Original Publication

OPEN ACCESS

Pedigree Analysis: A Team-Based Learning Activity

Jack Thatcher, PhD, Patty Canfield, MD, Laurie Bauer, DO, Brian N. Griffith, PhD*

*Corresponding author: bgriffith@osteo.wvsom.edu

Citation: Thatcher J, Canfield P, Bauer L, Griffith BN. Pedigree analysis: a team-based learning activity. *MedEdPORTAL*. 2017;13:10557. https://doi.org/10.15766/mep_2374-8265.10557

Copyright: © 2017 Thatcher et al. This is an open-access publication distributed under the terms of the Creative Commons Attribution-NonCommercial-Share Alike license.

Abstract

Introduction: Regardless of their specialty, physicians, particularly those practicing primary care in rural areas, typically encounter genetic conditions. Therefore, it is important to incorporate genetic principles into medical training prior to students' clinical rotations. **Methods:** The advance preparation assignment for this team-based learning (TBL) resource includes lectures and directed study assignments on the following topics: Mendelian genetics, sexual genetics, population genetics, and pedigree analysis. Students then demonstrate their understanding of the content through a formal TBL lesson utilizing a gallery walk for group application exercises. **Results:** Course evaluations clearly indicated that students enjoyed and learned from this TBL. Student performance on the team readiness assurance test and the summative exam scores showed significant signs of improvement when compared to individual readiness assurance test performance. **Discussion:** In this TBL module, students develop an understanding of basic genetics, pedigree analysis, and calculation of risks for inheriting autosomal recessive and X-linked recessive diseases. This TBL can be easily adapted to other allied health programs.

Keywords

Team-Based Learning, Pedigree, Genetics, Population Genetics, Population

Appendices

- A. IRAT and TRAT.docx
- B. Application Exercises.docx
- C. Instructor's Guide.docx
- D. Advance Preparation Reading.pdf

All appendices are peer reviewed as integral parts of the Original Publication.

Educational Objectives

At the end of this team-based learning module, students will be able to:

1. Define the terminology of basic genetics.
2. List the genotypic and phenotypic ratios for autosomal recessive, heterozygous monohybrid and dihybrid crosses, and test crosses.
3. Use genetic ratios to calculate the risk that an individual will be homozygous or heterozygous for a specific allele.
4. Explain the characteristics of X-linkage.
5. Define the variables of the Hardy-Weinberg equations.
6. Be able to use Hardy-Weinberg equations to compute probabilities of inheriting given genotypes.
7. Construct a pedigree chart.
8. Use pedigrees to compute a patient's risk for being afflicted with a genetic disease.

Introduction

Genetics plays a major role in a multitude of diseases, thus making it a foundational discipline for all health professionals.¹ Since 1990, there has been a significant increase in our genetic knowledge.^{2,3} Due to this rapid increase in the understanding of genetics and diseases, it is imperative that the standards of training for physicians and other medical professionals be adjusted to incorporate these new findings. In addition, there is a huge deficiency in genetic knowledge across all health professional schools.¹⁻⁶ A study in 2013 surveying American and Canadian medical schools revealed that only 26% of third- and fourth-year students had exposure to genetics as part of their formal curriculum, and more than half (54%) of these students felt that they lacked basic fundamental genetic knowledge.⁷ Furthermore, a study of internal

medical residency programs in 2004 revealed an increased need for genetics education within residency training programs.⁸ Despite these needs, it has become even more difficult to add this content given demanding medical school curricula.

This team-based learning (TBL) exercise was developed as a component of the Blood & Lymph course at the West Virginia School of Osteopathic Medicine. This TBL differs from other MedEdPORTAL-published TBLs^{9,10} for a number of reasons. First, it incorporates the following genetic principles: hemizygoty and the use of monohybrid and dihybrid ratios to deduce risks. Second, the pedigrees in our application exercises are more advanced than those found in these other MedEdPORTAL TBLs, as our exercises require students to evaluate the inheritance of two genes in the same family's history: one autosomal recessive and the other X-linked recessive. Third, the third application exercise requires students to integrate patterns and consider population genetics. Fourth, our TBL is unique compared with other genetic TBLs because it incorporates basic genetics along with biochemistry, pathology, and clinical decision-making skills. Not only does this point reinforce each discipline, it also helps medical students appreciate the application of genetics to their future medical practice. Finally, our TBL is distinctive because we use a gallery walk as a component of the application exercise. In summary, this TBL module provides students with the opportunity to integrate and apply basic genetic concepts.

