# The Silent Gap
# What We Are Still Learning About Autism

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## Relevant Disclosure

- Co-author of the Autism Spectrum Rating Scales (MHS, 2009).
- Co-author of the Autism Spectrum Rating Scales Adult (MHS, 2025).
- Co-author of Assessment of Autism Spectrum Disorders text (Guilford, 2009).
- Co-author of Assessment of Autism Spectrum Disorders Second Edition (Guilford, 2016).
- Co-author/presenter Assessment of Autism Spectrum Disorders CEU (APA, 2009).
- Co-author of Raising a Resilient Child With Autism Spectrum Disorders (2011, McGraw Hill).
- Co-author of Treatment of Autism Spectrum Disorders (2012, Springer).
- My expanse are paid my MHS.
- AI note-taking is fine.

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# Goals

- **Provide a brief historical overview of Autism Spectrum Disorder (ASD)**
  Touch on key early theories and evolving perspectives that shaped our understanding of ASD.
- **Define ASD using the DSM-5 criteria**
  Clarify the diagnostic definition and how it's currently classified in clinical settings.
- **Describe common ASD symptoms across the lifespan**
  Highlight how symptoms can present in early childhood, adolescence, and adulthood.
- **Introduce a central theory of ASD**
  Summarize one core theoretical model that helps explain ASD features (e.g., Theory of Mind or Executive Dysfunction).
- **Review key findings from the ASRS Adult (Autism Spectrum Rating Scales Adult)**
  Share data from one of the largest standardization samples to show how typical and ASD populations compare.
- **Examine the benefits and limitations of traditional diagnostic approaches**, such as ADOS and ASRS, while introducing novel tools like EarliPoint.
- **Discuss current assessment tools and approaches**
  Explain how ASD is assessed today, including traditional tools like the ASRS and newer digital methods.
- **Explore emerging technologies in ASD evaluation**
  Examine how innovations, like EarliPoint and AI-driven diagnostics, are shaping the future of autism assessment.

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In 1911, Swiss psychiatrist Eugen Bleuler used the term “autism” in his book on Dementia Praecox or the Group of Schizophrenias. He used it to describe a symptom of schizophrenia in adults. By “autism,” he meant a withdrawal into one’s inner world and a detachment from reality. It was not yet a separate diagnosis.

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# Kanner's Description (1943)

- first physician in the world to be identified as a child psychiatrist
- founder of the first child psychiatry department at Johns Hopkins University Hospital
- Wrote *Child Psychiatry* (1935), the first English language textbook to focus on the psychiatric problems of children.

Leo Kanner who introduced the label *early infantile autism* in 1943 in his paper : Kanner, L. (1943). Autistic disturbances of affective contact. Nervous Child, 2, 217-250.

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# Kanner's Description (1943)

- His seminal 1943 paper, "Autistic Disturbances of Affective Contact", together with the work of Hans Asperger, forms the basis of the modern study of autism.
- Leo Kanner was the Editor for *Journal of Autism and Developmental Disorders*, then called *Journal of Autism and Childhood Schizophrenia*

Leo Kanner who introduced the label *early infantile autism* in 1943 in his paper : Kanner, L. (1943). Autistic disturbances of affective contact. Nervous Child, 2, 217-250.

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# Kanner's Description (1943)

- Inability to relate to others
- Disinterest in parents and people
- Language difficulties
- Fascination with inanimate objects
- Resistance to change in routine
- "early infantile autism."
- Purposeless repetitive movements
- A wide range of cognitive skills
- Where they possess an innate inability for emotional contact

Leo Kanner who introduced the label *early infantile autism* in 1943 in his paper : Kanner, L. (1943). Autistic disturbances of affective contact. Nervous Child, 2, 217-250.

