

humans, this means that female heterozygotes for colour blindness have patches of normal and colour blind cells in their retinas. Another example of this in humans is Duchenne Muscular Dystrophy (DMD). A heterozygous female for DMD will likely possess some of the characteristics of it, especially if the proportion of affected X chromosome cells is higher than not because as an X-linked disorder, some cells will have the affected X and others won't.

Inbreeding - when relatives mate. It gives a much higher chance of homozygosity by relatedness. For many extremely rare disorders it has been found that parents of affected child are cousins (for example, Garrod found that for the disorder alkaptonuria which affects 1 in 20,000, more than half the patients were offspring of cousins). Another example of inbreeding affecting health is that it has been established that Tutankhamon had a congenital foot defect - this is likely because his parents were brother and sister. Smaller populations tend to have more inbreeding - a classic example is that of vLINCL in Finland. There are about 20 known cases and they are all clustered in the same area in Finland. The majority of the parents are also known to be related to others. More distant inbreeding is exemplified through cystic fibrosis. Whilst there are many different mutations (over 1000), 70% of cases in Europe come from the same mutation.

Intermediate dominance is when the heterozygote is an intermediate between two homozygotes. In human disease an example is Tay Sachs disease. Hets for Tay Sachs have an intermediate level of hexosaminidase enzyme activity, whereas a heterozygote for the recessive allele will have no activity and vice versa.