

Genomics and Human Identity

Grades 7-12

Lesson 1



GENOME
UNLOCKING
LIFE'S
CODE

Inspired by the museum exhibit
Genome: Unlocking Life's Code
unlockinglifescode.org

Table of Contents ---

Table of Contents.....	2
About the Genomics and Human Identity lessons	3
Introduction to Lesson 1 – Alike and Different.....	3
Lesson Components.....	4
Time	4
Key Concepts.....	4
Learning Objectives.....	4
Next Generation Science Standards.....	4
Prerequisite Knowledge.....	5
Materials and Handouts.....	5
Preparation	6
Procedure	6
<i>Activity 1 – Alike and Different: Trait Survey</i>	6
<i>Activity 2 – Alike and Different: DNA Exploration</i>	9
Glossary	15
Acknowledgments.....	16

Genomics and Human Identity Lesson 1 – Alike and Different

About the Genomics and Human Identity Lessons

This is one of a series of inquiry-based lessons from the National Human Genome Research Institute inspired by the Smithsonian National Museum of Natural History exhibit Genome: Unlocking Life's Code and the website <http://unlockinglifescode.org>. NHGRI prepared this educational resource to bring the genome research featured in the exhibit into high school classrooms and other venues. These lessons are developed collaboratively with educators, scientists, teachers, and students nationwide.

The lessons address the genetic concepts of inheritance and variation of traits by engaging students in the process and application of genomic research on human genetic variation, identity, and ancestry. Students participate in hands-on inquiry-based learning that emphasizes collecting data, analyzing and comparing data, and drawing conclusions. The lessons are guided by the Next Generation Science Standards and address the concepts of inheritance and variation of traits. Genomics and Human Identity is also designed to raise awareness of the career opportunities in genomics, emphasize the importance of multidisciplinary collaboration in driving discoveries in science, and address common misconceptions in genetics and genomics.

Introduction to Lesson 1 – Alike and Different

Lesson 1 is comprised of two activities where students explore how we are alike and different.

Activity 1 - The Trait Survey is based on a display in the Smithsonian Museum exhibit that examines characteristics/phenotypes we can see. Students consider how we are alike and how we are different when they determine to which one of 16 groups they belong based on the combination of four common traits (dimples, widow's peak, attached earlobes, and tongue roll). A photo collage of museum visitors within these groups shows the collective diversity of others who share these traits. Statistical analysis allows students to compare the prevalence of each group in their classroom with the groups of museum visitors.

In Activity 2 - DNA Exploration, students examine single nucleotide differences between DNA sequences from two individuals to uncover the variation inside our cells at the molecular level.

Lesson Components

- Teacher's Guide - detailed description of lesson objectives, procedures, and materials needed
- Student Handouts - these worksheets are included at the end of each lesson description in the teacher's guide
- PowerPoint Slides - Each lesson has a PowerPoint presentation to guide you and your students through the activities
- Web - <http://unlockinglifescode.org> Teacher background information and supplementary activities for students are available online. However, none of the student lessons in this Teachers Guide requires computers or internet access.

Time

This lesson is expected to take 45 to 60 minutes.

Key Concepts

This lesson explores variation among humans - we share many traits (phenotypes), but have differences, too. This combination of traits is part of our identity. Genomic studies have found that humans are 99.5% identical at the DNA level. The 0.5% of difference in human DNA sequences is part of what makes us unique.

Learning Objectives

After completing this lesson, students will:

- Recognize variation of phenotypes among humans and know that human genomes are 99.5% the same.
- Collect and analyze phenotypic data from class mates and observe and discuss that some combinations of traits are more common than others in a given population.
- Discover how comparing DNA sequence differences in the genome of humans can explain human diversity.
- Know and explain that the 0.5% genetic variation between individuals is the result of sequence differences in their DNA due to mutations - deletions, insertions, single base pair differences, and numbers of repeated short base sequences.