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Around the same time, in 1944, Hans Asperger described a similar pattern in children who had strong language skills but social difficulties. His work later became associated with what was called Asperger's syndrome

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# Autism's First Child

AS NEW CASES OF AUTISM HAVE EXPLODED IN RECENT YEARS—SOME FORM OF THE CONDITION AFFECTS ABOUT ONE IN 110 CHILDREN TODAY—EFFORTS HAVE MULTIPLIED TO UNDERSTAND AND ACCOMMODATE THE CONDITION IN CHILDHOOD. BUT CHILDREN WITH AUTISM WILL BECOME ADULTS WITH AUTISM, SOME 500,000 OF THEM IN THIS DECADE ALONE. WHAT THEN? MEET DONALD GRAY TRIPLETT, 77, OF FOREST, MISSISSIPPI. HE WAS THE FIRST PERSON EVER DIAGNOSED WITH AUTISM. AND HIS LONG, HAPPY, SURPRISING LIFE MAY HOLD SOME ANSWERS.

*By John Donvan and Caren Zucker*

Atlantic Monthly, October 2010

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## SPARK = Simons Foundation Powering Autism Research for Knowledge

Key Facts:

- Launched by: Simons Foundation Autism Research Initiative (SFARI)
- Goal: Recruit and study 100,000+ individuals with autism and their family members.
- Current size (2025): Over 300,000 participants, including genetic data (whole exome and genome sequencing), behavioral traits, developmental history, and family medical history.
- Participants: Individuals diagnosed with ASD and their biological parents/siblings (triads).

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# Exome Versus Genome

| Feature | Exome | Genome |
|---|---|---|
| Size | ~30 million base pairs | ~3 billion base pairs |
| % of genome | ~1–2% | 100% |
| Includes | Protein-coding regions (exons) | All DNA (coding + non-coding) |
| Used for | Identifying disease mutations | Comprehensive genetic analysis |

The **exome** is the portion of the genome that consists of **exons. T**hese are the **protein-coding regions** of genes. While small in size compared to the whole genome, the exome contains the **majority of known disease-causing genetic mutations**.

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# Key Details

- Litman, A., Sauerwald, N., Snyder, L. G., Foss-Feig, J., Park, C. Y., Hao, Y., Dinstein, I., Theesfeld, C. L., & Troyanskaya, O. G. (2025). *Decomposition of phenotypic heterogeneity in autism reveals underlying genetic programs*. *Nature Genetics*. https://doi.org/10.1038/s41588-025-02224-z

- **Cohort:** Over 5,000 children with autism from the SPARK study (ages 4–18), with analyses validated in an independent cohort.
- **Approach:** A **person-centered**, generative mixture modeling methodology analyzing both phenotypic and genotypic data across 230+ traits per individual

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**Findings:** Four distinct subtypes of autism, each with unique clinical presentations, genetic profiles, and developmental trajectories:

- **Social and Behavioral Challenges** (~37%)
  Core autism traits, intact developmental milestones, frequent co-occurring conditions like ADHD, anxiety, or OCD
- **Mixed ASD with Developmental Delay** (~19%)
  Delayed early milestones, fewer psychiatric co-morbidities, a mix of de novo and inherited genetic mutations.
- **Moderate Challenges** (~34%)
  Milder core ASD traits, typical developmental progress, minimal psychiatric co-morbidities.
- **Broadly Affected** (~10%)
  Severe core traits, high levels of co-occurring conditions (e.g., intellectual disability, psychiatric issues), and greater burden of rare de novo mutations

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## Key Findings

- Used generative mixture modeling on a large SPARK cohort of children with autism.
- Identified four robust autism subtypes based on genetic and phenotypic patterns.
- Subtypes corresponded with different developmental, psychiatric, and genetic profiles.
- Each subtype is linked to distinct patterns of common, inherited, and de novo mutations.
- The developmental timing of disrupted genes aligned with subtype-specific clinical outcomes.

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# ASD Background

- Autism Spectrum Disorder (ASD) is a neurodevelopmental condition marked by deficits in social interaction, communication, and repetitive behaviors.
- The etiology of ASD is complex, involving both genetic and environmental factors.
- Recent studies emphasize the need for individualized and technology-driven interventions to improve quality of life and functional outcomes (Qin et al., 2024).
- Despite progress in understanding ASD, challenges remain in diagnosis and treatment, mainly due to the disorder's heterogeneity and co-occurring conditions, which complicate the diagnostic process (Hus & Segal, 2021).

