

(1) A homozygote is ... [1]

- an individual with two identical alleles at a single locus
- an individual with two different alleles at a single locus
- always expressing dominance
- all of the above

(2) Mutations can be considered as one of the raw materials of evolution because they ... [1]

- contribute to new variations in organisms
- are usually related to the environment in which they appear
- are usually beneficial to the organism in which they appear
- usually cause species of organisms to become extinct

(3) Which statement best represents the meaning of the term evolution: [1]

- changes in species toward greater complexity over time
- changes in gene frequencies in a population over time
- the strongest individuals survive and produce the most offspring
- changes in an individual over time in response to natural selection

(4) Which cross could produce a child with type O blood? [1]

- AO x BB
- AA x BO
- AB x OO
- AO x BO

(5) Because the gene for hemophilia is located on the X-chromosome, it is normally impossible for a ... [1]

- carrier mother to pass the gene to her son
- hemophiliac father to pass the gene on to his son
- hemophiliac father to pass the gene to his daughter
- carrier mother to pass the gene to her daughter

(6) Why are recessive X-linked syndromes such as haemophilia seen more often in males than in females? [1]

It is a recessive sex-linked (chromosome  $X^{\text{haem}}$ ) syndromes and males ( $X^{\text{haem}}Y$ ) are hemizygote for the X chromosome - so males will always show the phenotype while only homozygote women ( $X^{\text{haem}}X^{\text{haem}}$ ) will show it ( $p \gg p^2$ ).

(7) What are the possible offspring genotypes (e.g. carrier son, affected daughter) of a X-linked recessive disease in humans with an unaffected father and a carrier mother? [1.5]

unaffected son / unaffected daughter / carrier daughter / affected son

(8) One pair of genes for coat color in cats is sex-linked. The gene  $B$  produces yellow coat,  $b$  produces black coat, and the heterozygous  $Bb$  produces tortoise-shell coat. What kind of offspring will result from the mating of a black male and a tortoiseshell female? Please write down genotype of the parents and use a "Punnett square" to determine the genotype and phenotype of the offspring. [1.5]

$X^B$  yellow coat,  $X^b$  black coat  $\rightarrow X^bY$  black male,  $X^BX^b$  tortoiseshell female

	$X^b$	Y
$X^B$	$X^BX^b$	$X^BY$
$X^b$	$X^bX^b$	$X^bY$

$\Rightarrow X^BX^b$  tortoiseshell female,  $X^bX^b$  black female

$\Rightarrow X^BY$  yellow male,  $X^bY$  black male

(9) What does it mean if the inbreeding coefficient equals zero ( $F=0$ )? What can you say about the heterozygosity frequency ( $H$ ) in the population and the expected frequency ( $H_e$ ) under Hardy-Weinberg Equilibrium in a case where  $F$  equals zero. [2]

$F=0$  means no inbreeding only outbreeding - random mating  
 If  $F = 1 - (H_{obs}/H_{exp}) = 0$  then  $(H_{obs}/H_{exp}) = 1$  i.e.  $H_{obs}=H_{exp}$  [1]

(10) Modified from Arcellana et al. 2011. Distribution of MN blood group types in local populations in Philippines. *J. Genet.* 90, e90–e93. [9]

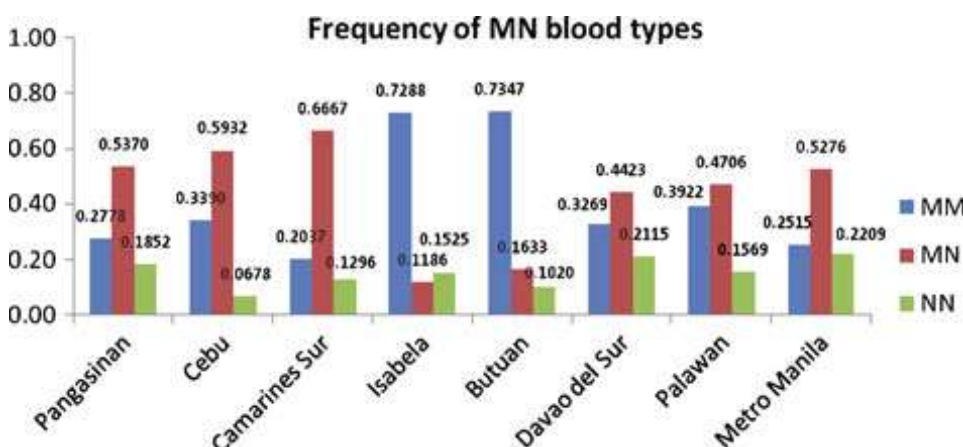
The classification of MN system of human blood type is based on the presence of either glycoprotein A (GPA) or glycoprotein B (GPB) in the erythrocyte membrane, leading to the expression of the M or N antigen for GPA and only the N antigen for GPB. *M* and *N* alleles are codominant to each other; this allows the heterozygote *MN* to have a distinguishable phenotype from the homozygous *MM* and *NN*. The codominant nature of the *M* and *N* alleles makes it convenient to test whether a population exhibits Hardy–Weinberg equilibrium (HWE) at the MN blood group locus. Eight populations and 541 individuals, representing the major regions of Philippines were selected for standard blood sampling.

MN blood groups have shown to exhibit differences in allele frequency in different populations, but are typically in HWE.

