

Objectives

- To compare normal and abnormal child development with respect to autism.
- To encourage students' tolerance for ambiguity of explanation, when it is inappropriate to accept a single hypothesis as true.
- To analyze and communicate information through visual displays.
- To develop reasoning skills, especially to learn to synthesize information from several sources and plan a course of action that is responsive to and appropriate in light of the information given.
- To introduce students to the tasks and concerns of several biomedical professionals.
- Optional Exercise: To promote appreciation for the value of the diversity of human experience.

Part 1 – Meet the Greens

Narrator:

As Mr. and Mrs. Green finished signing the papers to take their newborn son, Michael, home, the nurse said to them, "What a precious baby." The Greens couldn't have agreed more. Mr. and Mrs. Green went on to raise their baby as any caring parents would. At 23 months old, Michael was a healthy-looking boy. The Greens took him everywhere and encouraged him to explore his world. On the beach one day, Mrs. Green patiently held up a red pail and a shovel. "See, Michael," she said. "Pail. Pretty pail. Just say pail, darling." Michael stared past her, apparently watching Mr. Green, who was writing the child's name in the sand. That gave Mrs. Green an idea. Maybe "pail" was too hard. What about trying his own name again? "Look, that's your name." She pointed to herself and said "Mama." Then she pointed to the little boy and said "Michael. Say 'Michael,' honey." She stroked his hair. "Michael." The child continued to stare past her. Mrs. Green looked sad.

Mr. Green: "Oh well, he'll get it eventually. Some children take longer to learn how to speak than others."

Mrs. Green: "I don't know, he's almost two and he can't even speak common words. I've read and read, and made notes, and I even made a video the other day of Michael at the playground. I know you think I'm being too nervous, but I really think we should call Dr. Klotz and make an appointment for another checkup."

Mr. Green: "Well okay, if you think it might help."

Narrator: Mrs. Green wrote down all the symptoms that she had noticed in Michael that seemed to be a bit unusual for his age. She then called Dr. Klotz, Michael's pediatrician, and set up an appointment for the next day.

Dr. Klotz: "Come in Mrs. Green. Have a seat. And let's get Michael settled in the kid's corner. Fine. Now, what seems to be the problem?"

Mrs. Green: "Well, doctor, I know we've discussed this before, but now Mark and I both think something is wrong with Michael. He doesn't seem to be developing like the other children. We've set up play dates for the

children within our neighborhood and he is very withdrawn. He doesn't seem to want anything to do with them. He also doesn't talk; he only uses gestures to tell us when he needs or wants something. I didn't think anything of it for a while, but he is going to turn two soon, and it just seems like something is wrong."

Dr. Klotz: "Go on. Tell me more about it."

Narrator: As Mrs. Green spoke about Michael's puzzling behaviors, Dr. Klotz observed both mother and child

keenly. Mrs. Green repeated Michael's name frequently, and turned to smile at him, but the child did not respond and did not meet her gaze. The little boy sat quietly on a little chair gazing out the window. Beside him was a child's table full of colorful books and puzzles. Michael ignored them. Mrs. Green gestured frequently and wrung her hands. From time to time, she scratched a raw spot on her wrist. She appeared distracted and very troubled. Michael's father watched his wife and child unhappily, but it was difficult to discover what he thought by reading his expression.

Dr. Klotz: "Mrs. Green, how are you yourself feeling?"

Mrs. Green: "Oh, not too bad, Doctor. I have a little psoriasis. Sometimes it seems like it gets worse when I'm feeling

stressed. And I do feel stressed. I'm so worried about my son."

Dr. Klotz: "What do you think is wrong with Michael, Mr. Green?"

Mr. Green: "I don't want to scare anyone, but I've been thinking maybe he's, well, slow."

Mrs. Green: "Michael is not slow!"

Narrator: For a moment, she seemed reluctant to go on.

Mrs. Green: "He has a cousin that was diagnosed with autism a while back. Could he be autistic?"