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# Broadening the Spectrum

- There’s no single published tally of all autism-related meta-analyses since 1966, but the peer-reviewed literature contains many dozens likely over 100 meta-analyses covering prevalence, interventions, cognitive/behavioral profiles, biomarkers, and other aspects of ASD. Some reviews of subdomains have explicitly counted dozens of meta-analyses within those narrower focuses. .
- Likely over 100,000 total subjects from around the world.
- Five psychosocial dimensions identified: emotion recognition, theory of mind, cognitive flexibility, planning and inhibition.
- For all 5 dimensions group differences between normal and those with ASD have declined since 2000.
- This is attributed to differences in diagnostic criteria, assessment practices and community awareness.

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## Diagnosis

- ASD diagnosis typically involves using standardized tools such as the Autism Diagnostic Observation Schedule (ADOS-2) and Autism Spectrum rating Scales (ASRS).However, these tools do not specifically diagnose
- This leads to potential misdiagnosis, especially in those with co-occurring cognitive or sensory impairments (Bishop & Lord, 2023).
- Early detection is critical, as timely intervention can significantly influence developmental outcomes.
- Advances in diagnostic technologies, including machine learning and biomarkers, enhance the precision of ASD diagnoses (Yu et al., 2024; Rasul et al., 2024).

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## Treatment

- The treatment of ASD is highly individualized, with a range of behavioral, educational, and pharmacological interventions available.
- Applied Behavior Analysis (ABA) remains one of the most well-established therapies, particularly for improving children's intellectual functioning and adaptive behaviors (Eckes et al., 2023).
- Other interventions, such as Cognitive Behavioral Therapy (CBT), have proven effective in managing emotional and social challenges (You et al., 2023).
- Emerging therapies, including transcranial pulse stimulation and virtual reality-based interventions, offer promising alternatives for addressing the core symptoms of ASD and improving social skills (Cheung et al., 2023; Dechsling et al., 2021).

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# Outcome

- Long-term outcomes for individuals with ASD vary widely, influenced by early intervention, co-occurring conditions, and the level of intellectual functioning.
- Early comprehensive treatment models have improved cognitive, language, and adaptive functioning, especially when intensive interventions involve parental participation (Shi et al., 2021).
- However, many individuals with ASD continue to face challenges in adulthood, particularly in areas such as employment and independent living (Scheeren et al., 2022).
- The outcomes' trajectory highly depends on the severity of symptoms and access to sustained, individualized support (Elias & Lord, 2021).

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# The Autism Spectrum by Cognition & Language

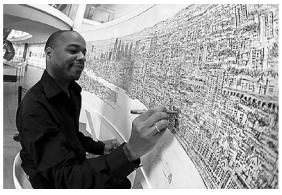
*Levels of Cognitive Functioning*

*(~30-35%)* ← → *(~65-70%)*

*Cognitive Impairment*
*Language Impairment*
*Seizures & Medical Co-Occurring Conditions*
*Severe/Profound Autism*

*In-tact Cognition & Language*
*Asperger Syndrome*
*Psychiatric Co-Occurring Conditions*
*Neurodivergent*



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# Current Stats on Autism (CDC)

**IN THE GENERAL POPULATION:**

- 1 in 44 8-year-old children are identified with ASD
- Male-Female Ratio:
  - 4 times higher in boys
- Median Age of Diagnosis: 4-5 years
  - Much later for disadvantaged populations
- When ASD can be reliably diagnosed:
  - 18-24 months when diagnosed by experienced clinicians
- Co-Occurring Intellectual Disability:
  - 35% with ID

**GENETIC LIABILITY:**

- ASD in Subsequent Biological Siblings: 1 in 5 (~20% risk)
- Broader Autism Phenotype ("shadow symptoms*"): 1 in 5 Siblings*
- *Non-ASD developmental delays: 1 in 10 Siblings*

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# Autism in Females

- Females often misdiagnosed or missed to diagnosis
- Females may present with stronger social skills (Kreiser & White, 2014):
  - Intact symbolic and imaginary play
  - Larger emotional vocabulary
  - Greater awareness and desire for social interaction
  - Ability to mimic others in social situations
  - May develop one or two close friends
- Restricted interests tend to be related to people/animals rather than inanimate objects (Lai & Baron-Cohen, 2015)
- Research points to a "protective effect" in females (Satterstrom et al., 2020)
- "Camouflaging Effect": Females are more likely to use coping strategies to hide ASD behaviors – likely due to social pressures (Hull et al., 2017)
- Higher rates of internalizing disorders (anxiety, depression, eating disorders)


