UPDATE:
AUTISM SPECTRUM DISORDER IN CHILDREN
HISTORY OF AUTISM

• ABLE TO TRACE THE CHANGING APPROACHES TO AUTISM BY LOOKING AT DSM CRITERIA OVER TIME

• IN THE BEGINNING, IT WAS PERCEIVED AS A SINGULAR DISORDER CALLED “AUTISM”

• **1943**: FIRST DESCRIBED BY LEO KANNER
  - PUBLISHED “DISTURBANCES OF AFFECTIVE CONTACT”
HISTORY OF AUTISM

• 1950-1970: PSYCHOANalytic Approach
  ❖ Mothers Blamed
  ❖ Categorized as “Cold”
  ❖ Called the Refrigerator Parent
HISTORY OF AUTISM

• 1952: DSM-I
  ❖ AUTISM LISTED WITH SCHIZOPHRENIA

• 1968: DSM-II
  ❖ AUTISM GIVEN BRIEFER CONNECTION TO SCHIZOPHRENIA
HISTORY OF AUTISM

1980: DSM-III

- LISTED AUTISM UNDER PDD
  (PERVASIVE DEVELOPMENT DISORDER)
- ONSET BEFORE 30 MONTHS
- DELAYED OR ABNORMAL SPEECH MUST BE PRESENT
HISTORY OF AUTISM

• 1987: DSM-III REVISION
  - CHANGED INFANTILE AUTISM TO “AUTISTIC DISORDER”
  - ADDED “PDD NOS”
HISTORY OF ASD

• 1994: DSM-IV
  ❖ ADDED TERM “ASPERGER SYNDROME”
  ❖ DID NOT RECOGNIZE SPEECH DELAY ASSOCIATION
  ❖ INTRODUCED A “SPECTRUM” OF SYMPTOMS, RATHER THAN A SINGULAR DEFINITION
HISTORY OF ASD

• 2013: DSM-V
  ❖ ELIMINATED ASPERGER SYNDROME & PDD NOS
  ❖ PLACED ALL VARIETIES OF PDD UNDER THE UMBRELLA OF “AUTISM SPECTRUM”
HISTORY OF ASD

• AUTISM NOW REFERRED TO “AUTISM SPECTRUM DISORDER”

• INCLUDES:
  • AUTISTIC DISORDER
  • ASPERGER SYNDROME
  • PERSVATIVE DEVELOPMENT DISORDER
MYTHS ABOUT AUTISM
MANY MYTHS ABOUT AUTISM

THE ORGANIZATION “AUTISM SPEAKS” HAS IDENTIFIED 11 MYTHS. WE WILL LOOK AT 5.
1. **Autism is over diagnosed.**

- 2014: 1/68 children diagnosed with ASD
- 1992: 1/600 children diagnosed with Autism
- Increased # of children with Autism?
- Diagnosing better?
- Diagnosis has widened

Centers for Disease Control & Prevention (2012); Autism Speaks 11 Myths of Autism
1. **THE CHILD WITH AUTISM DOES NOT WANT FRIENDS**

**TRUTH:** THE CHILD WITH AUTISM MAY STRUGGLE WITH SOCIAL SKILLS, WHICH MAKES IT DIFFICULT FOR THEM TO KNOW HOW TO INTERACT WITH PEERS.
2. **THE CHILD WITH AUTISM CAN'T FEEL OR EXPRESS EMOTION-HAPPY OR SAD**

**TRUTH**: CHILDREN WITH AUTISM MAY COMMUNICATE AND PERCEIVE EMOTIONS DIFFERENTLY.
3. THE CHILD WITH AUTISM CAN'T UNDERSTAND THE EMOTIONS OF OTHERS

**TRUTH:** CHILDREN WITH AUTISM MAY NOT UNDERSTANDING TONE OR NON-VERBAL CUES, BUT THEY ARE OFTEN MORE LIKELY TO FEEL COMPASSION FOR OTHERS.
4. **CHILDREN WITH AUTISM ARE INTELLECTUALLY DISABLED.**

**TRUTH:** MOST CHILDREN WITH AUTISM HAVE NORMAL TO HIGH IQ’S AND OFTEN EXCEL IN CERTAIN AREAS.
MYTHS

4. THE CHILD WITH AUTISM CANNOT PROGRESS TOWARDS IMPROVED FUNCTION.

**TRUTH:** RECENT RESEARCH HAS SHOWN THAT CHILDREN WITH AUTISM CAN MAKE SIGNIFICANT GAINS AFTER EARLY INTENSIVE INTERVENTION.
5. **MYTHS**

THE CHILD WITH AUTISM HAS SIGNS & SYMPTOMS THAT SHOW AUTISM ONLY AFFECTS THE BRAIN.