The primary goal of this resource is to offer faculty an opportunity to create an active and engaging TBL where health profession learners can apply their skills to real-life genetics and pedigree problems that physicians may experience in a wide variety of health specialties. At the same time, additional goals include facilitating the development of collaborative team skills and implementing a hands-on learning experience for real-life genetic/pathology clinical case presentations.

Methods

Advance Preparation Resources

The advance preparation assignment is located in Appendix D. This appendix contains background information on basic, sexual, and population genetics, as well as information on pedigree analysis. The first three topics (basic Mendelian genetics, sexual genetics, and population genetics) are lectures. Pedigree analysis is a self-study assignment. As a component of their directed study assignment, students are asked to work out multiple pedigree problems individually. After a few days, students have the opportunity to attend a Q&A session with the instructor. During this session, students are encouraged to ask the instructor specific questions about any of the homework assignments. The final activity of this lesson is the TBL session. In total, this entire TBL module requires a minimum of 4 hours of advance preparation time, 1 hour for a Q&A session, and 2.5 hours for the TBL session.

If a faculty member chooses to replace the handouts in Appendix D, the following genetics textbooks can serve as an adequate replacement:

- Turnpenny and Ellard, *Emery's Elements of Medical Genetics*.¹¹
- Russell, *Genetics*.¹²

Team Formation

Large class sizes of 200+ students may be divided into approximately 30 groups of six or seven students per group. At our institution, this TBL occurs in the seventh month of the students' medical curriculum. Students may be randomly selected for teams at the beginning of medical school based on the criteria published by Michaelsen and colleagues.^{13,14} Students stay on the same teams for their first year of medical school.

Calculators

Many types of testing software, such as Exam Soft, have built-in scientific calculators, which students will need to use for the individual readiness assurance test (IRAT). Alternatively, calculators with restricted functionality may be used in instances where testing software lacks a built-in calculator or if a paper examination format is utilized.

Facilitators

This TBL is designed to be administered to 200 students but could be easily modified for smaller groups. At our institution, the TBL room contains eight projector screens and room for all 200 students. Each group is provided a large card (approximately 8 by 11 inches) with the group number on it to help the facilitator identify the group. For this TBL module, if used in a large-group setting, a content expert in pathology and genetics, as well as a facilitator with experience in pathology, genetics, biochemistry, and extensive experience in TBL pedagogy, should be present to administer the resource.

Readiness Assurance Questions

Seven multiple-choice questions constitute both the IRAT and the team readiness assurance test (TRAT). These tests are located in Appendix A. Facilitator notes are included in the instructor guide (Appendix C). Appendix C also contains the correct IRAT/TRAT answers and potential talking points. Regarding group appeals, a group may file an appeal if it has missed a particular question and may do so before the end of the group discussion. If an appeal is accepted, then those points are awarded to the group that appealed unless the appeal is based on grammar or question syntax, in which case the entire class receives points.

Immediate Feedback

Immediate feedback is provided on the TRAT through the use of Immediate Feedback–Assessment Technique cards. The cards can be purchased from <http://www.epsteineducation.com/home/about/default.aspx>.

Group Application Exercise

The application part of the exercise (Appendix B) starts immediately after the TRAT and can be either open or closed book based on instructor preference. Answers and rationales for each exercise are located in Appendix B. Students work out the first two pedigree problems on large 3M Post-it notes. Once the groups are finished, they display their work around the classroom. A gallery walk is conducted by providing each team member with one sticker to vote for the best pedigree based on, but not limited, to the following criteria: clarity, explanation, and correctness. Students should not vote for their own project.

At the conclusion of the gallery walk, students go back into their groups and complete the third application exercise. Students are provided real-life scenarios that they rank based on which is the most concerning due to pathology and probability of disease manifestation. For the group discussion, each team responds simultaneously using a series of laminated, color-coded flash cards (lettered A, B, C, D, and E) that should be created beforehand. The first team to explain its answers is randomly selected. This discussion continues until a consensus answer is reached or until the instructor provides a definitive answer.

While students are discussing the third application exercise, an instructor reviews the first two application exercise calculations displayed on the sticky notes, looking for recurrent errors. Typically, there are one or two common mistakes. The most common mistakes include errors in the pedigree format, mathematical errors, or forgetting that for X-linked traits, when the sex of a progeny is not specified, one must multiply by .5 for the chance that the offspring will be a boy. Once the discussion of the third exercise question is concluded, the second exercise is discussed. To do this, the instructor selects a group to come up to the front of the room and describe how its members calculated the genetic frequency for the exercise. After the group finishes, the room is asked if anyone disagrees. In this manner, the instructor guides the students to the correct calculation, thus revealing recurrent errors. The instructor then moves onto the first application exercise, which is conducted similarly to the second application exercise.

