

JUNE 4 – JULY 29, 2014

Course Syllabus

GeorgetownX MEDX202-01: Genomic Medicine Gets Personal

5/28/2014

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COURSE DESCRIPTION

While the advances in genomics promise to usher in a new era in medical practice and create a major paradigm shift in patient care, the ethical, legal, and social impact of genomic medicine will be equally significant. The information and potential use of genomic discoveries are no longer issues left for scientists and medical professionals to handle, but have become ones for the public at large. Rarely a day passes without a genomics-related story reported in the media. By the end of this course, students will be able to better understand the field of genomics; be familiar with various online databases and resources; and understand and appreciate the medical, social, ethical, and legal issues associated with the availability of personal genomic information.

Given the diversity of the topics and the specific expertise required to cover each, this is a unique cross-disciplinary course where faculty from different disciplines including genetics, computational sciences, bioinformatics, genetic counseling, bioethics, law, and business will participate in lecturing. We have assembled a team of experts from various departments at Georgetown University and other institutions, to teach this comprehensive online genomics course.

KEY LEARNING OUTCOMES

Upon completing this course, students should have an understanding of these course-learning outcomes:

- 1. The basics of genetic abnormalities and disease
- 2. What we can learn from genetic testing both pre- and post- birth, and in oncology
- 3. The basic science behind how the genetic tests are done
- 4. The ethical, legal, and social implications of genomic discoveries; the genetic counseling issues; and the new trends in direct-to-consumer marketing of genetic tests
- 5. Critical information medical professionals as well as patients and their families need to know to be current in the field
- 6. Available Resources (education, patient support, general information, etc.)

The course contains 5 main themes that, in conjunction, work to support the course-learning outcomes. The themes are:

Theme 1: The Clinic

Theme 2: The Diagnostic Lab

Theme 3: Minding The Business of Genomics

Theme 4: Genomics and The Patient, The Family and The Society

Theme 5: Looking to the Future of Medicine in the Genomic Era

FACULTY

LEAD FACULTY

Bassem R. Haddad, MD, Associate Professor at Georgetown University and Co-Director of the Medical School Molecular and Human Genetics course, will direct this course. Dr. Haddad has an active molecular cytogenetics research laboratory working in the area of translational research and biomarker discovery. His research focuses on understanding the genetic aberrations and instability that occur in genetic diseases, particularly cancer. Dr. Haddad received his MD and Residency training in Obstetrics and Gynecology from the American University of

Beirut, and completed his fellowship training in Molecular Cytogenetics at Boston University School of Medicine, Baylor College of Medicine, and the NIH. He is a longtime member of the Georgetown University Medical School faculty and has been involved in genetic teaching for many years.

GUEST LECTURERS

Michael B. Atkins, MD, is the Deputy Director of the Georgetown-Lombardi Comprehensive Cancer Center and Professor of Oncology and Medicine (Hematology/Oncology) at Georgetown University School of Medicine. Dr. Atkins' major research interests are cancer immunotherapy, treatment of melanoma and renal cell carcinoma, predictive markers for response to biologic therapy, and antiangiogenic and targeted therapies. Dr. Atkins received his MD from Tufts University School of Medicine, and postgraduate training at the Cancer Center at Tufts New England Medical Center Hospital.

Robert Clarke, Ph.D., D.Sc., is the Dean for Research at Georgetown University Medical Center and co-Director of the Breast Cancer Program at the Georgetown-Lombardi Comprehensive Cancer Center. Dr. Clarke's research interests include the cellular and molecular mechanisms driving endocrine resistance in breast cancer as well as drug resistance, drug/hormone interactions, and the application of molecular profiling to predict breast cancer phenotypes and identify functionally relevant gene signaling networks. Dr. Clarke leads several multinational molecular medicine studies in breast cancer, in collaboration with colleagues at Lombardi Comprehensive Cancer Center, Virginia Tech and the University of Edinburgh. Dr. Clarke received a PhD and DSc in Biochemistry from The Queen's University of Belfast, UK.

Kevin Fitzgerald, S.J., Ph.D., a Jesuit priest and a geneticist, is a Research Associate Professor at Georgetown University and the Dr. David P. Lauler Chair for Catholic Health Care Ethics. His research interests have included the investigation of abnormal gene regulation in cancer and ethical issues in human genetics, including the ethical and social ramifications of molecular genetics research. Dr. FitzGerald received a BA in Biology from Cornell, a M.Div. from the Jesuit School of Theology, and a PhD in both Bioethics and Molecular Biology from Georgetown University.

Alessandro Ghidini, M.D., is a Professor at Georgetown University. He is an obstetrician and specializes in maternal-fetal medicine and medical genetics. His areas of focus are in amniocentesis, genetic counseling, and high-risk pregnancies. Dr. Ghidini received his MD from the University of Milan, completed his residency training at the New Haven Medical Center – Yale University, and a fellowship at the Mount Sinai Medical Center.

Eric Green, M.D., Ph.D., is the Director of the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH). NHGRI is the largest organization in the world dedicated solely to genomics research, and aims to advance human health through genomics research. For the past ~25 years, Dr. Green has been at the forefront of efforts to map, sequence, and understand the human genome, including significant start-to-finish involvement in the Human Genome Project. He is responsible for providing overall leadership of NHGRI's research portfolio and other initiatives. In 2011, he led NHGRI to the completion of a strategic planning process that yielded a new vision for the future of genomics research, with particular emphasis on clinical applications of genomics.

Eden Haverfield, Ph.D., FACMG, is an ABMG-certified Clinical Molecular Geneticist and the director of the Whole Exome Sequencing Program at GeneDx. Eden was previously the Director of the Microarray Sequencing Diagnostics and Quantitative Testing groups as well as the Hematology/Immunology program at GeneDx. She received her B.A. in Biological Anthropology from the University of Pennsylvania and her M.Sc. and Ph.D. in Human Genetics from the University of Oxford in Oxford, UK. Eden completed a post-doctoral fellowship in Clinical Pharmacology and Pharmacogenomics before completing her ABMG-accredited clinical molecular genetics training at the University of Chicago. Prior to joining GeneDx, Eden was the assistant director at the University of Chicago Genetic Services Laboratory.

