Chromosomal Abnormality Investigation

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Primary Learning Outcomes
Students will research a variety of chromosomal abnormalities and resulting syndromes to determine risk factors and contributors of the abnormality, and characteristics of the syndromes.

Additional Learning Outcomes
Students will increase their knowledge of sources/information available on the Internet, especially the resources of the National Center on Biotechnology Information, advance their PowerPoint presentation skills, and learn about genetic karyotyping and genetic counseling.

Materials and Equipment
1. Computers with Internet access
2. LCD projector or CDL scan converter or smart board
3. Computer disk or CD
4. Microsoft Word or other word processing program
5. PowerPoint or other presentation software

Technology Connection
Students will conduct research on the Internet and will then develop a PowerPoint presentation to convey their findings to their classmates.

Total Duration
4 hours and 25 minutes

Procedures

Step 1 **Duration: Varies**
Before getting started, students should have experience with the Internet and familiarity with how to create a PowerPoint presentation. A tutorial is attached below for students, who have not used PowerPoint before. In addition, teachers should prepare for this lesson by reading the background articles on birth defects provided in Step 2.

Web Resources
Title: PowerPoint Tutorial
URL: http://www.bcschools.net/staff/PowerPointHelp.htm
Description: This tutorial created by the Bay City Public School Technology Team is a helpful resource for students that are not familiar with how to construct a PowerPoint presentation.

Title: Florida Gulf Coast University's PowerPoint Tutorial
URL: http://www.fgcu.edu/support/office2000/ppt/
Description: This is a PowerPoint tutorial from the Florida Gulf Coast University.

Step 2 **Duration: 30 minutes**

To begin this lesson, have students look through the information provided on the two Web resources provided. Then have the students write down three interesting things that they have learned from these Web sites and share their thoughts with another student. Using the "Chromosomal Abnormality Discussion Questions", the teacher should guide the class in a discuss what they have learned. Encourage the students to share their ideas and help them arrive at the correct answers. After the discussion, have the students complete the "Chromosomal Abnormality Quiz." This will help them solidify the concepts they have just learned. The students will be graded on this quiz using the indicated point values using the "Chromosomal Abnormality Quiz Answer Key."

Web Resources

Title: NCBDDD's What is a birth defect?
URL: http://www.cdc.gov/ncbddd/bd/faq1.htm#Whatisabirthdefect
Description: National Center on Birth Defects and Developmental Disabilities provides general information on birth defects on this site.

Step 3 **Duration: 30 minutes**

In Step 4, students will be conducting research using the Internet, specifically resources from the National Center on Biotechnology Information called PubMed. To familiarize students with this Web site, have students go through the PubMed tutorial provided in the Web resources. In addition, draw their attention to Entrez and have them call up this Web resource. Here the students can find nucleotide, protein, and genome sequence information by entering a medical topic and clicking on the respective database for a list of the results. If they are not familiar with these resources, allow the students to complete the tutorial and explore both the Entrez and PubMed sites before moving on to Step 4.

Web Resources

Title: PubMed Home Page
URL: http://www.ncbi.nlm.nih.gov/entrez/query.fcgi
Description: PubMed allows the students to search for relevant journal articles and information about a specified health topic.

Title: PubMed tutorial
URL: http://www.nlm.nih.gov/bsd/pubmed_tutorial/m2001.html
Description: This tutorial for the PubMed Web site will familiarize students with how to effectively search for their topic of interest on PubMed.

Title: Entrez Cross-Database Search Page
URL: http://www.ncbi.nlm.nih.gov/gquery/gquery.fcgi?itool=toolbar
Description: Entrez allows the students to conduct a search for a specific topic in many different databases at once. Here students can access sequence information for their PowerPoint presentations in Step 4.

Step 4 **Duration: 3 hours**

Break the students in groups of three and give students a chromosomal abnormality from the "Possible Chromosomal Abnormalities Search Topics" list. Web resources for researching topics are listed in this document as well as in this step's Web resources. The first six web resources are general and the remaining sites are for specific topics. Students can propose to research a chromosomal abnormality not on the list; however, for approval there must be adequate information about the chromosomal abnormality on several of the general Web resources provided.