After the TBL session, the instructor collects all the sticky notes and analyzes each group's project. The sticky notes are then displayed in the classroom, which allows students to compare and contrast the strengths and weaknesses of each group's project. Students are allowed to compare each other's work for several days prior to the final summative exam for the course. This learning strategy also makes our TBL unique. It should be noted that in this group application exercise, any of the pedigrees, cases, and/or individual problems may be removed from the exercise to simplify and/or shorten the exercise.

Facilitation Schema

The time breakdown is 2.5 hours. See Appendix C for a detailed diagram of our facilitation schema.

- IRAT: 15 minutes (2 minutes per question).
- TRAT: 20 minutes.
- Group discussion: 5 minutes.
- Application exercises 1 and 2: 30-40 minutes.
- Gallery walk: 30 minutes.
- Application exercise 3 administration and discussion: 35 minutes.
- Group discussion for application exercises 1 and 2: 5 minutes.

Results

This TBL module was piloted for 2 years at the West Virginia School of Osteopathic Medicine. We have 400 students in our data set (see the [Figure](#)). All but four groups from the class of 2015 scored 100% on the TRAT, and all 31 groups from the class of 2016 scored 100%. The IRAT scores are very similar for the two classes, with a mean increase of 25.6% from IRAT to TRAT. The summative course exam score for genetics questions increased by 16.5% from the class of 2015 to the class of 2016. Moreover, student performance increased by 13.1% when comparing the mean IRAT scores to the mean score on the genetic portion of the final exam. These data confirm that the TBL experience improved student learning. The following excerpts from course evaluations clearly show that students enjoyed the genetics TBL:

Figure. Individual readiness assurance test (IRAT), team readiness assurance test (TRAT), and Blood & Lymph course exam scores are expressed as a percentage of items correct ($N = 400$). Error bars denote high and low scores. All groups from the class of 2016 scored a 100% on the TRAT, thus collapsing that error bar.

- “I think the genetics TBL strongly added to my understanding.”
- “The genetics TBL was useful in [that] it created an open forum to discuss inheritance and get the group working on problems.”
- “The TBL was well organized and added to my understanding. While genetics is a difficult subject to study in a group, it was helpful to get feedback from other students and tips on how to approach problems in easier, quicker ways.”
- “The material provided for the genetics TBL was detailed and thorough and adequately prepared me for the TBL.”

Discussion

This genetics TBL is a component of the Blood & Lymph course. Based on the data, the TBL clearly shows improvement on both the TRAT and the summative exam and based on feedback has been well received by students. This particular TBL includes a gallery walk component. To our knowledge, this is the only TBL

publication in MedEdPORTAL using a gallery walk in a large-group setting. In addition to the gallery walk, the instructors also provide critiques of each group's pedigree problems, which are then reposted in the classroom prior to the final exam to give students additional opportunities to learn the content. The other major contribution of this TBL is the final application exercise, which requires students to rank clinical cases from most severe to least severe. This real-life application exercise allows students to put everything they have learned into practice.

The one problem we encountered with this TBL session is time. First, students require more time to work out a pedigree problem than a typical multiple-choice problem. Therefore, we increased the time per question from 1.5 minutes to 2 minutes. It is important to instruct students ahead of time that to complete the problems during the time constraints of an examination, they must become proficient at working them out in a timely manner. A second time constraint is the gallery walk. To compensate, we increased this TBL module by half an hour, to 2.5 hrs.

A strength of this genetic TBL is peer learning. While peer learning is a benefit of any TBL, it is particularly advantageous for the topic of pedigrees because peer learning complements the advance preparation reading that students undertake on their own. After students familiarize themselves with the process by working out practice problems, their skills are bolstered by comparing and contrasting each other's grasp of the content. Thus, this TBL solidifies the competency of the majority of a class to work out pedigree problems.

