Teaching the Genome Generation – Wojiski, et al.

Appendix.

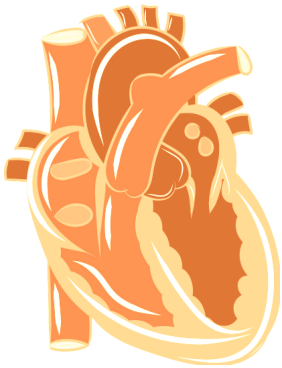
GENE INFO SHEET

ANGIOTENSIN I CONVERTING ENZYME (ACE)

ACE and Winning the Race

Biology Background

- The ACE gene produces the protein angiotensin-converting enzyme (ACE), which functions as a protease that cuts other proteins.
- ACE plays a central role in the system that controls blood pressure by regulating the volume of fluids in the body.
- ACE is located within the cell membrane.
- ACE seems to be made in nearly all tissues of the human body, but appears to be most strongly expressed in capillaries.

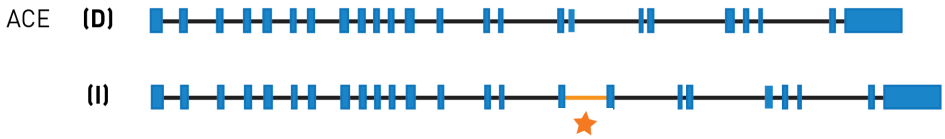


Blood Vessels

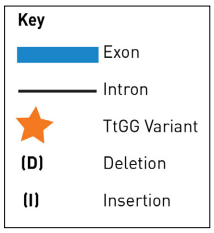
Genomic Locus

The ACE gene is located on chromosome 17 of the human genome. The ACE gene is 21,310 base pairs in length and consists of 25 exons and 24 introns.

The TtGG Variant



- In this assay, you are studying one polymorphism, or variant, in ACE called an INDEL (short for **In**sertion/**Del**etion) that is situated within intron 16 of the ACE gene.
- The common allele is considered the deletion (D), and the variant is an insertion of 287 base pair transposable element (I).
- Since this insertion is in the intron of the gene, it does not directly affect the amino acid sequence of the protein.

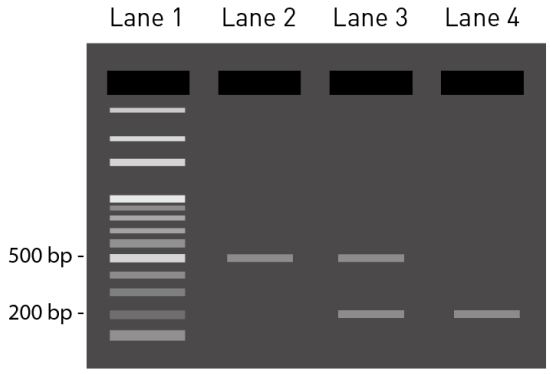


1 · TEACHING THE GENOME GENERATION THE JACKSON LABORATORY

Figure A1. TtGG Gene Sheet. Sample gene sheet for the *ACE* gene, one of five genes available for analysis through the TtGG program. Each gene sheet is structured identically, containing information on the biological relevance, genomic location and gene structure, and nature of the variant being analyzed. Additionally, sample gel electrophoresis images are provided as a reference for students and teachers who perform these assays in their classroom. Information on population genetics and any connections to human health or phenotypes is also provided.

GENE INFO SHEET

ACE Gel



Lane 1: DNA ladder
Lane 2: Homozygous I genotype, 500 bp
Lane 3: Heterozygous I/D genotype, 200 bp, 500 bp
Lane 4: Homozygous D genotype, 200 bp

Population Genetics

- The insertion (I)/deletion (D) polymorphism located within intron 16 of the ACE gene (see star on page 1) has been studied for its contribution to physical endurance.
- This variant seems to reduce enzymatic activity, possibly due to a decrease in protein levels circulating in blood plasma.

Influence on Human Health

- The presence of the insertion allele has been associated with improved endurance performance in studies of mountaineers and soldiers.
- These effects are attributed to increased mechanical efficiency in muscles, possibly due to an increase in type 1 muscle fibers.
- Variants in ACE have also been associated with heart disease, but have not been proven to cause heart disease. Heart disease is a complex disorder that has many different genetic and environmental influences.
- Take this information with a grain of salt, as the presence of either ACE allele in no way causes the aforementioned physiological states.

Sources

» Online Mendelian Inheritance in Man (OMIM) http://www.omim.org/entry/106180#0001
» National Center for Biotechnology Information (NCBI) Gene http://www.ncbi.nlm.nih.gov/gene/1636
» Review on ACE and link to athletic performance: Puthucheary et al. The ACE Gene and Human Performance. Sports Medicine (2011)
» Human Protein Atlas
» UCSC Genome Browser

Figure A1. (cont.)

