

a

expected proportion of individuals in the general population who carry fully penetrant *PRNP* variants

incidence of
prion disease

proportion
genetic cases

life expectancy
of genetic cases

EXPECTED
CASES

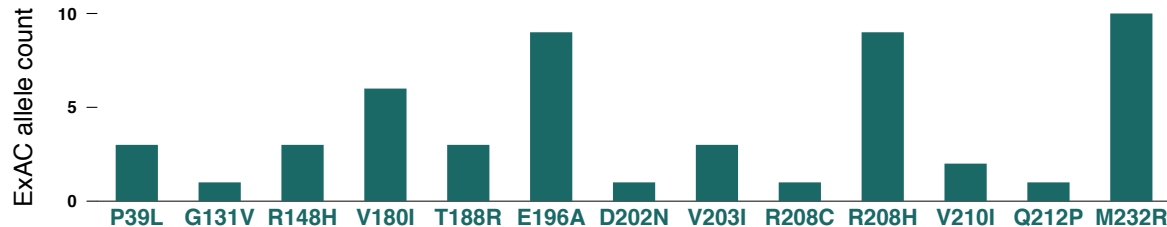
$$= \frac{1}{1,000,000 \text{ cases/person/year}} \times 15\% \times 50 \text{ years/case} \times 60,706 \text{ people} \approx 0.5 \text{ cases}$$

individuals in ExAC

b

OBSERVED
REPORTEDLY
PATHOGENIC
ALLELES

=



= 52
alleles