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# Racial & Ethnic Disparities

www.cdc.gov/ncbddd/autism/addm

- Prevalence rates are FINALLY identical for non-Hispanic white, non-Hispanic black, and Asian/Pacific Islander children but continue to be LOWER for Hispanic children
- 47% of Black children and 36% of Hispanic children are more likely to have Intellectual Disability with ASD compared to 27% of White children
- Black children with ASD are are less likely to have a first evaluation by age 3 than White children

CDC
CENTERS FOR DISEASE CONTROL AND PREVENTION


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# Assessment of ASD in Late Adolescence and Adults

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## DSM 5 Autism Spectrum Disorder

- Combined social and communication categories.
- Tightened required criteria reducing the number of symptom combinations leading to a diagnosis.
- Omitted Retts and Childhood Disintegrative Disorders.
- Clarifies co-morbidity issues.
- Eliminated PDD NOS and Aspergers in favor of Autism Spectrum Disorder.
- Created Social Pragmatic Communication Disorder.
- Still no specified profile for adults, just guidelines.

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## DSM 5 Versus DSM 5 TR

- The criteria for diagnosing ASD including the two main domains:
  - Social communication/interaction
  - Restricted, repetitive behaviors/interests
- The requirement for symptoms to be present in early development
- The specifiers (e.g., intellectual impairment, language level, comorbid conditions)
- DSM-5-TR did not revise the diagnostic criteria for Autism Spectrum Disorder, but it did expand and refine the surrounding descriptions, with more up-to-date evidence and better consideration of cultural, gender, and mental health contexts.

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# DSM 5 Versus DSM 5 TR

**Clarification of Language and Terminology**

•The DSM-5-TR refined the language in the text sections accompanying the diagnostic criteria.

•These changes aimed to improve clarity, reduce ambiguity, and provide updated guidance for clinicians.

•For example, the text was updated to reflect current research on autism and neurodiversity perspectives, though the criteria themselves were not altered.

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ASRS®
ADULT
AUTISM SPECTRUM RATING SCALES™ ADULT

# Psychometric Properties of ASRS Adult

MHS
Beyond Assessments

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## Overall psychometric findings!

**The ASRS Adult shows strong evidence of:**

- **Reliability** – are we getting the same results every time?
- **Validity** – is the test measuring what we want it to every time?
- **Fairness** – are we measuring the same way for everyone?

This slideshow provides examples and summarizes findings related to the **full-length ASRS Adult.**

These findings are also the same for **ASRS Adult–Short**

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## Standardization

**ASRS Adult Normative Samples**

| SELF-REPORT | OBSERVER | TOTAL |
|---|---|---|
| *N* = 1,000 | *N* = 1,000 | *N* = 2,000 |

Normative samples differ less then **1.7%** from actual proportions to the **2023 U.S. Census in terms of:**

- **Gender**
- **Age**
- **Race/Ethnicity** (*White, Black, Hispanic, Asian, Other)*
- **Region** (*Northeast, Midwest, South, West*)
- **Education Level** (*Less than High School to Graduate/Professional degree*)

****Though normed on a U.S. sample, ASRS Adult scores are validated for use in Canada (refer to manual – appendix D, *Cross-National Validation Study [Canada vs. United States])***

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## Standardization

**8.3% for Self-Report and 8.2% for Observer of the normative sample includes individuals with one or more clinical diagnosis:**

- Autism Spectrum Disorder (ASD),
- Attention-Deficit/Hyperactive Disorder (ADHD),
- Generalized Anxiety Disorder (GAD), and/or
- Major Depressive Disorder (MDD).

**Trends in raw scores by scales were inspected to determine our normative groups.**

Gender differences were minimal, so **combined gender norms** were used**;** age differences were more pronounced, leading to **five distinct age bands:**

- 18 to 24
- 25 to 34
- 35-44
- 45-54
- 55+

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## Reliability

The reliability of an instrument describes how **precise** and **consistent** the scores are. This section provides a summary of results for the reliability analyses of the ASRS Adult, including **internal consistency**, **test-retest reliability**, and **inter-rater reliability**.