**TRUTH**: RESEARCH SHOWS US THAT MANY CHILDREN WITH AUTISM ALSO HAVE GI DISORDERS, FOOD SENSITIVITIES AND MANY ALLERGIES.
NIH Definition of ASD

• The name for a group of developmental disorders.

• Includes a wide range, “a spectrum,” of symptoms, skills, and levels of disability.

• Problems with social, emotional and communication skills.
NORMAL VARIATION VS ABNORMALITY?

• **EINSTEIN** - DID NOT SPEAK UNTIL AGE 3

• **STEVE JOBS** - A LONER WHO BROUGHT SNAKES TO SCHOOL

• WERE THEY AUTISTIC OR JUST DIFFERENT?
3 KEY FEATURES IN ASD

1. DEFICIT IN SOCIAL RECIPROCITY

2. COMMUNICATION IMPAIRMENT

3. RESTRICTED OR REPETITIVE PATTERNS OF BEHAVIOR, INTERESTS OR ACTIVITIES
DEFICIT IN RECIPROCAL SOCIAL INTERACTION

- Marked impairment of nonverbal behaviors
- Failure to develop age appropriate peer relationships
- Lack of social-emotional reciprocity
- Lack of spontaneous seeking to share interests, achievements or enjoyments
COMMUNICATION IMPAIRMENT

• ABSENT OR DELAYED LANGUAGE WITHOUT ATTEMPTING TO COMPENSATE
• MARKED IMPAIRMENT IN ABILITY TO SUSTAIN CONVERSATION
• STEREOTYPIC OR REPETITIVE LANGUAGE USAGE
  • ECHOLALIA
• LACK OF FANTASY OR MAKE BELIEVE
RESTRICTED OR REPETITIVE BEHAVIORS

- **RESTRICTED INTEREST** with abnormal focus or intensity
- INSISTENCE ON **ROUTINES**
- STEREOTYPIC MOTOR **MANNERISMS**
- HYPER OR HYPO RESPONSE TO **SENSORY INFORMATION**
  - MAY NOT FEEL PAIN BUT MAY HAVE INTENSE RESPONSE TO SMELL
THERE ARE ALSO SUB-TYPES OF ASD

IDIOPATHIC
SECONDARY
IDIOPATHIC

• MEETS CRITERIA FOR ASD WITH NO COMORBID MEDICAL CONDITIONS

• LESS LIKELY TO HAVE GDD, MR, OR DYSMORPHIC FEATURES

• ACCOUNTS FOR MOST CHILDREN WITH ASD
SECONDARY

• HAVE AN IDENTIFIABLE CONGENITAL “SYNDROME”

• PART OF ANOTHER MEDICAL DISORDER

• LESS THAN 10% OF ASD CHILDREN
DISORDERS WITH SECONDARY ASD

- **FRAGILE X**: RANGE OF DEVELOPMENTAL & COGNITIVE
- **PKU**: PHENYLKETONURIA
- **FAS**: FETAL ALCOHOL SYNDROME
- **RETTS DISORDER**: LOSS OF DEVELOPMENTAL MILESTONES AFTER AGE 6
- **ANGELMAN SYNDROME**: CONGENITAL SYNDROME WITH GDD, SEIZURES, BALANCE DISTURBANCE
INTERESTING RESEARCH BY TEMPLE GRANDLIN
WHO IS DR. TEMPLE GRANDLIN

• DID NOT SPEAK UNTIL AGE 3 AND WAS DIAGNOSED WITH AUTISM

• WROTE A BOOK - MERGENCE: LABELED AUTISTIC

FIRST BOOK WRITTEN FROM AN INSIDE NARRATIVE OF AUTISM
WHO IS DR. TEMPLE GRANDLIN?

