Lab VI-1

Exploring Mendelian Genetics

Equipment and Materials

You'll need the following item to complete this lab session. (The standard kit for this book, available from *www.thehomescientist.com*, includes the only item needed for this session.

Materials from Kit
□ Phenylthiocarbamide (PTC) test strips
Materials You Provide
□ None

Background

In 1930, the American chemist Arthur Fox had a lab accident while he was purifying an organic compound called *phenylthiocarbamide* that he had just synthesized. Some of the finely powdered phenylthiocarbamide (also called *PTC* or *phenylthiourea*) escaped as a cloud of dust. Fox thought nothing of it until a colleague working nearby complained about the bitter taste of the material. Fox, who had been exposed to much more of the compound than his colleague, had tasted nothing.

Being scientists, they immediately verified their observation by re-tasting the PTC, confirming that Fox was unable to detect any bitterness while his colleague nearly gagged at the bitterness. Fox soon extended the experiment to determine which of his family, friends, and colleagues at DuPont were able to taste PTC. Some of them were unable to detect any taste, others detected a slight bitterness, and still others found the taste of PTC to be extremely bitter. As it turns out, about 70% of people can taste PTC, but that percentage differs significantly in different populations, ranging from about 58% among Australian Aborigines to about 98% among American Indians.

> Until well into the 20th century, chemists commonly tasted new compounds they had synthesized. Also well into the 20th century, accidental poisoning was a common cause of death among chemists. As a college freshman chemistry major in 1971, Robert was told never to taste any compound unless it was known to be safe, but many of his older professors were survivors from a generation that had routinely tasted compounds whose toxicity was unknown.

Geneticists soon determined that the ability to taste PTC is an inherited trait. In fact, the genetic correlation is so strong that, until the advent of DNA analysis, PTC testing was used (along with blood type) in paternity testing. But it wasn't until 2003 that geneticists discovered exactly what determined the ability of an individual to taste or not taste PTC. That ability is determined by one gene, designated TAS2R38.

There are two common *alleles* (forms) of that gene—one for tasting and one for non-tasting—along with several rare alleles. Everyone has two copies of each gene, so an individual may have two copies of the tasting gene, two copies of the non-tasting gene, or one copy of each gene. *Homozygous* individuals—those who have either two copies of the tasting gene or two copies of the non-tasting gene—find PTC extremely bitter or entirely tasteless, respectively. *Heterozygous* individuals—those who have one copy of each allele—can taste PTC, but only as a mild bitterness.

> Actually, there is some debate about whether the ability to taste PTC is a purely Mendelian trait, associated only with the TAS2R38 gene. Humans have about 30 other genes that code for the ability to taste various other bitter compounds. It is possible that the tasting TAS2R38 allele exhibits either *complete dominance*—in which case the partial ability to taste PTC could be coded for by one or more of the other tasting genes—or *incomplete dominance*, in which case the partial ability to taste PTC would manifest in individuals who are heterozygous with respect to the TAS2R38 gene.

PTC is very uncommon in nature—occurring only at levels near or below the detection threshold of even homozygously-sensitive individuals—but the ability to detect PTC is nonetheless a selective advantage because the structure of PTC is similar to the structures of the bitter alkaloids produced as defense mechanisms by many plants. Accordingly individuals who are PTC tasters can detect the bitterness of many alkaloids, many of which are extremely toxic.

You might wonder, then, why the non-tasting gene was not eliminated long ago by non-tasting individuals unintentionally poisoning themselves by consuming toxic plants. There is still some debate among geneticists as to why this has not occurred. Prevailing opinion is that non-tasting individuals are instead able to taste other toxic compounds that tasting individuals are not able to taste. If that is the correct explanation, one would expect individuals who are heterozygous with respect to the TAS2R38 gene to have a competitive advantage because they are able to taste both classes of toxic compounds, albeit not as strongly as homozygous individuals are able to taste one or the other class. If true, this would tend to preserve the recessive non-tasting allele in heterozygous form. From that, it follows that a minority of individuals in a large population should be homozygous with respect to the recessive (non-tasting) TAS2R38 gene, which is exactly what we observe.

In this lab session, we'll use test paper strips impregnated with tiny amounts of PTC to test as many individual volunteers as possible for their ability to taste PTC. You can begin with your own family, of course, but your friends, neighbors, church, sports teams, and other social groups that you belong to should provide a ready source of volunteers if you present your project properly to them.

> The kit contains a vial of 100 PTC test strips, which you can cut into halves or thirds if you have more than 100 test subjects. If necessary, you can purchase additional PTC test strips from nearly any lab supplies vendor.

Testing unrelated individuals provides some useful data, but ideally you want to test as many related individuals as possible so that you can follow inheritance of the PTC tasting and non-tasting alleles through generations of families. Testing both parents and their children is good; testing parents, children, and all four grandparents is better still. Best of all is testing the full extended family, with aunts and uncles and cousins.

Any human genetic testing, including this lab session, potentially has serious ethical implications. Many families have at least one “skeleton in the closet” that they'd prefer to keep hidden from the world at large. You are obligated—morally, ethically, and possibly legally—to maintain the absolute privacy of your test subjects by refusing to disclose the data you obtain to anyone else, including the test subjects themselves.

For example, in one of your family groups of test subjects you might find that both parents are non-tasters, as are all of their children except the eldest. You might conclude that that child was adopted or had a different father. Disclosing that conclusion **TO ANYONE, INCLUDING THE TEST SUBJECT**, is a serious ethical violation, and may have direct and indirect consequences you cannot imagine. If you discover such an anomaly, **KEEP IT TO YOURSELF**.

Also consider this: you are not a geneticist, so you may be wrong.

Procedure VI-1-1: Testing subjects for the ability to taste PTC

Do not tell your test subjects what to expect, lest you skew your results. If asked, you can assure your test subjects that there is no hazard involved in this experiment, but do not do so unless asked.

1. Working one-on-one if possible, ask your first test subject to place a PTC test strip on his or her tongue. Once they have done so, ask them to describe what, if anything, they taste. Some of your subjects will probably report tasting nothing, and others may spontaneously report strong bitterness. (YUCK!) If a subject reports a bitter taste, ask the subject to characterize the bitterness as extremely bitter, strongly bitter, moderately bitter, or slightly bitter.
2. Record the identity and relationships of that test subject in your lab notebook, along with the results of the taste test for that subject.
3. Repeat steps 1 and 2 for your other test subjects.

Procedure VI-1-2: Charting inheritance of PTC-tasting ability

1. Either with pen and paper or with your computer and graphing software, make up a genealogy chart for each of your family groupings, indicating tasting, non-tasting, or partial-tasting for each member of the chart by color-coding or other visual means.
2. Analyze the chart to track the effect of the PTC tasting allele through the generations.

Review Questions

??? Production: Please strip the answers (formatted as comments) from the final layout. RBT

1. Based on your data, can you conclude with reasonable certainty that the ability to taste PTC is a full Mendelian trait? Why or why not?

   Answers may vary, but it is very likely that student data will confirm PTC tasting to be a full Mendelian trait, with perhaps some uncertainly due to test subjects who were partial tasters.

2. In human studies, it is common to anonymize the data, which is to say to remove any information that would allow another party to identify the test subjects. Propose a method for anonymizing your own data, both in raw form and in the final genealogy charts. What overriding factors must you take into account when anonymizing your data?

   Answers may vary as to method, but the two overriding factors are that the data must not identify individuals but that the relationships between and among those individuals must be preserved. The most common method for anonymizing data is to assign test subjects a random number or code name and to record relationships by those numbers or code names.