

Autistic Spectrum Disorders

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Autistic Spectrum Disorders

- A set of syndromes distinguished by:
 - impairments in social interaction.
 - impairments in communication
 - Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities.
- About 1 in 150 children has some form of ASD; overall, the ASD's occur 5-10 X more often in boys than in girls.

Commonly Seen in Individuals with ASD:

- Lack of “joint attention”
- Withdrawn behavior
- Unsocial behavior
- Problems or delays in using language to communicate
- Repetitive patterns of behavior
- Problems with imaginative play
- Difficulties in deviating from schedule
- Self-stimulating behavior

Medical Aspects of ASD

- In up to 30 % of ASD cases, ASD is diagnosed in DSM-5 as “ASD with known genetic or medical condition,” such as:
 - Fragile X syndrome (most common genetic cause of ASD and mental retardation; mutation in FMR1 gene on X chromosome)
 - Epilepsy
 - Cerebral palsy
 - Phenylketonuria (PKU)
 - Neurofibromatosis (“Elephant Man’s Disease”)
 - Trisomy 21 (Down Syndrome)
 - Congenital rubella (German measles)
 - Rett Syndrome (genetic cond., infantile deterioration)
- In 70% of ASD cases, no known medical disorder is apparent.

Varieties of ASD:

No Longer Diagnosed Separately in DSM-5

Normal Early Development, Then Deterioration

- Childhood Disintegrative Disorder
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Abnormal Early Development

- Autism
- Asperger's Syndrome
- PDD-Not Otherwise Specified (PDD-NOS); refers to a PDD that does not fit any of the previous three.

ASD: Childhood Disintegrative Disorder

- Normal development to age 3 or 4 or even later (up to age 10), then the child rapidly loses acquired skills such as language and self-care.
- Control over bowel and bladder habits may be lost.
- Declines may occur rapidly (over days or weeks) or more slowly.
- Older children may be able to vocalize concern (“Mommy, what’s happening to me?”)
- Condition is very rare (< 2 per 100,000 children), and no cause has been found.
- Result is profound autism and mental retardation, and patients become custodial; lifespan is normal if not complicated by seizures.

“Early Infantile Autism”