Next Generation Science Standards

This lesson addresses the NGSS life science standards HS-LS3 “Heredity: Inheritance and Variation of Traits” and HS-LS4 “Biological Evolution: Unity and Diversity.” Specifically, the lesson addresses these performance expectations:

Core Idea: HS-LS3 Heredity: Inheritance and Variation of Traits

HS-LS3-1: Ask questions to clarify the relationships about the role of DNA and chromosomes in coding the instructions for characteristic traits passed from parents to offspring

HS-LS3-2: Make and defend a claim based on evidence that inheritable genetic variation may result from: (1) new genetic combinations through meiosis, (2) viable errors occurring during replication, and/or (3) mutations caused by environmental factors

HS-LS3-3: Apply concepts of statistics and probability to explain the variation and distribution of expressed traits in a population.

Core Idea: HS-LS4 Biological Evolution: Unity and Diversity

HS-LS4-2: Construct an explanation based on evidence that the process of evolution primarily results from four factors: (1) potential for a species to increase in number, (2) the heritable genetic variation of individuals in a species due to mutation and sexual reproduction (3) competition for limited resources, and (4) the proliferation of those organisms able to survive and reproduce in the environment.

Science Practices:

Asking Questions and Defining Problems
 Analyzing and Interpreting Data
 Engaging in Argument from Evidence
 Using Mathematics and Computational Thinking
 Constructing Explanations and Designing Solutions
 Obtaining, Evaluating and Communicating Information

Prerequisite Knowledge

Students should know that we inherit our genetic traits from the DNA in the nucleus of the egg and sperm of our parents. They should also be familiar with the structure of DNA and that our chromosomes are made up of long stretches of the four DNA base pairs in different combinations. Mutations are changes in DNA.

Materials and Handouts

Lesson 1 PowerPoint

Handout 1-1: Survey of Four Traits	1 copy per student
Handout 1-2: Trait Mosaics (12 pages)	1 set per class
Handout 1-3: Smithsonian Trait Survey Results	1 copy per student
Handout 1-4: DNA Sequence Comparison	1 copy per two students
Handout 1-5: DNA Sequence Comparison Key	1 copy per group (optional)

Preparation

Print one color (if possible) copy of each of the 16 trait-group photos in Handout 1-2. Write the group number on the back and place them face down evenly around the room.

Procedure

Activity 1 - Alike and Different: Trait Survey

1. Tell students that the goal of this lesson is to explore how we are the same and how we are different.

Ask the class to identify some ways that people are alike and ways that they are different. What defines our identity? What makes us who we are? Which of these characteristics are inherited?

Guide the students to recognizing that part of our identity comes from our environment; our culture defines how we dress and how we communicate and behave. Part of who we are also comes from parents through the DNA that we inherit. These lessons focus on the genetic component of our identity.

2. Ask students to estimate how genetically similar two humans are. Tell students that scientists have found that humans are genetically 99.5% identical.

Ask students if some groups of people are even more genetically identical than 99.5%.

