Autism: Myth, Legend & Science

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Modern Study of Autism

Leo Kanner: 1943 paper "Autistic Disturbances of Affective Contact"

Hans Asperger: 1944 paper “Autistic Psychopathy”
What is Autism?

- Autism is a complex *developmental disability* that causes problems with social interaction and communication.

- Symptoms usually start *before age three* and can cause delays or problems in many different skills that develop from infancy to adulthood.

- Different people with autism can have very different symptoms.

- Health care providers think of autism as a “spectrum” disorder, a group of disorders with similar features. For example, one person may exhibit mild symptoms, while another may show more serious symptoms. But they both have an autism spectrum disorder.
Autism: Just the facts

- Autism affects one in every 150 newborns.
- By 2015, an estimated 500,000 U. S. residents will be affected.
- Males predominate classic autism by 4:3 ratio.
- Males predominate the spectrum by up to a 14:1 ratio.
- Every person diagnosed with an Autism Spectrum Disease is different.
- Is disease without an obvious cause, creating wild and bizarre speculation.
- Autism may be treated, but not cured.
What is “the spectrum?”

Autism Spectrum Disorders

- **PDD-NOS**
  - Impaired social interaction
  - Impaired communication
  - Restricted repetitive and stereotyped patterns or behaviors, interests and activities

- **Autistic Disorder**
  - Impaired social interaction
  - Impaired communication
  - Restricted repetitive and stereotyped patterns or behaviors, interests and activities

- **Asperger’s Disorder**
  - Impaired social interaction
  - Normal communication/language development
  - Restricted repetitive and stereotyped patterns or behaviors, interests and activities

Burell Autism Center
Symptoms of autism

The main signs and symptoms of autism involve problems in the following areas:

**Communication** - both verbal (spoken) and non-verbal (unspoken, such as pointing, eye contact, and smiling)

**Social** - such as sharing emotions, understanding how others think and feel, and holding a conversation

**Routines or repetitive behaviors** (also called stereotyped behaviors) - such as repeating words or actions, obsessively following routines or schedules, and playing in repetitive ways
Ends of the spectrum: Classic autism

Rain Man (1988)
Ends of the spectrum: Asperger’s Syndrome

Parenthood (2010)
Successful people on the spectrum

Pokemon creator
Satoshi Tajiri

Nobel Prize Laureate
Vernon Smith, Ph.D.

Actress Daryl Hannah

Academy Award
winning
director/producer
Steven Spielberg

Comedian/Actor
Dan Aykroyd
**Legend:** Some historic figures exhibited behaviors consistent with autistic spectrum

Sir Isaac Newton  
Emily Dickenson  
Andy Warhol  
Albert Einstein  
W. B Yeats  
Charles Darwin

Lumrix.net/medial/autism
Diagnosis of autism spectrum disorders (including Asperger’s Syndrome)

- Is both clinical and syndromic.

- Protocol only recently codified, despite increasing numbers of people seeking treatment since 1960.

- **STAGE 1:** Developmental screenings begin with well-child checkups. More sensitive diagnostic screenings may be recommended, such as CHAT, M-CHAT, STAT, or SCQ.

- **STAGE 2:** Comprehensive Diagnostic Evaluation with a multidisciplinary team. Neurologic and genetic assessment, along with in-depth cognitive and language testing. Measures developed specifically for diagnosing autism are often used.
Before the dawn of modern science...

Coping with what humans could not understand often meant placing blame for unsociable behavior on:

- Witchcraft or demonic possession
- Displeased god(s)

Such behavior resulted in social marginalization, or perhaps death.

Even in an advanced scientific environment...

Beware of crackpots promising cure-alls, and spouting conspiracy theories...
Unsubstantiated cause of autism: cold parenting *(refrigerator mother)*

- Postulated by Kanner, popularized by Bruno Bettelheim.
- Expressed the belief that parents should not be involved with the therapy of autistic children as it would lead to parental guilt.
- Theory debunked by the work of Dr. Bernard Rimland and others.
- By the mid-1970’s, the “refrigerator mother” fell out of fashion.
Unsubstantiated cause of autism: vaccines

- 1998 article in *The Lancet* by Dr. Andrew Wakefield postulated that MMR vaccines caused higher rates of autism.
- Study could not be replicated, encompassed just 12 children and was financed by lawyers intending to file lawsuits against vaccine manufacturers.
- Created worldwide panic about the mercury-based preservative thimerisol (never in MMR vaccine).
- Anti-vaccination groups formed, including one lead by Robert F. Kennedy, Jr., have claimed a government cover-up of vaccine induced autism.
Unsubstantiated cause of autism: vaccines