Dr. Klotz: "Hmm... a cousin? Well, it seems that there is a genetic component to autism, but ..."

Mr. Green: "I've heard it runs in families sometimes."

Dr. Klotz: "Yes. And when you have twins, the incidence of both having autism is far higher if they are identical

twins, rather than fraternal. But we don't know which genes are involved, whether there is more than one

gene, and if it's the same gene or genes that are involved in each case."

Mrs. Green: "I don't understand that."

Dr. Klotz: "Well, suppose you get a cold. There are several different viruses that can cause what we call a cold.

So, there's more than one possible causative agent—in other words, more than one way to get a cold. Sometimes you might have two different viruses at the same time in your upper respiratory tract, and they combine to cause multiple different cold symptoms. So, they are working together to cause that cold. And again, not every cold is alike. Sometimes your cold is mild, and sometimes it's a humdinger. Autism is a little like that. It can be mild, moderate, or quite severe. In fact, when it comes to autism, we usually talk about ASD, or an autism spectrum of disorders. Individuals with ASD have some common behaviors and

challenges."

Mr. Green: "So, now you're saying it's viral? I thought you just said it was genetic."

Dr. Klotz: "No, no, I bring up the cold virus just as an analogy. Most researchers are looking at a dozen or so genes

for proteins that may not be working properly in autistic children. But there are some people that think autism might be caused by a virus or viruses. And some people think it could be caused by environmental

factors, like toxins, allergens, or pollutants."

Mrs. Green: "How about another type of environmental factor—how about poor mothering?"

Dr. Klotz and Mr. Green: "Not in this case."

Dr. Klotz: "There is very little support for that theory, even in general. ... But, we're putting the cart before the horse

here. I told you to keep an eye on Michael last time you were here, and maybe make some notes. Well, from my observations, and what you've described today, it seems that Michael could possibly be autistic.

But we're going to want to do some tests before we say that."

Mrs. Green: "What kind of tests?"

Dr. Klotz: "Well, behavioral tests, to see what he can and can't do. If it seems warranted, I'll recommend a pediatric

neurologist who will investigate further and either rule out autism spectrum disorders or make a formal

diagnosis and suggest a plan tailored to help Michael."

Mrs. Green: "Okay—anything we can do to help him. I just want him to grow and be as normal as he can be."

Dr. Klotz: "Let's try not to worry. Michael may not have autism. And if he does, it may very well be mild. And there

are lots of things you can do to help a child with autism."

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Activity 1.1

Make a growth chart showing the approximate age at which children reach developmental milestones like sitting up without support, reaching for objects, first words, first phrases, and other events. If you like, you may include infant, toddler, kindergarten pictures of yourself, or of a member of your family.

With reference to your chart, answer the following question: Are Mr. and Mrs. Green's concerns about Michael's speech and social development well-grounded?

Activity 1.2

Develop a list of behaviors associated with autism spectrum disorders.

Part 2 — What Causes Autism Spectrum Disorders?

Mr. Green left Dr. Klotz's office with a bee in his bonnet. Over the next couple of weeks, while Mrs. Green methodically charted her observations of Michael in her notebook, he began a systematic study of the known causes of the autism spectrum disorders.

He found that scientists and doctors have lots of questions about the organic basis of the syndrome and many promising leads, but no definite answers. Studies have found the following possible causes.

Activity 2.1

Irregularities in some areas of the brain which can be detected by imaging techniques like MRI.
 Draw a picture of the brain showing the frontal and temporal lobes, amygdala, and cerebellum. Tell the function of these areas. How do you think abnormalities in these brain structures or in their functions could produce some of the symptoms of autism?

Activity 2.2

• Irregularities in levels or functionality of the neurotransmitters serotonin and GABA, or the presence of unusual alleles of several genes that encode proteins like shank3 and neuroligins, which are involved in formation and maintenance of synapses.

Draw a generalized neural synapse and describe how neurotransmitters work at the synapse to connect one neuron to the next. Explain how inappropriate neural transmission could disrupt neural pathways and cause autistic behaviors.