After the different chromosomal abnormalities have been decided, pass out the attached list of PowerPoint requirements and topics to be covered. The students should use the nine topics to be covered as a basis for their research. Then the students should construct a PowerPoint presentation, including notes, based on their research and the requirements. A sample of a presentation for Tay-Sachs is provided below as a reference. When the groups have finished constructing their presentations, they should practice their delivery in preparation for Step 5.

Web Resources

Title: National Center on Birth Defects and Developmental Disabilities
URL: http://www.cdc.gov/ncbddd
Description: The Centers for Disease Control and Prevention's National Center on Birth Defects and Developmental Disabilities' home page provides a starting point for students to search for their topic. In particular, students should explore the Search Health Topics listed in the left sidebar.

Title: PubMed Home Page
URL: http://www.ncbi.nlm.nih.gov/entrez/query.fcgi
Description: From the National Center on Biotechnology Information, PubMed allows the students to search for relevant journal articles and information about a specified health topic.

Title: Entrez Cross-Database Search Page
URL: http://www.ncbi.nlm.nih.gov/gquery/gquery.fcgi?itool=toolbar
Description: Entrez, another part of the National Center on Biotechnology Information, allows the students to conduct a search for a specific topic in many different databases at once. Here students can access sequence information for their PowerPoint presentations.

Title: National Organization for Rare Diseases
URL: http://www.rarediseases.org
Description: This is the National Organization for Rare Diseases Homepage. It contains information about uncommon diseases and conditions, including descriptions and lists of supporting organizations.

Title: American Association for Klinefelter Information and Support
URL: http://www.aaksis.org/
Description: This is the homepage for the American Association for Klinefelter Information and Support. It includes descriptions Klinefelter Syndrome, current studies, and connections to support groups for those affected.

Title: Tay Sachs Disease – National Organization for Rare Diseases
URL: http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Tay%20Sachs%20Disease
Description: This National Organization for Rare Diseases on Tay Sachs disease contains a description and list of supporting organizations.

Title: Triple X Syndrome
URL: http://www.triplo-x.org/
Description: This site is run by group at Albert Einstein College of Medicine in New York City. It contains information on the Triple X Syndrome including articles, information on prenatal diagnosis, and relevant contacts.

Title: Trisomy 18 – National Organization for Rare Diseases
URL: http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Trisomy%2018%20Syndrome
Description: This National Organization for Rare Diseases on Trisomy 18 contains a description and list of supporting organizations.

Title: Turner Syndrome
URL: http://turners.nichd.nih.gov/
Description: This National Institutes of Health site contains information on Turner Syndrome's clinical and genetic features as well as current research begin conducted.

Step 5 **Duration: 55 minutes**

To conclude this lesson, students will present their PowerPoint presentations and answer questions about the information they presented. Each presentation should last between five and 10 minutes and should cover the information listed on the "PowerPoint Slide Requirements" handout. Please share a copy of the attached rubric with the students so that they will know the criteria for their presentation evaluation.

Assessment

Teachers will assess what the students have learned about chromosomal abnormalities through the "Chromosomal Abnormality Quiz" taken in the Step 2. Additionally, the teacher will be assessing the PowerPoint presentations with the "Chromosomal Abnormality PowerPoint Rubric" included in Step 5.

Modifications

Extension

Allow students to do further research on specific chromosomal abnormalities in which they are interested. Have them answer a specific question or questions. For example, students could report on current research and new technology that helps treat conditions associated with the selected chromosomal abnormality. The National Center for Biotechnology Information Web resource can assist the students in their research.

Web Resource for Extension
Title: National Center for Biotechnology Information
URL: http://www.ncbi.nlm.nih.gov/
Annotation: This site compiles and organizes current research in the area of health. This site is very large, so students might need some guidance during this extension. The NCBI site search engine would be great for extension activities.

Remediation
Students might need assistance in the PowerPoint program navigation or creation. If possible, create groups with varying PowerPoint skills. Students might also need help in Internet searching. Assistance can be accomplished by giving them very specific sites or limited topics.

