

actionbioscience.org lesson

To accompany the peer-reviewed article by Michael J. Dougherty, Ph.D.:

“The Genetics of Autism” (Sept. 2000)

<http://www.actionbioscience.org/genomic/dougherty.html>

Autism: A Different World (Mar. 2002)

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Educator’s section: *p. 1-3*
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Grades & Levels:

- **Handout 1:** senior high school (advanced/AP) – undergraduate (year 1)
- **Handout 2:** high school (general)

Time Recommendations:

- Two, 50-minute class periods
- 2-3 hours homework time

NSES (USA) Content Standards, 9-12:

- NSES 2.2. Science as Inquiry: understanding about scientific inquiry
- NSES 4.2. Life Science: molecular basis of heredity
- NSES 7.1. Science in Personal & Social Perspectives: personal & community health

Note: View the NSES content standards on this site to choose other curricular applications for additional activities at:

<http://www.actionbioscience.org/educators/correlationcharts.html>

Learning Objectives: Students will ...

- be able to describe the symptoms of autism as a syndrome
- outline the history of our understanding of the disorder
- explain briefly why scientists now believe that the disorder is biological in origin
- discuss evidence for the genetic basis of the syndrome
- suggest further lines of investigation or research

Key Words Include:

alleles, autism, autistic savant, autistic spectrum disorders, Asperger’s Syndrome, genetics, genetic disorder, gene therapy, Mendelian inheritance, Behavioral Genetics (see definitions provided in the Supplementary Handout)

Preparation:

Article Discussion: Have students read the online article “The Genetics of Autism” by Michael J. Dougherty, Ph.D. at www.actionbioscience.org/genomic/dougherty.html. A list of questions is provided on page 3. The discussion can be conducted as a class or you can distribute the questions to students and have them address the questions in groups. Discussion may be concentrated in one time period or dispersed into smaller periods of time involving consideration of 2-3 questions at a time.

Handout 1: senior high school (advanced/AP) – undergraduate (year 1)

- Distribute or have students print out Student Handout 1 (pages 4-6).
- The activities that follow the reading are divided into three sections: Group Discussion, What if?, and Projects:
 - **What if?** (pages 4-5): This is part of Student Handout 1 and provides scenarios for consideration. After reading them, students should list the issues involved, and after discussion (recommended: 15-20 minutes on each scenario) suggest a resolution or solution. You may suggest that each team present their findings to the class for general discussion. If so, then allocate another 10 minutes for each team's report and 5-10 minutes for questions.
 - **Projects** (page 6): This is also part of Student Handout 1. Each project requires teams of up to five students to plan, research, and construct or carry out a project followed by a presentation or demonstration. It is suggested that from two to six weeks be allowed for completion of each project and that firm deadlines or progress report dates be set.

Handout 2: high school (general)

- Distribute or have students print out Student Handout 2 (page 7).
- Ask students to choose one activity to perform individually or with a classmate(s).
- Assign these activities as homework or work to be done during study periods.
- Inform students when the assignment is due and/or their presentation date.

Supplementary Handout:

To accompany Handout 1 or 2, this supplement contains a bibliography and glossary. Distribute or have students print out this handout (page 9). They can use the information on this handout to help them with their research.

For Educators: Article Discussion

About the article by Michael J. Dougherty, Ph.D.: “The Genetics of Autism”

www.actionbioscience.org/genomic/dougherty.html

Article Content Questions:

1. What is an autistic savant? Are they typical of people with autistic syndrome? What is the more typical situation of people with autism?
2. What are the defining characteristics of autism?
3. Why do scientists now believe that autism is a biological disorder and not entirely environmentally based, as had been previously supposed?
4. Give one example of a situation that illustrates that even complex behavior patterns can be strongly influenced by genetics.
5. What is “reductionist thinking” and why might it not be the best approach to understanding autism?
6. Why isn’t “Mendelian inheritance” adequate to explain the inheritance of behavioral syndromes such as Autism?
7. Give one example of a “natural experiment in shared inheritance” that may shed light on how behavior patterns are inherited.
8. Why are family doctors unlikely to see enough cases of two affected siblings in the same family to suspect a genetic influence in autism?
9. Give one example of evidence provided by twin studies, which implies a strong role for genetics in autism.
10. Why does the author believe that we will someday have a better understanding of the brain differences that lead to autism?

