This introductory seminar course and the materials presented here focus on the science underlying personal genome analysis, comparison of tests available from the varied companies that dominate the direct-to-consumer genetic marketplace (e.g., Ancestry DNA, Family Tree DNA, The Genographic Project, and 23andMe), and building the knowledge to navigate the results obtained from the analysis of your own DNA sample. Materials are organized to support this course, aimed at introducing someone with only a basic understanding of biology to The Personal Genome and the discoveries that lie within, but are available to anyone. The site is actively "under construction".

**COURSE OBJECTIVES:** In this course you will:

- Contrast different types of genetic information indicative of ancestral relationships
- Recognize the existence of genetic structure among human populations across the globe
- Build proficiency in the conceptual foundation for the methods that underlie tests of human ancestry
- Navigate the 23andMe web platform to view the various interpretations of genome data
- Investigate the relationship between genotype and phenotypic characteristics with known genetic basis
- Evaluate the personal and the potential societal impacts from commercialization of genetic tests
- Develop skills for contributing to a productive group discussion about science and humanity

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<thead>
<tr>
<th>Date</th>
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| Week 1 | Consequences of DNA Testing | Consider the possible outcomes of DNA testing  
Evaluate the value of the results versus potential unforeseen consequences  
Identify the potential impacts of DNA testing on you and your relatives |
| Week 2 | DNA and the Human Genome | Registering a 23andMe Kit  
Relate the molecular structure of DNA to genome sequence  
Recognize the presence of a shared genome among cells of your body  
Prepare authorization forms and samples for DNA testing |

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**Resources**

- Genetic discovery holds implications for better immunity, longer life  
  Dimitrov, Marija posted on Dec 09, 2019
  Researchers from Washington State University have found a transparent nematode that is found in soil (caenorhabditis elegans). The researchers found that the immune system of the worm, controls the worm cuticles. Humans share the elegans nematode, ours has a very similar structure. Sun used gene silencing and CRISPR to show that a gene called npr-8 and a G-protein regulate collagen. Nematodes that had the npr-8 receptor removed were seen to survive longer when exposed to pneumonia. The nematodes without the receptor remained smooth, unlike the others whose cuticles wrinkled when exposed to the same pathogen. Declining collagen levels are associated with aging and could also be harmful to organs if they are too loose or too stiff. This study indicates that collagen has an important role in defending pathogen infections.

- Genetic discovery holds implications for better immunity and longer life  
  Kwiatkowski, Kathleen posted on Nov 25, 2019
  How do wrinkles on a microscopic worm, a transparent nematode found in the soil, provide a longer and healthier life for humans? Scientists found that nematodes, even though they are very small animals, share very similar genetics with larger mammals, including humans. Because of this, properties that nematodes have may be tested to see if they are the same in humans and in this case possibly provide information for human health. The specific property looked at in this article was a worm’s cuticle and how it compared to a human’s skin. The cuticle, the same as human skin, is the protectant layer and first line of defense against pathogens. When nematodes get infected, their cuticle changes shape to protect it and further defend it from other pathogens. This response is controlled by the nervous system. Many pathogens produce wicked proteins that try to destroy the barrier and cause an infection and according to research, the nervous system can detect these attacks and respond by strengthening the protective structure. The team of researchers found that the gene called npr-8 regulates collagens, which are proteins that are the key structural components of the nematode’s cuticle. They also tested what happened when the npr-8 protein was removed, and the cuticle was smooth and survived longer when exposed to pathogens as opposed to the wild ones that shriveled up. Gene technology shows that the npr-8 receptor negatively regulates defense against pathogenic infections and inhibits the expression of the collagen in the cuticle. Collagen is the most abundant protein found in mammals and declining collagen levels are associated with aging, but for humans, it can create more problems than just wrinkles. Collagen plays an important role in defense of infection and pathogen prevention and researchers suggest that because a nematode’s nervous system controls their cuticle response, neural regulation in humans may provide more of a defense and overall improve longevity.

