

## Faculty of Science

Laboratory Manual Human Genetics

Bachelor of Biotechnology (Hons.)

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## Human Genetics

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LINCOLN UNIVERSITY COLLEGE FACULTY OF SCIENCE (DEPARTMENT OF BIOTECHNOLOGY) LABORATORY SAFETY RULES

The following rules must be obeyed by all students in the science laboratory of the faculty. Wilful or repeated in advertent non-compliance may result in dismissal or suspension from the laboratories

- No entry without permission:
- Outsiders are not allowed to enter the laboratory without permission.
- No student is allowed to enter the laboratory unless permission has been given by a laboratory assistant or a lecturer.
- At work in the laboratory:
- No experiment may be attempted without the knowledge and permission of a lecturer.
- Students must wear shoes in the laboratory. Students wearing slippers or sandals are not allowed to work in the laboratory.
- Lab coat must be worn at all times during practical work in the laboratory.
- Do not mouth pipette chemicals.
- Do not eat or smoke in the laboratory.
- Do not taste any chemicals, including dilute solutions. If any acid or alkali accidentally enters your eyes or mouth, wash immediately with plenty of tap water. Inform your lecturer, and seek medical attention if necessary.
- Paper should be used to light up the Bunsen burners.
- Used match sticks, filter papers, and other solid waste must never be thrown into the sinks. They must be thrown into the dustbins provided. Lighted match sticks and smoldering materials must be extinguished with tap water before thrown in to the dustbins.
- Any equipment broken or damaged must be reported to the laboratory assistant.


## - Before leaving the laboratory:

- All the equipment and benches must be cleaned at the end of each practical session.
- Wash hands and arms with soap and water before leaving the laboratory.
- No student is allowed to take away any chemicals, equipment or other property of the laboratory.


## INTRODUCTION

## 1. The Scientific Method

- Making observations
- Generating hypotheses
- Making predictions
- Designing and carrying out experiments
- Constructing scientific models


## 2. Practical Exercises

To get the most out of the practical exercises, you need to follow carefully the instructions given. These instructions have been designed to provide you with the experience in the following skills:

- Following instructors
- Handling apparatus
- Having due regard for safely
- Making accurate observations
- Recording results in an appropriate form
- Presenting quantitative results
- Drawing conclusions


## 3. Following Instructions

Instructions are provided in the order in which you need to carry them out. We would advise that before carrying out the instructions, you read through the entire exercise. This will help you to remember what you have learned.

Each practical exercise in the book begins with a few lines describing its purpose in most cases the following headings are also used:

- Procedure-numbered steps that need to be carried out.
- For consideration -some questions to help you think carefully about the results you have obtained.
- Materials-a list of the apparatus, chemicals and biological materials you need.


## 4. Handling apparatus

Biologists need to able to use many different types of apparatus, for example, photometers (to measure water uptake by plants), respirometers (to measure oxygen uptake or carbon dioxide production), Petri dishes (for plating out bacteria and other microorganisms) and the light microscope (to magnify specimens). Many of the practical exercises are designed to help you derive the maximum benefit from a piece of apparatus.

## 5. Having Due Regard for Safety

Surveys have been shown that science laboratories are among the safest places to be. Nevertheless, this is no cause for complacency.

- Always move slowly and carefully in a laboratory.
- Never put your fingers in your mouth or eyes after using chemicals or touching biological specimens until you have washed your hands thoroughly with soap and warm water, and dried them.
- Make sure glass objects (e.g, thermometers, beakers) cannot roll off tables or be knocked onto the floor.
- Wear safely goggles whenever there is a risk of damage to the eyes.