Education Standards

National Science Education Standards
SCIENCE AS INQUIRY, CONTENT STANDARD A:
As a result of activities in grades 5-8, all students should develop:

- **Abilities necessary to do scientific inquiry**
- **Understandings about scientific inquiry**

LIFE SCIENCE, CONTENT STANDARD C:
As a result of activities in grades 5-8, all students should develop understanding of:

- The cell
- **Molecular basis of heredity**
- Biological evolution
- Interdependence of organisms
- Matter, energy, and organization in living systems
- Behavior of organisms

Georgia State Science Standards
Grade: 9-12, Science, Applied Biology/Chemistry II 2
Topic: Inquiry, Process and Problem Solving
Standard: Uses traditional reference materials to explore background and historical information regarding a scientific concept.

Grade: 9-12, Science, Applied Biology/Chemistry II 4
Topic: Continuity of Life
Standard: Relates the continuation of life to the cell's chemical code.

Grade: 9-12, Science, Applied Biology/Chemistry II 11
Topic: Continuity of Life
Standard: Predicts how genetics engineering might affect society during your lifetime.

Grade: 9-12, Science, Biology 11
Topic: Genetics (Patterns of Inheritance)
Standard: Describes patterns of inheritance and genetic engineering.

Grade: 9-12, Healthcare Science Technology, Introduction to HSTE 68
Topic: Introduction to Life Changes – The Process of Change - Disabilities and Role Changes
Standard: Compare five debilitating illnesses including respective classification(s), causes, disorders, therapies, and care/rehabilitation to include biotechnological applications.

Introduction to Birth Defects and Chromosomal Abnormalities
Questions for Classroom Discussion

Chromosomal Abnormality Investigation
April Jones, CDC's 2004 Science Ambassador Program

1. **What is a birth defect?**
   A birth defect is an abnormality of structure, function, or body chemistry present in a baby at birth. Birth defects happen while the baby is developing in the mother's body. A birth defect can result in physical or mental disability, or can even be fatal. A birth defect can affect the way a baby's body looks, functions, or both. (1,2)

2. **How many babies are born with birth defects each year?**
   Each year, out of about 4 million babies born in the U.S., about 150,000 babies are born with birth defects. This means about 1 out of 28-33 babies is affected by one or more birth defects. They are the leading cause of death in the first year of life. (1,2)

3. **What causes birth defects?**
   The cause of most birth defects (about 60-70 percent) is unknown. The rest are caused by genetic factors, environmental factors, or a combination of the two. For example, Down syndrome is a genetic syndrome that occurs when a baby has an extra copy of chromosome 21. Children affected with Down syndrome have varying degrees of mental retardation, characteristic facial features, and heart defects. An example of an environmental factor is heavy alcohol use during pregnancy. Alcohol can cause an altered environment for the developing fetus and can result in Fetal Alcohol Syndrome, a syndrome characterized by a set of structural abnormalities. Clef Lip/Palate is an example of a birth defect that is caused by a variety of genetic and environmental factors. (1,2)

4. **Are some birth defects treatable?**
   Some physical birth defects, such as cleft lip/palate are treatable with surgery. Mental retardation or developmental delays can be treated with special schooling and training. However, some birth defects do not have effective treatments. (1,2)

5. **Are all birth defects detrimental?**
   Birth defects can vary greatly in severity. Some birth defects are not noticeable while others are very serious and can lead to death before or shortly after birth. (1,2)

6. **What are chromosomal abnormalities?**
   A chromosomal abnormality is a genetic defect in the number of complete chromosomes present in cells. In other words, the number of chromosomes deviates from the usual number of 46. A chromosomal deletion refers to when a piece of or an entire chromosome is missing. A chromosomal insertion refers to when a piece of or an entire chromosome is added. About 1 in 200 babies is born with a chromosomal abnormality. These abnormalities generally result from an error that occurred when the egg or sperm cell was developing.

These errors can affect the number of chromosomes present in the cells of the developing baby. (3)

Note to teachers

Feel free to add questions and answers to this list. The Web sites in the references section below are excellent! When adding questions and answers, please use these Web sites to ensure that students are provided with scientifically accurate information.

References

1. March of Dimes. Birth Defects [online]. 2004. [Date Citied 23 June 2004]. Available from URL: http://www.modimes.org/professionals/681_1206.asp .