Claudine Isaacs, MD, is a Professor of Medicine and Oncology and the co-Director of the Breast Cancer Program at the Lombardi Comprehensive Cancer Center at Georgetown University. She is also the Medical Director of the Jess and Mildred Fisher Center for Familial Cancer Research. Her research interests include cancer risk assessment and medical management and prevention strategies for women at high risk for breast cancer. Dr. Isaacs received her MD and residency training at McGill University (Canada). She completed fellowship training in the Division of Hematology and Oncology at McGill University and a Fellowship in Breast Medical Oncology in the Division of Medical Oncology at Georgetown University.

Michael Johnson, PhD, is an Associate professor of Oncology at Georgetown University. His research Interests are proteases, invasion and metastasis and the progression of breast cancer. He also focuses on the pharmacogenetics of drugs used in cancer therapy. Dr. Johnson received his BSc and PhD in Biochemistry from the University of Newcastle upon Tyne, UK. He completed a postdoctoral fellowship in Cancer Biology at the Lombardi Cancer Center at Georgetown University.

Anne Deslattes Mays, is a mathematician, software engineer and computational biologist currently working in the field of applied systems biology. Working for Craig Venter at Celera, her Software Systems team together with the Informatics Research team led by Gene Myers, and the Sequencing team led by Mark Adams, sequenced, assembled, mapped and annotated the Celera Genome in the private effort that together with the Human Genome Project completed the sequencing of the human genome. Anne also works with data generated from 2nd and 3rd generation sequencing machines, focusing primarily on RNA-Seq data and seeks to discover full-length transcripts and create predictive models capable of determining causality in an organisms' response to ever changing environmental and evolutionary pressures. Anne received her BSc in Mathematics from McGill University, MSc in Computer Science from The Johns Hopkins University, and a Masters in Tumor Biology from Georgetown University.

Jeanne Meck, Ph.D., FACMG, is the Director of Cytogenomics and Prenatal Diagnostic Services at GeneDx. Her interests are in constitutional cytogenetic diagnosis using classical and molecular methods, as well as in medical genetics education. She has maintained her faculty appointment as Professor at Georgetown and is actively involved in training medical genetics fellows through the NHGRI/NIH fellowship program. Dr. Meck received her MS in Behavior Genetics and PhD in Human Genetics from the University of Connecticut as well as a postdoctoral fellowship in cytogenetics and immunogenetics at the University of Miami School of Medicine.

Cynthia Casson Morton, **PhD**, is the William Lambert Richardson Professor of Obstetrics, Gynecology and Reproductive Biology and Professor of Pathology at Harvard Medical School, Director of Cytogenetics and Past Director of the Biomedical Research Institute at Brigham and Women's Hospital. Her research interests are in molecular cytogenetics, hereditary deafness, genetics of uterine leiomyomata and human developmental disorders. Dr. Morton is currently President of the American Society of Human Genetics. She received her Bachelor of Science degree from the College of William and Mary in Virginia and her Ph.D. in Human Genetics from the Medical College of Virginia in Richmond.

Beth N. Peshkin, MS, CGC, is a Professor of Oncology and the Senior Genetic Counselor at Georgetown-Lombardi Comprehensive Cancer Center. She is also the Education Director for the Jess and Mildred Fisher Center for Familial Cancer Research. Her research focuses on genetic counseling and testing for individuals with an inherited susceptibility to breast/ovarian cancer, such as outcomes in newly diagnosed cancer patients and the effectiveness of telephone genetic counseling as well as internet-based adjuncts to genetic counseling. Ms. Peshkin received her MS in Medical Genetics from the University of Wisconsin-Madison, and is certified by the American Board of Genetic Counseling. She also holds a Certificate in Bioethics and Health Policy from the Loyola School of Medicine in Chicago.

Reem Saadeh, MD, is an Assistant Professor of Pediatrics at Georgetown University. Her interests include dysmorphology, developmental delay, congenital anomalies, and pediatric and adult cancers. She is Board certified

in both pediatrics and clinical genetics. Dr. Saadeh completed her MD at Georgetown University and her pediatric residency at NYU and a Clinical Genetics Fellowship at Johns Hopkins Hospital.

Congresswoman Debbie Wasserman Schultz, is a Member of Congress from South Florida, representing parts of Broward and Miami-Dade Counties. Congresswoman Wasserman Schultz was elected to Congress in 2004, the first Jewish woman to be elected from Florida. Access to quality, affordable health care and the safety and health of children and families are among her top policy priorities. In 2008, her personal and professional lives collided when she was diagnosed with breast cancer at age 41. Shortly thereafter, Congresswoman Wasserman Schultz learned she was a carrier of the BRCA-2 gene mutation. Seven surgeries and six years later the Congresswoman is cancer free, and used her own experience battling the disease to create policy that helps young women access the tools and resources they need to detect breast cancer early. Her Education and Awareness Requires Learning Young Act, or EARLY Act, became law as part of the Affordable Care Act in 2010.

Luc Wathieu, is Professor of Marketing and Deputy Dean at Georgetown University's McDonough School of Business. His research combines economics and psychology to understand consumer empowerment and the adoption of new technologies. His papers have addressed a variety of specific topics including habit formation, brand loyalty, pricing psychology, and privacy preferences. Dr. Wathieu received his B.A. in Economics and M.Sc. in Economic Theory from the University of Namur (Belgium), and received his PhD in Decision Sciences from INSEAD (France).

Louis Weiner, MD, is the Director of the Georgetown-Lombardi Comprehensive Cancer Center and holds the Francis L. and Charlotte G. Gragnani Chair and Professor of Oncology at Georgetown University. His research focuses on new therapeutic approaches that mobilize the patient's immune system to fight cancer using monoclonal antibodies and other modalities of therapy. Dr. Weiner earned his bachelor degree in biology with honors from the University of Pennsylvania and his medical degree from Mount Sinai School of Medicine. He completed his residency training at the University of Vermont's Medical Center Hospital and fellowship in hematology and oncology at Tufts University School of Medicine.