## Situations of risk include:

- Heating anything with a Bunsen burner (even heating water has its dangers')
- Handling many liquids, particularly those identified as corrosive, irritant, toxic or harmful
- Handling corrosive or irritant solids
- Some dissection work
- Allow Bunsen burners, tripods, gauzez and beakers to cool down before handling them.
- Never allow your own body fluids (especially blood and saliva) to come into contact with someone else, or theirs into contact with you.
- Keep long hair tied back and do not wear dangly earrings.
- Do not allow electrical equipment to come into contact with water.
- If you are unsure how to carry out a scientific procedure, ask.
- Make sure you understand why you are going to do something before you do it.
- Wear a lab coat when using chemicals or handling any biological specimens.
- Follow exactly agreed procedures with regard to cuts, burns, electric shocks and other accidents (e.g. with chemicals).
- Follow exactly all specific safely instructions given in this book or provided by your teacher for particular practical exercises (e.g. use of gloves, disinfection)

With practice, these procedures should become second nature to you. They will enable you to carry out practical work in safety.
6. Making Accurate Observations

In most cases the practical exercise will make it clear what you need to observe, e.g. the time taken for a certain volume of gas to be evolved or the width of a sample cells. Ensure that you know how to use any necessary equipment before starting practical. Think carefully about the precision with which you will make your observations.

## 7. Recording Results in an Appropriate Form

Results can be recorded in various ways. Often it is helpful to record raw data in a table. Most data will be in the form of numbers, i.e. they will be quantitative data (also known as numerical data). However, some data, e.g. flower colour, will be qualitative data.

One form in which some biological findings can be recorded is a drawing. You don't need to be professional artist to make worthwhile biological drawings. If you follow the following guidelines, a drawing can be of considerable biological value:

- Ensure that your completed drawing will cover at least a third of A4 page.
- Plan your drawing so that the various parts are is proportion and will not be drawn too small. Small marks to indicate the length and breadth of the drawing are a great help in planning and a faint outline can be rapidly drawn to show the relative positions of the parts.
- The final drawing should be made with clean, firm lines using a sharp HB pencil and, if needed, a good quality eraser (not a fluid).If important details are too small to be shown in proportion, they can be put in an enlarged drawing at the side of the main drawing.
- Avoid shading and the use of colour unless you are an excellent artist and they really help, for example when drawing soil profiles.
- When drawing structures seen with the naked eye or hand lens, use two lines to delineate such things as blood vessels and petioles. This will help you to indicate the relative widths of such structures.
- When drawing low power plan drawings from the light microscope, do not attempt to draw individual cells-just different tissues.
- When drawing plant cells at high power under the light microscope, use two lines to indicate the width of cell walls, but a single line to indicate a membrane.
- Always put a scale on each drawing.


## 8. Presenting Quantitative Results

Presentation of data is all about using graphs or other visual means to make it easier to see what your results tell you. The following four ways of presenting data are the most frequently used in biology: line graphs, bar charts, histograms and scatter graphs (Figure 1).





Figure 1: Line graphs, bar charts, histograms and scatter graphs

## 9. Drawing Conclusions

Finally, you will need to draw conclusions. If your practical exercise has involved the testing of a hypothesis, for example that the enzyme pepsin works better at low pH than in neutral or alkaline conditions, your conclusion should indicate whether the hypothesis has been refuted (i.e. shown not to be the case) or supported. Of course, even if your hypothesis has been supported, it doesn't mean that it has been confirmed with $100 \%$ certainty- in other words it isn't proved. Science proceeds more by showing that certain ideas are wrong than by showing that others are right (think about that!). Your conclusion might therefore include further ways of testing the original hypothesis, or might raise new possibilities to be investigated.

Often you will only be able to arrive at your conclusions after statistically analysing your data.

## 10. Writing a Scientific Lab Report

Title

- Communicate the subject investigated in the paper.


## Introduction

- State the hypothesis.
- Give well-defined reasons for making the hypothesis.
- Explain the biological basis of the experiment.
- Cite sources to substantiate background information.
- Explain how the method used will produce information relevant to your hypothesis.
- State a prediction based on your hypothesis. (If the hypotheis is supported, then the results will be.)


## Materials and Methods

- Use the appropriate style.
- Give enough detail so the reader could duplicate your experiment
- State the control treatment, replication and standardized variables that were used.