### INTERNAL CONSISTENCY

- Excellent internal consistency for both forms, and across all age groups.

| FORM | MEDIAN ALPHA |
|---|---|
| SELF-REPORT | .88 |
| OBSERVER | .91 |

### TEST-RETEST RELIABILITY

- Excellent test-retest reliability across all forms
- Demonstrates stability in scores over time

| FORM | MEDIAN CORRELATION (*r*) |
|---|---|
| SELF-REPORT | .79 |
| OBSERVER | .82 |

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# Reliability

**Inter-Rater Reliability**

**STUDY 1 (SELF-REPORT VS. OBSERVER)**

- Moderate/strong levels of consistency across scales
- Self-report can provide greater insight into internal processes (e.g., thoughts, emotions)

| PAIRS | MEDIAN CORRELATION (*r*) |
|---|---|
| SELF-REPORT/OBSERVER | .69 |

**STUDY 2 (TWO DIFFERENT OBSERVERS)**

- Moderate/strong levels of consistency
- Different settings, level of insight, nature of the relationship could all be used to explain the level of agreement between different raters

| PAIRS | MEDIAN CORRELATION (*r*) |
|---|---|
| TWO OBSERVERS (DIFFERENT TYPE) | .65 |

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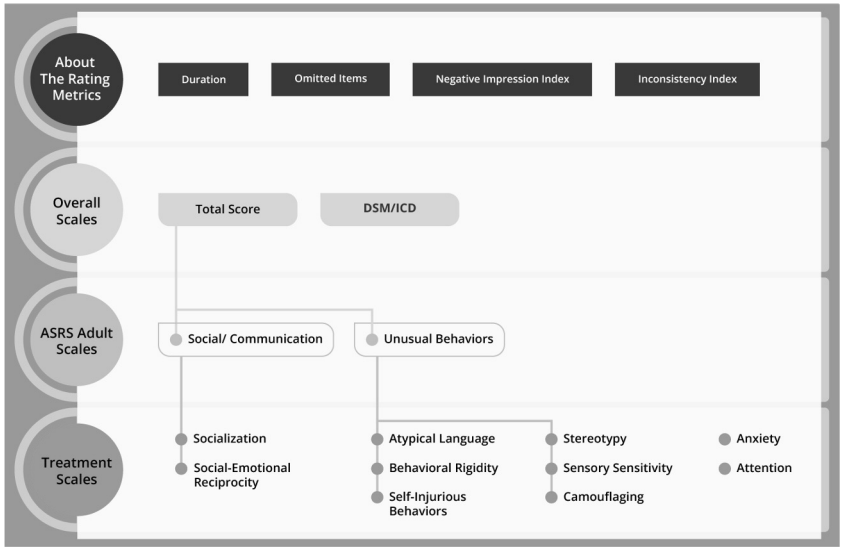
# ASRS Adult Scale structure

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# Validity – internal structure

**Unidimensionality of Treatment Scales**

- Each of the 10 Treatment Scales is supported by confirmatory factor analysis (CFA) as unidimensional.

**Higher-Order Structure**

- Scale-level factor analysis supports a two-factor model: (1) Social/Communication and (2) Unusual Behaviors. These domains are highly correlated ($r \approx .76–.77$), also justifying the use of a **Total Score**.
- All factor loadings were positive and significant, with median values for Social/Communication and Unusual Behaviors of .587 and .719 (Self-Report), and .647 and .735 (Observer), respectively.

| | | MEDIAN | |
|---|---|---|---|
| | Fit Indices (CFA) | SELF-REPORT | OBSERVER |
| Treatment Scales (Median Values) | CFI | .979 | .979 |
| | TLI | .968 | .971 |
| | SRMR | .039 | .041 |
| | RMSEA | .093 | .079 |
| Higher-Order Model (2-factor) | CFI | .934 | .932 |
| | TLI | .931 | .930 |
| | SRMR | .060 | .060 |
| | RMSEA | .042 | .046 |

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# Validity

**CONVERGENT VALIDITY**

ASRS Adult scores show strong correlations with established ASD measures:

- Social Responsiveness Scale, Second Edition (SRS-2)
- Autism Spectrum Quotient (AQ)
- Camouflaging Autistic Traits Questionnaire (CAT-Q)

| ASSESSMENT | MEDIAN CORRELATION (*r*) | |
|---|---|---|
| | SELF-REPORT | OBSERVER |
| SRS-2 | .77 | .85 |
| AQ | .58 | - |
| CAT-Q | .75 | - |

**CLASSIFICATON ACCURACY**

- Using the Total Score *T*-score cutoff of 60, the ASRS Adult correctly classifies ASD vs. general population with high accuracy (Self: 80.7%, Observer: 76.2%), balancing sensitivity and specificity.

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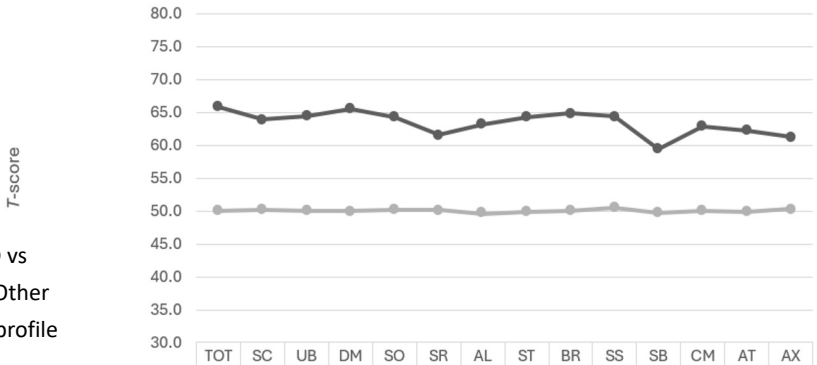
# Validity: Criterion-related

**Findings:**

- ASRS Adult effectively distinguishes ASD from both the general population and other clinical groups.
- The ASRS Adult manual documents ASD vs general population differences across all scales.

- Large effect sizes support robust differentiation:
  - Self-Report: *d* = 0.98–1.89
  - Observer: *d* = 0.47–1.52

- Forthcoming publication presents ASD vs other clinical groups (ADHD, GAD/MDD, Other clinical groups); results show clear score profile differentiation




| | TOT | SC | UB | DM | SO | SR | AL | ST | BR | SS | SB | CM | AT | AX |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| ASD | 65.9 | 63.9 | 64.4 | 65.6 | 64.3 | 61.5 | 63.2 | 64.3 | 64.9 | 64.4 | 59.5 | 62.9 | 62.2 | 61.2 |
| General Population | 50.0 | 50.2 | 50.0 | 50.0 | 50.2 | 50.1 | 49.6 | 49.9 | 50.0 | 50.5 | 49.6 | 50.1 | 49.9 | 50.4 |

***Note***. TOT = Total Score, SC = Social/Communication, UB = Unusual Behaviors, DM = DSM/ICD Scale, SO = Socialization, SR = Social/Emotional Reciprocity, AL = Atypical Language, ST = Stereotypy, BR = Behavioral Rigidity, SS = Sensory Sensitivity, SB = Self-Injurious Behaviors, CM = Camouflaging, AT = Attention, AX = Anxiety.

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# Fairness

**FAIRNESS**

- **GOAL:** Ensuring that the measurement is sensitive to individual characteristics of the intended audience, and that the intended use and interpretation of scores are valid and just across relevant subgroups

Characteristics of the person (other than the construct being measured – symptoms of ASD!) should not influence their responses to the test

To investigate these differences, we conducted these analyses:

1. **Differential Test Functioning** – tests whether the ASRS Adult scales operate in the same way across groups
2. **Mean Group Differences** – compares the size of differences between each group's average score

- Explored differences between groups: ***gender, race/ethnicity, country of residence, and education level***

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# Fairness

## DIFFERENTIAL TEST FUNCTIONING:

**GENDER -** No evidence of differential test functioning was found

- Negligible measurement bias (median ETSSD = .04)
- E.g., overlapping curve evidence of no DTF