- German Police Testing DNA of 900 Men for Unsolved Murder Case  
  Eadler, Julia M posted on Dec 10, 2019
  German police testing DNA of 900 men for unsolved murder cold case
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<td>Genealogy to Trees: Inheritance and Tracing Ancestry Near and Far</td>
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<td><strong>Reading:</strong> Genetic Connections Between Organisms from the Tree of Life Project</td>
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<td>Integrate parent-offspring relationships into the broader Tree of Life</td>
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<td>Apply a ‘tree thinking’ framework to individuals, populations and species</td>
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<td>Recognize the utility of shared features, such as SNPs, as indicators of common ancestry</td>
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<td>Mitochondrial Eve &amp; Y-Chromosome Adam</td>
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<td><strong>Reading:</strong> The Recent African Genesis of Humans, Cann &amp; Wilson, Scientific American (ICON link)</td>
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<td>Trace the uni-parental history of the mitochondrial genome and Y chromosome</td>
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<td>Contrast between unique mutations and common SNPs in ancestry analysis</td>
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<td>Recognize the geographic patterns and the prevalence of haplogroups</td>
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<td>Ancestry Composition</td>
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<td>Identify your placement within the Y chromosome and/or mtDNA tree of humanity</td>
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<td>Interpret the different predictions of the ancestry composition</td>
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<td>Recognize the presence of ancient genetic variants revealed by Neanderthal ancestry</td>
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<td>Geographic Variation Among Humans</td>
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<td><strong>Reading:</strong> assigned on ICON</td>
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<td>Evaluate the importance of reference populations in ancestry analysis</td>
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<td>Contrast geographic patterns of shared and private variation in humans</td>
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<td>Relate patterns of genetic variation in modern human populations to global colonization</td>
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<td>Autosomal Inheritance</td>
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<td>Contrast different patterns of inheritance; mtDNA, Y, X, and autosomes</td>
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<td>Consider the role of recombination in shuffling autosomes and X chromosomes</td>
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<td>Calculate degrees of relatedness and expected similarity in autosomal DNA</td>
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<td>8</td>
<td>Genetic Genealogy: It’s All Relative(s)!</td>
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<td>Evaluate the relationships revealed with other users indicated by shared genome segments</td>
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<td>Recognize the value of testing known relatives to partition branches of your family tree</td>
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<td>Predict the percent similarity expected based on degree of family relationship</td>
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German police asked about 900 men to have their DNA tested to help solve the cold case murder of then 11-year-old Claudia Ruf, who was killed 23 years ago. In May 1996, Claudia was kidnapped while walking a neighbor’s dog in the western city of Grevenbroich. 2 days later, her body was found 43 miles south of her home having been strangled, raped, covered in gas, and partially burned.

This is just one of many waves of testing investigators are pursuing to find Claudia’s killer. The men selected in this wave were between 14 and 70 at her time of death. Because DNA traces of her murderer were found on Ruf, investigators and family members of Claudia hope that this mass testing will help find the culprit. According to one source, there has been renewed interest in the case and men were voluntarily lined up by 10 a.m. the first day of testing to help find Claudia’s killer.

The renewed interest comes years after a first test was used on local men. Since then, “new hints...could help them find the murderer” due to advancing technology. The new samples from late November will take 4 to 8 weeks to be analyzed.

The method used by German Police and Investigators in this cold-case could be a helpful way for the average citizen to help in brutal cases such as this one. These volunteers sign over their DNA without knowing if it could potentially connect them to a relative. Personally, I like this tactic. It is a great publicity move to spark increased interest and keep victim’s names a household occurrence for longer. Additionally, with new and improving technology and science, mass tests like these could be started weeks or months after killings instead of years later, potentially saving lives of potential victims.

For example, if the technology was available at the time of Joseph James DeAngelo’s (Golden State Killer) burglaries, perhaps DeAngelo could have been caught sooner. The truth is, we won't know until law enforcement either uses a tactic similar to this one or creates a better one in the ever-changing fields of genetics and forensics.

_A Virus in Koala DNA Shows Evolution in Action_ posted on Dec 10, 2019

Gomez, Camila S
Week 9
Genetic Variation and Phenotypic Diversity
Recognize the influence of the genotype on the appearance of a phenotype
Contrast between traits with simple versus complex genetic causation
Evaluate the association between genetic variants and complex phenotypes

Week 10
Navigating Your Genome
Compare regions of your genome shared with relatives
Use SNPedia to identify variants of interest and explore your genotype

Week 11
Test Results and Health Risk
Interpret the meaning of increased health risks associated with genetic variants
Recognize the impact of genetic variants on the effectiveness of pharmaceuticals

Week 12
FDA Regulation of DTC Genetic Tests
Readings: Green & Farahany Nature 2014 (ICON link), Swann Genetics in Medicine 2010 (ICON link)
Other coverage: The New Yorker, NY Times
Evaluate potential outcomes of learning about disease risks
Recognize the current level of impression in risk assessment from genetic data

Week 13
Downloading and Using Your Genome Data
Download your DNA test results and identify fields of the text file
Identify tools available for further analysis and interpretation of genome data

Week 14
Direct-to-Consumer Genetic Tests; Which Test To Do?
DTC Genetic Testing Companies Compared
Consider the different uses of direct-to-consumer genetic tests
Compare the results and platforms provided by different companies
Identify relatives that can be tested to enhance studies of ancestral relationships

Week 15
The Future of Genetic Testing
Reading: Perfect Genetic Knowledge by Dawn Field
Evaluate the value of personal genetic information relative to its costs
Identify societal impacts of widespread genetic testing