Personal Viewpoint Questions:

1. Autistic savants often demonstrate amazing musical or artistic talents. Some, like the character, Raymond Babbit in the movie *Rainman*, have peculiar abilities in counting or mathematics. If autism could be cured, then these savants might lose such abilities. Do you think that if a cure were available, that we should proceed in such cases? Why or why not?
2. A significant proportion of autistic people are termed “high functioning;” that is, although autistic, they can function well-enough to live in and contribute to society. For example:
 - Dr. Temple Grandin is a world-renowned animal scientist who has designed one third of all the livestock handling facilities in the United States.^{1,2,3}
 - Donna Williams, born in Australia and now living in England, is the author of four books that explain what it is like to be autistic. Diagnosed only as an adult, she travels extensively, speaks at conferences, corresponds with other autistic people and their families, and works with professionals to help others understand and deal with autism.^{6,7,8,9}If you were to meet one of these people what would you most like to ask them and why?
3. In your view, if autism does turn out to have a strong genetic component, then should we seek to cure autism through gene therapy? Why or why not?
4. Behavioral modification approaches have helped to alleviate the worst symptoms of autism in many cases. Some examples exist of autistic children who, through such therapy, have become apparently normal adults. Do these situations indicate that autism is not genetic in origin? Why or why not?
5. In your view, does the information that Dr. Dougherty presents in his article offer hope for parents of young children diagnosed as autistic?
6. Do you think that autism is entirely genetically based? Why or why not?

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Student Handout 1

What if? Scenarios

Instructions:

- Conduct the “What if?” activities as a group.
- Choose *one* “What if?” scenario of the three listed for your activity.
- After reading the scenario you have chosen, list the issues involved.
- Then, discuss the questions presented at the end of each scenario.
- Suggest a resolution or solution to these questions.

Notes:

- To conduct research on the Internet follow the links provided at the end of Dr. Dougherty’s article to other web sites, some of which will also have links that you can follow.
- To conduct research in literature, such as books and journals, see the bibliography list in the “Supplementary Handout.”

Scenario #1:

You and your family have noticed that your 18-month-old daughter assumes a rigid posture when held and avoids direct eye contact. Nor does she respond to tickling and other games with smiles and giggles as did her older brothers at the same age. You are concerned because of her apparent lack of sociability and interest in her environment. Your pediatrician minimizes your concerns, suggesting that some children just take a little longer to develop such interests. However, he suggests a hearing test because your daughter’s inattention to voices and noise may indicate deafness. This you arrange but your daughter turns out to have normal hearing.

By the age of three, your daughter has developed peculiar mannerisms, such as tearing paper up into pieces, twisting them, and then “flicking” them back and forth rapidly in front of her eyes. She walks on her toes most of the time, and has lost any inclination to talk under normal circumstances. Single words are spoken only at moments of emotional stress. Again you visit the same pediatrician. Again he seems unconcerned, suggesting that your daughter is only taking a little longer than normal to develop.

Not satisfied with this apparent lack of medical concern and help, you arrange to have your daughter undergo a thorough series of tests at a major children’s hospital. The result is a diagnosis of classical infantile autism. Furthermore, the neurologists at the hospital suggest that there is really no hope for a cure or improvement, implying that the best approach would be to have your daughter institutionalized. One of them suggests that the problem may have started through lack of love and caring at home, muttering something about “a refrigerator mother.”

Discussion:

- Is there anything about the described behavior of the daughter in this scenario that is consistent with a genetic component to autism?
- If, as Dr. Dougherty suggests in his article, there is a genetic component to autism, then what would you do next if this were your daughter?
- Is there anything that you, as one of the parents in this scenario, could do to help your child?

Student Handout 1 (cont.)

Scenario #2:

The article “The Genetics of Autism” indicates that the field of Behavioral Genetics holds great promise in increasing our understanding and treatment of complex behavioral syndromes like autism. Much of the pioneer and most fundamental research in this emerging field was carried out by Dr. Seymour Benzer and his research associates. As described in the recent book by Jonathan Weiner,⁵ Dr. Benzer began this work in the 1960s to search out the links between genes and behavior using mutant strains of fruit flies. They demonstrated that a single gene and even a change in a single letter of the genetic code in key genes could change a fly’s behavior. Could this happen in humans?

It seems likely, as Dr. Dougherty suggests that a genetic influence exists for autism. However, it is very likely that in the case of autism multiple genes may be involved and they may be located on a number of chromosomes. Suppose that in 10 years time a prenatal test is available to identify the risk of a child developing autism based on the number of autism-linked genes and their autism-related functioning. However, the test has an error rate of false-positive results of 10%. Based on the results of such a test, a couple could decide whether to have such a child or to abort it.

Suppose also that gene therapy techniques and behavioral modification approaches have improved so that there is a 75% chance that a child born and diagnosed with autism at up to 5 years of age will develop into a normally functioning adult. The earlier that gene therapy is performed after birth, the better the child's prognosis.

However, if gene therapy is attempted on a normal child, falsely identified as autistic according to the prenatal test (false-positive result), then there is a 60% risk of inducing an autistic-like condition by adulthood. In such cases, the parents or government-run health agencies would have to bear the cost of behavioral or medical therapy or institutional care.