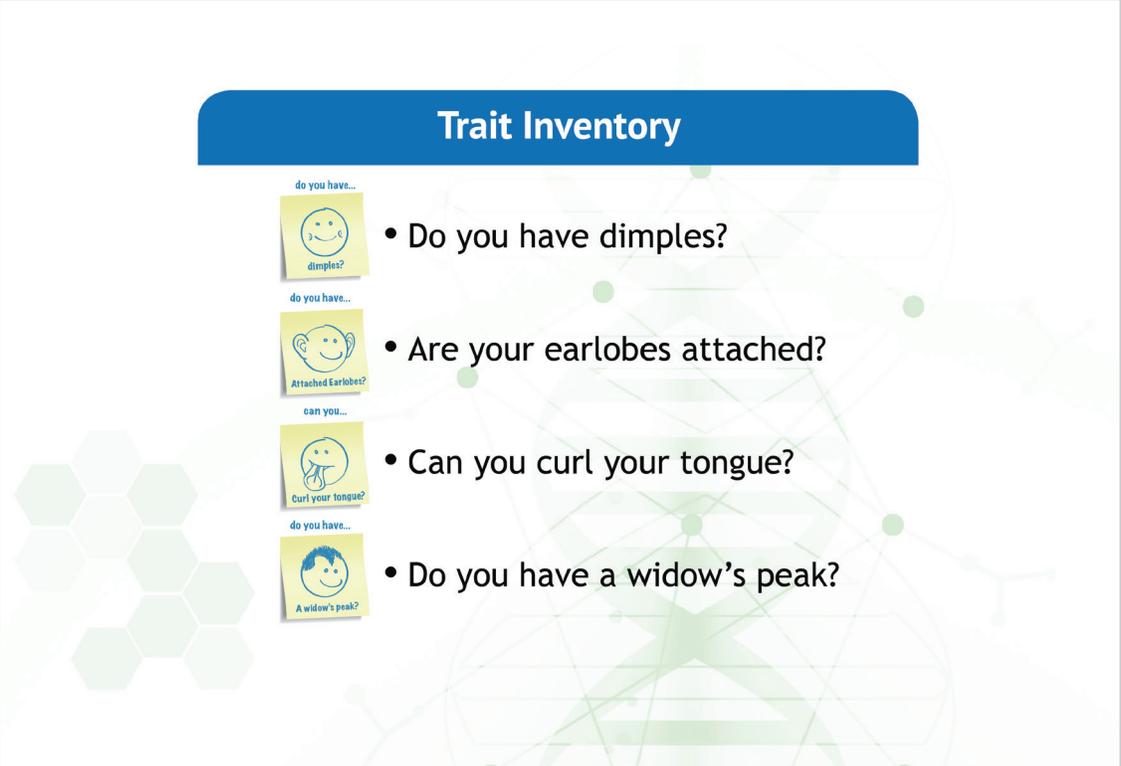
Students should note (or be guided to realize) that family members are more genetically identical to each other than to another random person.

3. Tell students they will now do an activity to see how similar they are to one another through a survey of easily visible traits. The inventory will put students into one of 16 groups based on similarities.

Ask students to look around the room and quickly note the person/people who they think will be in their group. That is, who in the room are they most “identical” to?

Note from Field Test - Students may write down the name of the person who they feel are most identical to them, so they don't forget later in the activity.

4. Show **Slide 1-1: Trait Inventory** and pass one copy of **Handout 1-1 Survey of Four Traits** to each student.



Trait Inventory

- do you have...

dimples? • Do you have dimples?
- do you have...

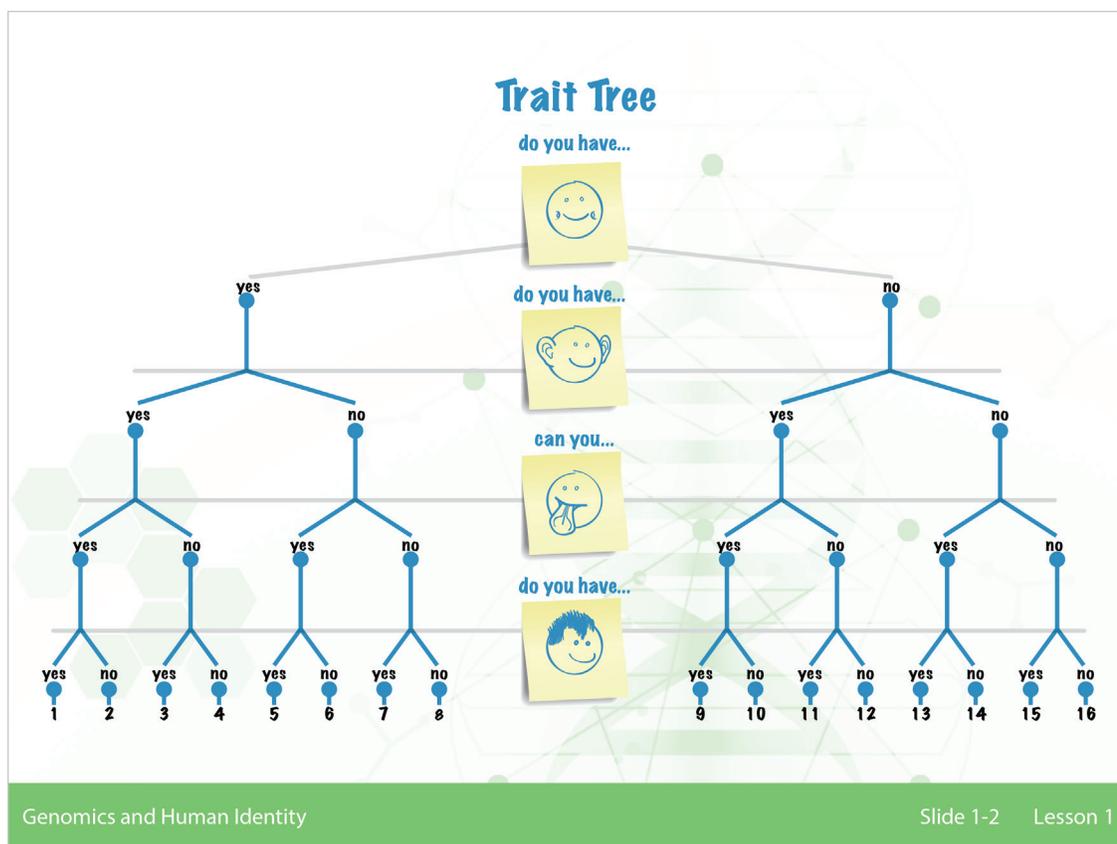
Attached Earlobes? • Are your earlobes attached?
- can you...