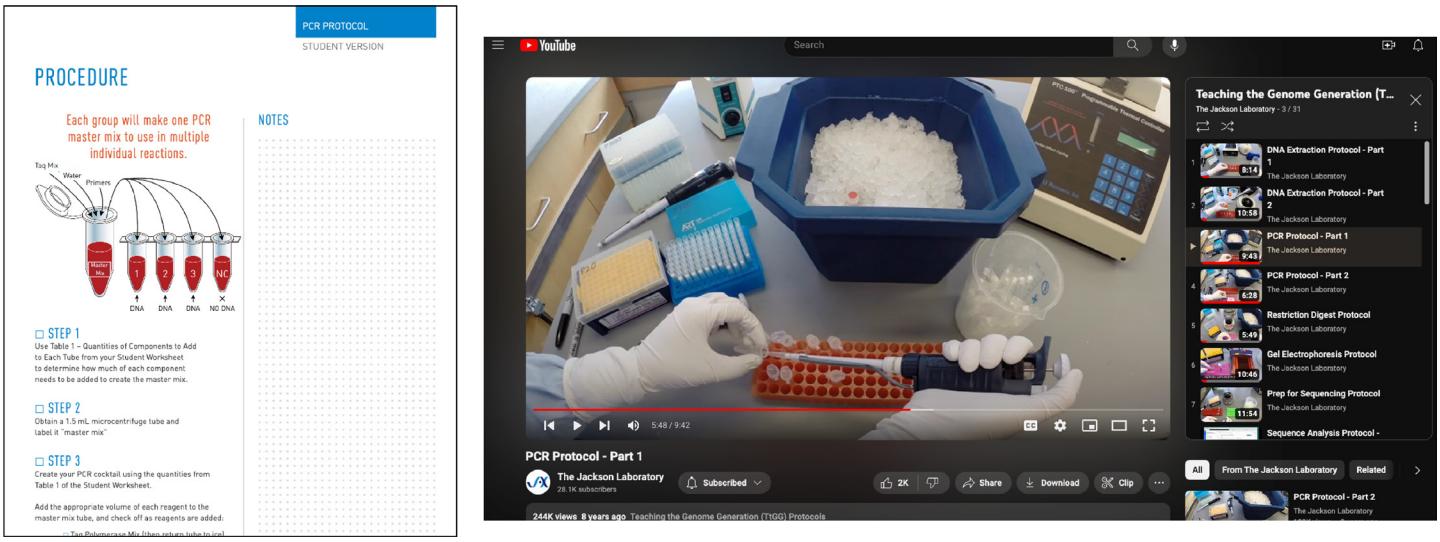


Figure A2. TtGG Protocols and Video Tutorials. Detailed protocols (left) are provided for each laboratory assay. Protocols contain step-by-step instructions, supporting images and diagrams, and a column for students (and teachers) to record notes and comments. Each protocol also has an accompanying video tutorial (right) to demonstrate how to perform each step in the lab.

