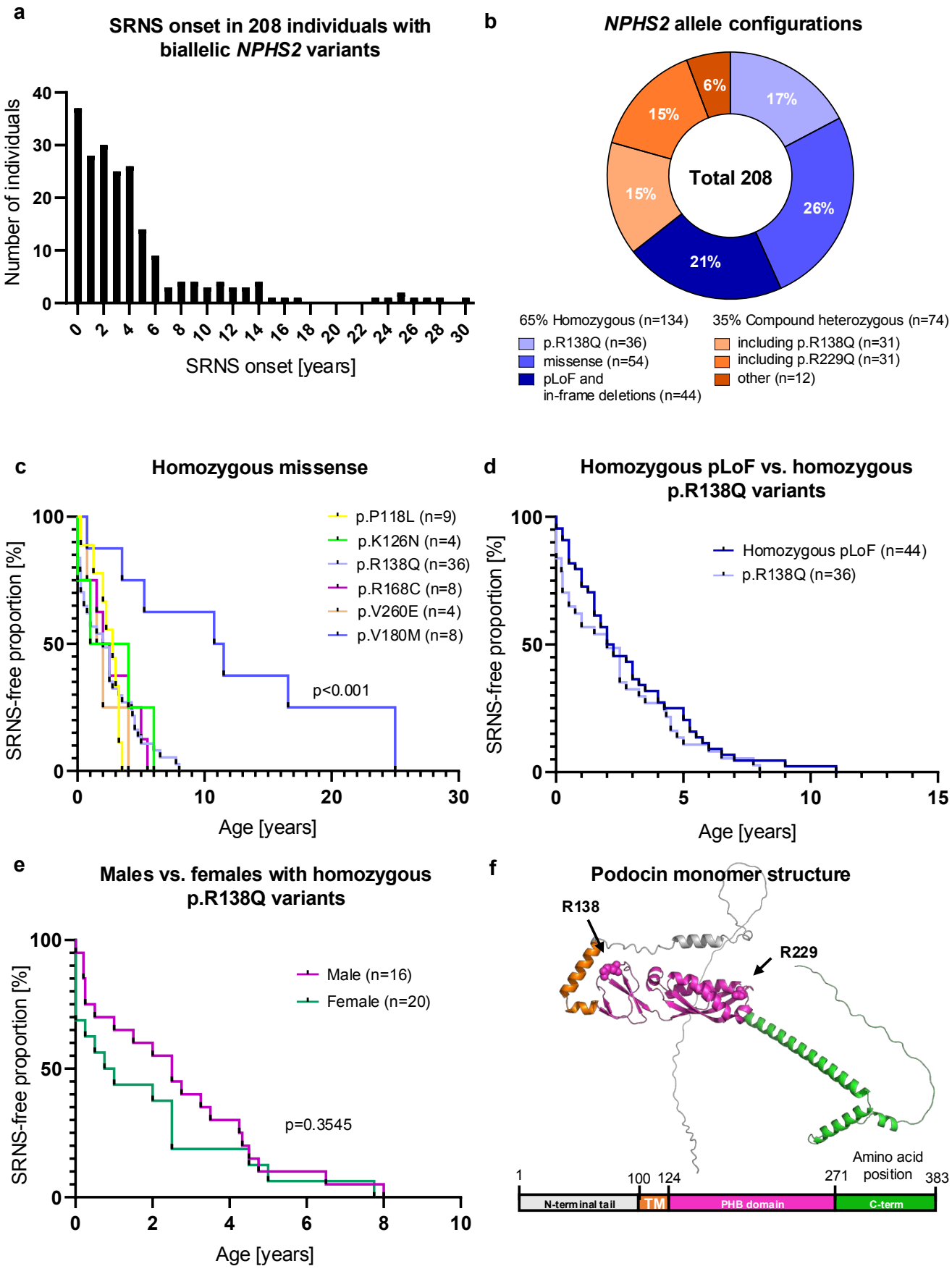
a
SRNS onset in 208 individuals with biallelic NPHS2 variants
Number of individuals
SRNS onset [years]
b
NPHS2 allele configurations
Total 208
17%
26%
21%
15%
15%
6%
65% Homozygous (n=134)
p.R138Q (n=36)
missense (n=54)
pLoF and in-frame deletions (n=44)
35% Compound heterozygous (n=74)
including p.R138Q (n=31)
including p.R229Q (n=31)
other (n=12)
c
Homozygous missense
SRNS-free proportion [%]
Age [years]
p.P118L (n=9)
p.K126N (n=4)
p.R138Q (n=36)
p.R168C (n=8)
p.V260E (n=4)
p.V180M (n=8)
p<0.001
d
Homozygous pLoF vs. homozygous p.R138Q variants
SRNS-free proportion [%]
Age [years]
Homozygous pLoF (n=44)
p.R138Q (n=36)
e
Males vs. females with homozygous p.R138Q variants
SRNS-free proportion [%]
Age [years]
Male (n=16)
Female (n=20)
p=0.3545
f
Podocin monomer structure
R138
R229
Amino acid position
1
100
124
271
383
N-terminal tail
TM
PHB domain
C-term