Sheila Cohen Zimmet, BSN, JD, is the Senior Associate Vice President for Regulatory Affairs at Georgetown University Medical Center, an Adjunct Associate Professor in the Department of Pharmacology and Physiology, a Faculty Associate in the Georgetown University Center for Clinical Bioethics, and a member of the National Advisory Child Health and Human Development Council of the National Institutes of Health. She holds a BS in Nursing degree from Georgetown University School of Nursing and a juris doctor degree from Georgetown University Law Center.

DISCUSSION BOARD MODERATORS

A team of geneticists will moderate the discussion boards:

Donna Brown, MD

An Dang Do, MD, PhD

Mauricio De Castro, MD

Maria Guillen-Sacoto, MD

Kristin Gunderson, MS

TEACHING ASSISTANTS

Merrick Tan, B.S. (M.S. expected June 2014), Merrick earned a bachelor's degree in biology from California State University, East Bay in 2009 and will complete his master's degree in Physiology from Georgetown University in

2014. Merrick will also begin medical school in Summer 2014 at The George Washington University School of Medicine.

Darren Jackson, B.S., M.P.H., Darren earned a bachelors of science degree in biology from Morgan State University, Baltimore, MD in 2007. In 2010, he received his Master's of Public Health from the Mailman School of Public Health at Columbia University. Darren will begin the Georgetown Experimental Medical Studies (GEMS) program at Georgetown School of Medicine this summer.

FACULTY SUPPORT TEAM

Barrinton Baynes, B.A, Videographer
Rebecca Berg, B.F.A., Graphic Designer
Dedra Demaree, Ph.D., Learning Designer
Cornelia Gustafson, BSLA, Videography Coordinator
Anna Kruse, M.A., MOOC Consultant
Susan Pennestri, M.A., Project Coordinator
Janet Russell, Ph.D., Team Lead
Alfred Schoeninger, B.A., Videographer
Yianna Vovides, Ph.D., Learning Design Consultant
Ryan Walter, M.A., Videographer

COMMUNICATING WITH THE PROFESSOR:

Please use the following email address for any communication regarding this course:

genomicsmooc@georgetown.edu

Students are requested not to email the lecturers directly. Faculty <u>will not</u> respond to individual emails about this course.

FORMAT

The official start of the course is June 4, and each weekly unit be released at noon Eastern Daylight Time (EDT) on Wednesday. The weekly unit should be completed by the following Tuesday to stay on track.

Students are expected to complete the readings that are part of each class session, to watch the videos, to take the formative assessment problems after each video, to take the weekly quizzes, to take the final exam, and to otherwise engage with the material presented on the class website (such as the polling questions, and the pre- and post-course survey).

Students are also encouraged to form discussion groups and to ask and respond to questions regarding the course material via the discussion board.

COURSE CONTENT OUTLINE

Overview

a. What is the course about?

- b. What does the course include?
- c. What will I learn in the course?
- d. How do I use the course features?

Week 1: Genomic Medicine Gets Personal

- a. Overview of the Course: Navigating the World of Genomic Medicine [Haddad]
- b. Introduction to the Lectures [Haddad]
- c. Introductory Remarks to the Course by Congresswoman Debbie Wasserman Schultz: Putting a Human Face on the Complexity of Genomics [Wasserman Schultz]
- d. The Human Genomics Landscape: Bringing Genomic Medicine into Focus [Green]
- e. Week 1 Quiz

Week 2: Theme 1: The Clinic: Making Precision Medicine a Reality Through Genomics - Part 1: Chromosomal and Mendelian Disorders

- a. Introduction to the Lecture [Haddad]
- b. A More Precise Diagnosis of Chromosomal and Mendelian Disorders [Saadeh]
- c. Week 2 Quiz

Week 3: Theme 1: The Clinic: Making Precision Medicine a Reality Through Genomics - Part 2: Prenatal Care

- a. Introduction to the Lecture [Haddad]
- b. Changing the Way Prenatal Diagnosis and Screening Are Done [Ghidini]
- c. Week 3 Quiz

Week 4: Theme 1: The Clinic: Making Precision Medicine a Reality Through Genomics - Part 3: Cancer Care

- a. Introduction to the Lectures [Haddad]
- b. Changing the Landscape of Cancer Care Through Genomics [Weiner]
- c. Targeted Therapy for Melanoma [Atkins]
- d. Managing Hereditary Breast Cancer [Isaacs]
- e. Week 4 Quiz

Week 5: Theme 2: The Diagnostic Lab: Making Precision Diagnosis With Next Generation Tools (Part 1)

- a. Introduction to the Lectures [Haddad]
- b. Novel Approaches to Cytogenetic Diagnosis [Meck]
- c. Molecular Genetics and Next Generation Sequencing [Haverfield]
- d. Week 5 Quiz

Week 6: Theme 2: The Diagnostic Lab: Making Precision Diagnosis With Next Generation Tools (Part 2) and Theme 3: Minding The Business of Genomics

- a. Introduction to the Lectures (Theme 2) [Haddad]
- b. Pharmacogenetics [Johnson]
- c. Handling Big Data [Mays]
- d. Introduction to the Lecture (Theme 3) [Haddad]
- e. A New Business Model in Medicine [Wathieu]
- f. Week 6 Quiz

Week 7: Theme 4: Genomics and The Patient, The Family and The Society

- a. Introduction to the round table discussion [Haddad]
- b. A Round Table Discussion addressing Counseling, Ethical and Legal Issues In The Genomic Era [FitzGerald, Peshkin, Zimmet, and Haddad]
- c. Week 7 Quiz

Week 8: Theme 5: Looking to the Future of Medicine in the Genomic Era

- a. Introduction to the Lectures [Haddad]
- b. The Time of Our Lives [Morton]
- c. Personalized Medicine: Changing the Landscape of Medicine [Clarke]
- d. Week 8 Quiz
- e. Conclusion & Looking Ahead [Haddad]

Final Exam

WHAT DOES THE COURSE INCLUDE?

Each section of the course listed in the Course Content Outline above begins with an Introduction subsection where professor Haddad provides an orientation as to how the section's topic fits within the broad scope of the course, and he introduces the speaker(s) for the week. After the introduction, the topics are organized sequentially as they are related to each other. These topics include lectures highlighting specific learning outcomes, graded formative assessment questions, ungraded polls, ungraded discussion prompts, and graded quizzes. There are lecture notes provided at the bottom of most of the videos, and a glossary of terms and a list of useful resources available in the course panel. The lecture video sub-pages include learning objectives for each video, lists of readings and/or resources that may be linked to external websites or PDF files.