Activity 2.3

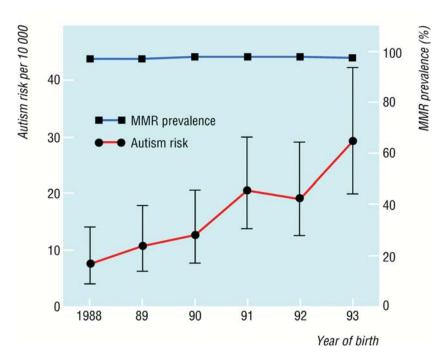
Influence of environmental factors like organophosphates and polychlorinated biphyenyls (PCBs); it is believed
that the genetic makeup of autistic individuals may render them more sensitive to the presence of environmental
toxins that affect neurophysiology.

Explain how the release of high levels of environmental toxins could generate "clusters" of autism, similar to cancer clusters. Use a Venn diagram to show how genetic susceptibility inherited from both parents, along with environmental toxicity could produce autistic children.

Activity 2.4

A link with vaccination against viral diseases, especially (i) MMR (measles-mumps-rubella), (ii) a set of vaccines
that include the mercuric compound thimerosol as a preservative, and (iii) the practice of delivering multiple
vaccines simultaneously to an infant, which, while efficient for the parents and health care people, might seriously
weaken the juvenile immune system (Gerber & Offit 2009).

While researchers continue to investigate a possible link between vaccination processes and autism, many scientists point to a recent study in the United Kingdom of the relationship between the practice of infant MMR vaccination and the rate of autism diagnoses. Look at the graph on the next page: what is the "take home message" of this graph (i.e., the main idea), and how does that message tend to disprove the idea that MMR vaccines are a cause of autism?



Reproduced from "Mumps, measles, and rubella vaccine and the incidence of autism recorded by general practitioners: A time trend analysis," by Kaye, J.A., Melero-Montes, M., and Jick, H., *British Medical Journal*, 322(7284), pp. 460–463, ©2001, with permission from BMJ Publishing Croup Ltd.

Mr. Green read and read, and though his head was full of interesting ideas and hypotheses, his final conclusion could be summed up in his comment to Mrs. Green, "Well, dear, it looks like the doctors don't know what causes autism; they're trying pretty hard to figure it out though. But I guess that right now the best they can do is to detect the syndrome, describe each child's unique presentation of the disease, and figure out an individual treatment plan that is customized just for that child and his or her family."

Part 3 — Diagnosing Autism and Parent Education

Several weeks later ...

Narrator:

Dr. Klotz compared Michael's speech and other behavioral skills to those expected for children his age and decided to refer the Greens to a pediatric neurology unit at a large regional teaching hospital and medical center. There, a specially-trained multidisciplinary team gave the boy a comprehensive physical exam, with special emphasis on motor, coordination, reflex, and cognitive skills, as well as a hearing test. Blood was drawn and he was screened for several disorders that are sometimes associated with autism. Michael also received an EEG, which monitors electrical activity in the brain, and an MRI, which provides high-quality diagnostic images of the central nervous system.

Activity 3.1

The multidisciplinary team at the pediatric neurology unit included the following medical specialists. Select one professional from the list below, do some research, and prepare a presentation that describes: (i) the special area of expertise of that professional, (ii) what sort of training he/she receives, and (iii) how his/her measurements, observations, or analysis can contribute to the diagnosis of autism in a child. *Think:* Would any of these professions suit you?

- Neurologist
- Psychologist
- Speech/Language Pathologist
- Audiologist
- Radiologist
- Nurse Practitioner
- Pediatric Nurse
- Biomedical Laboratory Technician

Narrator: Dr. Katrine O'Rourke, a pediatric neurologist, met with her team, reviewed their results and suggestions, and then asked her secretary to schedule an appointment with the Greens to discuss Michael.

Dr. O'Rourke: "Please have a seat."

Mr. Green: "Thank you, Doctor. Do you know if Michael is autistic?"