Leo Kanner

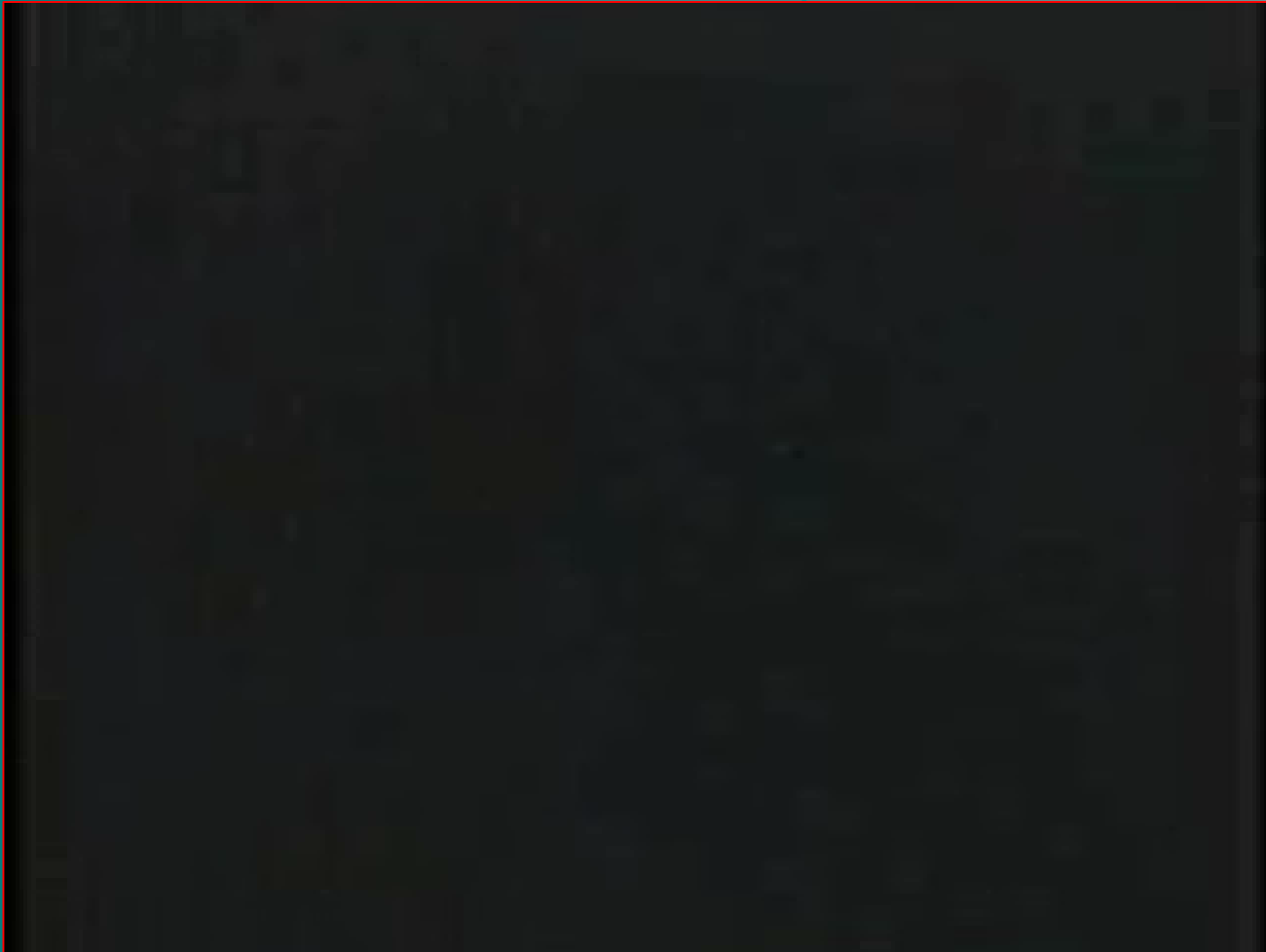


- The diagnosis of “Early Infantile Autism” was first used by psychiatrist Leo Kanner at Johns Hopkins University in 1943.
- Kanner, like many psychiatrists, believed “autism” to be an escape from reality, and so through the 1960’s, parents of autistic children were often blamed for their children’s disorder. The parents were accused of being contradictory communicators (“double-binding”) and the mothers were emotionally withholding (“refrigerator mothers”).
- The disorder was often confused with “childhood schizophrenia.”
- We now know that autism is a neurodevelopmental disorder that has nothing to do with child-rearing.

Signs of ASD: Autism

- Problems in verbal and nonverbal communication
 - No babbling, gesturing or pointing by 12 months
 - Failure to use single words by 16 months or 2-word phrases by 24 months
 - Echolalic (repeat) speech
 - Social withdrawal; no eye contact, social smiling or “joint attention”
- Sensory problems
 - Hyper-sensitive to touch, normal sounds
 - Absence of startle to loud noises
- Repetition and preference for sameness
 - Repetitive body movements (hand-flapping, finger-flicking)
 - Perseveration (gets stuck on a specific topic or task)
 - Very narrow set of interests
 - Sometimes, self-injurious behavior (head-banging, skin-chewing)

ASD: Autism as a Congenital Disorder



ASD: Adult Autism in *Rain Man* (1988)



What Goes Wrong in ASD?

- Probably a number of different syndromes with various etiologies.
- Between 600-1200 genes may contribute to the risk for autism, as well as gene copy number variants (CNV's) and chromosomal abnormalities.
- Concordance of 70-95% for MZ twins, and 0-24% for DZ twins.
- *De novo* gene mutations, especially in the sperm of older fathers, also raise the risk of autism.
- Brain findings show an over-abundance of neurons in the autistic brain beginning *in utero*; in the prefrontal cortex, autistic brains have 67% more neurons than average, and there is over-wiring of local connections among the neurons.

Controversies in ASD

Q: Is there an Autism Epidemic?

A: In 2000, the estimated prevalence of autism was 1 in 500 children; by 2010 it was 1 in 110. Most experts, however, do not believe there is an actual increase. Instead, they believe that more children are being diagnosed with autism instead of other disorders such as “mental retardation” because of increased awareness of the disorder.

