Teaching and Learning Grant Application Form

Small Grant (up to $2000)

Project Title: Engaging students in learning genetics and genomics principles using PGT.

Project Supervisor: Dr. Kimberley Dej

Department/School: Biology

Faculty: Science

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Collaborator(s): Dr. Mihaela Georgescu, Dr. Bhagwati Gupta, Dr. Xu-Dong Zhu

Please complete the areas below:

I. Principle course(s) for which this project is/are intended:

Course(s)	Title	Enrolment
MOLBIO2C03	Genetics (Pilot project, if funded by MIIETL)	40
BIOL2C03	Genetics (Extended application of project resources)	600+
LIFSCI2G03	Genes, Genomes, and Society (Use of modules)	100+

II. Briefly outline the teaching and learning issue(s) addressed by your proposed project, and how the results might benefit student learning: *(max 1200 characters)*

Genetics is the study of the inheritance of traits and diseases. At many universities the topic causes student anxiety. Students struggle with the breadth of information and the added frustration of the mathematics of statistical calculations. We now add to our list of expectations, an understanding of the study and application of genomics, a subfield of genetics that looks at all of the genes in an organism.

We do and should expect that our Science students graduate with an understanding of genetic and genomic principles and the ability to engage in constructive discourse about the applications of genomic data. Together, these form "genomic literacy". Limits to genomic literacy in our society may compromise the positive impacts that the power of genetics and genomics will have on health care. As future policy makers, health care practitioners, and indeed patients, we hope that our students leave McMaster University with a well-informed understanding of the possibilities and limitations of genomic information.

Indicate/describe the role of the Project Supervisor as it relates to the project and the course(s) it affects:

Dr. Kimberley Dej, Project Supervisor:
Co-instructor of MOLBIOL2C03 and BIOL2C03. Instructor of LIFESCI2G03.

Amount requested from MIIETL: $2 000

Signature of applicant*: Kimberley Dej

Date: February 9, 2015

**Signatures are not required if submitting the application electronically; however, in order for the application to be complete, the applicant must 'cc' the chair/director/dean when submitting via email. For electronic submissions, please type the name and date of the applicant/chair/director/dean into the appropriate fields.*

Comments of Chair/Director of School (in Health Sciences, Program Director) *max 1800 characters*

The Teaching & Learning Grant Peer Review Committee would appreciate your comments on the following:

1. What is the department's contribution to this project?
2. How will the department ensure the continuation of this project after the initial grant?
3. Where relevant, please comment on the relationship between this project and other initiatives in the department.

The Department of Biology is supportive of this new direction in teaching genetics and genomics. It is consistent with our mandate to engage students in active-learning and research opportunities.

1. Three faculty and one IA have agreed to contribute to the development of learning modules for analysis of PGT data. These modules will be used in two genetics courses, MOLBIO2C03 (40 students) and BIOL2C03 (600+ students), in the analysis of either PGT data or anonymized data sets. The Biology department will provide lab and computer support to implement and interpret the study. Biology will subsidize release time and travel costs to meetings to communicate SoTL results.

2. Biology's academic plan includes introducing more active learning experiences into our programs. We are therefore dedicated to allocating resources to support this project. A cost analysis will enable us to decide if we can fully support this through normal course expenses, or whether students will be asked to purchase their data sets, like courseware. Completed learning modules will continue to be a part of both genetics courses after the study.

3. Our level II Genetics offerings are an ideal target for more active learning experiences, as these courses have no lab component. This initiative will develop proficiency in a number of our program Learning Objectives, including to (1) analyze and interpret data regarding inheritance, (2) demonstrate computer literacy, (3) translate scientific knowledge to a real world application, (4) exhibit an ethical approach to science. This project highlights and contributes to the Department of Biology's strengths in genetics, bioinformatics, and molecular genetics. We believe that the incorporation of these research-based, active-learning modules in core courses will contribute to the enrolment and retention of students in the Molecular Biology Program. Success of this project may lead to the incorporation of similar data sets in upper year courses in our department. The project may also contribute to novel teaching resources in graduate courses and potential new graduate programs.

I have had the opportunity to read the Teaching & Learning Grant written application, and ask any relevant questions to the applicant.

Name of Chair/Director: Dr. Roger Jacobs	
Signature*: Roger Jacobs	**Date:** February 9, 2015

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ENGAGING STUDENTS IN LEARNING PRINCIPLES OF GENETICS AND GENOMICS USING PERSONAL GENOME TESTING.

