

# Autism: Myth, Legend & Science

Fifth Annual John and Valerie Rowe Lecture

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University of Connecticut

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Presented by J. Robert Galvin, MD, MPH, MBA

Commissioner, Connecticut Department of Public Health

Hartford, Connecticut



# Modern Study of Autism

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



Hans Asperger: 1944 paper "Autistic Psychopathy"



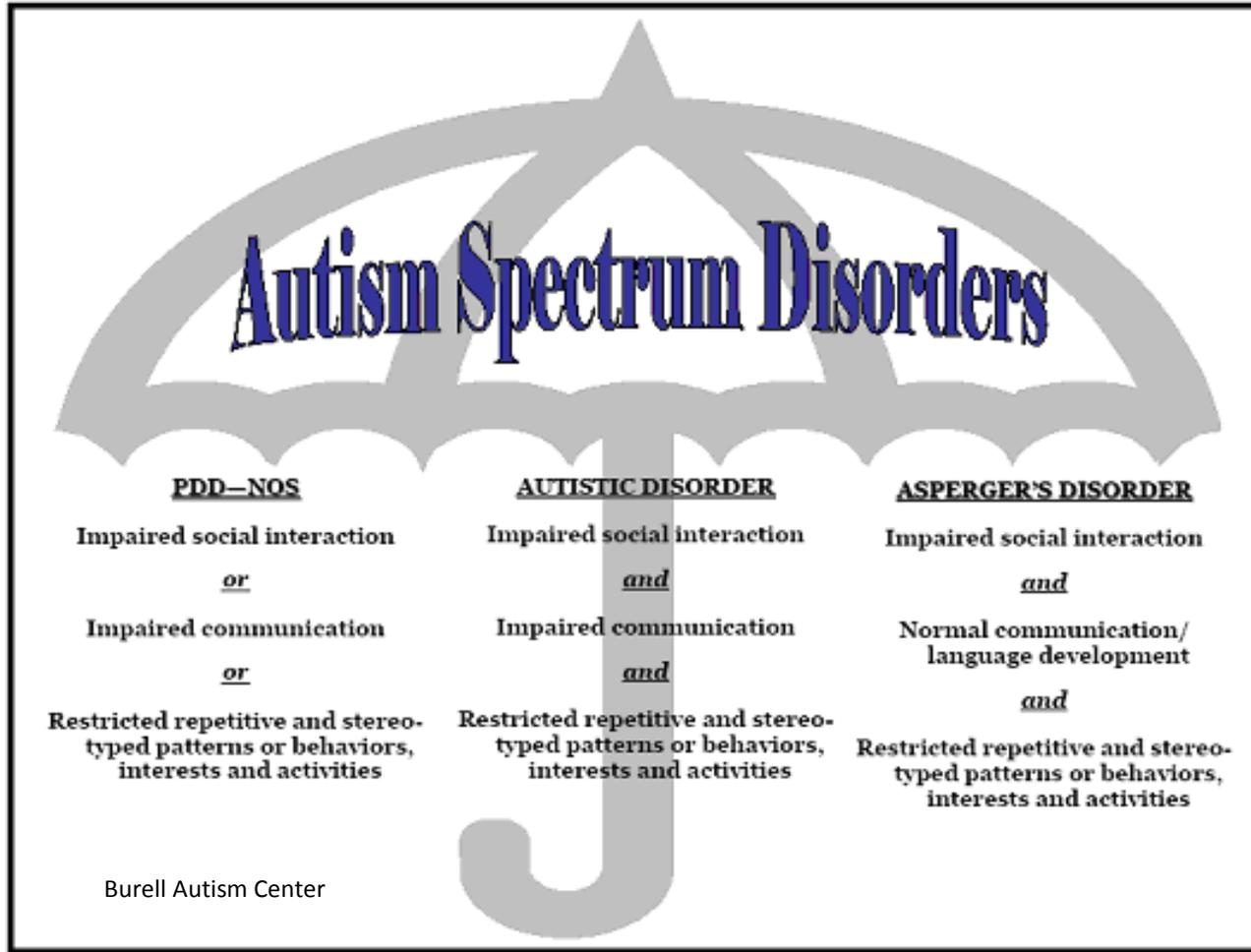
# *What is Autism?*

- 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?”