Dr. O'Rourke: "Mr. and Mrs. Green, first let me say that all tests for biochemical markers and illnesses that are frequently associated with autism were negative. No abnormalities were observed in his brain. However, our behavioral observations suggest that Michael is indeed autistic. So we have no definite physical cause here. In cases like Michael's, we say the child has idiopathic autism rather than primary autism. That means we're not sure what causes it. It could be one gene or many genes. It could be some substance from his environment. Or it could be an interaction between genes and environment."

Mrs. Green: "There's no cure for autism, is there? And since we can't see anything wrong with Michael, there's nothing to be done, right?"

Dr. O'Rourke: "You're right to say there's no cure. But you are wrong when you say there's nothing to be done."

Mr. Green: "Where do we start?"

Dr. O'Rourke: "Well, there is someone I'd like you to meet. Let's go down the hall and visit with Mary Matthews. She'll be coordinating Michael's treatment team. Based upon Michael's specific needs, she'll make therapeutic recommendations to one of our programs or to other programs in your community. These will probably include individual or group therapies, speech and language therapy, and some training in social skills. And when the time comes, she'll help you to work with your school district to obtain specialized educational services that are appropriate."

Activity 3.2

Michael's treatment team also included a diverse group of psycho/social and education professionals. Select one professional from the list below, do some research, and prepare a presentation that describes: (i) the special area of expertise of that professional, (ii) what sort of training he/she receives, and (iii) how his/her measurements, observations, or analysis can contribute to a therapeutic plan for a child with autism. *Think:* Would any of these professions suit you?

- Social Worker
- Occupational Therapist
- Physical Therapist
- Child Psychologist
- Dietician

Activity 3.3

When you have finished describing the roles of all of these professionals, take a few minutes to think about and describe the role of Michael's parents in his treatment.

Part 4 — Choosing a Treatment Plan

There are several types of treatments that Mr. and Mrs. Green must consider when selecting the types of treatments that are best for Michael. The social worker explained to them that the treatment plans are dynamic and are tailored to severity and the characteristics of the individual child with autism.

"Each child is different, and therefore must undergo different treatments. The treatments may change over short or long periods of time as Michael develops," explained Miss Mathews.

She went on to explain that there are several types of treatments and educational programs that the child can undergo. It is critical that each treatment is selected for the child according to his/her own weaknesses and strengths. Each child may have a different treatment, and while one treatment may work for one child, it may not for another. This will allow the child to move towards his/her own certain goals and for progress to be made.

Activity 4.1

- a. There are educational programs available for children 3 and under as well as programs for school-aged children. List specific programs that fall into each of these categories. Which educational program may be possible for Michael and why might his parents select that one?
- b. There are also many types of medical and dietary treatments available for children who have autism, each of which is selected according to each child's needs. What medications are available for children with autism? Some parents may have their child switch to a gluten/casein free diet. Why would this help the autistic child, and what precautions should be taken when switching diets?

Part 5 — Optional Creative Writing Exercises

Optional Exercise 5.1

Investigate and learn more about low-functioning autism, Kanner's syndrome, and Asperger's syndrome. Perhaps these developmental neurological disorders represent a continuum of defects, from nuances to profound impairments. Autism is frequently heritable, yet the parents of autistic children have managed to hold down jobs, function, attract mates, have children. Still, many of the parents appear socially awkward or lacking in affect, and it is likely that they have some nuance of autism. In fact, many believe that computer programmers, some engineers, mathematicians, physicists, and other scientists tend to have autistic traits and are more likely to produce autistic children. Do you think you have any traits like these? How might they hurt/help you in your studies, personal life, and eventual career? Consider the two short paragraphs below written by autistic educator/animal scientist Dr. Temple Grandlin and poet Frank Gallimore, a hearing son born into a deaf family, respectively.