Curl your tongue? • Can you curl your tongue?
- do you have...

A widow's peak? • Do you have a widow's peak?

Genomics and Human Identity Slide 1-1 Lesson 1

With Handout 1-1 in front of them, have each student circle yes or no as they follow the question tree down to assign themselves into one of the 16 groups based on their traits. Students may ask one another if they have attached earlobes or a peaked hairline. Each student should circle the group they fall into.



5. Show Slide 1-2: Which Group Are You In?

Survey the class to see how many students are in each of the sixteen categories. Have students raise their hands as you call out the group numbers.

Ask students to look around the room and see if the person they thought was most identical to them is in their same group, What group is that person in?

Have students record the results individually in the table on Handout 1-1 and write the results on the board for all to see.

6. One of each of the 16 **Handout 1-2 Trait Group Photos** should be spread evenly around the room face down with the group number written on the back. When the Genome exhibit opened at the Smithsonian's National Museum of Natural History in 2013, 293 visitors completed the same trait survey and a professional photographer, Rick Giudatti, took their pictures outside the museum.

Point out how the photos are ordered around the room. Have students get up and go to their group photo and turn it over.

Ask students to discuss their reactions to the photos in their groups.

Ask students to make some observations about the photos. What they want to know about people in their group.

Show Slide 1-3: Trait Inventory and use these questions to prompt the class discussion.

Trait Inventory




- What do you have in common with others in your group?
- What differences do you have?
- How are you unique? (we are genetically >99.5% identical)
- Is the person you identified as 'most identical' in your group?



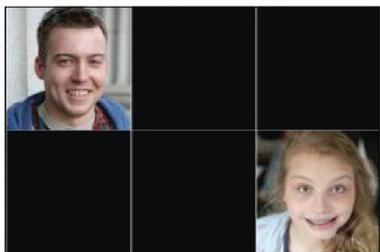

Genomics and Human Identity
Slide 1-3 Lesson 1

7. Provide each student with a copy of **Handout 1-3: Smithsonian Trait Survey Results** and show **Slide 1-4: Smithsonian Survey Results**

Smithsonian Survey Results



Group 1 (6%)



Group 3 (0.3%) Smallest group in survey



Group 14 (15%) Largest group in survey



Group 16 (5%)

Genomics and Human Identity
Slide 1-4 Lesson 1

Have students write their answers to Handout 1-3, Question 1 and then discuss as a group:

Ask students why do some groups have many people, while others have only a few?

The differing numbers of people in each group relate to patterns of inheritance and frequency of traits between different groups. It does not necessarily mean that certain traits are dominant or recessive.

8. Ask students to refer to the results of their classroom survey and write them down on the left-most column on Handout 1-3, next to the results from the Smithsonian survey.

Have students write their answers to Handout 1-3's question 2 and then discuss as a group:

Ask students how do the Smithsonian data compare to the results from your class?