Discussion:

- Can we reasonably expect that behavioral genetic research, such as Dr. Benzer’s on fruit flies, would help us understand the basis of human behavior patterns, such as autism?
- If the situation described in the scenario were true, then should couples base their decision whether to have or not have a child that may be autistic based on prenatal tests?
- Should parents risk gene therapy on a child that is identified as autistic, knowing that it could harm a normal child that has been falsely identified?
- Should the possible financial costs to society be weighed in the balance of such decisions?

Student Handout 1 (cont.)

Scenario #3:

In a recent article in *Wired Magazine Online*,⁴ Steve Silberman describes how autism - and “its milder cousin, Asperger’s Syndrome - is surging among the children of Silicon Valley.” Since at least the 1960s it has been noted that the incidence of autism spectrum disorders seems greater in families with a higher-than-average number of scientists and engineers. It has been suggested that if behavior has a genetic component, there may be genes that provide abilities to concentrate on a problem and follow it through to a resolution in a “single-track” fashion. This is often the strength in scientific research or engineering endeavors. Many talented engineers, famous scientists, and high-tech workers, who are very successful in their professional careers, are noted to be socially inept or somewhat eccentric. It has been suggested that many fit the criteria of Asperger’s Syndrome, so much so that it has been called “the engineers disorder”. Autistic spectrum disorders may occur when children inherit a higher than normal proportion of the “problem-solving” alleles for these genes.

Autism and related disorders such as Asperger’s Syndrome seem to be much more common today than in the past. This has been blamed on a kind of “diagnostic fuzziness” because the diagnostic criteria are somewhat subjective and open to interpretation in each case. However, many educators of learning disabled children and social workers are convinced that the statistics do indicate a real increase in the frequency of these disorders.

There is some concern that the genes that make one susceptible to autism may be becoming more common in the general population. One reason may be that unlike the past, more and more mildly autistic people are attracted to the same areas such as Silicon Valley, are meeting, marrying, and having children of their own. The likelihood that two technologically-inclined people who carry the autism alleles (if these exist) meet and have children with autism spectrum disorders will increase under these circumstances. This will result in greater demands on the educational system for special education classes and teacher’s aides to help these children learn to the best of their ability.

Discussion:

- List and discuss the positive and negative aspects of the increasing incidence of autistic spectrum disorders as noted in Silicon Valley.
- Do some research to determine if the statement that “the incidence of autism spectrum disorders seems greater in families with a higher than average number of scientists and engineers” describes a real or imagined situation? Present your findings to the class.
- What are the diagnostic criteria for autism and related disorders? Should we be concerned that the diagnosis is often somewhat “fuzzy”?
- Should we be concerned that any possible genes or alleles that contribute to autism may become more common in the population? Suggest possible actions society could take to remedy the situation.

Student Handout 1 (cont.)

Projects

Instructions:

- Review the projects below and find classmates to form a team to do the project.
- Choose *one* project from any of the three groups listed.
- Follow the instructions given for each group of projects.

Notes:

- To conduct research on the Internet follow the links provided at the end of Dr. Dougherty's article to other web sites, some of which will also have links that you can follow.
- To conduct research in literature, such as books and journals, see the bibliography list in the "Supplementary Handout."

Exhibit

With a partner or two, create a poster or kiosk exhibit on autism. Your exhibit should consist of one or more posters with text, graphics, photographs, etc. Some suggested topics:

- Possible causes of autism: genes or environment?
- Autistic spectrum disorders
- Hypothesis on the genetics of autism
- Diagnostic criteria for autism
- Therapies for autism
- Asperger's Syndrome: gift or curse?
- The Autistic Savant: windows to the mind

Be prepared to present it to the class at a time and place indicated by your instructor.

Role-play

Your team will plan a skit or play about an autistic child. Have one member of the group take on the role of an autistic child, while the others play parents, family members, and other people. To present an authentic skit, your team will have to research the behavior of autistic children. Choose one skit scenario:

- a visit to the dentist (or doctor or optometrist)
- first day at school
- meeting with school principal to design an education plan for the child
- discussing with a social worker, special educator, or teacher the resolution of a difficult situation that has arisen at school or at home
- an appointment with a doctor or pediatrician during which the parents raise questions about their child's behavior and autism (the physician may be helpful or doubtful)

Report

Complete a research project on an issue related to the genetics of autism arising from Dr. Dougherty's article or one of the other activities in this handout. This activity will require a research proposal outlining the nature of your topic and subquestions. Research the topic on the Internet and in other sources to produce one of the following:

- Individually: a research report of 1500 words (6-8 pages), which may include illustrations and tables
- Group: a 10-minute video, computer-based tutorial, or web site of about 6 component pages
- Pairs: a brief outline (2-3 pages) and a 10-minute oral presentation

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Student Handout 2

Instructions:

- Choose one of the projects listed below to do in teams or pairs.
- To conduct research on the Internet follow the links provided at the end of Dr. Dougherty's article to other web sites, some of which will also have links that you can follow.
- To conduct research in literature, such as books and journals, see the bibliography list in the "Supplementary Handout."