• INTERESTING:
  • HAS STUDIED AUTISM THROUGHOUT HER LIFETIME
  • NATIONALLY RECOGNIZED SPEAKER ON AUTISM
  • INDUCTED INTO THE AMERICAN ACADEMY OF ARTS & SCIENCES IN 2016 FOR HER WORK
MAJOR CONSTRUCT IN HER THEORY

• BELIEVES THAT MOST CHILDREN WITH AUTISM HAVE "SPECIALIZED BRAINS"

• IDENTIFIES 4 DIFFERENT SPECIALIZED BRAIN TYPES

• BELIEVES THERE CAN BE A MIX OF THE 4 TYPES
MAJOR CONSTRUCT IN HER THEORY

• 4 “SPECIALIZED BRAIN TYPES”:
  1. PHOTO REALISTIC THINKING
  2. PATTERN THINKING
  3. VERBAL FACTS LANGUAGE TRANSLATION THINKING
  4. AUDITORY THINKER
PHOTO REALISTIC THINKING

- VIEW THE WORLD THROUGH PICTURES
- EVERYTHING THEY THINK ABOUT HAS A PHOTO REALISTIC PICTURE
- LEARN BY SPECIFIC EXAMPLES THAT ARE PLACED IN CATEGORIES
- LOVE ART; POOR AT ALGEBRA
- EXCELLENT AT SKILLED TRADES...AUTO REPAIR EX.
PATTERN THINKER

- Spatial Visualization of World
- Love Patterns
- Great at Music and Math
- Poor at Reading and Art
VERBAL FACTS LANGUAGE TRANSLATION

• GOOD AT LANGUAGE COMPREHENSION
• LOVE TO DESCRIBE INTERESTS IN GREAT DETAIL
• LOVE READING
• LOVE LISTS & NUMBERS
• POOR AT DRAWING
• MEMORIZE TABLES & EVENTS IN HISTORY
AUDITORY THINKER

• VISUAL PERCEPTION IS FRAGMENTED

• SEE WORDS LITERALLY VIBRATING & JUGGLING ON PAGE

• RESPOND DRAMATICALLY TO LOUD NOISES AND SOUNDS
MORE FROM TEMPLE GRANDLIN’S RESEARCH

• MOST CHILDREN WITH AUTISM:
  ❖ HAVE **DIFFICULTY SHIFTING ATTENTION** BACK AND FORTH BETWEEN 2 DIFFERENT THINGS
  ❖ **STRUGGLE WITH SEVERE ANXIETY** BECAUSE THE AMYGDALA OF THE BRAIN IS IMPACTED BY AUTISM...
MORE FROM TEMPLE GRANDLIN’S RESEARCH

AMYGDALA:

• The center of the fear response in our brain
MORE FROM TEMPLE GRANDLIN’S RESEARCH

• MANY CHILDREN WITH AUTISM ALSO HAVE SENSORY PROCESSING DISORDERS
  ✤ DYSLEXIA
  ✤ LEARNING DISABILITIES
  ✤ ADHD
MORE FROM TEMPLE GRANDLIN’S RESEARCH

• MANY CHILDREN WITH AUTISM HAVE AUDITORY PROCESSING DISORDERS
  ✿ LOUD SOUNDS HURT THEIR EARS
  ✿ HEARING MAY FADE IN AND OUT LIKE A BAD PHONE
  ✿ DIFFICULTY HEARING HARD CONSONANTS
MORE RESEARCH NEEDED

• SHE GIVES SOME INTERESTING THINGS FOR ALL OF US TO CONSIDER WITH ASD.

• OBVIOUSLY… MORE RESEARCH NEEDED
WHAT CAUSES ASD?
WHAT WE KNOW FOR SURE: VACCINES

IT IS NOT CAUSED BY IMMUNIZATIONS!
WHAT WE KNOW FOR SURE

• 1993: DR. ANDREW WAKEFIELD (& COLLEAGUES) WROTE A PAPER THAT IMPLIED A SERIES OF EVENTS LED TO THE DEVELOPMENT OF AUTISM AFTER MMR

• DR. WAKEFIELD WAS AN ADULT GASTROENTEROLOGIST
WHAT WE KNOW FOR SURE

- THIS SERIES OF EVENTS INCLUDED:
  - DEVELOPMENT OF INFLAMMATORY BOWEL DISEASE
  - LOSS OF INTESTINAL BARRIER FUNCTION
  - ENTRANCE INTO THE BLOODSTREAM OF ENCEPHALOPATHIC PROTEINS
MMR CAUSES AUTISM MYTH
CRITICAL FLAWS IN STUDY!

- SMALL SAMPLE IN STUDY

- NO BLINDING IN THE STUDY

- EXPOSED & CONTROL GROUP DID NOT COME FROM SAME POPULATION
MMR CAUSES AUTISM MYTH CRITICAL FLAWS IN STUDY!

