Autism Spectrum Disorder

*Early Diagnosis & Early Intervention*

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Today we will talk about

- Defining ASD
- New Numbers/Prevalence
- DSM-5
- Core Symptoms/ Co-occurring Disorders
- Identification of ASD
- Treatment of ASD
  - Applied Behavior Analysis (ABA)
  - Relationship-Based Methods
Defining: Autism Spectrum Disorder

1943 – Leo Kanner – Infantile autism
1944 – Hans Asperger
1960s – Separation from schizophrenia
1970s – Biology / Genetic Underpinnings
1980 – DSM-III – Pervasive Developmental Disorders
1987 – DSM-III-R - Autistic Disorder / PDD-NOS
1994 – DSM-IV – Asperger’s Disorder
2013 – DSM-5 – Autism Spectrum Disorder
The nature of Autism Spectrum Disorder

Stayed tuned for more on DSM-5 later!
Prevalence:

What once was rare...

- Old estimate for autism:
  - ~ 1/2500 (1985)

- Recent estimates for autism:
  - ~ 1/500 (1995)

- Newest estimates for ASD:
  - 1/150 (CDC, 2002)
  - 1/110 (CDC, 2006)
  - 1/88 (CDC, 2008)

- NOW - 1/68 (CDC, 2010)
(March 27, 2014) – Today, the Centers for Disease Control and Prevention (CDC) released new data on the prevalence of autism in the United States. This surveillance study identified 1 in 68 children (1 in 42 boys and 1 in 189 girls) as having autism spectrum disorder (ASD).
Distinctions can be difficult both within the spectrum and across other disorders
Deficits in social communication (all 3):
- Deficits in nonverbal communication
- Deficits in social and emotional reciprocity
- Deficits in maintaining relationships

Restricted, repetitive patterns of behavior, interest, and activities (2)
- Stereotyped motor or verbal behavior
- Unusual sensory behavior
- Excessive adherence to routines and ritualized behaviors
- Restricted, fixated interests

Symptoms present in early childhood (manifest when social demands exceed capabilities)
Core Symptom Domains
PLUS Associated Medical Features

AUTISM SPECTRUM DISORDERS
& Restricted Interests

Speech/Communication Deficits

Social Impairment

Intellectual Disabilities

Epilepsy-EEG abnormalities

Social Anxiety

OCD

Language Disorders

Motor problems: Apraxia

Sleep Disturbance

Gastro-intestinal Dysfunction

Aggression

ADHD

Immune Dysfunction

OCD

Obsessive Compulsive Disorder

Epilepsy-EEG abnormalities

Social Anxiety

OCD

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Epilepsy-EEG abnormalities

Social Anxiety

OCD

Obsessive Compulsive Disorder
Autism Spectrum Disorders (ASD)

Core autism spectrum disorder symptoms

- Impaired social communication/interaction (3/3):
  - Social reciprocity
  - Nonverbal communication
  - Relationships

- Restricted/repetitive behavior (2/4):
  - Repetitive speech/behavior
  - Insistence on sameness
  - Restricted interests
  - Sensory abnormalities

BEHAVIORAL COMORBIDITIES
- Hyperactivity/impulsivity
- Agitation/aggression
- Anxiety
- Severe constipation
- Seizure disorder
- Abnormal EEG

COGNITIVE COMORBIDITIES
- Language impairment
- Intellectual disability

GENETICS
- Simple genetic disorders: fragile X, TS, Rett, etc.
- Copy number variants: 16p11-p12, 15q11-q13, 22q13, etc.
- Rare variants: NRXN1, NLGN4, Shank3, Sert, etc.