# *Symptoms of autism*

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

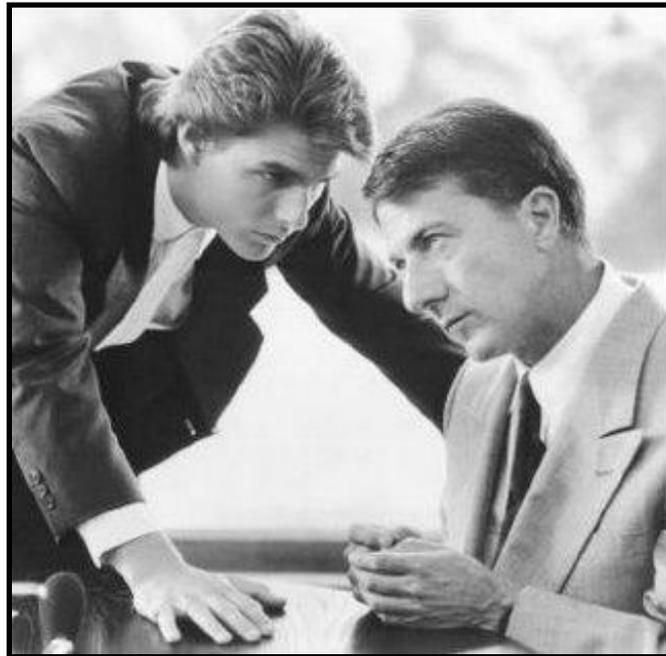
**Communication** - both verbal (spoken) and non-verbal (unspoken, such as pointing, eye contact, and smiling)

**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)

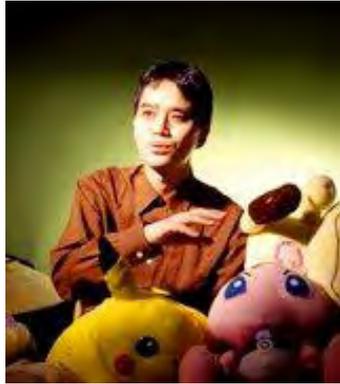


# *Ends of the spectrum: Asperger's Syndrome*

Parenthood (2010)



# *Successful people on the spectrum*



Pokemon creator  
Satoshi Tajiri



Nobel Prize Laureate  
Vernon Smith, Ph.D.