Discussion prompts - You may also want to record these answers on the board for all to see:

- Group 14 was the most common in the Smithsonian visitors - was this also the most common in your class?
- Group 3 was the smallest group from the Smithsonian visitors - was this the smallest for your class, too?
- Why are there differences between the Smithsonian visitors and your class?

Students should realize that the two sets of results are from very different populations. The Smithsonian visitors happened to be at the museum, an international tourist attraction, at the time of the survey and probably do not live near one another. Your students may have more of a family history in the area surrounding your school.

You may wish to point out to students that this activity uses simple mathematical analysis to compare variation between two populations - the visitors to the Smithsonian and the students in your class.

9. This trait activity has students considering how we are alike and how we are different based on things we can see.

Ask why do different people have different combinations of traits? Where does this variation come from?

Students should recognize that there is a genetic component to these traits that lies in the DNA we inherit from our parents. The next activity will look at variation between people at the DNA level.

Note - The four traits just examined have a genetic component. While some of these traits are considered dominant or recessive, the complete story may not be so simple genetically. The Online Mendelian Inheritance in Man database, part of the NIH National Library of Medicine, has more details:

Facial Dimples <http://omim.org/entry/126100>

Hairline peak <http://omim.org/entry/194000>

Earlobes - attached versus unattached <http://omim.org/entry/128900>

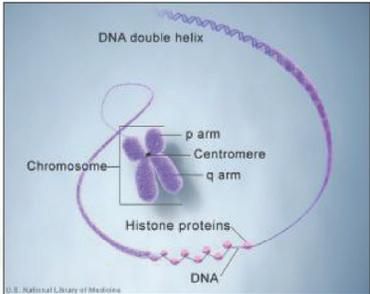
Tongue curling, folding, or rolling <http://omim.org/entry/189300>

Activity 2 - Alike and Different: DNA Exploration

1. Remind students that the previous activity focused on human variation that we can see. There is a genetic component that underlies these phenotypes based on differences in DNA sequences. Activity 2 focuses on the variation inside our cells at the DNA sequence level.

2. Show Slide 1-5: Genetics and Human Identity from the Lesson 1 PowerPoint

Genetics and Human Identity



U.S. National Library of Medicine

- What is genetics?
- How does it help us understand who we are?

Genomics and Human Identity
Slide 1-5 Lesson 1

Ask students what is genetics and how does it help us understand who we are?

The responses to these questions will allow you to gauge what the students already know and identify any misconceptions that they have about genetics and identity. Students will now focus on the DNA variation between two people.

3. Students will work in pairs or groups of three for this activity. Give each group one copy of **Handout 1-4: DNA Sequence Comparison**. Show **Slide 1-6: DNA Sequence Analysis**.

DNA Sequence Analysis

Find the differences in DNA sequence
between persons A and B

```
GGTGTGTGCTTATAGTTCAGCTACGTGGGAGGTTGAGGCGGGAGGATCGCTTGAGC
CTAGGAGGTCAGGGCTGCAGTGAGCTATGATCATGCCACTTGCACTCCAGCCTGGTT
GACAGAGCAAGACCATATCCCACAAAAAAGAAAAAGAAAAAAGAAAAAAGAAAAA
CCCTCTATAACCAATCACATAGGCTATACTGCCTGCCTGCTAACACCTGATAGTTCTG
TCCTGGTCTGAGTATCCATTCCAGGCCAAGCAACTCTCTGCATAGCAAGAGCTCT
GAGTTATTGAGGAGGAAGAGTCAACTGCTGACCCTCGCTAGCTTTTGAACAACAC
```

Genomics and Human Identity Slide 1-6 Lesson 1

Give the teams three minutes to find as many sequence differences as they can between Person A and Person B. Students should highlight the differences on handout 1-4.

Tip from Field Test: Ask students to start at top left of sequence, like they are reading words, and scan across hunting for differences.