Kimberley Dej, Mihaela Georgescu, Bhagwati Gupta, and Xu-Dong Zhu, Department of Biology

INTRODUCTION

Understanding genetics is essential not just for every student in Biology, but also for every citizen. The volume and complexity of information available to individuals in the daily news and at the doctor's office can be overwhelming. Moreover, many of our students are interested in various careers in medical research and health care. It is hard to imagine any future career in medicine that does not involve some degree of genetic testing either as diagnosis or as personalized pharmaceutical treatments. More recently, the use of the term 'genomics' has entered into common parlance. Genomics is a sub-discipline of genetics that involves the study of all of the genes in an individual, multiplying the complexity by thousands. In addition to understanding the scientific principles in the fields of genetics and genomics, we want our students to develop the ability to engage in constructive scientific discourse about the realistic and ethical applications of genomic data. Together, these form genomic literacy. Limits to genomic literacy in our society may compromise the positive impacts that the power of genetics and genomics will have on health care. As future policy makers, healthcare practitioners, and patients, we hope that our students leave McMaster University with an informed understanding of the power and limitations of genomic information.

Genetics has historically been a difficult class to teach. Genetics is a field that requires a comfort with biological principles in addition to the mathematical skills to interpret the data. This is more so in the younger field of genomics. Students struggle with both the depth and breadth of information that they need to learn, and the application and statistical analysis of the data. While data are becoming fast and inexpensive to acquire for many citizens, the knowledge and skills to interpret the data is lagging behind. To prepare our students for their futures, we need to find the best ways for students to learn how to analyze and interpret genomic data.

Many private companies are starting to offer Personal Genome Testing (PGT) in the form of direct-to-customer genome testing. PGT is the collection of many genetic markers across the genome of an individual. PGT provides the information, but the interpretation of this information is the most important part of the process. How can we best teach our students the skills to calculate risk assessment and patterns of inheritance? Engaging students in the lab or classroom by using themselves as subjects has proven to be successful in many disciplines. In our own Biology 1A03 labs, we have engaged students by having them sample their own DNA and proteins to examine the relationship between amylase gene copy number and levels of enzyme expression. This project, piloted in Spring 2014 and offered to over 1200 students in 2014/15 has met with great success. Students are excited about the lab and skill acquisition comes much more naturally as it is simply a necessary part of the thrill of the research.

The primary hypothesis of this study is that the analysis of personal genomic testing (PGT) data will enhance student engagement and the understanding of the principles and applications of genomics.

RATIONALE AND PROJECT GOALS

Outcomes:

- Students will show a greater level of engagement.
- Students will illustrate a better understanding of the risks and challenges of PGT and be able to make informed decisions.
- Students will demonstrate deeper learning of the principles.

Proposed Teaching and Learning Activities:

Currently available PGT covers an array of hundreds of single nucleotide polymorphisms or changes in genetics sequences that are associated with variation in the appearance of traits. The "Health" component includes genes for inherited characteristics (e.g. cystic fibrosis), genetic risk factors (e.g. celiac disease), drug responses, and traits. We wish to work with the 23andMe Academic program to develop a miniature version of the PGT kit that contains only non-clinical traits. This would include:

- *Bitter taste perception:* A common test in a genetics class is the ability to taste a harmless chemical called PTC. Such studies can be partnered with information about gene sequence.
- *Eye colour:* Eye colour is based upon sequence variation at several genetic loci. It is the combination of genetic variation that ultimately leads to the colour that we see. The test provides a statistic called "likelihood" for different eye colours.
- *Asparagus metabolite detection:* The ability to detect an odor in urine after consuming asparagus.
- *Photic sneeze reflex:* The tendency to sneeze when moving from relative darkness into bright light. Each copy of a particular genetic variant increases the odds of exhibiting this response by 1.3 times.
- *Earwax type:* Earwax may be described as wet or dry. This trait displays a classic Mendelian pattern of inheritance at one gene, with no effect from the environment.
- *Alcohol flush reaction:* A common test in physiology and genetics labs in the 1970's and 80's was the alcohol flush reaction – the tendency to turn bright red upon consuming alcohol. This test is inappropriate for undergraduates, but we can revisit it at the genetic level. The response is due almost entirely to two genes.
- *Height:* Students can make predictions on their expected height based upon PGT results and compare that prediction to their actual height.