## Results

- Summarize the data (do not include raw data).
- Present the data in an appropriate format (table or graph).
- Present tables and figures neatly so they are easily read.
- Label the axes of each graph completely.
- Give units of measurement where appropriate.
- Write a descriptive caption for each table and figure.
- Include a short paragraph pointing out important results but do not interpret the data.


## Discussion

- State whether the hypothesis was supported or proven false by the results, or else state that the results were inconclusive.
- Cite specific results that support your conclusions.
- Give the reasoning for your conclusions.
- Demonstrate that you understand the biological meaning of your results.
- Compare the results, with your predictions and explain any unexpected results.
- Compare the results to other research or information available to you.
- Discuss any weaknesses in your experimental design or problems with the execution of the experiment.
- Discuss how you might extend or improve your experiment.


## Conclusion

- Restate your conclusion.
- Restate important results.


## Literature Cited

- Use the proper citation form in the text.
- Use proper citation form in the Literature Cited section.
- Refer in the text to any source listed in this section.


## Acknowledgement

- State any appropriate acknowledgement that you think is necessary.


## Practical 1

Title: Multiple allele

## Objective:

After completing the practical, you will be able:

1. To determine compatible blood types for transfusions

## Introduction:

Red blood cells have molecules called antigens on their surface. An antigen is a molecule that causes the immune system to produce antibodies against it. When a foreign antigen enters the body the immune system will build antibodies against it. ABO antigens are attached to human red blood cells. The surface antigens on red blood cells are coded for by one gene that has three different alleles. This is an example of a multiple allelic system. The IA allele determines the A antigen, the IB allele determines the B antigen, and the " i " allele determines no antigens (type O ).

An individual carries a matched pair of chromosomes and thus has two alleles for the ABO blood groups. Two alleles may be expressed at the same time. If an individual has IA and IB, they will have type $A B$ blood. Since both alleles are expressed, this is an example of codominance. The possible genotypes and phenotypes are listed in Table 2. If red blood cells with foreign antigens on them enter the body the antibodies produced against them will cause the blood to clump (not clotclotting is something quite different). A normal person never makes antibodies against his own antigens. If this occurred it would be disastrous! Imagine a person with A antigen making antibodies against A and clumping his own blood cells. Instead, a person with A would make antibodies against foreign antigens such as B (anti-B antibodies). This is why only a blood type that is compatible can be used during blood transfusions. For example, type A blood can only be given to persons with type A or type AB blood (See Table 2). The Rh system is completely separate, but works in much the same way. If you have the Rh antigen present on your red blood cells, you are Rh+. If it is absent, you are Rh-.

| Table 2. Human Blood Groups |  |  |  |
| :---: | :---: | :---: | :---: |
| Genotype | Phenotype | Antigen present | Antibody produced |
| ${ }^{\text {A }}{ }^{\text {A }}$ | A | A | Anti-B |
| $A_{i}$ | A | A | Anti-B |
| $\left.{ }^{18}\right\|^{\text {B }}$ | B | B | Anti-A |
| IBi | B | B | Anti-A |
| ${ }^{\text {A }}{ }^{\text {B }}$ | AB | A \& B | None |
| ii | 0 | None | Anti-A \& Anti-B |
| Rh+, Rh+ | Rh+ | Rh | None |
| Rh+, Rh- | Rh+ | Rh | None |
| Rh-, Rh- | Rh- | None | Anti-Rh (after exposure) |

## Procedure:

1. Conduct a blood typing exercise as per instructions. (using blood cards)

| Table 3. Blood Antigen Test |  |  |
| :--- | :--- | :--- |
| Sample | Result |  |
| e.g. A | Clump at anti-A and not Rh | Type A Rh- |


| Blood sample 1 |  |  |
| :--- | :--- | :--- |
| Blood sample 2 |  |  |

What blood type are you? $\qquad$ What are your possible genotypes?

What type(s) of blood could you safely receive in a transfusion? $\qquad$
To which type(s) of blood could you safely donate in a transfusion? $\qquad$

## Questions:

1. Blood type O is sometimes called the universal donor. Why?
2. Type $A B$ is sometimes called the universal recipient. Why?