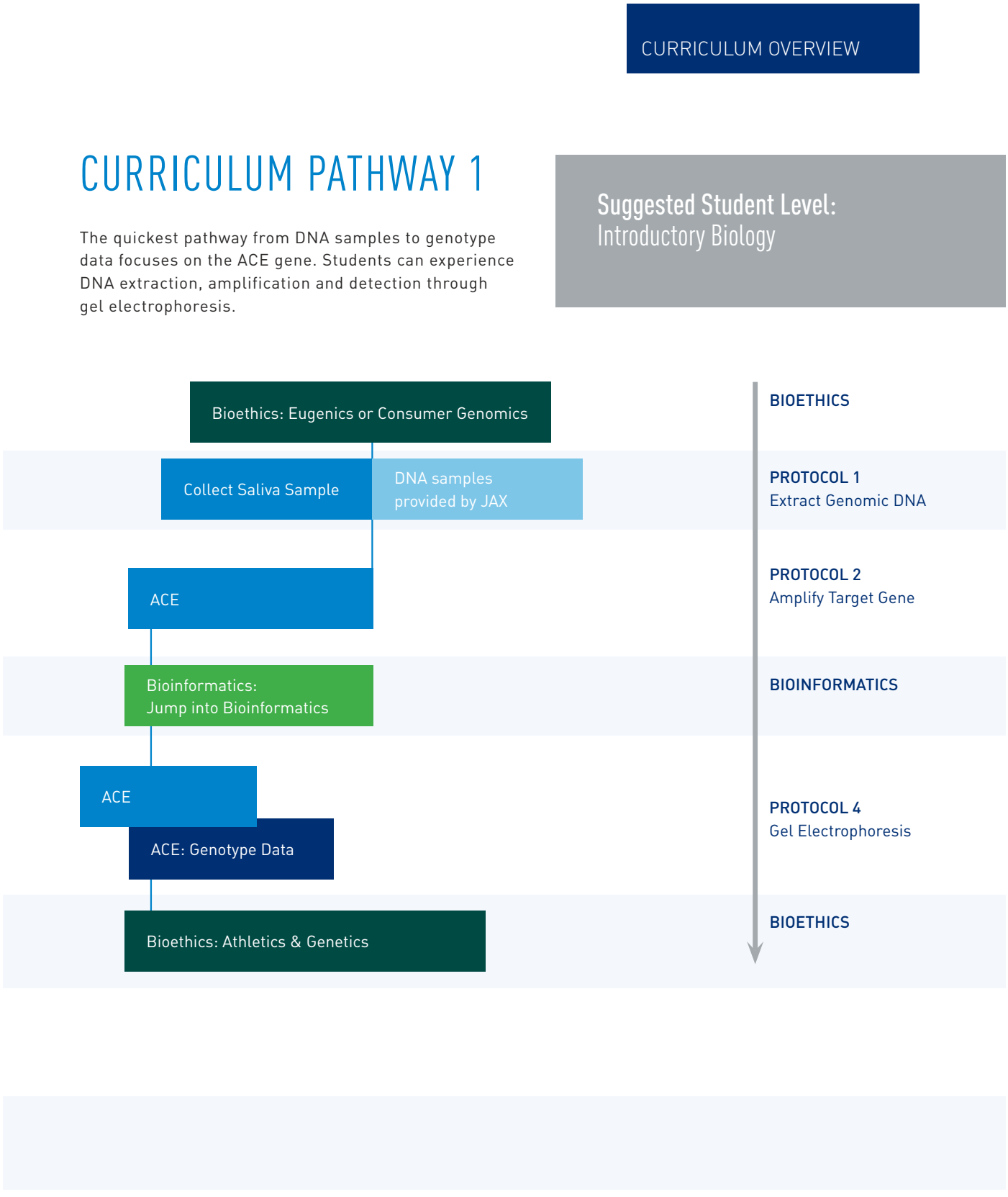


Figure A3. TtGG Curriculum Map. A sample curriculum map is shown that provides suggestions regarding which lessons and lab activities to use together, and in what order. These maps are provided for introductory and advanced courses.