EXPECTATIONS

PARTICIPATION TOWARD THE CERTIFICATE OF COMPLETION, AND CERTIFICATE OF COMPLETION REQUIREMENTS

At the start of the course you will be emailed two links – one for a pre-course demographic survey and one for a pre-course education survey. **Neither of these surveys will be counted toward your course grade**, but they are very valuable for the instructional team and both of these surveys are short. Your completion of these is expected as part of your course participation. There will also be a brief post-course survey emailed after the completion of the course.

To receive a certificate of mastery, students must complete the formative assessment questions after each video lecture and take the short quizzes at the end of each week of the course. These quizzes consist of multiple-choice, multiple-select, True/False, dropdown, and self-assessment questions. There is also a final exam consisting of multiple-choice questions. The formative assessment questions at the end of each video count for a total of 20 percent of the final course grade. The short quizzes at the end of each week count for a total of 40 percent of the course grade. The final exam accounts for the remaining 40 percent.

A total score of 70 percent or higher overall qualifies as a passing grade for the course.

Formative assessment questions can be attempted three times, while quiz questions and final exam questions can be attempted only once. While you are encouraged, throughout the semester, to discuss the topics of the course with your friends and fellow students, you must do the formative assessment questions, quizzes, and the final exam on your own, without consulting others. We strongly recommend that students take the assessments in sequence, to assess their individual progress.

Please note that the polling questions and discussion board prompts are very useful aspects to your learning and we look forward to seeing your contributions, but these do not count toward your grade or certification for this course.

WHAT YOU CAN EXPECT FROM THE COURSE TEAM

The teaching assistants and some of the faculty support team will be moderating the course discussion forum (on the edX platform), the Facebook group (GUXGenomics), and the Twitter feed (#GUXGenomics). Though these will be monitored for academic integrity, ONLY questions posted on the discussion boards that receive the most upvotes each week will be addressed by the teaching assistants. Up-voting can be done by clicking on the green plus sign within the edX discussion board; the more up-votes a question receives, the higher it will appear in the overall

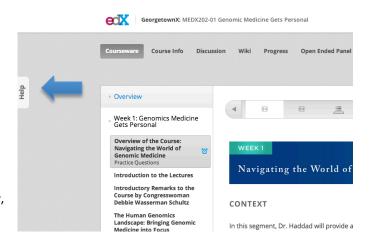
discussion thread. **Make sure you post any questions on the discussion board and not on Facebook.** We will also provide regular updates and reminders in the course info page and through weekly email updates.

WHAT YOU CAN EXPECT FROM EDX

In the event of a technical problem, you should click the "Help" tab located on the left border of the screen within the edX platform. This "Help" tab opens an instruction box that directs you to student Frequently Asked Questions (FAQs) for general edX questions. You can also:

- Report a problem
- Make a suggestion
- Ask a question

You may post technical problems to the "Technical" thread of the discussion board. Finally, you may also contact technical@edx.org directly to report technical problems.



WHAT WE EXPECT FROM YOU

NETIQUETTE GUIDELINES

Please be respectful

To promote the highest degree of education possible, we ask each student to respect the opinions and thoughts of other students and be courteous in the way that you choose to express yourself. Some topics may be controversial and promote debate. Students in this course should be respectful and considerate of all opinions.

In order for us to have meaningful discussions, we must learn to really try to understand what others are saying and be open-minded about others' opinions. If you want to persuade someone to see things differently, it is much more effective to do so in a polite, non-threatening way rather than to do so antagonistically. Everyone has insights to offer based on his/her experiences, and we can all learn from each other. Civility is essential.

Look before you write

Prior to posting a question or comment on the discussion board, the Georgetown course team asks that you look to see if any of your classmates have the same question. Up-vote questions that are similar to your own or that are also of interest to you, instead of starting a new thread. Up-voting can be done by clicking on the green plus sign within the edX discussion board; the more up-votes a question receives, the higher it will appear in the overall discussion thread. This will greatly help our Georgetown teaching assistants to best monitor the discussions and will bring the most popular questions to their attention.

Use the discussion board for course-related posts only

While we encourage students to get to know each other, please use the discussion board for course content conversations only and NOT for personal messages or discussions unrelated to the course.

Properly and promptly notify us of technical issues

While we do not predict technical issues, they can and may happen. To make sure these receive prompt attention, post details about any technical issues directly on the "Technical" discussion thread or email technical@edx.org directly.

ACADEMIC INTEGRITY

Observe edX and GeorgetownX's honor policies

While collaboration and conversation are encouraged and will certainly contribute to your learning during the course, we ask students to refrain from collaborating with or consulting one another on any graded material for the course. Violations of the honor policy undermine the purpose of education and the academic integrity of the course. We expect that all work submitted will be a reflection of one's own original work and thoughts.

GeorgetownX faculty and staff expect all members of the community to strive for excellence in scholarship and character.

APPENDIX A: DETAILED COURSE OUTLINE

WEEK 1: GENOMIC MEDICINE GETS PERSONAL, JUNE 4 TO JUNE 10

Overview of the Course: Navigating the World of Genomic Medicine

- Introduction
- The Human Genome Project
 - o 2a) Students will be able to recall basic facts about the Human Genome Project
 - 2b) Students will be able to state the outcomes that are hoped for from the Human Genome Project
 - 2c) Students will be able to list benefits of being able to sequence the whole genome for the predicted cost of \$1000
- Personalized Medicine
 - 3a) Students will understand the basics of how medicine can become more personalized through the use of genetics and genomics
 - o 3b) Students will be able to list some medical benefits from genomic breakthroughs
 - o 3c) Students will be able to list the ways in which genomics and genetics have impacted medicine
 - o 3d) Students will be able to briefly discuss the role of genetics and genomics in risk prediction
 - 3e) Students will be able to briefly discuss the role of genetics and genomics in making an accurate diagnosis
 - 3f) Students will be able to briefly discuss the role of genetics and genomics in choosing the right treatment
 - 3g) Students will be able to briefly discuss the role of genetics and genomics in determining more accurately disease prognosis
- Implications of Genomic Discoveries Beyond the Clinic and Lab
 - 4a) Students will be able to recognize that genomic medicine has ethical, legal, and social, implications
 - 4b) Students will become aware that direct-to-consumer marketing of genetic tests is a current topic of interest
 - o 4c) Students will be able to state the importance of being educated about genomic medicine
- Genomic Medicine Gets Personal Course Overview

