The Chosen Child – Screening Genetic Content

Secrets of the Sequence Video Series on the Life Sciences • Grades 9 – 12 Teaching materials developed by VCU Life Sciences.

Virginia Commonwealth University

Classroom Tested Lesson

Video Description

"Secrets of the Sequence," Show 127, Episode 1 "The Chosen Child" – approximately 9 minutes viewing time

Our newfound ability to screen the genetic content of human embryos can protect against passing on a number of inherited disorders. The procedure known as pre-implantation genetic diagnosis (or PGD) is offered in a small number of fertility clinics today to parents who can afford it. This is a life-enhancing preemptive therapy. But is it also the beginning of designer babies?

Ward Television

Producer: Dale Minor

Associate Producer: Mara Mlyn

Featuring: Dr. James Guifo, Reproductive Endocrinology, New York University School of Medicine; Francis Fukuyama, International Economy, Johns-Hopkins University; Arthur Kaplan, Center for Bioethics, University of Pennsylvania School of Medicine

Lesson Author; Reviewers: Stephanie Estes; Cathie Alder, Catherine Dahl, Richard Rezba, and Selvi Sriranganathan

Trial Testing Teachers: Lyn Slygh, Mike Comet, Jen Adams

National and State Science Standards of Learning

National Science Education Standards Connection

Content Standard C: Life Science

As a result of their activities in grades 9-12, all students should develop an understanding of:

- the cell and
- the molecular basis of heredity.

Content Standard E: Science and Technology

As a result of their activities in grades 9-12, all students should develop:

- abilities of technological design and
- understandings about science and technology.

Content Standard F: Science in Personal and Social Perspectives

As a result of their activities in grades 9-12, all students should develop an understanding of:

personal and community health, and population growth.

Selected State Science Standards Connection

Use <u>http://www.eduhound.com</u> (click on "Standards by State") or a search engine to access additional state science standards.

Virginia

BIO.6 The student will investigate and understand common mechanisms of inheritance and protein synthesis. Key concepts include:

- use, limitations and misuse of genetic information; and
- exploration of the impact of DNA technologies

Maryland

Expectation 3.3: The student will analyze how traits are inherited and passed on from one generation to another.

- Indicator 3.3.1: The student will demonstrate that the sorting and recombination of genes during sexual reproduction has an effect on variation in offspring.
- Indicator 3.3.2: The student will illustrate and explain how expressed traits are passed from parent to
 offspring.

Expectation 1.7: The student will show that connections exist both within the various fields of science and among science and other disciplines including mathematics, social studies, language arts, fine arts, and technology.

 Indicator 1.7.2: The student will identify and evaluate the impact of scientific ideas and/or advancements in technology on society.

Overview

Genetic research involving human embryos has been a sensitive area of investigation. A process called Pre-implantation Genetic Diagnosis, or PGD, is a procedure available at a few infertility clinics that allows parents to screen future embryos for potential disorders. While scientists still cannot select characteristics such as eye or hair color for a child, they can prevent the birth of a child with a known genetic condition. This episode of Secrets of the Sequence looks at the implications of PGD and the prospect of creating "designer babies."

In this lesson students explore the ethical and moral considerations regarding the application of technology to the creation of human life. PGD is only possible to couples who have already elected *in vitro* fertilization, or IVF, a procedure that may involve the possible destruction of embryos. Some students may have moral, religious, or political objections to some of the information and viewpoints presented in this video about PGD and IVF. Further, students will learn in this lesson how karyotyping is used to identify human genetic disorders, and that this can be used to select against embryos with these genetic changes. These issues have ethical and social implications that you may wish to address with your class. See the general lesson, Introduction to Ethics, available on the Secrets of Sequence Website, for suggestions on how to conduct discussions of ethical issues with high school students.

Note: One trial testing teacher commented that controversial issues may arise unexpectedly, such as questions about the possible euthanasia of children affected with severe genetic conditions. Be prepared to facilitate a reasoned discussion among your students, should such topics arise.