4. Show Slide 1-7: DNA Sequence Analysis.

DNA Sequence Analysis

- What variations did you find?
- Where do these differences come from?

```
GGTGTGTGCTTATAGTTCAGCTACGTGGGAGGTTGAGGCGGGAGGATCGCTTGAGC
CTAGGAGGTCAGGGCTGCAGTGAGCTATGATCATGCCACTTGCACTCCAGCCTGGTT
GACAGAGCAAGACCATATCCCACAAAAAAGAAAAAGAAAAGAAAACTTGCTGAGAG
CCCTCTATACCAATCACATAGGCTATACTGCGTGCCTGCTAACACCTGATAGTTCTG
TCCTGGTCTGAGTATCCATTCCCAGGCCAAGCAACTCTCTGCATAGCAAGAGCTCT
GAGTTATTGAGGAGGAAGAGTCAACTGCTGACCCTCGCTAGCTTTTGAAAACAAACAC
```

Genomics and Human Identity Slide 1-7 Lesson 1

Ask students what differences (variations) they noticed between Person A and Person B. Refer to **Handout 1-5** that points out the variations. Distributing this to the students is optional.

The students should note the following differences between the sequences:

- **Single Nucleotide Changes:** there are five single changes between Sequences A and B. Note that genomic studies have found that among the 3 billion base pairs in the human genome there are as many as 10 to 15 million base pair single nucleotide differences between one individual and another. These changes are spread all over our chromosomes. Most have no effect on us since they are in regions that do not encode proteins or regulate other genes.
- **Insertion:** The short sequence TCTG is inserted into B at position 345
- **Deletion:** Sequence B is missing 20 nucleotides that are in A
- **Short Tandem Repeats:** students may also notice that there are short repeated sequences or Short Tandem Repeats. For example, TCTA is repeated 15 times in A and only 8 times in B

5. Ask students the second question on **Slide 1-7**: *Where do these differences come from?*

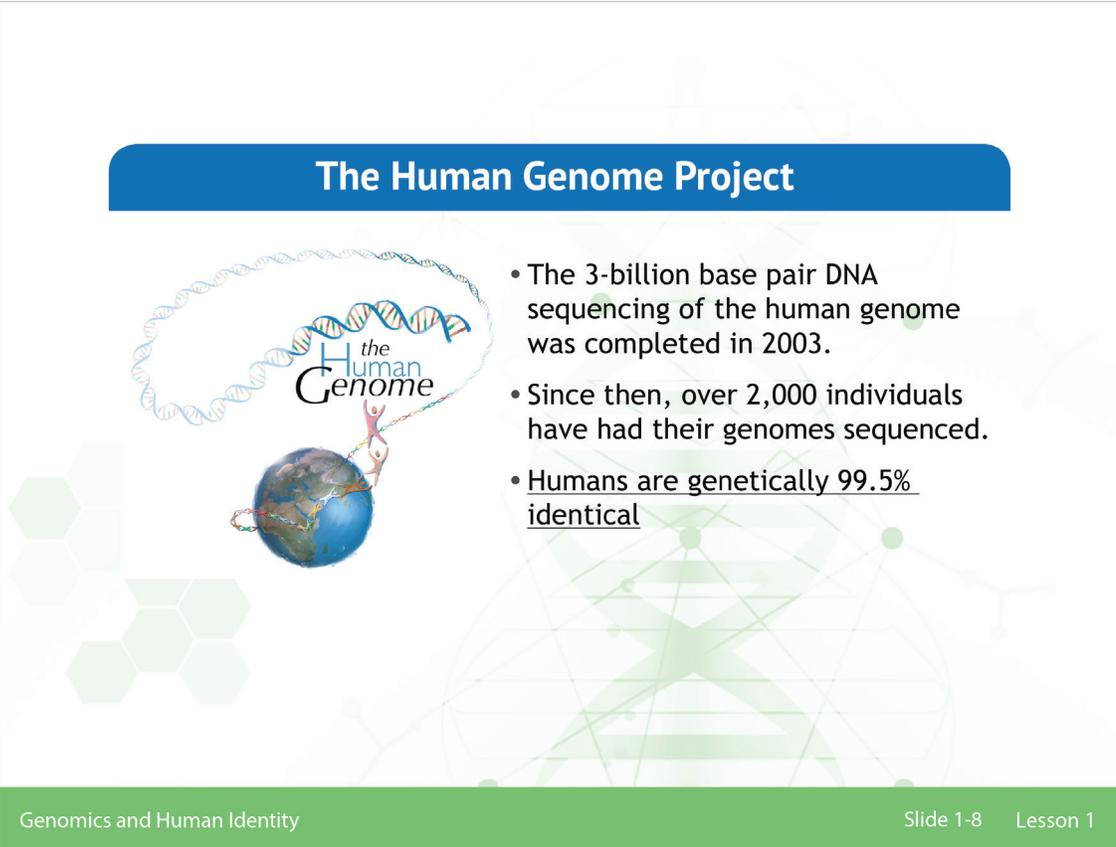
Most variations in DNA are **inherited from our parents**. The sequence shown is a short stretch from one copy of chromosome 1. Person A and B have two copies of chromosome 1, one from the mother and the other one from the father. There will likely be variation between these copies, too.

