

(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

MAXIMUM  
EXPECTED  
CASES

$$= \frac{2}{1,000,000} \times 18\% \times 80 \times 60,706 \approx 1.7$$

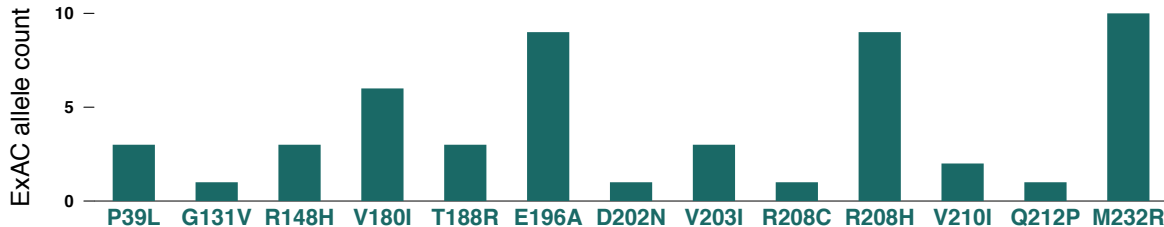
cases/person/year      years/case      people      cases

Individuals in ExAC

(B)

OBSERVED  
REPORTEDLY  
PATHOGENIC  
ALLELES

=



= 52  
alleles