Testing: A sample related multiple choice item from State Standardized Exams

Human Karyotype

ļ	Ĩ	پر	4	1
11	ц,	8	ព្	1 1 10
ង្គ	12	13	11 14	11 15
1	17	Å 3 18	19	[] 20
1.1 21	1	[] ××		

A chart of human chromosome pairs is called a karyotype. What information is revealed in the karyotype above?

- F The sex*
- G The age
- H Trisomy
- J Gene dominance

(Source: Standards of Learning End of Course Test, Biology, Virginia State Department of Education, 2001).

Video Preparation

Preview the video and make note of the locations at which you will later pause the video for discussion.

Before Viewing

- 1. Go to the Web site http://www.cff.org on your classroom computer and open the <u>About Cystic</u> <u>Fibrosis and "65 roses"</u> page, or have your students go to this site on their computers. If this technology is not available in your classroom, print out the relevant material to distribute in class.
- 2. Read the description of the disease to your students (or have your students read from their computers).
- 3. Ask your students the following:
 - How does Cystic Fibrosis affect the lifestyle of a person who has it?
 - What is the likelihood of a child having Cystic Fibrosis if the parents are carriers?
 - How would you feel as a parent if one of your children was born with a disorder that you carried, but that did not affect you?
 - If your first born child had cystic fibrosis, would it change your feelings about having more children? What if you could guarantee that the next child would not have cystic fibrosis – would it make a difference?
- 4. Explain that this episode shows how infertility clinics are able to assist parents in determining whether embryos carry the genes for a genetic disease. One trial test teacher suggested inviting a guest speaker, such as a genetic counselor, for additional genetic and career information.

During Viewing

- 1. **START** the video.
- 2. **PAUSE** the video at 3:13 minutes, after Dr. Levy discusses the method used for testing embryos for genetic disorders.

Ask the following questions:

- Do you think testing embryos for genetic conditions is a good idea?"
- What are some examples of conditions or disorders an embryo might carry?
- Are all conditions equally severe? Are some conditions more severe than others?
- What do you think are some genetic conditions that are very severe? ...less severe? (Responses might include: Cystic Fibrosis, Huntington's disease, Muscular Dystrophy, Down's syndrome, hemophilia, deafness, nearsightedness, etc.)
- What choices are available if the embryo has a genetic condition? Students' responses may lead to a discussion of possibly different choices based on severity of the genetic condition along with the fact that their "ranking" of severity may differ. They may also evolve into an ethical debate – be prepared for this and be able to deflect a full-scale discussion until the end of the video.
- 3. **RESUME** the video.
- 4. PAUSE the video (6:50 minutes into the video) after the segment with Dr. Fukuyama

Ask the following questions:

- Do you think sex selection is an acceptable reason for selecting an embryo?
- Do you think there should be guidelines for making these and other decisions about the selection or termination of embryos?
- What kind of guidelines might distinguish between selections for 'good' reasons versus 'bad' reasons?
- Who decides what the 'good' and 'bad' reasons are?
- 5. **RESUME** the video and play to the end.

After Viewing

- 1. Explain that one way Dr. Levy's lab can tell if an embryo has a genetic disorder is to examine the pairs of chromosomes from one of the embryo's cells through a process called *karyotyping*. Tell your students that today they are going to observe chromosomes and pair them up, just as a geneticist would, to determine if an embryo has the expected number of chromosomes.
- 2. Conduct Student Activity: Karyotyping Reading Chromosomes.

Teacher Notes for the Student Activity: Karyotyping – Reading Chromosomes

Materials

- Copies of the student handout, *Karyotyping Reading Chromosomes*, Version A or B Student Handout A (if your classroom has Internet access) Student Handout B (if your classroom does not have internet access)
- Computers with Internet access (If computers are unavailable, you can print copies of Appendix B, which includes 4 sections:

What Can Our Chromosomes Tell Us? What are chromosomes,? How do scientists read chromosomes? Making a Karyotype.

