QUICK FIX

Constructing & Using Case Studies in Genetics To Engage Students in Active Learning

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NABT BioClub RECOMMENDATION

One of the national goals in science education is to teach science in a way that mirrors the process of science as inquiry, described by the *National Science Education Standards* (NSES) Science Teaching Standard B and Content Standard A (NRC, 1996). Inquiry-based learning, including the use of case studies, is one of several types of active learning that allows students to experience critical thinking skills inherent in the science process (Handelsman et al., 2007). Using case studies also develops skills in group learning and personalizes and humanizes science, making it more relevant to students (National Center for Case Study Teaching in Science, 2008). Case studies involving inherited conditions can be used to learn Medelian genetics in freshman or honors biology classes. Case studies can also serve as formative assessments to see how well students have learned and can apply genetic principles to real-world situations.

○ Building a Case

The learning goals and the needs of the class should be taken into consideration when planning cases. For my lower level high school classes, cases are diagnostic and have one correct answer. The goal is for students to form hypotheses, apply their prior knowledge of Medelian genetics to analyze the information, and support their conclusions based on the data (Table 1).

Table 1. Learning goals for genetics case study in a sophomore biology class.

• Gain familiarity with the nature of science through hypothesis generation.
• Apply knowledge of human inheritance patterns by using pedigrees.
• Draw conclusions based on evidence.

Figure 1. An example of a case study in genetics with student prompts.

A 27-year-old investment banker called his doctor concerned about malodorous urine. When the patient urinated first thing that morning, he had noticed that his urine had a terrible sulfurous smell. It was now 10:00 am and his most recent urine still smelled. The patient was concerned that he might have food poisoning. The patient felt physically fine but was mentally distraught. He had gone out to eat with his girlfriend the evening before to celebrate her promotion. They drank a celebratory bottle of a new variety of red wine that, in retrospect, seemed to have an off-taste. In addition, the patient mentioned eating some foods for the first time, such as artisanal cheeses made with raw milk, guava, star fruit, asparagus, and parsnips. He had called his girlfriend but she reported no change in the smell of her urine.

Working with your group, think of three possible reasons (hypotheses) why this patient could be exhibiting these symptoms.

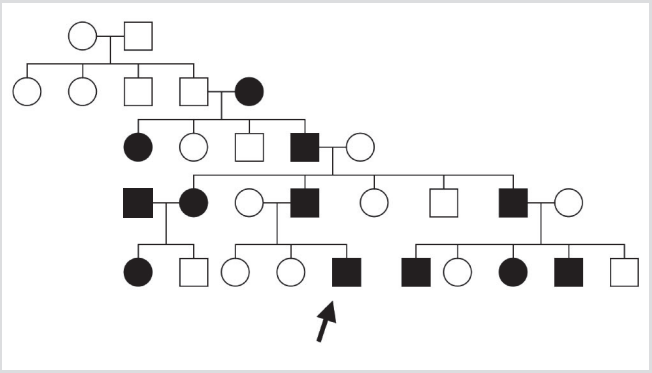
Describe how you would test each of the three hypotheses, including any further information you would need.

A case opens with a scenario presenting information about an individual who is displaying unusual symptoms, along with prompts for student discussion (Figure 1). To promote thinking skills in students, I initially do not give them enough information to conclude whether the symptoms are from an inherited condition or due to other causes. Students propose hypotheses for the symptoms and how these could be tested.

Next, students are given a pedigree that shows the prevalence of the symptoms in the family of the individual (Figure 2). Students examine the pedigree and use the prompts to discuss their reasoning and conclusion about the type of inheritance depicted. Students often need to be reminded that even if the pedigree could support several modes of inheritance, they need to look at the prevalence of the trait in each generation.

Students want to know if the condition described in their case study is real or not. I either reveal the information or ask students to do some sleuthing for homework and report back to the class on their findings.

Figure 2. A family pedigree of the individual (arrow) in the case described in Figure 1, with student prompts. The inheritance is autosomal dominant.



Evaluate your pedigree to determine if the person's symptoms are a heritable trait.

If this is a heritable trait, explain the pattern(s) of inheritance that is (are) compatible with the data. Provide evidence for your answer. Explain how you could verify the mode(s) of inheritance. Further, if you can rule out some patterns of inheritance, explain how you were able to do so.

If the symptoms do not appear to be inherited, describe other hypotheses that could support the data. Explain how you could test these hypotheses.

Table 2. Genetic conditions used for case studies. These conditions were chosen because they are not necessarily life-threatening or commonly found in biology textbooks.

Trait	Feature	Mode of inheritance	Resources for more information
Producing odorous urine after eating asparagus	About 79% of Americans excrete sulfur compounds in their urine after eating asparagus.	Autosomal Dominant	http://www.latimes.com/features/printedition/health/la-he-eat21jan21,0,317605.story
Ability to taste PTC/PROP	Many plant foods contain phytochemicals that are related to PTC, so tasters find these foods bitter.	Autosomal Dominant	Drewnowski, A., Henderson, S.A. & Barratt-Fornell, A. (2001). Genetic taste markers and food preference. *Drug Metabolism and Disposition* 29(4), 535-8. http://dmd.aspetjournals.org/cgi/reprint/29/4/535.pdf
Piebaldism	Piebald patterns in skin are caused by interference of the migration of melanocytes during development.	Autosomal Dominant	http://www.pubmedcentral.nih.gov/articlerender.fcgi?artid=295855
Earwax types	There are two types of earwax, wet and dry. The wet type is found in approximately 97% of people of European and African descent. The dry form is more common in East Asians.	Wet type: Autosomal Dominant	http://www.nytimes.com/2006/01/29/science/29cnd-ear.html?_r=1&oref=slogin
Pachyonychia Congenita	Thick skin and nails with skin blistering. Some forms have onset from late childhood to early middle age.	Autosomal Dominant	http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&partid=128
Trimethylaminuria	People are unable to break down trimethylamine, a compound in the diet, resulting in a fish-like odor in sweat, urine, and breath.	Autosomal Recessive	Source: http://ghr.nlm.nih.gov/condition=trimethylaminuria http://abcnews.go.com/print?id=2287206

I use genetic conditions not usually found in biology textbooks that follow a single gene inheritance pattern. These conditions are not usually life-threatening and some may manifest later in life (Table 2), making for interesting cases. The National Center for Case Study Teaching in Science has many resources and links for designing or using existing case studies in all areas of science. Additionally, there are many Web resources (such as Genetics Home Reference) for specific information on human genetic diseases and conditions. (Table 3).•

Table 3. Resources for creating genetics case studies.

Resource	Web Address
Genetics Home Reference	http://ghr.nlm.nih.gov/
Medline Plus Medical Encyclopedia	http://medlineplus.gov/
National Organization for Rare Disorders	http://www.rarediseases.org/
National Human Genome Research Institute	http://www.genome.gov/
Mayo Clinic	http://www.mayoclinic.org/
Merck & Co., Inc.	http://www.merck.com/

References

Handelsman, J., Miller, S. & Pfund, C. (2007). *Scientific Teaching.* New York, NY: W.H. Freeman and Co.

National Center for Case Study Teaching in Science. (2008). University at Buffalo, State University of New York. Available online at: http://ublib.buffalo.edu/libraries/projects/cases/case.html.

National Research Council. (1996). *National Science Education Standards.* Washington, DC: National Academy Press. Available online at: http://www.nap.edu/openbook.php?record_id=4962&page=R1.

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