Paired with the students' access to these data would be activities on data analysis including:

- *What are SNPs?* Single nucleotide polymorphisms are single changes in DNA sequence. Students will learn about SNPs in the human genome and then examine their own genome to see the identity and frequency of SNPs.
- *What is heritability?* Heritability is a measure of the proportion of the variation of the appearance of a trait in population that is due to genetics. Students will see first hand with their data sets how heritability is dependent upon variation in genes and the environment.
- *What is Mendelian inheritance?* Demonstration of the basic principles of inheritance using student data.
- *Relationship between genotype and phenotype?* The relationship between the genetic variation that we carry (genotype) and the expression of the trait or characteristic (phenotype) is an essential part of any genetics class. We will use examples from the student's data set to illustrate relationship between gene, protein, and trait.

- *Interpreting PGT data – what is risk assessment?* Genetic variation at many genes and variation in environment contribute to the propensity to show different traits. Data interpretation requires an understanding of conditional probabilities and risk analysis.
- *How "good" is PGT data?* Given that many genes and the environment contribute to a trait, what is the usefulness of PGT data? Students will make informed, personal decisions on the usefulness of PGT in today's society and in the healthcare system.

LITERATURE REVIEW AND ALTERNATIVES CONSIDERED

PGT at other institutions:

A few institutions have offered students the opportunity to learn and practice the art of PGT interpretation. One example comes from a small Stanford Medical School course that demonstrated enhanced student learning when students used their own genome for analysis (Salari *et al*, 2013). In the Stanford study, 46 medical and graduate students purchased PGT kits from the private company, 23andMe, and brought their data to the classroom. Students were surveyed before and after the course about their attitudes and knowledge of genomics and personalized medicine. The effect on attitudes was strong. One question was whether the students felt that patients and doctors could comfortably interpret information provided by PGT. Students who studied their own data as compared to a standardized data set were more likely to feel that individuals could perform the necessary analysis and interpretation of the data.

In terms of knowledge, both groups expressed in the pre-survey questionnaire that patients and doctors were ill-prepared to analyze the data. By the end of the course, students who analyzed their own data felt more competent to interpret PGT data than those who looked at standardized data sets. A short test of knowledge before and after the course substantiated this perception. Students using their own personalized data sets showed significant improvement in their knowledge scores after the course compared to before (31% improvement in scores). Students using the standardized data set showed little improvement in knowledge scores (1% improvement in scores).

Another study looked at the effect of incorporating PGT into the undergraduate classroom at two universities: Duke University and Penn State University (Daley et al, 2012). Students indicated in surveys that when course material is personalized, the course is more interesting (94.6%) and the material is easier to learn (87.3%). Professors reported that personalized data increases student learning (72.6%). On the various criteria of attitudes and knowledge, the researchers felt that the PGT augmented the educational value of teaching genomics, PGT, and personalized medicine.

Who else is using PGT in the classroom?

- Duke University (Nursing School, 2013)
- Duke and Penn State (Undergraduate Genetics, 2013)
- Mount Sinai Medical School (School of Medicine, 2013)
- Stanford University (Medical School, 2012)
- York University, Canada (Not course based - Innovation Project; 2012).

Other approaches:

The company 23andMe provides access to many educational resources as part of their 23andMe Academic Program. These include videos, activities, and discussion forums with other instructors. In addition, they will provide anonymized sample genetics profiles of 14 individuals

representing diverse ancestries. Students can use these data sets to analyze and explore questions about PGT data analysis and interpretation.

Studies have made comparisons between student cohorts using their own data versus anonymized data sets (Daley et al, 2013; Salari et al, 2013). The conclusions were that there was significantly increased engagement and significant increase in knowledge acquisition when students used their own data. Given the increased access an individual has to PGT and the increased role of PGT in human health care, teaching undergraduates about the psychological and ethical dimensions of human gene testing seems important and timely.

Ethical considerations:

There are many ethical considerations that these groups have met with before bringing the personal genome analysis to the classroom. At the forefront, with PGT comes the clinical and diagnostic information that it carries. We will work with 23andMe to assemble just a non-clinical array of genetics markers for a mini version of the PGT, as described above. Using other courses as models, students engage in exercises before deciding to participate in PGT about the risks and consequences. Students are provided with information about the safety, confidentiality, and privacy of the information. Students may choose not to disclose their own data at any time in the course. In addition, students will be allowed to opt out. Students enrolled in the course will have the option of not obtaining their personalized data set and instead receiving an anonymous or fabricated data set.