- DIFFERENT CONTROLS WERE USED FOR DIFFERENT TECHNIQUES IN THE STUDY

- SUBSEQUENT STUDIES USING MOLECULAR AMPLIFICATION TECHNIQUES FAILED TO DETECT MEASLES VIRUS GENES IN A SINGLE INFLAMMATORY BOWEL DISEASE PATIENT
MMR CAUSES AUTISM MYTH CRITICAL FLAWS IN STUDY!!!

- **DIAGNOSTIC CRITERIA WERE NOT DEFINED**

- **GI SYMPTOMS WERE OBSERVED** AFTER SX’S OF AUTISM PRESENT.
MMR CAUSES AUTISM MYTH CRITICAL FLAWS IN STUDY!!!

- The autistic children were reported to have low levels of IGA, when levels reported were actually in normal range using pediatric reference.

- Intestinal nodular hyperplasia reported in study is actually a variant of normal in young children.
MMR CAUSES AUTISM MYTH CRITICAL FLAWS IN STUDY!!!

- JOURNAL OF PEDIATRIC ALLERGIES 2-2-2015

CONCLUSIONS:

- INTESTINAL LNH SHOULD BE CONSIDERED A BENIGN FINDING IN CHILDREN WITHOUT RED FLAGS, BECAUSE THE SYMPTOMS ARE LARGELY SELF-LIMITING.

- JOHN WILEY & SONS A/S. PUBLISHED BY JOHN WILEY & SONS LTD.
MMR CAUSES AUTISM MYTH CRITICAL FLAWS IN STUDY!!!

RETRACTIONS

▪ 10 OUT OF 12 AUTHORS OF STUDY RETRACTED THE LINK TO AUTISM

▪ “WE WISH TO MAKE IT CLEAR THAT IN THIS PAPER NO CAUSAL LINK WAS ESTABLISHED BETWEEN MMR VACCINE AND AUTISM AS THE DATA WERE INSUFFICIENT.”

▪ LANCET VOL. 363, NUMBER 9411, 3/6/04
MMR CAUSES AUTISM MYTH CRITICAL FLAWS IN STUDY!!!

• DR. WAKEFIELD WOULD NOT RETRACT
  • HE LOST HIS MEDICAL LICENSE
  • AN INTERVIEW IN 2015: HE STILL MAINTAINED HIS STUDY WAS GOOD AND THE FINDINGS WERE RELIABLE
  • DIED IN 2016
  • SINGLE-HANDEDLY LED MILLIONS TO SUSPICION ABOUT IMMUNIZATIONS
WHAT CAUSES ASD?

**GENETICS:**
- YES!
- YES... A SINGLE GENE
- YES... A COMBINATION OF GENES INTERACTING TOGETHER
- YES... GENE MUTATIONS
- IMPLICATED GENETIC SITES ON CHROMOSOMES: 2, 3, 6, 7, 13, 15, 16, 17, 22
WHAT CAUSES ASD?

**GENETIC MUTATIONS**

• 73 SPECIFIC MUTATIONS CURRENTLY KNOWN
• THOUGHT TO BE MORE THAN 1000 POSSIBLE
• GENETIC MUTATIONS MAY NOT BE PRESENT IN OTHER FAMILY MEMBERS
• SIBLINGS WITH AUTISM MAY HAVE DIFFERENT MUTATIONS
WHAT CAUSES ASD?

OTHER RISK FACTORS

• INCREASING PATERNAL AGE & INFECTION DURING PREGNANCY GETTING MORE ATTENTION

• EXPOSURE TO SOME ANTI-CONVULSIVE MEDS & ANTIBIOTICS IN UTERO
WHAT CAUSES ASD?

• THERE IS MUCH WE DO NOT YET FULLY UNDERSTAND
DIAGNOSING AUTISM
EARLY IDENTIFICATION OF ASD
AAP POLICY STATEMENT 2007

• ASD IS PRESUMABLY PRESENT AT BIRTH
• ONSET OF SYMPTOMS BEFORE 36 MONTHS
• ACCURATE DX POSSIBLE AT 18-24 MOS
• PARENTS FIRST VOICE CONCERNS AROUND 18 MOS, BUT DX TYPICALLY NOT UNTIL AROUND 3 YRS.
• CHILD CAN BENEFIT FROM EARLY DX!
AAP GUIDELINES FOR EARLY IDENTIFICATION FOR AUTISM