Actress Daryl Hannah

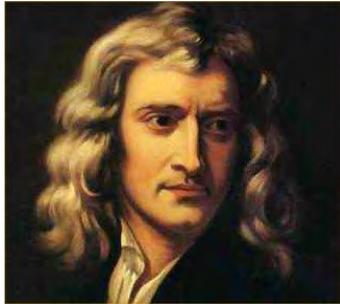


Academy Award  
winning  
director/producer  
Steven Spielberg



Comedian/Actor  
Dan Aykroyd

# **Legend:** Some historic figures exhibited behaviors consistent with autistic spectrum



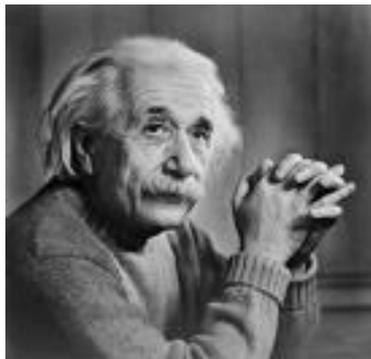
Sir Isaac Newton



Emily Dickenson



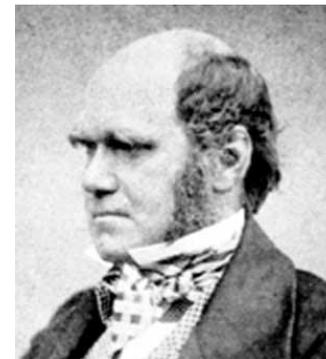
Andy Warhol



Albert Einstein



W. B. Yeats



Charles Darwin

# ***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

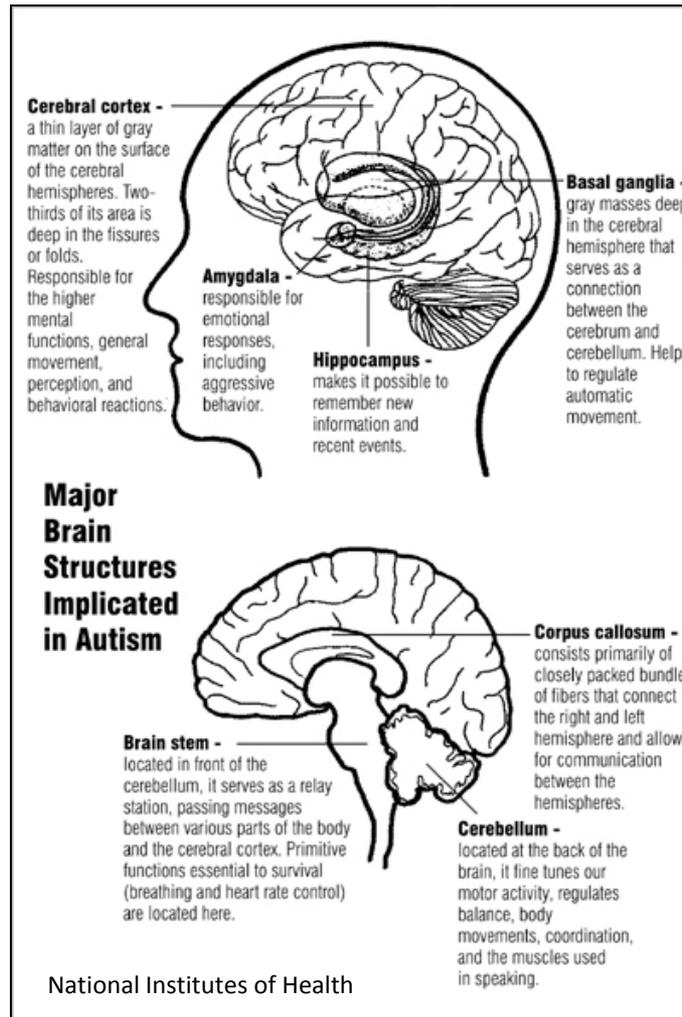
## ***More scientific studies required for these treatments:***

- 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



# ***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 a 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.

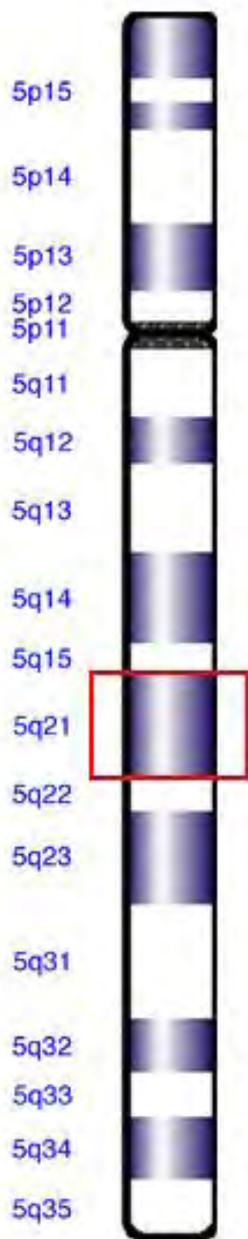
Key advances in genomic technologies are transforming all areas of human genetics.