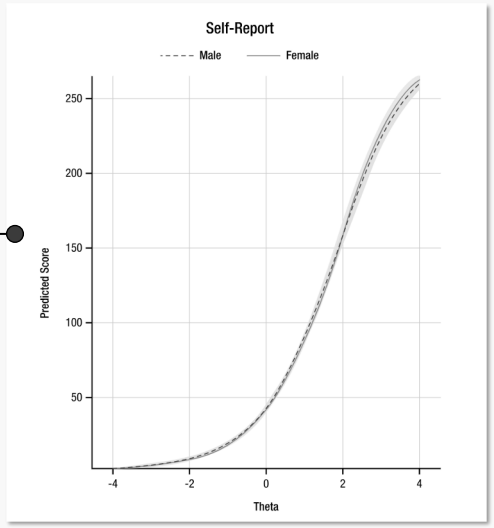
**RACE/ETHNICITY -** Minimal bias across Asian, Black, Hispanic, and White groups

- Median ETSSD = .05 (Self-Report), .04 (Observer); all scale-level ETSSD negligible

**EDUCATION LEVEL -** No evidence of differential test functioning was found

- Negligible measurement bias (median ETSSD = .03)

*Note, across groups there was minor divergence only for Self-Injurious behaviours at high trait levels

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# Fairness

## MEAN GROUP DIFFERENCES:

**GENDER -** After controlling for age, race/ethnicity, and education, mean score differences between males and females were negligible to small.

- Median Cohen's *d* = 0.16 (Self-Report), 0.08 (Observer).

**RACE/ETHNICITY -** Mean differences between Asian, Black, Hispanic, and White groups were negligible to small.

- Median Cohen's *d* = 0.09 (Self-Report), 0.10 (Observer).
- Maximum difference: ~3.5 points.
- No group was meaningfully advantaged or disadvantaged.

**EDUCATION LEVEL -** Mean differences between lower and higher education groups were negligible.

- Median Cohen's *d* = 0.05 (Self-Report), 0.01 (Observer).

**No practically meaningful differences in ASRS Adult scores by gender, race/ethnicity, or education level. All observed differences are well below thresholds for practical significance.**

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# OVERALL PSYCHOMETRIC FINDINGS!

The ASRS Adult shows strong evidence of:

- **Reliability**
- **Validity**
- **Fairness**

ASRS® ADULT

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MHS Beyond Assessments

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# EarliPoint™: An FDA-Authorized Tool to Accelerate Early and Efficient ASD Diagnosis & Assessment

May 2024

LBL2-1005, rev F

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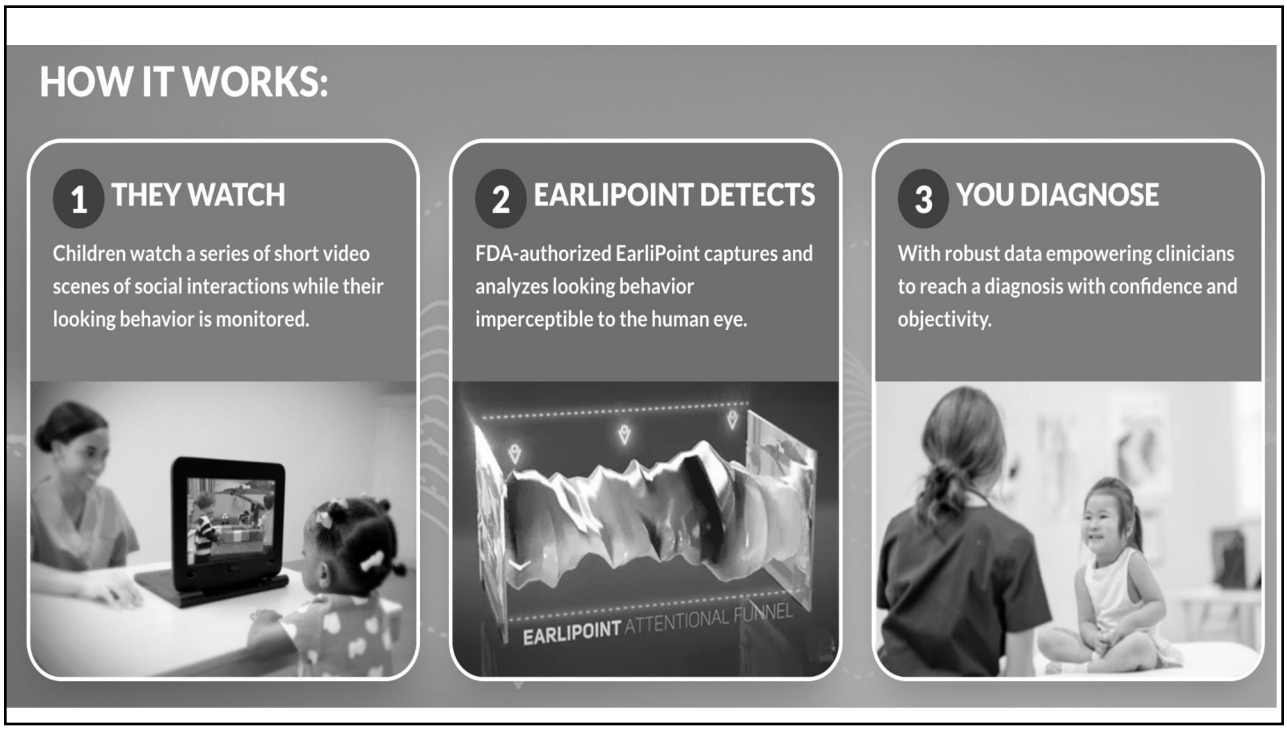
HOW IT WORKS:
1 THEY WATCH
Children watch a series of short video scenes of social interactions while their looking behavior is monitored.
2 EARLIPOINT DETECTS
FDA-authorized EarliPoint captures and analyzes looking behavior imperceptible to the human eye.
EARLIPOINT ATTENTIONAL FUNNEL
3 YOU DIAGNOSE
With robust data empowering clinicians to reach a diagnosis with confidence and objectivity.