Our genetic sequence is also affected by **mistakes in DNA replication** and the action of **DNA damaging agents** (mutagens). In some cases, accumulation of deleterious mutations leads to loss of cell function and/or uncontrolled cell growth, i.e., cancer.

Students should end this activity by recognizing that there is variation in the DNA sequences

inside our cells. A person will have less DNA variation differences with people that are closely related and more DNA variation differences with people who are not related. Although we each have a unique DNA sequence, we inherit most of our variation from our families. Our DNA sequences are most identical to near family members, such as our mother and father. We have slightly more variation between extended relatives, such as cousins.

6. Show Slide 1-8: The Human Genome Project



The Human Genome Project

- The 3-billion base pair DNA sequencing of the human genome was completed in 2003.
- Since then, over 2,000 individuals have had their genomes sequenced.
- Humans are genetically 99.5% identical

Genomics and Human Identity Slide 1-8 Lesson 1

Note to students that they just scanned a few hundred nucleotides of DNA for differences. Our genome, however, is made up of 3 billion nucleotide pairs of DNA. The Human Genome Project sought to sequence the 3 billion base pairs across our 23 chromosomes of one genome. This task was started in 1990 and completed in 2003. Since then, over 2,000 individuals have had their genomes sequenced.

Online Resource - Interactive Timeline of the Human Genome
<http://unlockinglifescode.org/human-genome-project>

A comparison of all of these sequences finds that humans are 99.5% genetically identical. If you compare the complete DNA sequences between two random people, there will be approximately 15 million differences pairs - or one difference in every 200 base pairs. In other words, 2.985 billion base pairs will be the same. These variations are all across the chromosomes, usually in areas that do not encode proteins.

5. Show Slide 1-9: Lessons from the Human Genome Sequence

Lessons from the Human Genome Sequence

- 3 billion base pairs of DNA in our genome
- 20,000 to 25,000 genes
- 2% of genome codes for proteins
- 80% looks like it does something else
- 15 million single base pair differences between individuals

Genomics and Human Identity Slide 1-9 Lesson 1

The sequencing of the human genome and subsequent comparisons between individual genomes provides scientists with a general idea of what our DNA does - but there is a lot more that needs to be uncovered.

There is a common misconception that the sequencing of the human genome told us what everything does. You may wish to point out to your students the current understanding of our genome:

- There are 20,000 to 25,000 potential genes. This is far fewer than the 100,000 estimated at the start of the Human Genome Project in 1993.
- Only 2% of the genome looks like it contains instructions for encoding proteins.
- 80% of the genome looks like it has some other function, such as regulating gene expression or affecting how DNA is folded.
- Of the 0.5% variation between individuals, there are 10 to 15 million single nucleotide polymorphisms. These variants are most likely in non-coding regions of the genome. Students will focus on variation in these regions in the next two lessons.

6. End the lesson by telling students that the variation in our DNA is part of what defines who we are. By comparing DNA between people, scientists can also tell how closely we are related. The next lessons in this series explore the connection between DNA and human identity further.

Glossary - For more terms, see the Talking Glossary of Genetic Terms, <http://www.genome.gov/Glossary/>

Base Pair: A base pair is two chemical bases bonded to one another forming a “rung of the DNA ladder.” The DNA molecule consists of two strands that wind around each other like a twisted ladder. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases--adenine (A), cytosine (C), guanine (G), or thymine (T). The two strands are held together by hydrogen bonds between the bases, with adenine forming a base pair with thymine, and cytosine forming a base pair with guanine.