2. CDC. Birth Defects: Frequently Asked Questions [online]. 2004 [Date Cited 23 June 2004]. Available from URL: http://www.cdc.gov/ncbddd/bd/faq1.htm#Whatisabirthdefect.

3. March of Dimes. Chromosomal Abnormalities [online]. 2004. [Date Cited 23 June 2004] Available from URL: http://www.modimes.org/professionals/681_1209.asp.

Chromosomal Abnormality Quiz

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Name:_____ Class period:_____ Date:_____ Grade: ____/ 10

Directions
Answer each question below. Each question is worth one point.

1. An abnormality in the structure, function, or body chemistry of a baby present at birth is called a _____ _____.

2. A birth defect may affect how the baby's body _____, _____, or both.

3. The cause of most birth defects is _____.

4. One out of _____ babies is born with a birth defect.

5. **True/False** Birth defects and developmental disabilities are treatable in some but not all circumstances.

6. Human cells usually have _____ chromosomes.

7. Trisomy 21 or _____ _____ is the most common known cause of mental retardation.

8. When a piece of or an entire chromosome is missing, this is called a chromosomal _____.

9. When a piece of or an entire chromosome is added, this is called chromosomal _____.

10. **True/False** Birth defects are the same for every child.

Chromosomal Abnormality Quiz Answer Key

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Name:_____ Class period:_____ Date:_____ Grade: ____/ 10

Directions
Answer each question below. Each question is worth one point.

11. An abnormality in the structure, function, or body chemistry of a baby present at birth is called a **birth defect. (1)**

12. A birth defect may affect how the baby's body **looks, functions,** or both. **(2)**

13. The cause of most birth defects is **unknown**. **(1)**

14. One out of **28 to 33** babies is born with a birth defect. **(1,2)**

15. **True**. Birth defects and developmental disabilities are treatable in some but not all circumstances. **(1,2)**

16. Human cells usually have **46** chromosomes. **(3)**

17. Trisomy 21 or **Down Syndrome** is the most common known cause of mental retardation. **(3)**

18. When a piece or entire chromosome is missing, this is called a chromosomal **deletion**. **(3)**

19. When a piece or entire chromosome is added, this is called chromosomal **insertion**. **(3)**

20. **False**. Birth defects are **not** the same for every child. **(1,2)**

References

1. March of Dimes. Birth Defects [online]. 2004. [Date Citied 23 June 2004]. Available from URL: http://www.modimes.org/professionals/681_1206.asp .

2. CDC. Birth Defects: Frequently Asked Questions [online]. 2004 [Date Cited 23 June 2004]. Available from URL: http://www.cdc.gov/ncbddd/bd/faq1.htm#Whatisabirthdefect.

3. March of Dimes. Chromosomal Abnormalities [online]. 2004. [Date Cited 23 June 2004] Available from URL: http://www.modimes.org/professionals/681_1209.asp.

Possible Chromosomal Abnormalities Search Topics

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General Web sites

Title: National Center on Birth Defects and Developmental Disabilities
URL: http://www.cdc.gov/ncbddd
Description: The Centers for Disease Control and Prevention's National Center on Birth Defects and Developmental Disabilities' home page provides a starting point for students to search for their topic. In particular, students should explore the Search Health Topics listed in the left sidebar.

Title: PubMed Home Page
URL: http://www.ncbi.nlm.nih.gov/entrez/query.fcgi
Description: From the National Center on Biotechnology Information, PubMed allows the students to search for relevant journal articles and information about a specified health topic.

Title: Entrez Cross-Database Search Page
URL: http://www.ncbi.nlm.nih.gov/gquery/gquery.fcgi?itool=toolbar
Description: Entrez, another part of the National Center on Biotechnology Information, allows the students to conduct a search for a specific topic in many different databases at once. Here students can access sequence information for their PowerPoint presentations.

Title: National Organization for Rare Diseases
URL: http://www.rarediseases.org
Description: This is the National Organization for Rare Diseases Homepage. It contains information about uncommon diseases and conditions, including descriptions and lists of supporting organizations.

Specific Web sites

Title: Down Syndrome – National Organization for Rare Diseases
URL:
http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Down%20Syndrome
Description: This National Organization for Rare Diseases on Down Syndrome contains a description and list of supporting organizations.