 5a) Students will understand what Dr. Haddad means by the continuum of the progression of our understanding of genomic medicine

Introduction to the Lectures

Introductory Remarks to the Course by Congresswoman Debbie Wasserman Schultz: Putting a Human Face on the Complexity of Genomics

 1a) Students will be able to appreciate the personal experience of the congresswoman and her views about the importance of genomic medicine and of educating the public about genomics

The Human Genomics Landscape: Bringing Genomic Medicine into Focus

1a) Students will hear from the Director of the National Human Genome Research Institute (NHGRI) at NIH about the current state of genomic medicine and how the future of medicine is being shaped by genomic disocveries

WEEK 2: THEME 1: THE CLINIC: MAKING PRECISION MEDICINE A REALITY THROUGH GENOMICS- PART 1: CHROMOSOMAL AND MENDELIAN DISORDERS, JUNE 11 TO JUNE 17

Introduction to the Lecture

A More Precise Diagnosis of Chromosomal and Mendelian Disorders

- Introduction to Genetics and Genomics
 - 1a) Students will be able to distinguish between genetics and genomics
 - 1b) Students will recognize that genomics is important for treatment of both rare and common medical conditions
 - 1c) Students will be able to identify main goals of genetics and genomics
 - 1d) Students will be able to recognize that there is an interplay between our genes and the environment
 - o 1e) Students will be familiar with the role of a geneticist
 - 1f) Students will be able to define mutations in terms of the structure of DNA/genes or of the chromosome
- Clinical Use of Chromosomal Microarray Analysis to Detect Chromosomal Disorders
 - 2a) Students will be able to learn what cytogenetic methods are available to detect chromosomal abnormalities
 - 2b) Students can discuss key differences between the two presented cases
 - 2c) Students can name a few cytogenetic tests (Karyotype, FISH, Chromosome Microarray)
 - 2d) Students can recognize how Chromosome Microarray has advanced the field of clinical cytogenetics
- Clinical Use of Whole Exome Sequencing
 - 3a) Students will be able to learn what molecular methods are available to detect mutations in genes
 - 3b) Students can recognize the role of Whole Exome Sequencing (WES) in the clinical setting
 - o 3c) Students can state how WES is refining the diagnosis of mendelian disorders
 - 3d) Students can identify limitations of WES (inconclusive findings) as a results of not knowing the function of all genes, lack of clinical data, and not understanding some gene interactions
- The Future: Where Genomics is Taking Us
 - 4a) Students will be able to identify future improvements in medical diagnosis and treatment as a result of genomics discoveries
- Questions and Answers with Dr. Saadeh
 - o Students will understand the overall concepts discussed in the lecture

WEEK 3: THEME 1: THE CLINIC: MAKING PRECISION MEDICINE A REALITY THROUGH GENOMICS - PART 2: PRENATAL CARE, JUNE 18 TO JUNE 24

Introduction to the Lecture

Changing the Way Prenatal Diagnosis and Screening Are Done

- Diagnosis Vs. Screening
 - o 1a) Students will be able to distinguish between screening and diagnosis
 - o 1b) Students will be able to define true positive, false positive, true negative, and false negative
 - 1c) Students will be able to define detection rate, false positive rate, positive predictive value, and negative predictive value
 - o 1d) Students will be able to recognize that accuracy is not the same as likelihood
- Impact of Genomics on Prenatal Screening for Chromosome Anomalies
 - 2a) Students will be able to recognize that trisomies account for the majority of all chromosome anomalies
 - 2b) Students will be able to state that, historically, screening relied on family history and maternal age, and more recently serum and ultrasonographic markers
 - 2c) Students will be able to recall that detection rates have increased from about 90% to over
 99% with cell-free fetal DNA analysis
 - 2d) Students will be able to briefly define the cell-free fetal DNA analysis
- Impact of Genomics on Prenatal Screening for Single Gene Disorders
 - 3a) Students will be able to state that historically, screening for single gene disorders was done based on ethnicity and family history
 - 3b) Students will be able to recognize that currently over 100 conditions can be screened for as a gene panel
 - 3d) Students will be able to discuss some limitations of screening for single gene disorders, and reasons why genetic counseling is beneficial
- Impact of Genomics on Prenatal Diagnosis
 - 4a) Students can identify procedures used to obtain fetal cells needed for prenatal genetic diagnosis
 - 4b) Students can recognize karyotype and microarray as two prenatal diagnostic testing techniques
 - 4c) Students can identify advantages and disadvantages of chromosome microarray vs. karyotype analysis
- Future of Prenatal Diagnosis and Screening
 - 5a) Students will be able to summarize Dr. Ghidini's forecast for the future of prenatal screening and diagnosis
- Questions and Answers with Dr. Ghidini
 - o Students will understand the overall concepts discussed in the lecture

WEEK 4: THEME 1: THE CLINIC: MAKING PRECISION MEDICINE A REALITY THROUGH GENOMICS - PART 3: CANCER CARE, JUNE 25 TO JULY 1

Introduction to the Lectures

Changing the Landscape of Cancer Care Through Genomics

- Historical Successes and Challenges of Treating Cancer
 - 1a) Students will recognize that though we are doing better at curing cancer than we were in the 1970's, it is still difficult to treat
 - o 1b) Students will be able to state what treatments are typically used for cancer

- 1c) Students will be able to recognize the limitations of our knowledge about cancer evolution and its molecular drivers
- 1d) Students will recognize that molecularly targeted treatments are rare, and also that the body typically builds resistance to them
- 1e) Students will be able to summarize the state of current medical practice as being similar to historical practice but with additional tools