Procedure

- Have students access the Genetic Science Learning Center Web site (at the University of Utah) http://gslc.genetics.utah.edu/units/disorders/karyotype/
- As a class read the introductory page at this site. If computers are not available, refer to Appendix B
- Distribute copies of the appropriate Student Handout. If your classroom has enough computers for all students, use Student Handout A. If your classroom does not have computers for all students, use Student Handout B.
- Students will read and answer the following questions: What are chromosomes? How do scientists read chromosomes?

Optional Extensions Activity:

- 1. The next section on the Web site ("Using karyotypes to predict genetic disorders") provides a description of how disorders result from nondisjunction and is a good extension for students who have understood the previous material. For classrooms without computers, this activity is not included on the Student Handout, as it is best viewed online. You should decide if this extension is optional or not.
- Have students read and comment on the following article: Newsweek, January 26, 2004. "Girl or Boy? Now You can Choose. But Should You?"
- 3. Students may also research a more current article on their own.

Student Handout A: Karyotyping – Reading Chromosomes

Geneticists are able to tell much about the genetic make-up of developing embryos using a technique called **karyotyping**. During this process, a cell is removed from an embryo during a particular phase of mitosis. The cell is treated with chemicals so that the chromosomes can be seen, then the chromosomes are observed and put into pairs. In this activity, you will be able to determine how karyotyping can show genetic disorders.

As a result of this activity, you will:

- Learn three ways scientists observe chromosomes to determine defects.
- Create a karyotype using unidentified chromosomes.
- Identify several disorders resulting from nondisjunction of chromosomes.
- Use karyotypes to determine genetic disorders.

Materials

Computer with Internet access

Procedure

 Open the browser on your computer and type the following URL in the address bar: http://gslc.genetics.utah.edu/units/disorders/karyotype/. You should be on the "What Can Our Chromosomes Tell Us" page at the Genetic Science Learning Center at the Eccles Institute of Human Genetics, University of Utah.

Read the introduction on this page, and then select the first activity, "What Are Chromosomes?" As you read the information on this page, answer the following questions:

- a) What is Giemsa?
- b) Why are some parts of chromosomes darker than others?_____
- c) During what part of the cell cycle can the chromosomes be seen clearly?______
- 2. Continue on to the next page, "How do scientists read chromosomes?" Then answer the following questions:
 - a) What are the three features that scientists use to read chromosomes?
 - b) Do the activity in the yellow box on the right: Click on the chromosome in Box B that matches the chromosome in Box A.
- 3. Continue on to the next page, "Making a Karyotype." Click on the gold bar "Organize the chromosomes into a karyotype!" to play the animation that pairs the chromosomes in a karyotype. Now try pairing the chromosomes yourself by clicking on the words, "Matching up chromosomes in a karyotype" in the yellow box.

Continue on to the final page, "Using karyotypes to predict genetic disorders." Play the animations on this page for an explanation of nondisjunction and how it may result in several forms of genetic disorders. To observe karyotypes for these disorders, click on the names of each of these disorders. Fill in the chart on Genetic Conditions prepared in Appendix A. Then complete the yellow box activity "Test your knowledge."

APPENDIX A (For use with Student Handout A only)

	How is it caused?	# chromosomes	Phenotypic effects
Genetic Conditions			V L
Down Syndrome			
Turner Syndrome			
Klinefelter			
Syndrome			
Cri du Chat			
Williams Syndrome			
Philadelphia			
Chromosome			
Robertsonian Translocation			
Transiocation			

Student Handout B: Karyotyping – Reading Chromosomes For use in classrooms without Internet access

Geneticists are able to tell much about the genetic make-up of developing embryos using a technique called **karyotyping**. During this process, a cell is removed from an embryo during a particular phase of mitosis. The cell is treated with chemicals so that the chromosomes can be seen, then the chromosomes are observed and put into pairs. In this activity, you will be able to determine how karyotyping can show genetic disorders.