## Practical 2

Title: Barr Bodies

## Objective:

After completing the practical, you will be able:

1. To know the significance of the Barr body

## Introduction:

The $X$ chromosome is fairly large and carries numerous genes necessary for life. The $Y$ chromosome is tiny and has only a few genes. Early in the development of the female embryo one of the X chromosomes becomes inactive in each cell. This means that only one of the two X chromosomes is actually working. The inactive $X$ becomes condensed and can be seen in certain cells as a Barr body. There is always only one active $X$ chromosome in human cells. Thus, if the person is a normal female there should be one Barr body present and a normal male should not have any. People with extra $X$ chromosomes will have more than one Barr body. For example an XXX female would have two Barr bodies. In the past Olympic athletes were required to have a Barr body test to determine their genetic sex before entering any events.

| Table 2. Human Blood Groups |  |  |  |
| :---: | :---: | :---: | :---: |
| Genotype | Phenotype | Antigen present | Antibody produced |
| $\\|^{\text {A }}{ }^{\text {A }}$ | A | A | Anti-B |
| ${ }^{\text {A }}$ i | A | A | Anti-B |
| \| ${ }^{1}{ }^{\text {B }}$ | B | B | Anti-A |
| \|Bi | B | B | Anti-A |
| ${ }^{\text {A }}{ }^{\text {B }}$ | AB | A \& B | None |
| ii | 0 | None | Anti-A \& Anti-B |
| Rh+, Rh+ | Rh+ | Rh | None |
| Rh+, Rh- | Rh+ | Rh | None |
| Rh-, Rh- | Rh- | None | Anti-Rh (after exposure) |

## Procedure:

1. Obtain a clean microscope slide.
2. Use the blunt end of a toothpick to scrape the inside of your cheek to obtain some epithelial cells.
3. Smear the cheek scraping onto the slide and add a drop of methylene blue.
4. Stir the dye and cells together and set aside for 2 minutes.
5. Add a coverslip, cover the slide with a piece of tissue and gently press straight down on the coverslip with your thumb to spread the cells.
6. Examine the slide with your microscope. Can you see any Barr bodies? The Barr body appears as a darkly stained structure against the nuclear envelope.
7. Exchange slides with someone of the opposite gender.
8. As an alternative, look at one of the prepared demonstration slides.

## Result:



## Sketch of a cheek cell with Barr body

Which sex has Barr bodies? $\qquad$ Why? $\qquad$

## Question:

## Sex Determination in Humans: $X$ and $Y$ chromosomes

Human cells contain 23 pairs of chromosomes and 46 chromosomes in all. One chromosome in each pair comes from each parent. Twenty-two of the 23 pairs are matched and are called autosomes. The chromosomes in these 22 pairs are similar in size, shape, and the genes that they carry. The 23rd pair, XX or XY , determines the sex of the individual and are called sex chromosomes. Each egg contains one $X$ chromosome. Each sperm contains either one $X$ or one $Y$ chromosome. If an " $X$ " sperm fertilizes the " $X$ " egg, the child is female ( $X X$ ). See Figure 2. If a " $Y$ " sperm fertilizes the " $X$ " egg, the child is male (XY). Notice that the only thing that determines sex is the presence or absence of the $Y$ chromosome. If you have a Y , you are male. If you lack the Y , you are female.

1. How many Barr bodies would you expect in a female with 4 XS (XXXX)?
2. How many in an XXY male?

## Practical 3

Title: Karyotyping lab

## Objective:

After completing the practical, you will be able:

1. To analyze a karyotype of an unborn baby's cells and use it to determine the sex of the baby and whether or not the baby has Down syndrome.

## Introduction:

Humans have 46 chromosomes in every diploid (2n) body cell. Figure 1 shows human chromosomes. The chromosomes of a diploid cell occur in homologous pairs, which are pairs of chromosomes that are similar in size, shape, and the position of the centromere. In humans, 22 homologous pairs of chromosomes are called autosomes. The twenty-third pair, which determines the individual's sex, make-up the sex chromosomes. Females have one type of sex chromosome, which is an $X$ chromosome. Males have two types of sex chromosomes, an $X$ chromosome and a much smaller Y chromosome. The diagram at the bottom of the page shows each of the 22 types of autosomes and the 2 types of sex chromosomes.