Making Sense of Genomic Data

- 2a) Students will recognize that 'omics profiles' such as genomics, proteomics, and metabolomics lead to gargantuan data sets
- 2b) Students will realize that there is too much patient information to sift through and that huge amounts of data are hard to interpret and can lead to mistakes in treatments
- 2c) Students will recognize a need for, what Dr. Weiner calls 'systems biomedicine,' to sift through the data to link information to outcomes
- 2d) Students will be able to state what TCGA is, and recognize that although it is an important step, there is still a need to identify the signals for guiding patient management

• Applying Genomic Information to Clinical Practice

- 3a) Students will recognize that not all mutations found in a genetic panel are clinically actionable
- 3b) Students will be able to identify the lack of understanding of when a mutation is 'driving a cancer' versus being 'along for the ride' as a limitation in using genetic information for cancer treatment
- 3c) Students will be able to recognize that while many mutations might contribute to the
 environment needed for cancer to develop, it is necessary to target the mutation that is driving
 the cancer in order to kill a cancer cell and prevent a cancer from being sustained
- 3d) Students will be able to explain that because some cancer prevention methods are costly or have significant side effects, it is important to be able to identify high-risk populations and apply certain procedures to only those at risk

Combating Resistance to Treatment

- o 4a) Students will be able to recognize that metastatic disease is rarely cured by targeted therapy
- 4b) Students will be able to recognize that cancer cells can undergo genetic changes or evolve variants, both of which cause resistance to treatment
- 4c) Students will be able to recognize that there is a need to understand the mechanisms behind cancer resistance in order to develop treatments
- 4d) Students will be able to describe what kinds of questions we would like to be able to answer about the relationships between variants in a cancer population and a person's cancer in order to treat a person more effectively

Administering Individualized Treatment for Colon Cancer

- 5a) Students will recognize that though 80% of early stage colon cancer is cured by surgery, only
 3% more are cured after adjuvant (postoperative) therapies
- 5b) Students will recognize that the adjuvant therapies are risky and therefore 97% of patients are undergoing a risk that has no benefit to them; it is important to understand who should be targeted for additional treatments
- 5c) Students will be able to summarize the finding of the research study reported from Georgetown University that was discussed by Dr. Weiner
- 5d) Students will be able to recognize that we need to be able to understand these predictive factors to target treatment in the future

• Questions and Answers with Dr. Weiner

o 6a) Students will understand the overall concepts discussed in the lecture

Targeted Therapy for Melanoma

- Introduction to Melanoma and Advanced Melanoma
 - o 1a) Students will be able to have a basic understanding of melanoma

- 1b) Students will be able to recall facts about the prevalence of melanoma and deaths from melanoma
- o 1c) Students will be able to relate the stage of a melanoma with the likely prognosis
- o 1d) Students will be able to recall information about current treatments for advanced melanoma
- Recent Treatment Approaches to Melanoma
 - o 2a) Students will be able to recall targets of treatment for metastatic melanoma
 - o 2b) Students will be able to recall drugs that inhibit RAS, KIT, and BRAF genes
 - o 2c) Students will be able to state the benefits of Vemurafenib for melanoma patients
 - 2d) Students will be able to recall the mutations that most likely cause mucosal and acral melanoma
- Applying Genomics in the Treatment of Melanoma: A Case Study
 - o 3a) Students will be able to describe the spread of melanoma in the particular case presented
 - o 3b) Students will be able to state benefits of genomic testing for melanoma patients
 - 3c) Students will be able to describe how clinicians can utilize the current literature to identify treatment plans for unusual melanoma cases
 - o 3d) Students will be able to discuss the impact of treatment in the particular case presented
- Questions and Answers with Dr. Atkins
 - 4a) Students will understand the overall concepts discussed in the lecture

Managing Hereditary Breast Cancer

- Introduction to Breast Cancer and Related Gene Mutations
 - o 1a) Students will be able to recall the percentage of breast cancer cases that are hereditary
 - 1b) Students will be able to briefly describe the difference between a hereditary predisposition and familial predisposition
 - o 1c) Students will be able to recall the two most common genes associated with breast cancer
 - o 1d) Students will have a basic understanding of the impact on a BRCA1/2 mutation carrier
- The Evolution and Impact of Genetic Testing on Breast Cancer: A Case Study
 - o 2a) Students will be able to describe a pedigree of a breast cancer patient
 - 2b) Students will be able to discuss the current impact on screening, prevention, and treatment for patients with BRCA1 and BRCA2 mutations
- Managing Cancer Risk through Screening and Prevention
 - 3a) Students will be able to recognize that BRCA1 and BRCA2 carriers have an increased risk of developing breast cancer in the contralateral breast, if they already have breast cancer in one breast
 - 3b) Students will be able to recognize that a mastectomy does not completely free a women from all possibility for developing a future breast cancer
 - 3c) Students will be able to state possible treatment options for women who are diagnosed with breast cancer and have BRCA1 mutations
 - 3d) Students will understand the relationship between BRCA1 and BRCA2 mutations and the risk for the development of other cancers
- Questions and Answers with Dr. Isaacs
 - o 4a) Students will understand the overall concepts discussed in the lecture

WEEK 5: THEME 2: THE DIAGNOSTIC LAB: MAKING PRECISION DIAGNOSIS WITH NEXT GENERATION TOOLS (PART 1), JULY 2 TO JULY 8

Introduction to the Lectures

Novel Approaches to Cytogenetic Diagnosis

- Introduction to Conventional Chromosome Analysis (Karyotyping)
 - o 1a) Students will be familiar with the basics of karyotype analysis
- Exploring Chromosomes in Depth Using FISH

- 2a) Students will be able to define FISH (Fluorescence in situ hybridization) and state when it is done and why
- Refining Cytogenetics with Chromosomal Microarray
 - o 3a) Students will be able to define Chromosomal Microarray and state when it is done and why
- Implementing Chromosomal Microarray in Prenatal Diagnosis
 - 4a) Students will have a basic understanding of prenatal use of chromosomal microarray
- A "tour" of the Conventional Cytogenetic Lab
 - o 5a) Students will have a basic understanding of how karyotyping is done
- A "tour" of the Molecular Cytogenetic (FISH) Lab
 - o 6a) Students will have a basic understanding of how FISH is done
- A "tour" of the Chromosomal Microarray Lab
 - o 7a) Students will have a basic understanding of how chromosomal microarray analysis is done
- Questions and Answers with Dr. Meck
 - o 8a) Students will understand the overall concepts discussed in the lecture