MEDICAL COMORBIDITIES
- Seizure disorder
- Abnormal EEG
- Neuroimaging: altered brain region size

BIOMARKERS
- Neuroimaging: altered brain region size
- Neurotransmitter alterations
- Altered immune/mitochondrial indices

Hyper-serotonemia

Altered immune/mitochondrial indices
Core Deficit
Core Deficit
Core Deficit
Frontal networks in adults with autism spectrum disorder


Figure 3: The anatomy of the limbic tracts in relation to childhood ASD symptoms. (A) Tractography reconstructions of the limbic pathways. (B) Negative correlation between the number of streamlines of the left uncinate fasciculus and the total ADI-R score for impaired sociomotoric reciprocity in the ADI-R (Pearson's correlation $r = -0.215; P = 0.01$) in the ASD group. ASD participants with a (C) significant history of impaired use of facial expression in childhood had a significantly lower fractional anisotropy (FA) and higher radial diffusivity in the left uncinate fasciculus.
Social Development

“Sistema Mirror”
Prospettiva nello Sviluppo Tipico

Livelli Progressivi di Sviluppo
LIVELLO III: Differenziazione di Prospettiva
Motor Imagery in Asperger Syndrome: Testing Action Simulation by the Hand Laterality Task

Massimiliano Conson¹*, Elisabetta Mazzarella², Alessandro Froli³, Dalila Esposito⁴, Nicoletta Marino¹, Luigi Trojano¹, Angelo Massagli⁴, Giovanna Gison⁵, Nellantonio Aprea⁵, Dario Grossi¹

Abstract

Asperger syndrome (AS) is a neurodevelopmental condition within the Autism Spectrum Disorders (ASD) characterized by specific difficulties in social interaction, communication and behavioural control. In recent years, it has been suggested that ASD is related to a dysfunction of action simulation processes, but studies employing imitation or action observation tasks provided mixed results. Here, we addressed action simulation processes in adolescents with AS by means of a motor imagery task, the classical hand laterality task (to decide whether a rotated hand image is left or right); mental rotation of letters was also evaluated. As a specific marker of action simulation in hand rotation, we assessed the so-called biomechanical effect, that is the advantage for judging hand pictures showing physically comfortable versus physically awkward positions. We found the biomechanical effect in typically-developing participants but not in participants with AS. Overall performance on both hand laterality and letter rotation tasks, instead, did not differ in the two groups. These findings demonstrated a specific alteration of motor imagery skills in AS. We suggest that impaired mental simulation and imitation of goal-less movements in ASD could be related to shared cognitive mechanisms.
EMBODIED SIMULATION AND VISUAL PERSPECTIVE TAKING IN AUTISM SPECTRUM DISORDERS

M. Conson, E. Mazzarella, D. Esposito, D. Grossi, N. Marino, A. Massagli, A. Frolli
The basis of social behavior

Ability to recognize a specific individual

<table>
<thead>
<tr>
<th>Non-primate mammals</th>
<th>Primates monkeys &amp; human</th>
</tr>
</thead>
<tbody>
<tr>
<td>SMELL</td>
<td>Eyes &amp; VOICES</td>
</tr>
</tbody>
</table>
« Eye Tracking »

Pelphrey et al., 2002
Why are numbers increasing: *Better tools?*

- New diagnostic measures
  - Autism Diagnostic Observation Schedule 2 (1989, 2000 ADOS2)

- Screening tools/algorithms/instruments in wide use:
  - MCHAT / AAP guidelines
  - SCQ
  - SRS
CHAT - Check list for autism in toddlers
(Baron-Cohen et al., 1992)

Sezione A. Domande ai genitori
1. Al vostro bambino piace essere cercato, fatto salire sulle ginocchia? Sì  No
2. Vostro figlio si interessa agli altri bambini? Sì  No
3. Vostro figlio piace amare i giocattoli, come le bambole o le macchinette? Sì  No
4. Si deve che il vostro figlio suoni? Sì  No
5. Quali giochi ha il piacere di fare lino? Sì  No
6. Ogni cosa che suona è a suo piacere di fare lino? Sì  No
7. Ogni cosa che il vostro figlio il piacere di prendere per interesse per qualcosa, cioè indurre a guardarlo? Sì  No
8. E in grado di giocare in modo appropriato con giocattoli, come macchinette o maschere? Sì  No
9. Il vostro bambino vi piace portare i suoi giochi e giocattoli? Sì  No