A karyotype is a diagram that shows all of the cell's chromosomes arranged in order from largest to smallest. A karyotype is made from a photomicrograph (photo taken by a microscope) of the chromosomes from a cell in metaphase. The photographic images of the chromosomes are cut and arranged in homologous pairs by their shape and size. The karyotype can be analyzed to determine the sex of the individual and whether there are any chromosomal abnormalities. For example, the karyotype of a female show two $X$ chromosomes, and the karyotype of a male show an $X$ and a $Y$ chromosome.

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Figure 1: Human chromosomes
Chromosomal abnormalities often result from nondisjunction, the failure of chromosomes to separate properly during meiosis. Nondisjunction results in cells that have too many or too few chromosomes. Trisomy is an abnormality in which a cell has an extra chromosome, or section of chromosome. This means that the cell contains 47 chromosomes instead of 46 . Down syndrome, or trisomy 21 , is a chromosomal abnormality that results from having an extra number 21 chromosome.

## Procedure:

You are a medical lab technician and one of your jobs is to assist with prenatal testing. Currently, you are working on the case of Mr. and Mrs. Smith. Mrs. Smith is pregnant, and her doctor has recommended amniocentesis, which is a type of prenatal testing. You have been given photomicrographs of the chromosomes in the unborn baby's cells, which were obtained through amniocentesis. Your job is to complete and analyze a karyotype of these cells to determine the sex of the Smith's baby and whether the baby is normal or has Down Syndrome.

1. Carefully cut out each chromosome from the chromosome spread. Be sure to leave a slight margin around each chromosome.
2. Arrange the chromosomes in homologous pairs. The members of each pair will be the same length and will have the centromere in the same location. Arrange the pairs according to their length, from largest to smallest. The banding patterns of the chromosomes may also help you to pair up the homologous chromosomes.
3. Glue each of the homologous pairs into your lab notebook. Place the pairs in order, with the longest pair at the upper left hand corner. Number the pairs starting with \#1, and the sex chromosomes \#23.
4. You just made a karyotype! Analyze the karyotype to determine the sex of the individual and whether or not the karyotype is normal.

## Questions:

1. Define chromosome.
2. What is the difference between a somatic cell and a gamete?
3. Describe trisomy. Why does this happen?

## Practical 4

Title: Human population genetics-The Hardy-Weinberg principle

## Objective:

After completing the practical, you will be able:

1. To compare phenotypic frequencies of inherited human traits in a class
2. To determine if the inherited traits conform to the Hardy-Weinberg equilibrium

## Introduction:

The Hardy-Weinberg Theorem deals with Mendelian genetics in the context of populations of diploid, sexually reproducing individuals. Given a set of assumptions (discussed below), this theorem states that:

1. allele frequencies in a population will not change from generation to generation.
2. if the allele frequencies in a population with two alleles at a locus are $p$ and $q$, then the expected genotype frequencies are $\mathrm{p} 2,2 \mathrm{pq}$, and q 2 . This frequency distribution will not change from generation to generation once a population is in Hardy-Weinberg equilibrium. For example, if the frequency of allele $A$ in the population is $p$ and the frequency of allele a in the population is $q$, then the frequency of genotype $A A=p 2$, the frequency of genotype $A a=2 p q$, and the frequency of genotype $a \mathrm{a}=\mathrm{q} 2$. If there are only two alleles at a locus, then $p+q$, by mathematical necessity, equals one. The Hardy-Weinberg genotype frequencies, $\mathrm{p} 2+2 \mathrm{pq}+$ q 2 , represent the binomial expansion of $(\mathrm{p}+\mathrm{q}) 2$, and also sum to one (as must the frequencies of all genotypes in any population, whether it is in Hardy-Weinberg equilibrium). It is possible to apply the Hardy-Weinberg Theorem to loci with more than two alleles, in which case the expected genotype frequencies are given by the multinomial expansion for all $k$ alleles segregating in the population: $\left(p_{1}+p_{2}+p_{3}+\ldots+p k\right)^{2}$.

