

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

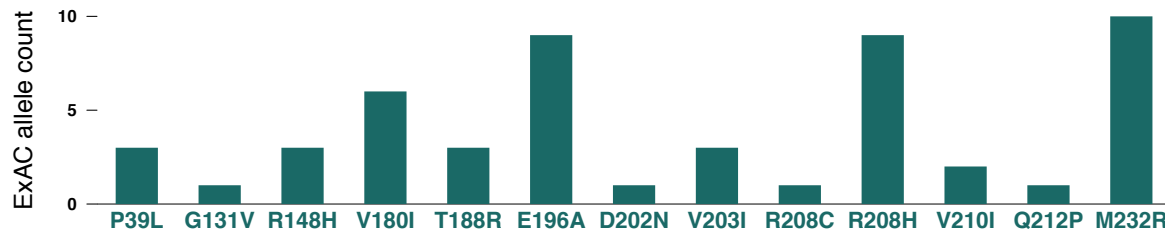
EXPECTED CASES

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

B

OBSERVED REPORTEDLY PATHOGENIC ALLELES

=



= 52 alleles