Molecular Genetics and Next Generation Sequencing

- Introduction
 - 1a) Students will be able to appreciate different techniques used for sequence based diagnostic tests
- Targeted Gene Testing
 - o 2a) Students will be able to define targeted testing for genes
 - 2b) Students will be able to state when targeted testing is recommended and its limitations
- Sanger Sequencing of Single Genes (the "Gold Standard" of Sequencing Technologies)
 - o 3a) Students will be able to describe how single gene sequencing is done
 - o 3b) Students will be able to define polymorphisms and how they relate to disease
 - o 3c) Students will be able to discuss what makes Sanger sequencing the "gold standard" for single gene sequencing
- Gene Panel Sequencing Using Next-Generation Sequencing
 - 4a) Students will be able to discuss the similarities and differences between Sanger sequencing and next generation sequencing
 - o 4b) Students will be able to identify when a physician will order a particular panel of genes
- The Potential of Whole Exome Sequencing
 - 5a) Students will be able to discuss the critical difference between gene panel testing and whole exome sequencing
 - 5b) Students will be able to recall how much of the genome is sequenced when performing whole exome sequencing
 - o 5c) Students will be able to compare whole exome sequencing and Sanger sequencing
- · Questions and Answers with Dr. Haverfield
 - o 6a) Students will understand the overall concepts discussed in the lecture

WEEK 6: THEME 2: THE DIAGNOSTIC LAB: MAKING PRECISION DIAGNOSIS WITH NEXT GENERATION TOOLS (PART 2) AND THEME 3: MINDING THE BUSINESS OF GENOMICS, JULY 9 TO JULY 15

THEME 2: The Diagnostic Lab Introduction to the Lectures Pharmacogenetics

- Introduction to Pharmacogenetics and pharmacogenomics
 - o 1a) Students will be able to define pharmacogenetics and pharmacogenomics
- Basics of Pharmacology

- o 2a) Students will be able to define pharmacokinetics and identify the aspects that impact it
- 2b) Students will be able to state the parameters necessary to consider when formulating drug dosage
- o 2c) Students will be able to explain some complications of drug metabolism
- o 2d) Students will recognize the role the gut plays in the excretion of drugs
- How Does Genetics Impact Pharmacology
 - 3a) Students will have a basic understanding of polymorphism and drug interactions
 - 3b) Students will be able to discuss the impact on patients who take Mercaptopurine but do not have the inactivating enzyme
 - o 3c) Students will be able to state the current ability to test for polymorphisms
 - 3d) Students will be able to describe how genetic testing can be useful in understanding the excretion of drugs
- Genotyping in Pharmacogenetics
 - 4a) Students will be able to state the concerns for the application of sequencing technology in pharmacogenetics and pharmacogenomics
- The Challenges and the Promise of Cancer Pharmacogenomics
 - o 5a) Students will be able to list areas of concern for pharmacology
 - 5b) Students will be able to state the relationship between genetics and choices made when prescribing drugs
 - 5c) Students will be able to describe the confounding factors that need to be considered for genetic testing and how it relates to predicting the outcomes for a patient taking a particular drug
- Questions and Answers with Dr. Johnson
 - o 6a) Students will understand the overall concepts discussed in the lecture

Handling Big Data

- A Discussion with Anne Deslattes Mays
 - 1a) Students will be able to appreciate the unprecedented amount of data that is being generated
 - o 1b) Students will be able to see the potential of having the data available and analyzed
 - 1c) Students will be able to summarize Ms. Deslattes Mays' forecast for how we will take advantage of big data in the future

THEME 3: Minding The Business of Genomics

Introduction to the Lecture

A New Business Model in Medicine

- The Commercialization of Genetic Testing
 - 1a) Students will be able to state what information they might be able to obtain from commercial genetic testing
- Obstacles in Establishing Genetic Testing as Consumer Product
 - o 2a) Students will be able to list obstacles in establishing genetic testing as a consumer product.
- Connecting Consumer Needs with Genetic Testing through Marketing
 - 3a) Students will be able to identify what important human needs genetic testing has the potential to speak to
 - o 3b) Students will be able to define marketing in terms of human pursuits and product features
 - o 3c) Students will be able to define value proposition
- Challenges to Marketing Genetic Testing
 - 4a) Students will be able to define a frame of reference for marketing and state how it relates to genetic testing
 - o 4b) Students will be able to define the concept of biological identity
 - 4c) Students will be able to identify reasons why genetic testing may cause controversy
- Stimulating Market Growth for Genetic Testing
 - o 5a) Students will be able to state what stimulates consumer adoption and market growth

- 5b) Students will be able to explain ways to build relationships between private companies and consumers
- o 5c) Students will be able to state what is needed for market growth to occur
- 5d) Students will be able to discuss the difficulties (for business) associated with the private nature of genetic information
- Integrating Genetic Testing with Clinical Practice
 - o 6a) Students will be able to explain how tension and anxiety can arise from results of a genetic test, in particular, with respect to the relationship with the medical industry
 - o 6b) Students will be able to summarize the three major hurdles to marketing genetic tests
- Questions and Answers with Dr. Wathieu
 - o 7a) Students will understand the overall concepts discussed in the lecture

WEEK 7: THEME 4: GENOMICS AND THE PATIENT, THE FAMILY AND THE SOCIETY, JULY 16 TO JULY 22

Introduction to the Round Table Discussion

A Round Table Discussion addressing Counseling, Ethical and Legal Issues In The Genomic Era