Klinefelter Syndrome
Title: American Association for Klinefelter Information and Support
URL: http://www.aaksis.org/
Description: This is the homepage for the American Association for Klinefelter Information and Support. It includes descriptions Klinefelter Syndrome, current studies, and connections to support groups for those affected.

Triple X Syndrome
Title: Triple X Syndrome
URL: http://www.triplo-x.org/
Description: This site is run by group at Albert Einstein College of Medicine in New York City. It contains information on the Triple X Syndrome including articles, information on prenatal diagnosis, and relevant contacts.

PowerPoint Slide Requirements and Topics to be Covered

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Students must have a minimum of 9 slides with backgrounds, at least one animation per slide, slide transitions, and different types of slide arrangements. At least two of the slides must contain a photo or graph (please make sure to cite appropriately). Please include something other than text on your slides, such as clip art, figures, or other visual aids. In general, try to put a minimal amount of text on your slides. You will expand upon the points highlighted on your slides when you give your presentation. Please use the notes section in your PowerPoint to write details about each slide. This will help you prepare for your presentation. Finally, you must also have a title slide, presentation outline slide, summary slide, and reference slide – these are not part of your 9 required slides.

Topics to be covered

1. Gene name and gene location on the chromosomes or chromosomal number
2. Karyotype or genome of syndrome (sequence of gene(s) that contribute to the syndrome you are researching)
3. An explanation of how the gene or chromosome is inherited
4. Possible factors that contribute to the chance of having an affected baby (mother's age, environmental factors, etc.)
5. A correctly quoted statement from a published scientist or researcher on the syndrome. (You should use www.ncbi.nlm.nih.gov/entrez/query.fcgi for this research.)
6. A brief description of a recent study regarding the specific chromosomal or genetic abnormality and the conclusions of that study. (Again, you will use www.ncbi.nlm.nih.gov/entrez/query.fcgi to find this information.)
7. Characteristics of the defect/syndrome
8. Possible detection methods or tests that can be used
9. Avenues or locations parents can take to get help understanding the syndrome

Chromosomal Abnormality PowerPoint Rubric

Chromosomal Abnormality Investigation

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Name: _____ Teacher: _____

Date of Presentation: _____ Title of Presentation: _____

Group members: _____
_____

Presentation title present: _____ (2 points each)
Presentation resources present: _____
Outline and Summary slides present: ____

	Criteria				Points
	1	2	3	4	
Organization	Audience cannot understand presentation because there is no sequence of information.	Audience has difficulty following presentation because student jumps around.	Student presents information in logical sequence which audience can follow.	Student presents information in logical, interesting sequence which audience can follow.	____
Content Knowledge	Student does not have grasp of information; student cannot answer questions about subject.	Student is uncomfortable with information and is able to answer only rudimentary questions.	Student is at ease with content, but fails to elaborate.	Student demonstrates full knowledge (more than required) with explanations and elaboration.	____
Visuals	Student used no visuals.	Student occasionally used visuals that rarely support text and presentation.	Visuals related to text and presentation.	Student used visuals to reinforce screen text and presentation.	____
Mechanics	Presentation had four or more spelling errors and/or grammatical errors.	Presentation had three misspellings and/or grammatical errors.	Presentation had no more than two misspellings and/or grammatical errors.	Presentation had no misspellings or grammatical errors.	____
Delivery	Student mumbles, incorrectly pronounces terms, and speaks too quietly for students in the back of class to hear.	Student incorrectly pronounces terms. Audience members have difficulty hearing presentation.	Student's voice is clear. Student pronounces most words correctly.	Student used a clear voice and correct, precise pronunciation of terms.	____
Power point specifications	Student included a background and no animation in the power point presentation.	Student included background and one animation in the power point presentation.	Students included backgrounds, animations and slide transitions in the presentation.	Students included backgrounds, animations, slide transitions, and text animation. Students went above and beyond standards.	____

Teacher Comments: **Total:** _____

Teach-nology, Inc. Rubric Makers [online]. 2003 [date cited 27 July 2004] Available at URL: http://teach-nology.com/web_tools/rubrics/.