As a result of this activity, you will:

- Learn three ways scientists observe chromosomes to determine defects.
- Create a karyotype using unidentified chromosomes.
- Identify several disorders resulting from nondisjunction of chromosomes.
- Use karyotypes to determine genetic disorders.

Materials

 Handout Appendix B: What Can Our Chromosomes Tell Us? What are chromosomes? How do scientists read chromosomes? Making a Karyotype

Procedure

1. Read the introduction "What Can Our Chromosomes Tell Us?" Continue to the second section "What Are Chromosomes?"

As you read the information on this section, answer the following questions::

- a) What is Giemsa?
- b) Why are some parts of chromosomes darker than others?_____
- c) During what part of the cell cycle can the chromosomes be seen clearly?
- 2. Continue on to the next section, "How do scientists read chromosomes?" Then answer the following question:
 - What are the three features that scientists use to read chromosomes?
- Continue on to the next section, "Making a Karyotype." Cut out the individual chromosomes on the bottom of the page and paste them next to their partners on the top. *Be sure of the match before you paste –* karyotyping is often not as simple as it seems.

APPENDIX B

INTRODUCTION:

What can our chromosomes tell us?

We can learn a lot by looking at chromosomes. They can tell us everything from the gender of a person to the likelihood that an unborn baby will have a genetic disorder. Scientists often analyze chromosomes in prenatal testing and in diagnosing specific diseases.

Examples of genetic disorders caused by chromosomal abnormalities:

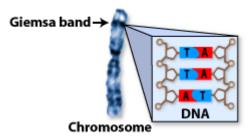
- Down Syndrome
- Turner Syndrome
- Klinefelter Syndrome
- Cri du chat Syndrome
- Willams Syndrome
- Reciprocal Translocation: Philadelphia Chromosome
- Robertsonian Translocation

SECTION ONE:

What are chromosomes and why do we need them?

Chromosomes are compact spools of DNA. If you were to stretch out all the DNA from just one of your cells, it would be over 3 feet (1 meter) long from end to end! You can think of chromosomes as "DNA packages" that enable all this DNA to fit in the nucleus of each cell. Normally, we have 46 of these packages in each cell; we received 23 from our mother and 23 from our father.

Why do chromosomes look like this?



Chromosomes are very small but can be specially prepared so we can see them using a microscope. Chromosomes are best seen during mitosis (cell division), when they are condensed into the fuzzy shapes you see here. Chromosomes taken from dividing cells are attached to a slide and stained with a dye called Giemsa (pronounced JEEM-suh). This dye gives chromosomes a striped appearance because it stains the regions of DNA that are rich in adenine (A) and thymine (T) base pairs. Why do scientists look at chromosomes?

Scientists can diagnose or predict genetic disorders by looking at chromosomes. This kind of analysis is used in prenatal testing and in diagnosing certain disorders, such as Down syndrome, or in diagnosing a specific type of leukemia. Such diagnosis can help patients with genetic disorders receive any medical treatment they need more quickly.

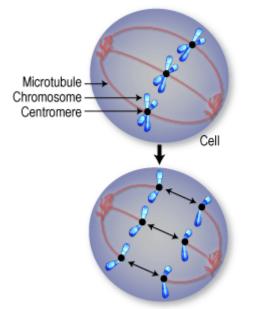
SECTION TWO:

How Do Scientists Read Chromosomes?

To "read" a set of human chromosomes, scientists first use three key features to identify their similarities and differences:

- 1. Size. This is the easiest way to tell two different chromosomes apart.
- 2. **Banding pattern**. The size and location of Giemsa bands on chromosomes make each chromosome pair unique.
- Centromere position. Centromeres are regions in chromosomes that appear as a constriction. They have a special role in the separation of chromosomes into daughter cells during mitosis cell division (mitosis and meiosis).