- Introduction
- What is Genetic Counseling and When to Offer it
 - o 2a) Students will be able to define genetic counseling and what it includes
 - 2b) Students will be able to state when genetic counseling is recommended
 - 2c) Students will be able to recall some of the factors that might increase demand for genetic counseling
 - o 2d) Students will be able to recall some challenges faced by genetic counselors
- Regulatory Considerations
 - o 3a) Students will be able to define a professional standard of care and how it is established
 - 3b) Students will be able to recall what litigation in the area of genetic testing has been focused on to date
 - o 3c) Students will be able to explain the relationship between "duty to warn" and "litigation"
 - 3d) Students will be able to explain the importance of having documentation of genetic counseling sessions
- Ethical Considerations when offering "Value Neutral" Counseling
 - 4a) Students will be able to define "value neutral" and "directive" counseling and discuss which is the role of a genetic counselor
 - 4b) Students will be able to state some of the benefits of genetic counseling prior to genetic testing
- Incidental Findings
 - o 5a) Students will be able to define incidental findings
 - 5b) Students will be able to state the role of a genetic counselor with regards to incidental findings
 - o 5c) Students will be able to define gene variants of uncertain significance
 - o 5d) Students will be able to discuss the ethical and legal concerns regarding incidental findings
- Genetic Counseling and Implications for Minors
 - 6a) Students will be able to discuss the legal rights of mature minors with regard to their own genetic information
 - 6b) Students will be able to recall the most critical ethical considerations when doing genetic testing on a fetus
 - 6c) Students will be able to discuss concerns about the ethics of prenatal whole exome sequencing becoming more common in the future
- Duty to Warn

- 7a) Students will be able to demonstrate a basic understanding of the Tarasoff ruling and its relationship to duty to warn and genetic testing
- o 7b) Students will be able to recall basic facts about genetic privacy laws
- 7c) Students will be able to state some general concerns for physicians (liability and ethics)
 regarding duty to warn
- o 7d) Students will be able to recognize the role of the genetic counselor in duty to warn
- Direct to Consumer Marketing of Genetic Tests
 - 8a) Students will be able to state some of the ethical and legal concerns regarding direct to consumer marketing of genetic tests
 - 8b) Students will be able to state the role the Food and Drug Administration (FDA) has in the
 jurisdiction over genetic testing in the USA
 - 8c) Students will be able to recall some of the concerns a genetic counselor might have if a
 patient has genetic testing done without proper counseling
- · Patenting Genes
 - 9a) Students will be able to state under what circumstances DNA can be patented
 - 9b) Students will be able to state some ethical concerns regarding the patenting of genetic information
 - 9c) Students will be able to identify if there is a relationship between patents and costs for genetic tests
- Protection against Discrimination based on Genetic Information: The Genetic Information Nondiscrimination Act (GINA)
 - 10a) Students will be able to define and state the implications of The Genetic Information Nondiscrimination Act (GINA)

WEEK 8: THEME 5: LOOKING TO THE FUTURE OF MEDICINE IN THE GENOMIC ERA, JULY 23 TO JULY 29

Introduction to the Lectures

The Time of our Lives

- Introduction: Opportunities and Challenges in the Genomic Era
 - 1a) Students will be able to appreciate the opportunities and challenges that surround the genomic era
- Improved Medical Therapies: Genetic Studies Leading to a Potential Treatment for Uterine Fibroids
 - 2a) Students will be able to appreciate that uterine fibroids are a major public health problem
 - 2b) Students will be able to learn about Dr. Morton's research aiming at finding genes for uterine fibroids
 - 2c) Students will be able to learn that the Fatty Acid Synthase (FASN) gene is a good candidate gene for fibroids
 - o 2d) Students will be able to learn that a Fatty Acid Synthase inhibitor (Orlistat) may be a potential therapeutic agent to investigate further for the medical treatment of uterine fibroids
- Improved Diagnostics: Deciphering Chromosomal Structural Rearrangements at the Nucleotide Level
 - o 3a) Students will be able to learn about chromosomal aberrations in uterine fibroids
 - 3b) Students will be able to learn about new approaches used in Dr. Morton's lab to refine the chromosomal breakpoints at the nucleotide level and pinpoint the exact genes that are disrupted
 - 3c) Students will be able to appreciate the need for a nomenclature system to refer to the new "refined" chromosomal breakpoints to achieve interoperability among researchers around the world
- Challenges: Data Sharing, Education of Health Care Professionals, Engaging The Public
 - 4a) Students will be able to discuss the challenges and benefits of sharing genomic data globally

- 4b) Students will learn about "Global Alliance", an initiative aimed at catalyzing data sharing projects
- 4c) Students will appreciate the need to reduce the knowledge chasm among healthcare professionals in order to facilitate the implementation of precision medicine into clinical practice
- 4d) Students will appreciate the presence of an opportunity to engage the public by Increasing Science Literacy
- Conclusion: This is the Time of Our Lives
- Questions and Answers with Dr. Morton
 - 6a) Students will be able to hear a recapitulation about the value of educating the medical professionals and the public in genomics

Personalized Medicine: Changing the Landscape of Medicine

- Lecture
 - o 1a) Students will be able to learn about Dr. Clarke's vision about the future of personalized medicine and the challenges that we face, in the new genomic era
- Questions and Answers with Dr. Clarke
- \circ 2a) Students will be able to hear a recapitulation about the main points discussed by Dr. Clarke. Conclusion and Looking Ahead

APPENDIX B: COURSE COMPLETION GUIDE

Date	Topics	Activities	Assignments
Overview (Available May 28 and throughout the course)	Information about the Course	 Complete the pre-course demographic survey Complete the pre-course education survey Read the provided information Watch the video(s) Respond to the Discussion Board prompt(s) 	
Week 1 (June 4 to June 10)	Navigating the World of Genomic Medicine	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 1 Quiz
Week 2 (June 11 to June 17)	Chromosomal and Mendelian Disorders	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 2 Quiz
Week 3 (June 18 to June 24)	Prenatal Care	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings 	Week 3 Quiz

Date	Topics	Activities	Assignments
		4. Complete the poll(s)5. Respond to the Discussion Board prompts	
Week 4 (June 25 to July 1)	Oncology Care	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 4 Quiz
Week 5 (July 2 to July 8)	Diagnostics Tests	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 5 Quiz
Week 6 (July 9 to July 15)	Pharmacogenetics, Big Data, and Marketing of Genetic Tests	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 6 Quiz
Week 7 (July 16 to July 22)	The Patient, The Family and The Society	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 7 Quiz
Week 8 (July 23 to July 29)	The Future of Medicine	 Watch the video lectures Do the formative assessment questions after the end of each video Read the required readings Complete the poll(s) Respond to the Discussion Board prompts 	Week 8 Quiz
Final Exam (July 30 to August 5)			Final Exam