Sezione B. Osservazione del pediatra
1. Durante l'osservazio del bambino vi fissa mai negli occhi? Sì  No
2. E possibile attirare l'attenzione del bambino, indicare poi un oggetto interessante, segnare col dito o nominarlo con un "oh, guarda..." e osservare che il bambino effettivamente si girà a guardare ciò che gli è stato indicato? Sì  No
3. E possibile interessare il bambino a un gioco di finzione, ad esempio preparare qualcosa da bere o da mangiare? Sì  No
4. Chiedendolo "dove si trova la bici" o "nuova la bici", risposto eventualmente la domanda con un altro oggetto conoscuto (es. sedia), il bambino risponde o indica con il dito e contemporaneamente a guardarlo in faccia? Sì  No
5. Rimane a fare una torre? Sì  No
6. Si, con quanti cubi? (n. di cubi)

Scoring
- alto rischio di autismo: insuccessi in A5, A7, B2, B3 e B4
- medio rischio di autismo: insuccesso solo in A7 e B4
- rischio di diversi disturbi di sviluppo: 3 insuccessi in qualsiasi item
- nei limiti di norma: 3 insuccessi in qualsiasi item

<diagram>
<table>
<thead>
<tr>
<th>Before 6 months</th>
<th>6-12 months</th>
<th>12-18 months</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Looking at faces</td>
<td>- responding to name</td>
<td>- Pointing</td>
</tr>
<tr>
<td>- Smiling at others</td>
<td>- babbling</td>
<td>- Using single words</td>
</tr>
<tr>
<td>- Cooing</td>
<td>- playing social games</td>
<td>- Using gestures</td>
</tr>
<tr>
<td></td>
<td>- Displaying bright affect</td>
<td>- Imitating</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- Interest in other children</td>
</tr>
</tbody>
</table>
Typical signs in ASD

- Eye contact
- Response to name
- Gestures
- Showing
- Imitation
- Pretend play
- Joint attention

Repetitive behaviors
- Spinning or lining up

Stereotyped behaviors
- Flapping
- Walk on tip toes

Sensory behaviors
- Visual examination
- Feeling material
Patterns of symptoms

Early onset and regression

- Lack of response to name
- No good eye contact
- No Joint attention
- No emotional behaviors sharing
- No imitation

between 12 and 24 months:

most strongest pattern of « early onset »

The Importance of Effective Early Diagnosis and Treatment: A public health perspective

- Earlier diagnosis = More intervention opportunities

- More opportunities = Optimal intervention benefit
  - Core features: social communication / atypical behaviors
  - Cognitive and adaptive functioning
  - Fully integrated classroom placements
  - Potentially promoting optimal adaptive independence
  - Potentially reducing considerable lifetime cost and service system demands associated with ASD and related care
Neurodevelopmental Underpinnings

- Core and associated vulnerabilities likely have complex neurogenetic origins:

**Evidence:**

- Maleness (3:1 to 4:1)

- Familial loading/risk:
  - MZ twins: 58-96%
  - DZ twins: 0-31%
  - Sibs: 5-20%
  - 1% - Population

*(18.7% - Ozonoff et al., Pediatrics, 2011)*
Born to Change!
Cerebral plasticity—critical period

Structural organisation = functional learning
• Learning and memory associated with multilevel brain changes

• Both dendritic development and pruning are strongly influenced by the use or disuse

• Behavioral treatments cause changes in brain structure, function and organization in brain

Keller et al, 2009; Spironelli et al, 2010
Multiple studies show that the diagnosis of autism when made in younger children is stable over time.