Koalas have suffered throughout the years, from habitat destruction, dog attacks, car accidents. But that's only the explicit stuff, they have been plagued by chlamydia and many cancers, like leukemia and lymphoma. As scientists researched these problems, they found a natural laboratory in which they can study one of the most controversial topics in biology: how viruses can insert themselves into animal's DNA and at times change the course of evolution. The target of this research project is the Koala retrovirus (KoRV), a segment of protein and genetic material in the same family as H.I.V., it began inserting itself into the koala genome about 40,000 ago and is currently passed down generations. It can also be transmitted between species, as a viral infection. Recently scientists have found that the insertion of viruses into animal genomes many times through evolution. It is estimated that 8% of the human genome is made up of viral DNA from previous infections, this occurred millions of years ago, many of them occurring in primate ancestors before the existence of humans. KoRV is peculiar because 40,000 years ago was an important period in evolutionary time, and because the process keeps occurring. A group of scientists reported they observed a genome immune system fighting to render the virus inactive now that has established itself in the Koala DNA. They also reported that KoRV might have activated other ancient viral DNA. This activity stirs the pot of mutation and variation that is essential for natural selection. William Therkauf, a professor in molecular medicine at the University of Massachusetts Medical School, says that "Koala genetics are a gold mine, what they are going through is the process of what's driven the evolution of every animal on the planet". Previous viral infections have led to major evolutionary changes, he said for example: "A gene that is absolutely essential for the placenta was derived from the shell of a virus millions of years ago." humans wouldn't exist without that specific retroviral infection. When the viral RNA infects a cell, it takes over the DNA machinery and replicates itself, which keeps the process going. This process is what causes diseases like AIDS, the most known retroviral disease. However, when the insertion of the retrovirus occurs in a sperm or egg cell, the changes become permanent, kept forever. When retroviruses become part of an animals DNA, they are called endogenous and eventually no longer cause the same type of original infection they once did. But they can still be used for other purposes, like making placenta. "It was long thought they were just junk DNA," said Shawn L. Chavez, a molecular biologist at the Oregon Health and Science University School of Medicine in Portland, who wrote a review of research on endogenous retroviruses in mammals . Now it is clear that some of them have changed the course of evolution. Exactly how is what scientists are trying to find out. "It seems like there's a new publication every day," she said.

Consequently, koalas are drawing a lot of attention from scientists who did not start out with an interest in the animal or its conservation. "I'm a fruit fly guy," Dr. Theurkauf said. He became interested after a reading a report in 2006 by Rachael Tarlinton of the University of Nottingham and other scientists about the invasion of the koala genome by the retrovirus. Dr. Tarlinton began her career in Australia as a veterinarian with an interest in infectious diseases in wildlife. She became involved in the study of koala genetics because of the big issue of chlamydia and because Jon Hanger, an independent researcher, had noticed very high death rates from leukemia and other cancers in koalas kept in zoos. Their research led to the discovery that koala retrovirus was causing some cancers, and that it was not only infecting the animals but also part of their genome. Dr. Tarlinton and her colleagues established the presence of the retrovirus in koalas in Queensland, however there is another, a southern population of koalas that at first seemed not to have the virus. These koalas also had fewer chlamydia infections. The genetics of the population in the south are different because most koalas in that region had been killed for the fur trade by the 1920s. Only a few them survived by being moved to small islands at the beginning of the 20th century. From that population, they have been reintroduced and those koalas have done extraordinarily well, even though they have a few genetic problems. There are over 10,000 of them, in some areas they even have been killed to keep the population down. The researchers expected the southern koalas to be less healthy overall than the northern ones but it was the other way around.
A closer look at the southerners' DNA showed that they weren't free from the inherited retrovirus as it was thought. The virus was there but it was damaged. The beginning and end of its genetic code were present, but the middle was missing. A report on this work is now in bioRxiv, an online database for papers that have been written but not yet accepted by claimed journals. It is thought that the missing segment could be the key to saving the koala population, this defective version can be protective. Dr. Tarlinton are planning an experiment, in which there are cells lines which can grow KoRV, then the defective version can be inserted, ans study the cells to determine if the full virus can successfully infect or inhibit the cells. Dr. Theurkauf and his colleagues found that in koalas there appears to be an initial first-line genome defense involving the piRNA snippets that responds to any virus trying to jump around the genome. Then, a more specific response built to a particular virus comes into play. This is something they want to test further, by looking at koalas in different populations. Dr. Theurkauf and his colleagues also discovered other intriguing clues to what happens as viruses become part of an animal’s DNA. “One thing that we found that is really curious,” Dr. Theurkauf said, is that koalas also have ancient retroviruses that became part of the genome millions of years ago and were presumably deactivated long ago. But at least four of them are just as active as koala retrovirus. They are moving around the genome. “KoRV may be activating these dormant old viruses. It may really stir the pot.” “It’s evolution in real time,” he said.

This is extraordinary to me, its evolution right in front of us, I believe in studying populations of animals and hope to do so in the future. This article was interesting to me because it gathered different scientists from different fields to study this phenomenon. I have been looking more and more into these experiments and I think this is a giant breakthrough in evolutionary and genetic biology. This will help us discover more about how retroviruses work and save the koala population which has previously had many issues perishing.