Pedigree problems typically are not conducive to large-group discussion because few errors persist after small-group discussions and these errors are usually resolved very quickly. The gallery walk addresses this limitation. It allows students to compare and contrast different practices. For instance, they see how shortcuts and inappropriate use of symbols make the pedigrees more difficult to keep track of, thereby increasing the potential for error as well as the time required to complete the problem. In order to answer genetic questions on standardized exams, it is important for students to be both expeditious and accurate. Thus, the gallery walk serves to promote best practices. Furthermore, the third application exercise allows students to apply genetic principles to clinical scenarios. Not only does this reinforce their appreciation of genetics, it allows them to integrate genetic and clinical principles. We conclude that this TBL is an effective tool for the teaching of pedigree analysis to medical students and that this resource offers students the opportunity to integrate topic-specific basic science and clinical science content.

Jack Thatcher, PhD: Associate Professor, Department of Biomedical Science, West Virginia School of Osteopathic Medicine

Patty Canfield, MD: Pathologist and Medical Director, Greene County General Hospital, Putnam County Hospital, and Sullivan County Community Hospital

Laurie Bauer, DO: Assistant Professor of Clinical Science, West Virginia School of Osteopathic Medicine

Brian N. Griffith, PhD: Associate Professor, Department of Biomedical Science, West Virginia School of Osteopathic Medicine

Disclosures

None to report.

Funding/Support

None to report.

Ethical Approval

Reported as not applicable.

References

1. Wolyniak MJ, Bemis LT, Prunuske AJ. Improving medical students' knowledge of genetic disease: a review of current and emerging pedagogical practices. *Adv Med Educ Pract*. 2015;6:597-607. <https://doi.org/10.2147/AMEP.S73644>

2. Feero WG, Green ED. Genomics education for health care professionals in the 21st century. *JAMA*. 2011;306(9):989-990. <https://doi.org/10.1001/jama.2011.1245>
3. Daack-Hirsch S, Jackson B, Belchez CA, Elder B, Hurley R, Kerr P. Integrating genetics and genomics into nursing curricula: you can do it too! *Nurs Clin North Am*. 2013;48(4):661-669. <https://doi.org/10.1016/j.cnur.2013.08.005>
4. Dudlicek LL, Gettig EA, Etzel KR, Hart TC. Status of genetics education in U.S. dental schools. *J Dent Educ*. 2004;68(8):809-818.
5. Goldgar C, Rackover M. Current status of genetics education and needs assessment of physician assistant programs: a nationwide survey. *J Physician Assist Educ*. 2007;18(2):53-59. <https://doi.org/10.1097/01367895-200718020-00011>
6. Cichon M, Feldman GL. Opportunities to improve recruitment into medical genetics residency programs: survey results of program directors and medical genetics residents. *Genet Med*. 2013;16(5):413-418. <https://doi.org/10.1038/gim.2013.161>
7. Mainous AG III, Johnson SP, Chirina S, Baker R. Academic family physicians' perception of genetic testing and integration into practice: a CERA study. *Fam Med*. 2013;45(4):257-262.
8. Riegert-Johnson DL, Korf BR, Alford RL, et al. Outline of a medical genetics curriculum for internal medicine residency training programs. *Genet Med*. 2004;6(6):543-547. <https://doi.org/10.1097/01.GIM.0000144561.77590.85>
9. McKee E. Risk analysis in human genetics—a team-based learning exercise. *MedEdPORTAL Publications*. 2011;7:8409. http://dx.doi.org/10.15766/mep_2374-8265.8409
10. Park V, Brescia W, Nishimoto S, et al. Genetic risk for recessively inherited disease (Duchenne muscular dystrophy and congenital hearing loss). *MedEdPORTAL Publications*. 2012;8:9211. <https://www.mededportal.org/publication/9211>
11. Turnpenny PD, Ellard S. *Emery's Elements of Medical Genetics*. 14th ed. Philadelphia, PA: Elsevier Churchill Livingstone; 2012.
12. Russell PJ. *Genetics*. 5th ed. San Francisco, CA: Benjamin-Cummings Publishing; 1998.
13. Michaelsen LK, Sweet M. The essential elements of team-based learning. *New Dir Teach Learn*. 2008;(116):7-27. <https://doi.org/10.1002/tl.330>
14. Johnson C. Review of: Michaelsen LK, Parmelee DX, McMahon KK, Levine RE, eds. *Team-Based Learning of Health Professions Education: A Guide to Using Small Groups for Improving Learning*. *J. Chiropr Educ*. 2009;23(1):47-48. <https://doi.org/10.7899/1042-5055-23.1.47>

Received: September 20, 2016 | **Accepted:** March 1, 2017 | **Published:** March 21, 2017