

Teacher Preparation Notes for Cookie-ase

By Drs. Ingrid Waldron and Jennifer Doherty, Department of Biology, University of Pennsylvania, 2011¹

Teaching points

- An enzyme acts on substrate(s) to produce product(s).
- An organism's genotype influences their phenotype through the processes of transcription (DNA to mRNA) and translation (mRNA to protein).
- Different types of mutations can cause phenotypic changes of different severities. For example, point mutations usually result in less severe phenotypic effects than deletion or insertion mutations, especially when the number of nucleotides deleted or inserted in the mRNA is not a multiple of three so there is a frameshift and none of the subsequent codons is read correctly during translation.
- Changes at the molecular level can cause macroscopic changes.
- A phenotype can be characterized at different levels of biological organization: molecular (amino acid sequence in protein), biochemical (e.g. level of enzyme function), and macroscopic (e.g. skin color).
- In a heterozygous individual, the allele for a defective enzyme often is recessive because the allele for the normal enzyme results in enough enzyme activity to produce normal phenotypic characteristics.

Equipment and Supplies

Stopwatch (1 per group)

Sandwich cookies (15-30 per group) (If you are not using name brand Oreos (either full size or mini) make sure you test the cookies before you use them in your classroom; some discount sandwich cookies tend to shatter instead of separating.)

Paper plates (3 per group) (Paper plates should be used so the cookies are never on the table and are always clean. Each student enzyme needs two plates: one to hold the pool of whole cookies and another to put the cookie halves on as they are separated. Each group needs another plate to collect the cookie halves in between rounds of opening.)

Soap, water and paper towels for each student to wash their hands

Job cards: enzyme, recorder, timekeeper

Instructional Suggestions

Posting class averages on the board can be very useful when students are interpreting their data, especially in the rare case when a student somehow opens more cookies with the defective enzyme than with the normal enzyme. To avoid this problem, we suggest that you allow the students simulating the enzyme to practice on a few cookies before beginning the actual simulation.

Biology Background

The first mutation shown illustrates a missense mutation since there is a change in one of the amino acids in the polypeptide. Some missense mutations have little effect, e.g. if an amino acid with similar chemical properties is substituted or the change in amino acid occurs in a region which is not crucial for

¹ These teacher notes and the student handout for this activity are available on the molecular biology page of <https://sites.google.com/site/biologypd/home>.

the function of the polypeptide. Other missense mutations have a substantial effect, e.g. the substitution of a single amino acid which results in the difference between normal hemoglobin and sickle cell hemoglobin (discussed in the hands-on activity "From Gene to Protein - Transcription and Translation", available at http://serendip.brynmawr.edu/sci_edu/waldron/#trans).

The information on albinism and muscular dystrophy illustrates the general point that alleles that code for defective enzymes are very often recessive, since the normal allele is generally able to code for enough functioning enzyme to produce a normal phenotype.

For more information on albinism, see <http://www.mayoclinic.com/health/albinism/DS00941>

Both Duchenne muscular dystrophy and Becker muscular dystrophy result from mutations of a gene on the X chromosome that codes for the dystrophin protein in muscle cells; this protein helps to stabilize the plasma membrane during the mechanical stresses of muscle contraction. About two-thirds of cases are due to deletion mutations. If the number of nucleotides deleted in the mRNA is not a multiple of three, this type of frameshift mutation results in a severely defective or absent version of the protein resulting in more rapid breakdown of muscle cells and the more severe Duchenne muscular dystrophy. If the number of nucleotides deleted in the mRNA is a multiple of three, the mutation does not cause a frameshift and this typically results in a less defective version of the protein, less rapid breakdown of muscle cells, and the milder Becker muscular dystrophy. (Notice that the crucial factor is whether the mutation is a frameshift mutation, not the overall length of the deletion which can be quite long for some cases of Becker muscular dystrophy and quite short for some cases of Duchenne muscular dystrophy.) Up to one-fifth of cases of Duchenne muscular dystrophy are caused by a nonsense mutation, a point mutation that results in a stop codon. (The type of dystrophy can also be influenced by additional factors such as the specific location of the mutation in the dystrophin gene.)

Because these recessive alleles are on the X chromosome, muscular dystrophy is much more common in boys than in girls. Duchenne muscular dystrophy affects one in every 3500 male births.

For more information on muscular dystrophy see:

- Your Genes, Your Health from the DOLAN DNA LEARNING CENTER, available at <http://www.ygyh.org/dmd/whatisit.htm>
- Muscular Dystrophy, available at <http://www.mayoclinic.com/health/muscular-dystrophy/DS00200>

Alternative Activity

"The Molecular Biology of Mutations and Muscular Dystrophy" is a discussion/worksheet activity in which students explore the effects of different types of point mutations and deletion mutations and relate different types of deletion mutations to more or less severe types of muscular dystrophy (available at <http://serendip.brynmawr.edu/exchange/bioactivities/mutation>).

Related Activity

"Toothpick-ase" is a simulation in which students learn about factors that influence the rate of the enzyme-facilitated reaction (available at <http://www.biologyjunction.com/toothpickase.htm>).