Deletion: Deletion is a type of mutation involving the loss of genetic material. It can be small, involving a single missing DNA base pair, or large, involving a piece of a chromosome.

DNA Sequencing: DNA sequencing is a laboratory technique used to determine the exact sequence of bases (A, C, G, and T) in a DNA molecule. The DNA base sequence carries the information a cell needs to assemble protein and RNA molecules. DNA sequence information is important to scientists investigating the functions of genes. The technology of DNA sequencing was made faster and less expensive as a part of the Human Genome Project.

Genome: The genome is the entire set of genetic instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes, found in the nucleus, as well as a small chromosome found in the cells’ mitochondria. Each set of 23 chromosomes contains approximately 3.1 billion bases of DNA sequence.

Genotype: A genotype is an individual’s collection of genes. The term also can refer to the two alleles inherited for a particular gene. The genotype is expressed when the information encoded in the genes’ DNA is used to make protein and RNA molecules. The expression of the genotype contributes to the individual’s observable traits, called the phenotype.

Insertion: Insertion is a type of mutation involving the addition of genetic material. An insertion mutation can be small, involving a single extra DNA base pair, or large, involving a piece of a chromosome.

Mutation: A mutation is a change in a DNA sequence. Mutations can result from DNA copying mistakes made during cell division, exposure to ionizing radiation, exposure to chemicals called mutagens, or infection by viruses. Germ line mutations occur in the eggs and sperm and can be passed on to offspring, while somatic mutations occur in body cells and are not passed on.

Nucleotide: A nucleotide is the basic building block of nucleic acids. RNA and DNA are polymers made of long chains of nucleotides. A nucleotide consists of a sugar molecule (either ribose in RNA or deoxyribose in DNA) attached to a phosphate group and a nitrogen-containing base. The bases used in DNA are adenine (A), cytosine (C), guanine (G), and thymine (T). In RNA, the base uracil (U) takes the place of thymine.

Phenotype: A phenotype is an individual’s observable traits, such as height, eye color, and blood type. The genetic contribution to the phenotype is called the genotype. Some traits are largely determined by the genotype, while other traits are largely determined by environmental factors.

Short Tandem Repeat: A tandem repeat is a sequence of two or more DNA base pairs that is repeated in such a way that the repeats lie adjacent to each other on the chromosome. Tandem repeats are generally associated with non-coding DNA. In some instances, the number of times the DNA sequence is repeated is variable. Such variable tandem repeats are used in DNA fingerprinting procedures.

Single Nucleotide Change/Polymorphism: Single nucleotide polymorphisms (SNPs) are a type of polymorphism involving variation of a single base pair. Scientists are studying how single nucleotide polymorphisms, or SNPs (pronounced “snips”), in the human genome correlate with disease, drug response, and other phenotypes.

Trait: A trait is a specific characteristic of an organism. Traits can be determined by genes or the environment, or more commonly by interactions between them. The genetic contribution to a trait is called the genotype. The outward expression of the genotype is called the phenotype.

Variation: Genetic variation refers to diversity in gene frequencies. Genetic variation can refer to differences between individuals or to differences between populations. Mutation is the ultimate source of genetic variation, but mechanisms such as sexual reproduction and genetic drift contribute to it as well.

Acknowledgements

NHGRI would like to thank those involved in the development of Genomics and Human Identity Lesson Plan 1.

Promega Corporation

The Hastings Center
Dave Vannier

Medgar Evers College, CUNY
Doris Withers

National Museum of Natural History
Pat Phelps

Positive Exposures
Rick Giudotti

Medgar Evers College Science and Technology Entry Program
John Brown
William Bailey

Science Skills Center High School
Deslyn Dover

Medgar Evers College Preparatory School
Albert Stoddart
Clifford Daniels
Delroy Burnett

The Brooklyn School for Math and Research
Padma Paramananda

Benjamin Banneker Academy for Community Development
Nicole Moore
Edwin Titley

Foundations Academy
Jite Lark
Mansur Ahmed