The conclusions of the Hardy-Weinberg Theorem apply only when the population conforms to the following assumptions:
1.Natural selection is not acting on the locus in question (i.e., there are no consistent differences in probabilities of survival or reproduction among genotypes).
2.Neither mutation (the origin of new alleles) nor migration (the movement of individuals and their genes into or out of the population) is introducing new alleles into the population.
3.Population size is infinite, which means that genetic drift is not causing random changes in allele frequencies due to sampling error from one generation to the next. Of course, all natural populations are finite and thus subject to drift, but we expect the effects of drift to be more pronounced in small than in large populations.
4. Individuals in the population mate randomly with respect to the locus in question.

Although non-random mating does not change allele frequencies from one generation to the next if the other assumptions hold, it can generate deviations from expected genotype frequencies, and it can set the stage for natural selection to cause evolutionary change.
If the genotype frequencies in a population deviate from Hardy-Weinberg expectations, it takes only one generation of random mating to bring them into the equilibrium proportions, provided that the above assumptions hold, that allele frequencies are equal in males and females (or else that individuals are hermaphrodites), and that the locus is autosomal. If allele frequencies differ between the sexes, it takes two generations of random mating to attain Hardy-Weinberg equilibrium. Sex-linked loci require multiple generations to attain equilibrium because one sex has two copies of the gene and the other sex has only one.

Given these conditions, it is easy to derive the expected Hardy-Weinberg genotype frequencies if we think about random mating in terms of the probability of producing each genotype via random union of gametes into zygotes (Table 1). If each allele occurs at the same frequencies in sperm and eggs, and gametes unite at random to produce zygotes, then the probability that any two alleles will combine to form a particular genotype equals the product of the allele frequencies. Since there are two ways of generating the heterozygous genotype (An egg and a sperm, or an egg and A sperm), we sum the probabilities of those two types of union to arrive at the expected Hardy-Weinberg frequency of the heterozygous genotype (2pq).

A Punnett square depicting the probabilities of generating all possible genotypes at a diallelic Mendelian locus in a population that conforms to Hardy-Weinberg assumptions.

Table 1: A Punnett square depicting the probabilities of generating all possible genotypes at a diallelic Mendelian locus in a population that conforms to Hardy-Weinberg assumptions.


It is important to recognize that the Hardy-Weinberg equilibrium is a neutral equilibrium, which means that a population perturbed from its Hardy-Weinberg genotype frequencies will indeed reach equilibrium after a single generation of random mating (if it conforms to the other assumptions of the theorem), but it will be a new equilibrium if allele frequencies have changed. This property distinguishes a neutral equilibrium from a stable equilibrium, in which a perturbed system returns to the same equilibrium state. It makes sense that the Hardy-Weinberg equilibrium is not stable, since a change from the equilibrium genotype frequencies will generally be associated with a change in allele frequencies ( $p$ and $q$ ), which will in turn lead to new values of $p^{2}, 2 p q$ and $q^{2}$. Thereafter, a population that meets Hardy-Weinberg assumptions will remain at the new equilibrium until perturbed again.

Given a population in which we know the number of individuals with each genotype, we can test for statistical deviation from Hardy-Weinberg equilibrium using a simple chi-square goodness-of-fit test or a more powerful exact test. The latter class of methods has proved particularly useful for large-scale genomic studies, in which scientists evaluate thousands of loci segregating for multiple alleles (Wiggington et al. 2005). Observed genotype proportions in natural populations typically conform to Hardy-Weinberg expectations, as we might expect given that a population perturbed from equilibrium can achieve new equilibrium frequencies after only one generation of random mating.