METHODS AND TIMELINE

1. Spring/Summer 2015: Analysis of 14 anonymized data sets available from 23andMe to establish lesson plans for the six activities described above. These activities will take place in both the lecture hall and the tutorial room.
2. Fall 2015: Pilot with approximately 40 students enrolled in MOLBIOL2C03: Genetics. Students will engage in exercises using their PGT data. Pre- and post-course surveys addressing engagement, perceptions, and knowledge acquisition will be employed. In parallel, the same activities and surveys will be used for students in BIOL2C03: Genetics (300+ students) who will be given the anonymized data sets. These two courses share lecture content, but differ in tutorial exercises.
3. Winter 2015: Evaluation of the efficacy of the use of participatory learning using PGT.
4. Spring 2015: Evaluation of the costs and benefits of expanding the PGT to the entire cohort of genetics students in Fall and Winter terms (600+ students in MOLBIOL2C03 and BIOL2C03).
5. Evaluation of how partial data sets may be incorporated into other classes including first year biology courses.

BUDGET

The requested funds would be used to pilot the course in Fall 2015 to approximately 40 students). We are working with the company 23andMe to come up with a suitable data set and a reduced, bulk cost per student PGT. The current estimate is $50 per student for the pilot. We request this money for the pilot since these students will be participating in a study of the efficacy of the learning tool. **Total funding requested: 40 students X $50 per student = $2000**

If successful, we would need to get the cost per student down to $10 to $20 per student. While we will consider the possibility of building this into the current courses budget (it is a cost

that is significantly lower than the cost of many undergraduate lab courses), we must also consider the possibility that students will be asked to purchase the PGT kit as part of their courseware in a manner similar to other lab supplies (lab coat, goggles), online lab simulations (currently used in genetics and purchased by students), or problems manuals. We will survey students during the pilot project to determine the willingness of students to carry this cost.

IMPACT

- The pilot will impact approximately 40 students engaged who have access to the PGT testing. About 300 students will be in the control group in this pilot year.
- Three faculty members and one instructional assistant will be engaged in the pilot and the long-term implementation of the PGT into the course.

  Project Lead: Dr. Kimberley Dej, Assistant Professor, Department of Biology and the Life Sciences Program

  Co-applicants: Dr. Miheala Georgescu, Instructional Assistant, Department of Biology
  Dr. Bhagwati Gupta, Professor, Department of Biology
  Dr. Xu-Dong Zhu, Professor, Department of Biology
- The potential impact on other courses is an exciting prospect.

Short-term:

1. Expansion of PGT beyond the pilot course to the full BIOL2C03 cohort (600+ students)
2. Expansion to LIFESCI2G03: Genes, Genomes, and Society (100+ students)

Long-term:

3. Possible expansion to first year biology courses, a smaller data set in BIOL1A03: Introduction to Cell and Molecular Biology and ancestry data sets may be incorporated into BIOL1M03: Ecology, Evolution and Biodiversity. Larger data sets that include clinical data sets may be incorporated into upper year courses, pending ethical considerations.
4. If successful, this approach may be of interest to graduate courses in Biology and Health Sciences, Molecular Anthropology courses, and Pharmacology courses.

EVALUATION AND DISSEMINATION

- Date on the effectiveness of PGT on student engagement and learning will use previously established pre- and post-course surveys (Salari etl, 2013). We are also interested to see if this course has an effect on enrolment and retention in our Molecular Biology and Genetics program.
- Data analysis, conclusions, and protocols will first be shared at the Departmental level to investigate further opportunities in our courses.
- Presentations at both the Mcmaster Research in Teaching & Learning conference and the McMaster Learning Technologies Symposium are anticipated.
- Presentation of our outcomes at STLHE (Society for Teaching and Learning in Higher Education) and at the Gordon Conference on Biology Education in 2016 will enable us to share our findings nationally and internationally.

REFERENCES:

Daley et al. 2013. Personal DNA Testing in College Classrooms: Perspectives of Students and Professors. Genetic Testing and Molecular Biomarkers. 17(6):446-452.

Eriksson et al, 2010. Web-based, participant-driven studies yield novel genetic associations for common traits. *PLoS Genet.* 6(6):e1000993.

Salari et al, 2013. Evidence that Personal Genome Testing Enhances Student Learning in a Course on Genomics and Personalized Medicine. *PLoS One*. 8(7):e68853.