1. Perform a web or literature search to find 2 or 3 examples of Autistic Savants, other than those mentioned in Dr. Dougherty's article. Explain briefly why each is considered autistic.
2. Imagine that you are the chairperson of a medical committee entrusted with preparing a list of symptoms of autism. List five to ten symptoms that you consider, taken together or in part, would indicate autism in a patient.
3. Write a brief essay (1 or 2 pages, up to 500 words) on the history of our understanding of the basis of autism.
4. Who was Gregor Mendel? What basic principles of inheritance did he discover? How did he discover them? Explain in a report why it is that even though Mendel's discoveries were fundamental to our modern understanding of inheritance, they cannot fully explain how a behavioral pattern like autism is inherited.
5. Does environment, either within the family or in the community, have an effect on development of autism or on its severity? If it does, is a genetic influence ruled out? Present your findings in a report or essay.
6. Twin studies are often used in behavioral genetic studies. Why? Give one or two examples other than those listed in Dr. Dougherty's article of twin studies in behavioral genetics. Create a visual presentation of your findings.
7. Briefly outline one or two examples of international collaboration between scientists investigating autism. Present your outline in a chart or other visual presentation.

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Supplementary Handout

Bibliography

The following books, articles, or web sites were used in the preparation of this lesson.

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4. Silberman, Steve. 2001. "The Geek Syndrome." *Wired*, Archive 9.12 (Dec 2001) at http://www.wired.com/wired/archive/9.12/aspergers_pr.html
5. Weiner, Jonathan. 1999. *Time, Love, Memory: A Great Biologist and His Quest for the Origins of Behavior*. Alfred A. Knopf, New York, NY, USA, [ISBN: 0-679-44435-1]
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8. Williams, Donna. 1996a. *Like Color to the Blind: Soul Searching and Soul Finding*. Doubleday Canada Ltd, Toronto, ONT, [ISBN: 0-385-25595-0]
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Autism Glossary

Alleles: Different forms of the same gene, which lead to different expression of a trait. For example, human eye color is determined by a gene that has two alleles, one determining blue eye color and the other brown eye color. Other alleles for other genes influence the intensity and shading of these two colors as well.

Asperger's Syndrome: One of the disorders on the autistic spectrum in which a person lacks basic social and motor skills, may be clumsy, seems unable to decode body language or to sense the feelings of others, and may avoid eye contact. Such individuals may have eccentric habits and periodically launch into monologues about very narrow and technical interests. Many, but not all, exhibit a high degree of language ability and intelligence but appear narrowly focused.

Autism: In its classic form, as first noted by Leo Kanner in 1943, it is a behavioral syndrome involving three areas of difficulty:

- limited social interaction and often complete avoidance of others with little eye contact
- communication and use of language is severely impaired and the individual is often nonverbal
- limited imagination reflected in repetitive and stereotypical patterns of behavior and activities

It is now generally accepted that autism is due to abnormal brain development and is not a mental illness or a psychological disorder. It affects people of all races and cultures. More males than females are affected, the ratio being 4:1 for classic autism (and 9:1 with Asperger's Syndrome). There appear to be both genetic and environmental influences in most cases, although often one or the other may appear stronger.

Autistic Savant: An individual with an often severe autistic spectrum disorder who shows evidence of a particular talent. Such talents often involve amazing feats of mathematics or precocious musical ability.

Autistic Spectrum Disorders: A wide array of disorders ranging from mild impairment, commonly labeled as "eccentricity," through Asperger's Syndrome to classic autism. All share some degree of language and/or social impairment often coupled with odd behavior.

Behavioral Genetics: The branch of genetics that seeks to discover the genetic basis of behavior in animals and humans. It is an experimental science that was pioneered by such researchers as Dr. Seymour Benzer.

Gene Therapy: A procedure designed to repair or replace a defective gene. This could involve introducing functional genes into individuals who have a genetic disorder in order to treat or correct that disorder.

Genetic Disorder: Any consistently abnormal function in health or behavior that has a genetic basis.

Genetics: The branch of biology, which seeks to establish and understand the principles and basis of heredity. It includes research on the inheritance of traits and the molecular functioning of genes.

Mendelian Inheritance: Inheritance of traits in patterns first discovered by Gregor Mendel in his classic series of genetic experiments with pea plants between 1850 and 1865. Generally this involves traits determined by one gene with two possible alleles, in a simple dominance/recessive relationship, which happens to be one of the simplest possible patterns of inheritance.