Although statistical deviation from Hardy-Weinberg expectations generally indicates violation of the assumptions of the theorem, the converse is not necessarily true. Some forms of natural selection (e.g., balancing selection, which maintains multiple alleles in a population) can generate genotypic frequency distributions that conform to Hardy-Weinberg expectations. It may also be true that migration or mutation is occurring, but at such low rates as to be undetectable using available statistical methods. And, of course, all real populations are finite and thus susceptible to at least some evolution via genetic drift.

## Materials:

- PTC-impregnated filter paper (food grade)
- A4 paper, pen, pencil and calculator


## (A) Allele Frequencies

1. To calculate allele frequencies for populations of diploid organisms, first multiply the number of individuals in the population by 2 to obtain the total number of alleles at that locus.
2. Select one allele for the first set of calculations, e.g., $A$ allele.
(a) Individuals homozygous for the $A$ allele will each have $2 A$ alleles. Multiply the number of $A A$ homozygotes by 2 to calculate the number of $A$ alleles.
(b) Heterozygotes will each possess only one $A$ allele.
(c) The total number of $A$ alleles in the population $=[$ (number of Aa heterozygotes $)+(2 X$ number of AA homozygotes)].
3. Frequency of the $A$ allele $=[$ (total number of $A$ alleles in the population)/ (total number of alleles in population for that locus)]
4. Frequency of the allele $=$ (1-frequency of the $A$ allele $)$

## (B) Genotype Frequencies

1. To calculate genotype frequencies for populations of diploid organism, first determine the number of individuals with each genotype in the population. In the earlier example, count the number of individuals with the following genotypes: $A A, A a$, and $a a$.
2. To determine the frequency of each genotype, divide the number of individuals with that particular genotype by the total number of individuals in the population.
(a) frequency of $A A$ genotype= \# AA individuals/ population size
(b) frequency of Aa genotype= \# Aa individuals/ population size
(c) Frequency of aa genotype= \# aa individuals/ population size

## Procedure:

The human traits you will examine and the phenotypes associated with the genotypes are described below. Test each phenotype and genotype in class by observing yourself and your classmates, and identifying the different characters. Tabulate your phenotype and possible genotype for each of the human characteristic studied, and also the frequencies of the students in your class in Table 2.

1. PTC-tasting-one of the most thoroughly documented genetic traits is the ability or inability to taste the chemical, phenylthiocarbamide (PTC, C7H8N2S), also known as phenylthiourea. The PTC taster gene is designated as $T$. Tasters are homozygous ( $T T$ ) or heterozygous dominant ( $T t$ ). Non-tasters carry the tt genotype.
2. Tongue-rolling-the inheritance of several tongue movement traits has been documented, researched and disputed throughout the last century. The ability to roll the tongue upwards from the sides has received the most emphasis. In 1940, A.H. Sturtevant reported two classes within the human population, roller and non-roller. The roller phenotype is dominant ( $R$ _) while individuals unable to perform the maneuver are homozygous recessive ( $r r$ ) for the trait.
3. Facial dimples-the occurrence of natural indentations at the corners of the mouth (dimples) is controlled by a dominant allele ( $D_{\_}$). Persons without facial dimples generally possess the
homozygous recessive phenotype; dd. Facial dimples are sometimes inherited as an irregular dominant.

Table 2: Record your data here for the inherited human traits.

|  | Phenotype | Homozygous Dominant <br> or Heterozygous | Homozygous <br> recessive |
| :--- | :--- | :--- | :--- |
| 1. | PTC taster <br> $(T)$ |  |  |
|  | Non-PTC taster <br> $(t))$ |  |  |
| 2. | Tongue roller (R_) |  |  |
|  | Non-tongue roller (rr) |  |  |
| 3. | Facial dimples (D_) |  |  |
|  | No facial dimples (D_) |  |  |

## Questions:

1. Calculate the frequencies of the dominant and recessive alleles for the ability of tasting PTC.
2. Calculate the frequencies of the dominant and recessive alleles for the ability of rolling tongue.
3. Calculate the frequencies of the dominant and recessive alleles for the occurrence of dimple dimples.




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