• 2007 AAP STATEMENT
  • DEVELOPMENTAL SURVEILLANCE AT EVERY VISIT
  • ROUTINE SCREENING AT 18 & 24 MONTHS USING **MCHAT** FOR YOUNGER CHILDREN
  • SENSITIVITY: 85% - TRUE POSITIVES
  • SPECIFICITY: 93%-TRUE NEGATIVES
DIAGNOSING ASD
AAP POLICY STATEMENT 2007

• "SURVEILLANCE" AT EVERY VISIT

• LOOKING FOR RISK FACTORS
  • SIBLINGS WITH ASD

• PARENT CONCERN

• PEDIATRICIAN CONCERN
DIAGNOSING ASD
AAP POLICY STATEMENT 2007

• ASD SCREENING:

  • **IF OLDER THAN 1**: USE SCREENING TOOL: MCHAT
  
  • **IF OLDER THAN 2**: REFER FOR ASD AND AUDIOLOGY EVALUATION SIMULTANEOUSLY
DIAGNOSING ASD
AAP POLICY STATEMENT 2007

**M-CHAT SCREENING:**

- FOR CHILDREN 16-48 MONTHS
- QUESTIONNAIRE COMPLETED BY PARENT
- SENSITIVITY: 85%
- SPECIFICITY: 93%
- AVAILABLE ONLINE WITH SCORING INSTRUCTIONS
DIAGNOSING ASD

OTHER DIAGNOSTIC TOOLS

• CARS: CHILDHOOD AUTISM RATING SCALE
  • FOR ≥ 2 YRS
  • 15 ITEM DIRECT OBSERVATION
  • TAKES 5-10 MINS
DIAGNOSING ASD
OTHER DIAGNOSTIC TOOLS

• ADOS: AUTISM DIAGNOSTIC OBSERVATION SCHEDULE
  • FOR TODDLERS TO ADULTS
  • DIRECT OBSERVATION
  • 30-45 MINS
DIAGNOSING ASD

OTHER DIAGNOSTIC TOOLS

• ADI-R: AUTISM DIAGNOSTIC INTERVIEW
  • FOR MENTAL AGE OVER 2 YRS
  • STRUCTURED INTERVIEW PROCESS
  • 1.5-2.5 HRS.
EARLY DIAGNOSIS OF AUTISM
KENNEDY KRIEGER INSTITUTE

- INSTITUTION DEDICATED "TO IMPROVING THE LIVES OF CHILDREN AND YOUNG ADULTS WITH PEDIATRIC DEVELOPMENTAL DISABILITIES AND DISORDERS OF THE BRAIN, SPINAL CORD AND MUSCULOSKELETAL SYSTEM"

- AUTISM FOCUS: EARLY DIAGNOSIS!
DIAGNOSING AUTISM

KENNEDY KREIGER NOTES CHANGES EVEN IN TODDLERS

• THE TODDLER NON ASD CHILD
  • WILL ENGAGE OTHERS IN PLAY
  • SHOW MEANINGFUL PURPOSEFUL AND PRETEND PLAY
  • SHARES ENJOYMENT BY SMILING
DIAGNOSING AUTISM

KENNEDY KREIGER NOTES CHANGES EVEN IN TODDLERS

• THE TODDLER NON ASD CHILD
  • SHIFTS GAZE FROM ONE PERSON TO ANOTHER
  • WILL LOOK IN THE DIRECTION THE SCREENER POINTS
  • COORDINATES GAZE, VOCALIZATION & GESTURE TO COMMUNICATE
EARLY DIAGNOSIS OF AUTISM

KENNEDY KREIGER NOTES DIFFERENCES EVEN IN TODDLERS

• THE TODDLER ASD CHILD
  • UNUSUALLY STRONG INTEREST IN OBJECT - NOT PERSON
• DOES NOT ENGAGE PEOPLE DURING PLAY
• NO RESPONSE TO NAME
EARLY DIAGNOSIS OF AUTISM

KENNEDY KREIGER NOTES CHANGES EVEN IN TODDLERS

• THE TODDLER ASD CHILD
  - No sharing of enjoyment
  - Hand flapping & unusual tensing of the body
  - Does not gaze in direction screener points
EARLY INTERVENTION IS SO IMPORTANT! THE SOONER WE BEGIN TO DO THIS, THE BETTER THE OUTCOME.

LAST… WE STILL HAVE SO MUCH TO LEARN ABOUT AUTISM
CARLY 20/20 VIDEO
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