Q: Do vaccines, or vaccines containing mercury, cause autism?

A: Despite the advocacy of certain Hollywood celebrities (Jenny McCarthy), the answer here is a definite NO! The major study on which this myth was based was discovered to be a fraud. The research article reporting it was retracted, and the author lost his medical license. Vaccines are safe, and thousands of children have suffered or died needlessly from measles, mumps, and other childhood diseases because their parents began to withhold childhood vaccinations.

ASD: Asperger's Syndrome

- First identified by Austrian pediatrician Hans Asperger in 1944, but not well-known until 1980's.
- Like autism (social impairment and repetitive interests and behavior), but intelligence is normal and speech is formally correct. However:
 - Empathic responding is deficient.
 - Social contacts are awkward and lacking in routine “manners.”
 - Routines are maintained insensibly and inflexibly.
 - Speech patterns are stilted and lecture-like, monotonous, or fixated on certain interests (air travel timetables, weather patterns, automobile engine details).
 - Sometimes: odd posturing, nervous tics, hand-flapping, violent outbursts, sensory hypersensitivity.
- No clear demarcation from “high-functioning autism,” hence Asperger's Syndrome was folded with autism into Autism Spectrum Disorders in DSM-5.

People Speculated to Have / Have Had Asperger's

- Isaac Newton
- Hans Christian Andersen
- Abraham Lincoln
- Alan Turing
- Albert Einstein
- Alexander Graham Bell
- Anton Bruckner
- Bela Bartok
- Benjamin Franklin
- Bertrand Russell
- Bobby Fischer
- Carl Jung
- Emily Dickinson
- George Orwell
- Erik Satie
- Franz Kafka
- Henry Ford
- Nikola Tesla
- Alfred Hitchcock
- Andy Kaufman
- John Nash
- Andy Warhol
- Charles Schulz
- Glenn Gould
- Hans Asperger
- Howard Hughes
- Isaac Asimov
- Jim Henson
- John Denver
- Bill Gates
- Bob Dylan
- Kevin Mitnick
- Keith Olbermann
- Oliver Sacks
- James Taylor
- Mark Zuckerberg

Asperger's Syndrome on TV



**the BIG
BANG
THEORY**





Temple Grandin, Ph.D.
Professor of Animal Science
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Temple Grandin
[HBO Films, 2010]

Temple Grandin
[TEDxDU, May 2011]



Go to www.templegrandin.com

How Are ASD's Treated?

- **Early Behavioral Intervention:**
 - Educational: structured, 1:1 teaching environment; initial focus on language then more general academics; high reinforcement density
 - Self-care
 - Social skills
 - Autism: play and group behavior, training to make eye-contact, facial expressions, and sustain attention
 - Asperger's: Social-script training, management of relationships, vocational and sexuality counseling
- **Medication:**
 - Ritalin for “ADHD”-like behavior
 - Risperdal for irritability
 - Sometimes, Depakote for violent outbursts
 - Medications for seizures and other medical problems (>30%)
 - No other medications show reliable effects
- **Outcomes of treatment are unpredictable and vary on case-by-case basis**
- **Long-term custodial care for lower-functioning cases**
- **Financial and emotional costs of interventions and continuing care are often overwhelming or impossible for families**

End

Rett's Syndrome

- Usually seen only in girls (boys with the disorder usually die before birth and the mother miscarries, or are born and die within 2 years).
- Caused by a spontaneous mutation in the MECP2 (methyl cytosine binding protein 2) gene on the X chromosome (parents are usually normal).
- Development is normal until 6-18 months, then language and motor abilities are lost:
 - Loss of use of hands
 - Loss of speech
 - Inconsolable crying and screaming fits
 - Withdrawal and loss of eye contact.
- People with Rett's soon become custodial and live to 40 years or more.
- Animal studies suggest that early detection and MECP2 therapy can reverse the deficits caused by the defective gene, and efforts to extend these findings to humans are now underway.

“Asperger” Speech: *Big Bang Theory* [S04E10]