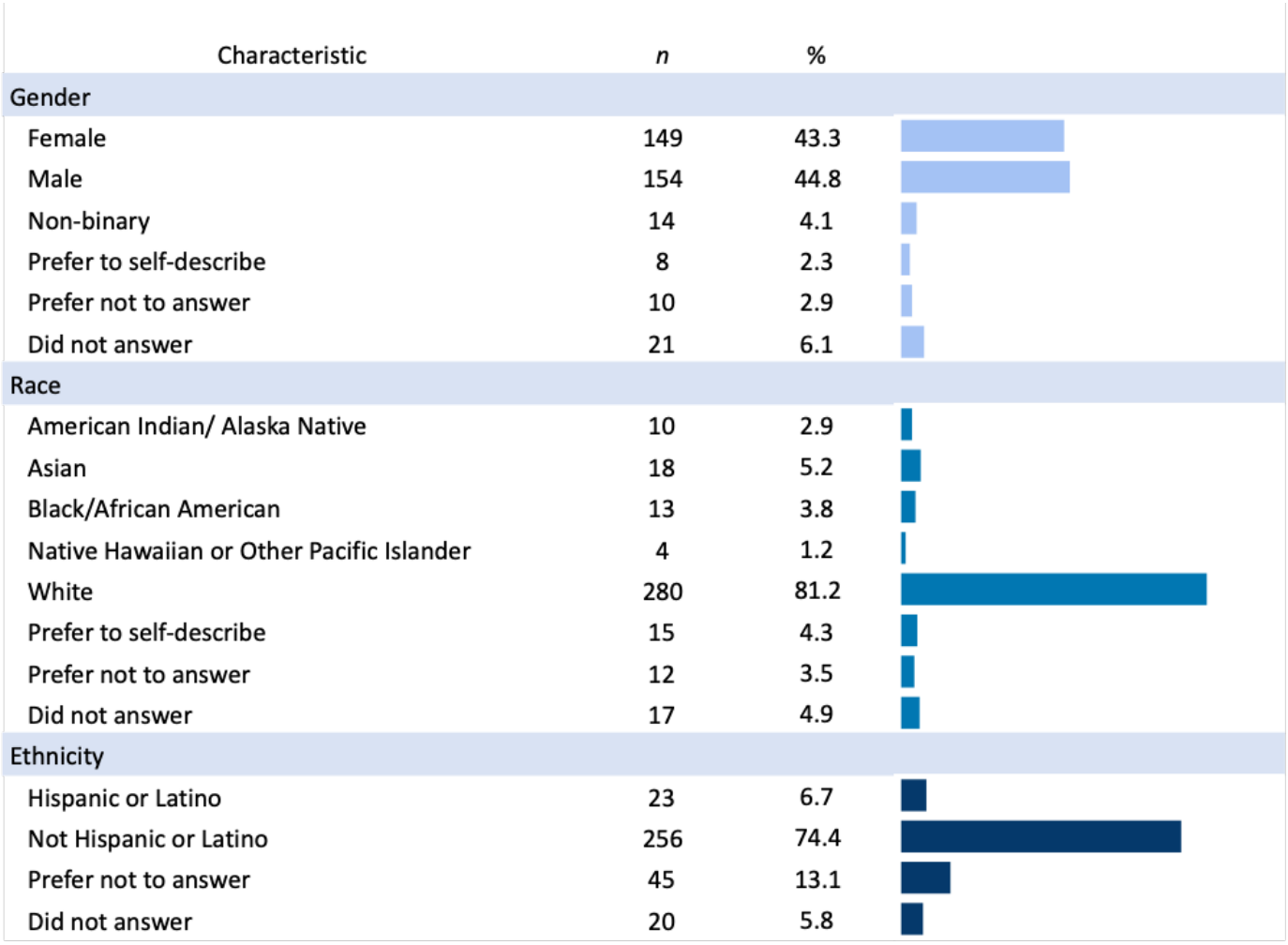


Figure A4. Self-reported gender, race, and ethnicity demographics of participating students (n = 344).

Table A1. *Percentage Correct Means, Standard Deviations, and Paired-Sample* t *Test Results for Lab Procedure Items, Pre and Post TtGG Lessons.*

Survey	Percentage Correct		$t(343)$	p	Effect size (Cohen's d)
	M	SD			
Pre	41.5	30.1	9.06	<.001	0.49
Post	60.7	34.1			

Table A3. *Percentage Correct Means, Standard Deviations, and Paired-Sample* t *Test Results for Molecular Genetics, Bioinformatics, and Bioethics Items, Pre and Post TtGG Lessons.*

Content	Percentage Correct				t	df	p	Effect size (Cohen's d)
	Pre		Post					
	M	SD	M	SD				
Molecular genetics	52.5	21.9	53.1	26.2	0.49	343	.626	0.03
Bioinformatics	40.4	38.4	26.3	28.5	1.64	18	.119	0.38
Bioethics	30.1	25.3	39.0	34.5	2.01	82	.048	0.22

Note. The bioinformatics and bioethics samples are restricted to the teachers who made those subjects a minor or major focus of any lessons. One teacher covered bioinformatics and three taught bioethics.

Table A2. *Means, Standard Deviations, and Independent-Samples* t *Test Results for Lab Procedure Item Gains by Teachers' TtGG Experience.*

Teacher Experience	n	Gains (Percentage Points)		$t(342)$	p	Effect size (Cohen's d)
		M	SD			
Novice (1st or 2nd time teaching TtGG)	90	9.63	40.05	2.71	.004	0.33
Experienced (3rd time or more teaching TtGG)	254	22.57	38.50			

Table A4. *Median Student Interest Ratings for Genetics-Related Activities Before and After the TtGG Lessons, with Wilcoxon Signed-Rank Test Results.*

Activity	*n*	Median (Max = 7)		*z*	*p*[1]	Effect Size (Cohen's *r*)[2]
		Pre	Post			
Taking a course in advanced biology	340	4.00	3.00	-4.16	<.001	-.23
Participating in a summer research internship in genetics	340	2.00	2.00	-0.98	.325	-.05
Watching a YouTube video about genetics	341	4.00	3.00	-3.38	<.001	-.18
Discussing what you've learned about genetics with family or friends	340	4.00	3.00	-2.33	.020	-.13
Paying attention to stories in the news about genetics	338	3.00	3.00	-1.27	.203	-.07
Talking to a health professional (e.g., nurse, doctor) about genetics or genetic testing	341	4.00	3.00	-0.64	.525	-.04
Participating in a voluntary genetic test like 23 and Me or Ancestry when you're 18 or older	340	6.00	5.00	-3.01	.003	-.16

[1]*Bonferroni-corrected alpha threshold for statistical significance = .007 (.05/7).*
[2]*Cohen's* r *values of 0.1, 0.3, and 0.5 are generally considered small, medium and large effects respectively, but interpretations dependent on context (Lipsey et al., 2012; Middlemis Maher et al., 2013).*

Program-related Websites

Link to the Teaching the Genome Generation webpage: www.jax.org/ttgg
Link to curricular resources: www.jax.org/stem-resources