• Lord 2006 Arch Gen Psychiatry 2006 June;63(6):694-701
Changing developmental trajectories

social Expertise

« Non – Social » Expertise
Historical Perspective

- Not far removed from an “untreatable” era

- Rutter (1970):
  - <2% functioning “normally”
  - 60% requiring institutional placement/support

- Lovaas (1987): UCLA Young Autism Project
  - Intensive ABA = 9 / 19 (47%) “recovered” or “normal functioning”
  - A breakthrough with major methodological concerns
Treatments and Therapies
78,300,000 results (0.08 seconds)

- Auditory Integration
- Sensory Integration
- ABA
- Discrete Trial Training
- Lovaas/UCLA Intervention
- Early Start Denver Model
- Holding Therapy
- Dolphin Assisted Therapy
- Facilitated Communication
- Augmentative Communication
- Vision Therapy
- Vitamins
- Hyperbaric Oxygen
- Psychoparmacological treatments
- Floortime
- Music Therapy
- Social Skills Training
- Incidental Teaching
- TEACCH
- PECS
- Pivotal Response Therapy
- Son-Rise
- RDI
- Chelation
- Diets
- Drugs
- Supplements
Evidence-Based Interventions*

Antecedent-Based Interventions (ABI)
Computer-Aided Instruction
Differential Reinforcement
Discrete Trial Training
Extinction
Functional Behavior Assessment
Functional Communication Training
Naturalistic Intervention
Parent-Implemented Intervention
Peer-Mediated Instruction and Intervention
Social Skills Groups
Speech Generating Devices/VOCA
Redirection - Self-Management
Redirection - Social Narratives

Picture Exchange Communication System (PECS)
Pivotal Response Training
Prompting
Reinforcement
Response Interruption/
Structured Work Systems
  Task Analysis
  Time Delay
  Video Modeling
  Visual Supports
Applied Behavior Analysis (ABA)

**Baer, Wolf, & Risley (1968)**

**APPLIED** - strives to produce rapid and clear benefit to problems of social importance;

**BEHAVIORAL** - uses objective and accurate measurement of the behavior of interest;

**ANALYSIS** - uses controlled (single-case) methods to understand the environmental variable(s) that influence an individual’s behavior.
Comportamentismo

Analisi del Comportamento

Analisi del Comportamento Applicata

- Ergonomia
- Organizzazioni
- Clinica
- Sviluppo
- Tipico
- Atipico
- Job Management, etc…
- OBM Performance Management, etc…
- ACT Acceptance Commitment and Therapy
- Educazione Speciale: ABA Autism, ABA Mental Retardation
- Educazione: Instructional Design and Technology
- Educazione: Instructional Design and Technology
COMPORTAMENTISMO

ANALISI SPERIMENTALE (BA)

ANALISI APPLICATA (ABA)

Discrete Trial Teaching

Insegnamento Incidentale

Verbal Behaviour

Precision Teaching

Pivotal Response Training (PRT)
Natural Enviroment Teaching (NET)
<table>
<thead>
<tr>
<th>Intervento</th>
<th>Evidenze di Efficacia</th>
<th>Presumibilmente Efficace</th>
<th>Non dimostrata Efficacia</th>
</tr>
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<tbody>
<tr>
<td>ABA/DTT</td>
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<tr>
<td>ABA/NET</td>
<td>X</td>
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<tr>
<td>PRT</td>
<td>X</td>
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<tr>
<td>DENVER</td>
<td>X</td>
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<tr>
<td>TEACCH</td>
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<td>X</td>
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<tr>
<td>DIR</td>
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<td>X</td>
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</table>
ABA/VB, ESDM, PRT
Grazie per l’Attenzione

Amarti…

È quando piove forte e guardi il mare vedi gli opposti dell’amore e nel mezzo lì trovo te che sei già parte di me se e solo se questo è amare…

È quando il vento ti spinge altrove sento un impulso ad andare avanti per ore e ad ogni costo andrò verso di te se quello che ho sei solo tu senza ragione.