"I think in pictures. Words are like a second language to me. I translate both spoken and written words into full-color movies, complete with sound, which run like a VCR tape in my head. When somebody speaks to me, his words are instantly translated into pictures. Language-based thinkers often find this phenomenon difficult to understand, but in my job as an equipment designer for the livestock industry, visual thinking is a tremendous advantage." (Thinking in Pictures. My Life with Autism, by Temple Grandin. Vintage Books, Random House NY 1995)

"Did I ever wish I was deaf?" he says. "Hell, yes. I wished that for most of my life. For most of my childhood, I thought the whole hearing world was for the birds and felt it was some kind of cruel joke that I was so unfortunate to be born a member of it. I always believed, and in some ways still believe, that the deaf world was so much more vivid, expansive, and beautiful than the hearing one. There is something about the way deaf people 'see' that is incredibly profound. In comparison, hearing people always seem rather nearsighted." (An Ear for Poetry, by Virginia Hughes. John Hopkins Magazine June 2006 58:3—Interview with Frank Gallimore)

Some Asperger's individuals say that they would not exchange their brain and its peculiarities of perception for a "normal" brain, preferring some aspects of their experience. This is similar to the idea expressed by many deaf people that theirs is not an "impairment" but a difference, and they find the deaf world rich and their experience beautiful and meaningful enough that they would not change it. What do you think about these statements? Have you or someone you know ever seemed to be enriched by what first appeared to be a horrible misfortune? Write/discuss.

Optional Exercise 5.2

Some people would say that the modern world, unlike the world of Odysseus, of Lewis and Clark, or even of Albert Schweitzer, lacks opportunities for adventure. Others would counter that Neil Armstrong's walk in space or Hillary Clinton's run for the U.S. presidency represent the real modern adventures of two extraordinary individuals. But can ordinary modern people have adventures? In the book *The Curious Incident of the Dog in the Night-time* by Mark Haddon, an autistic adolescent who is the child of divorced parents leaves the home of his father and travels alone across London to find his mother. This act, simple and accessible for most of us, is a tremendous challenge for the boy, but one he rises to with extraordinary courage. Have you ever had the experience of accomplishing something that would seem very ordinary for many people, but for you, was quite an adventure? Write/discuss.

Optional Exercise 5.3

We often hear about societal changes coming about due to an innovation like Gutenberg's printing press or Eli Whitney's cotton gin. Humans are social animals, and one thing leads to another. In the mid- to late 20th century, with the growing acceptance of feminism, large numbers of women appeared in laboratories and other high-tech workplaces formerly restricted to men (Baren-Cohen 2006). How might this trend lead to an increase in a population of individuals with both maternally-derived and paternally-derived genetic traits that predispose to autism, and therefore an "autism epidemic"? Write/discuss.

Resources

Autism Fact Sheet

 $http://www.ninds.nih.gov/disorders/autism/detail_autism.htm$

NIH informational site on autism; contains a wealth of detailed information on autism topics including treatments.

Autism Society

http://www.autism-society.org

Good site with information for/from voluntary organizations, including treatment options.

Developmental Milestones of Early Literacy

http://www.yic.gov/earlychildhood/learn2.html

Good resource for "normal" child development timeline.

Kimball's Biology Pages

http://users.rcn.com/jkimball.ma.ultranet/BiologyPages/

Very nice searchable version of introductory biology text, which has been adapted from print version by Dr. John W. Kimball, retired from Harvard University.

Society for Neuroscience—Autism in Children

http://www.sfn.org/index.aspx?pagename=brainBriefings_autismInChildren

Nice description of how autism is diagnosed.

AMA Health Care Careers Directory

http://www.ama-assn.org/ama/pub/education-careers/careers-health-care/directory.shtml

At this site, the American Medical Association presents free PDF files describing the expertise, day-to-day tasks, and training for various biomedical and medical professionals.

MEDLINE Plus Medical Dictionary

http://www.nlm.nih.gov/medlineplus/mplusdictionary.html

On-line medical dictionary.

On-line Biology Book: Glossary

http://www.emc.maricopa.edu/faculty/farabee/biobk/BioBookglossT.html

On-line biology glossary.

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