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

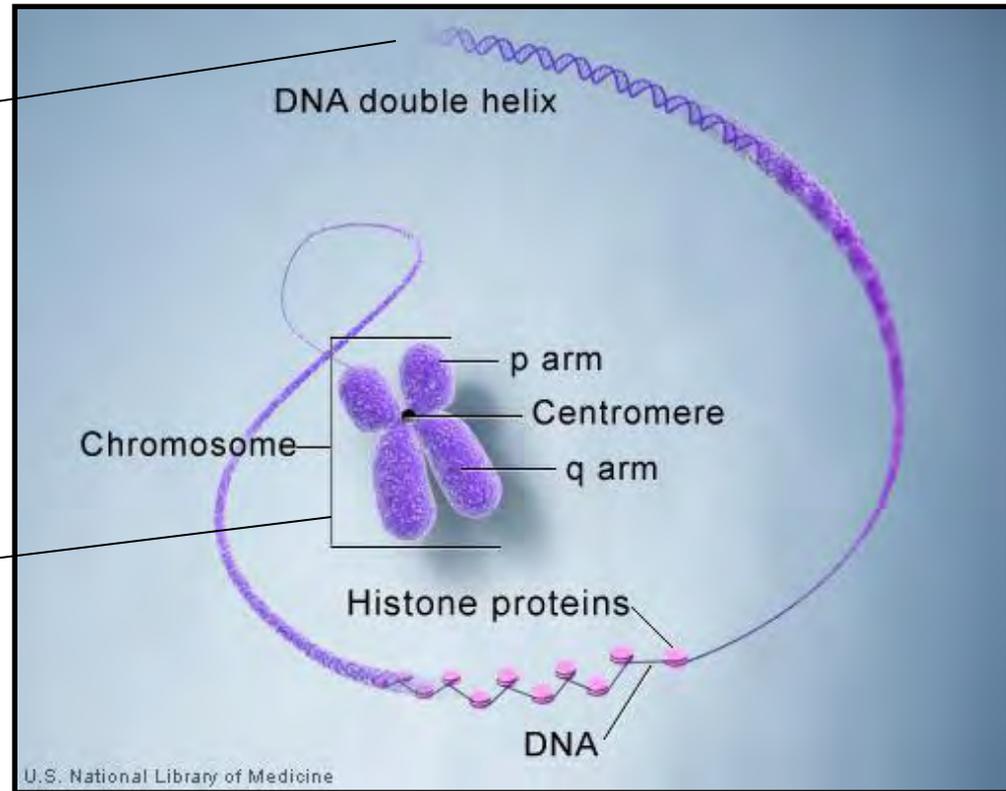


- There are 45 bands on Chromosome 5
- Chromosome 5 contains 1005 genes
- Chromosome 5 is ~181,000,000 bases long
- Genes are referred to by their chromosomal location
  - The *MCC* gene is located at 5q21
  - *MCC* is a tumor suppressor gene for colon cancer

# *Chromosomes are highly condensed DNA*

A chromosome is a single, very long piece of DNA

Each chromosome is replicated and supercoils before cell division

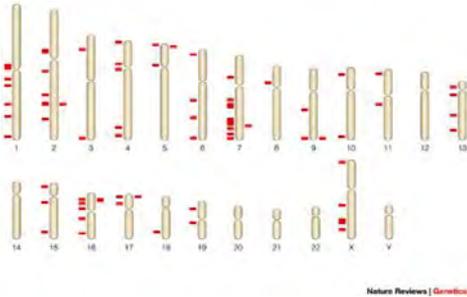


# *Autism: The genetics*

- 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](http://www.autismgenome.org))



**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.”*



- Karl Taro Greenfield, Boy Alone: A Brother's Memoir, HarperCollins, 2009

J. Robert Galvin, MD, MPH, MBA

Commissioner

Connecticut Department of Public Health

Hartford, Connecticut

(860) 509-7101

[Lynn.townshend@ct.gov](mailto:Lynn.townshend@ct.gov)



## ADDITIONAL CREDITS:

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