Using these key features, scientists match up the 23 pairs -- one set from the mother and one set from the father.



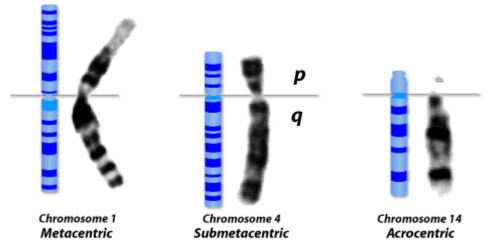
What are centromeres for?

During cell division, centromeres attach to microtubules which pull the chromosomes to opposite ends of the cell.

Centromeres are required for chromosome separation during cell division. The centromeres are attached to microtubules, which are proteins that can pull chromosomes toward opposite ends of each cell (the cell poles) before the cell divides. This ensures that each daughter cell will have a full set of chromosomes.

Normally, each chromosome has only one centromere.

The position of the centromere relative to the end of the chromosome helps scientists tell chromosomes apart. Centromere position can be described three ways: metacentric, submetracentric or acrocentric.



In metacentric (pronounced met-uh-CEN-trick) chromosomes, the centromere lies near the center of the chromosome.

Submetacentric (pronounced SUB-met-uh-CEN-trick) chromosomes have a centromere that is off-center, so that one chromosome arm is longer than the other. When chromosomes are aligned, they are oriented so that the short arm, designated "p" (for petite), is at the top, and the long arm, designated "q" (simply for what follows the letter "p"), is at the bottom.

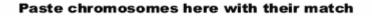
In acrocentric (pronounced ACK-ro-CEN-trick) chromosomes, the centromere resides very near one end.

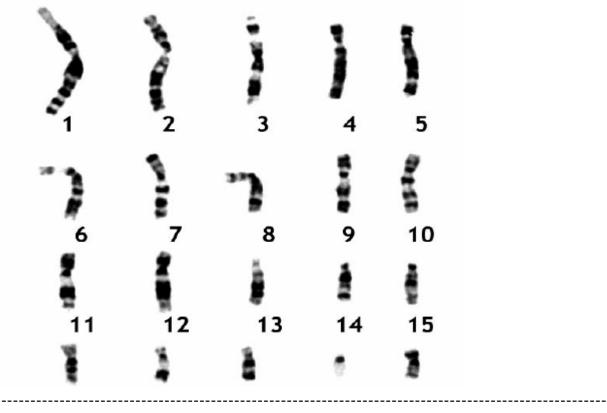
SECTION THREE:

Making a Karyotype

A karyotype is an organized profile of a person's chromosomes. In a karyotype, chromosomes are arranged and numbered by size, from largest to smallest. This arrangement helps scientists quickly identify chromosomal alterations that may result in a genetic disorder.

To make a karyotype, scientists take a picture of someone's chromosomes, cut them out and match them up using size, banding pattern, and centromere position as guides.





Cut out chromosomes here



Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation The Chosen Child – Screening Genetic Content • Page 12 of 13

Additional Resources

Because Web sites frequently change, some of these resources may no longer be available. Use a search engine and related key words to generate new Web sites.

http://www.cff.org

The official Web site of the Cystic Fibrosis Foundation. This informative site answers many questions about this inherited disorder.

http://http://gslc.genetics.utah.edu/

The Genetics Science Learning Center Web site; many links to other research being done in the field of genetics.

Genomic Revolution

http://www.ornl.gov/sci/techresources/Human_Genome/education/education.shtml The Web site to the government-funded Human Genome Project with links about genomics, the history of the project, and more.

Secrets of the Sequence Videos and Lessons

This video and 49 others with their accompanying lessons are available *at no charge* from <u>www.vcu.edu/lifesci/sosq</u>