(a) Cite and define a reason likely to explain the differences in the frequencies of the *M* and *N* alleles between human populations? [1]

Genetic drift...

**Figure 1**



(b) Describe the results of Figure 1. What can you conclude in terms of  $M$  and  $N$  allele frequency? [2]

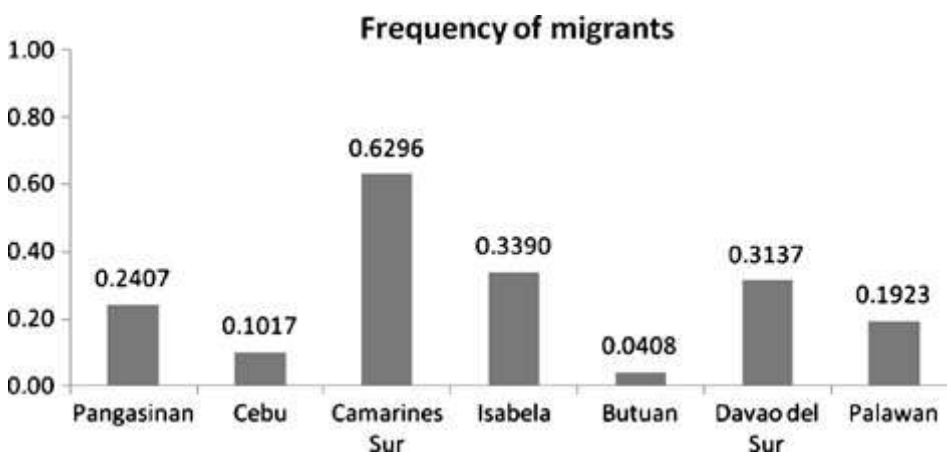
Excess of  $MN$  except for Isabela and Butuan populations ( $MM$ ), the  $NN$  genotype occurs the least, higher frequency of  $M$  allele over  $N$ , very high  $M$  allelic frequency in Isabela and Butuan.

All the populations observed in this study were in HWE except for the local populations of Camarines Sur, Isabela and Butuan based on Pearson's chi-square tests. Figure 2 gives the percentage of migrants for each population. Departure from Hardy-Weinberg equilibrium (DHW) was also tested using Wright's inbreeding coefficient. A positive value indicates a deficit of heterozygotes and a negative value indicates an excess of heterozygotes compared to a population at HWE (Figure 3).

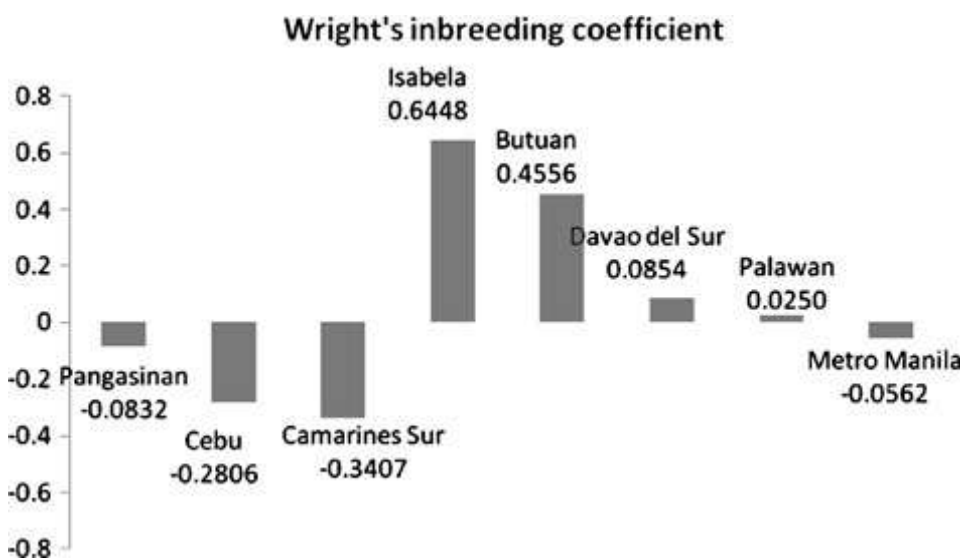
(c) In general, what can be the cause(s) of a departure from HWE? [2]

Departures from HWE could be caused by many factors, including migration, nonrandom mating, small population size, selection and mutations.

**Figure 2**



**Figure 3**



(d) Based on Figures 2 & 3, what are likely the reasons why the above cited 3 populations of Philippines deviate from HWE? [2]

In case of Camarines Sur population, the very high migration rate could account for the DHW. Isabela also has a relatively high migration rate. In case of Butuan, this is reflected by the lowest rate of migration across the

eight populations. Wright's inbreeding coefficient demonstrated extreme heterozygote deficit for Isabela and Butuan and excess of heterozygote for Camarines Sur. Altogether, migration in Camarines Sur and inbreeding in Isabela and Butuan populations are likely to cause DHW.

(e) Given that Isabela and Butuan populations belong to specific ethnolinguistic groups (minority) with very little outbreeding taking place, what can you conclude for these two populations? [2]

Both populations approximate isolated populations and nonrandom mating. This is clearly evident with the very small migration rate for Butuan, thereby limiting the probability of mating of completely unrelated individuals. Thereby 'social pressures' in humans can account for DHW.