- In February 2010, the British Medical Council ruled that Wakefield acted “dishonestly and irresponsibly.”
- Editors of The Lancet retracted the paper.
- Wakefield was forced to resign from his U. S. Institute that was the vehicle for promoting his disproved theories.
- “(Wakefield’s) improbable sequences of cause and effect were outclassed in every request by the impressive assembly of true experts in their respective field,” said Legal Special Master Denise Vowell.
Scientifically unsupported treatments for autism

- Secretin
- Chelation therapy
- Vision therapy
- Iridology

More scientific studies required for these treatments:

- Hyperbaric oxygen therapy
- Gluten-free/casein-free diet (not recommended by AAP)
- Vitamin & supplemental therapy
Treatment plans for autism should be individualized in consultation with a multidisciplinary team of family members and licensed, experienced care providers and educators, making use of scientifically proven methods for improvement of the condition.
The Science of Autism

Cerebral cortex - a thin layer of gray matter on the surface of the cerebral hemispheres. Two-thirds of its area is deep in the tissues of folds. Responsible for the higher mental functions, general movement, perception, and behavioral reactions.

Amygdala - responsible for emotional responses, including aggressive behavior.

Hippocampus - makes it possible to remember new information and recent events.

Basal ganglia - gray masses deep in the cerebral hemispheres that serve as a connection between the cerebrum and cerebellum. Helps to regulate automatic movement.

Major Brain Structures Implicated in Autism

Brain stem - located in front of the cerebellum. It serves as a relay station, passing messages between various parts of the body and the cerebral cortex. Primitive functions essential to survival (breathing and heart rate control) are located here.

Corpus callosum - consists primarily of closely packed bundles of fibers that connect the right and left hemisphere and allows for communication between the hemispheres.

Cerebellum - located at the back of the brain, it fine tunes our motor activity, regulates balance, body movements, coordination, and the muscles used in speaking.

National Institutes of Health
So what REALLY causes autism?

- No single cause has been identified.
- No single “trigger” that causes autism to develop.
- May be a series of events.
- Among the causes being investigated:
  - Viral infections
  - Environmental
  - Metabolic disorders, such as PKU
  - Genetics

Finding the cause of autism must be based upon scientifically valid investigation and research.
Genetics & Genomics

Genetics is the study of inheritance, or the way traits are passed down from one generation to another. Genes carry the instructions for making proteins, which in turn direct the activities of cells and functions of the body that influence traits such as hair and eye color.

Genomics is a newer term that describes the study of all the genes in a person, as well as interactions of those genes with each other and with that person's environment.

Key advances in genomic technologies are transforming all areas of human genetics.

Genetics and genomics are both considered opportunities (and challenges) for autism research.
What is a gene?

➢ A gene is a DNA sequence that contains the coding for making a particular protein.

➢ The average gene is ~3000 bases long.

➢ Some of the DNA sequence of a gene helps regulate the expression of the gene in our cells.
Genes are located on Chromosomes

- There are 45 bands on Chromosome 5
- Chromosome 5 contains 1005 genes
- Chromosome 5 is ~181,000,000 bases long
- Genes are referred to by their chromosomal location
  - The MCC gene is located at 5q21
  - MCC is a tumor suppressor gene for colon cancer
Chromosomes are highly condensed DNA

A chromosome is a single, very long piece of DNA.

Each chromosome is replicated and supercoils before cell division.
**Autism: The genetics**

- Genetic architecture of autism is complex, and not yet fully known.

- No single gene has been identified as the “autism” gene.

- Strongest finding for a genetic basis for autism:
  - **Identical twins:** If one is diagnosed autistic, the other has a 70%-90% chance of also being autistic.
  - **Fraternal twins:** Less than a 10% chance of the second twin being diagnosed with autism.
The Autism Genome Project (www.autismgenome.org) is a large-scale, collaborative genetics research project that aims to identify autism susceptibility genes.

The identification of autism risk factors requires large samples of well characterized individuals, and strong scientific cooperation between clinical and laboratory researchers.

Includes researchers from the US, Canada, UK, Portugal, Italy, Ireland, France and Germany.
Autism & Schizophrenia: Genetically Linked?

University of Leeds: Genetic studies so far are suggesting a common cause for both schizophrenia and autism (2010).

National Institute of Mental Health: brains of children with early-onset schizophrenia are much larger than normal in the first few years of life, for instance. Children with autism also have an unusual amount of brain growth before age 3. NIMH found two places where variations in genes tended to cluster in people with schizophrenia were also more common in people with autism (2008).
“A boy sits by himself on a stained white carpet, the corner of a frayed blanket stuffed into his mouth, his head bobbing, the fingers of both hands twiddling at ear level. He seems neither to miss company nor show any eagerness to seek it. If you say hello, he will not look at you nor turn in the direction of your voice. If you take up position in his line of sight, he will look away.”

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*Autism Spectrum Disorders in Early Childhood*, Carr, J. E., Primary care, June 2000 34(2); 343-59.