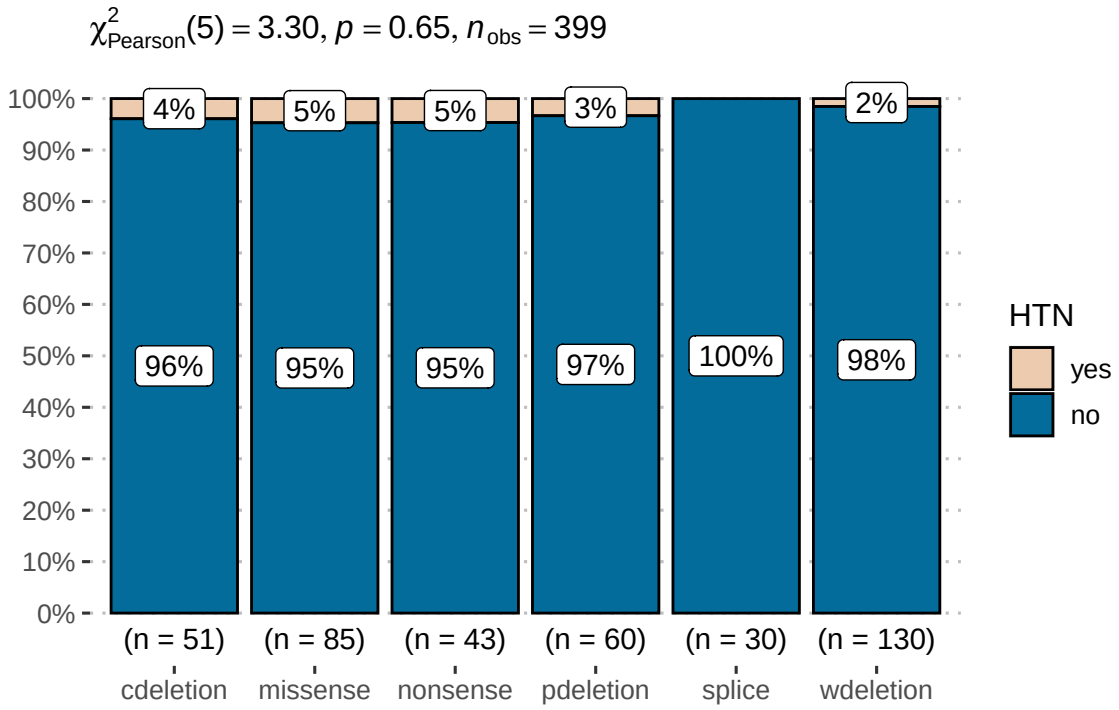
7.2 Proteinuria



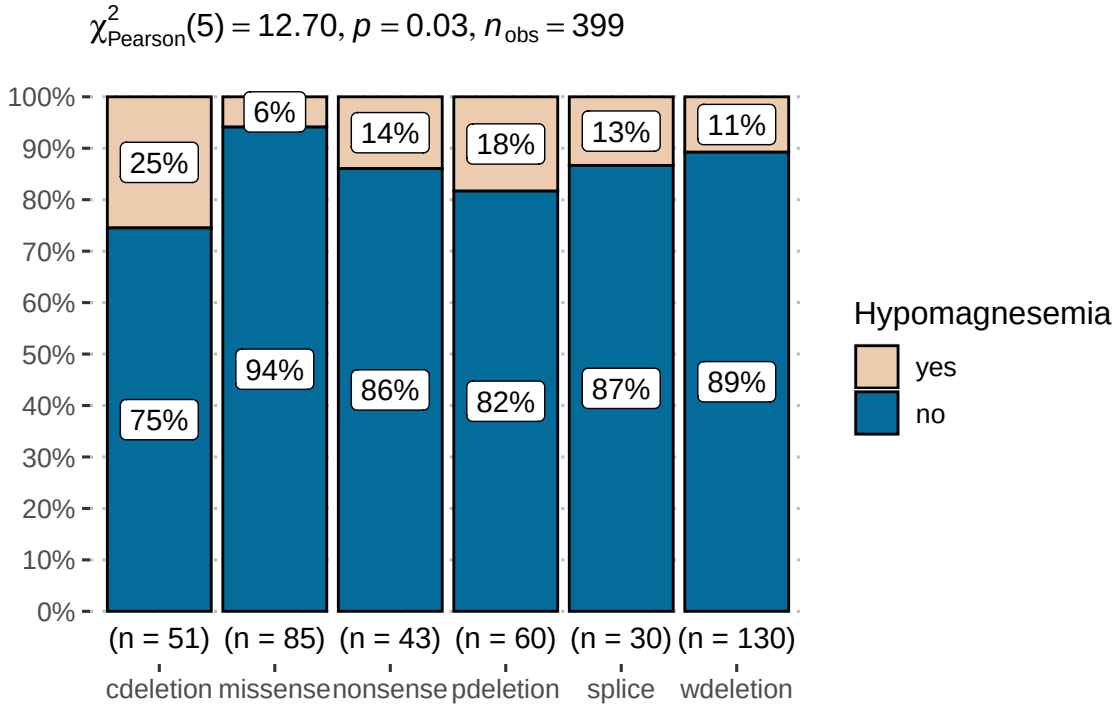
7.3 Hyperuricemia



7.4 HTN


$\chi^2_{\text{Pearson}}(5) = 3.30, p = 0.65, n_{\text{obs}} = 399$
100%
90%
80%
70%
60%
50%
40%
30%
20%
10%
0%
4%
5%
5%
3%
2%
96%
95%
95%
97%
100%
98%
(n = 51)
(n = 85)
(n = 43)
(n = 60)
(n = 30)
(n = 130)
cdeletion
missense
nonsense
pdeletion
splice
wdeletion
HTN
yes
no

7.5 Hypomagnesemia



7.6 Age ~ Mutation Type