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The EarliPoint™ Evaluation for Autism
Copy link
Play
EARLIPOINT™ EVALUATION
Watch on YouTube

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## Performance-Based Measures of Strengths and Vulnerabilities

**Example: Nonverbal Communication & Gestures**

Environmental Context

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## Performance-Based Measures of Strengths and Vulnerabilities

**Example: Nonverbal Communication & Gestures**

Quantitative Reference Metric: Age-Expected Social Visual Engagement

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## Performance-Based Measures of Strengths and Vulnerabilities


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## Performance-Based Measures of Strengths and Vulnerabilities

Example:
Facial Affect

Quantitative Reference Metric: Age-Expected Social Visual Engagement

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## Performance-Based Measures of Strengths and Vulnerabilities


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## Performance-Based Measures of Strengths and Vulnerabilities

Example:
Pointing &
Social
Monitoring

Quantitative Reference Metric: Age-Expected Social Visual Engagement

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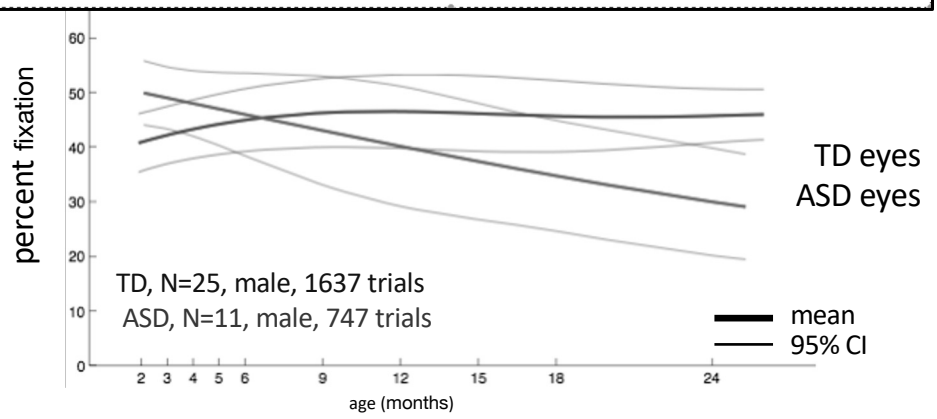
LETTER

doi:10.1038/nature12715

# Attention to eyes is present but in decline in 2–6-month-old infants later diagnosed with autism

Warren Jones[1,2,3] & Ami Klin[1,2,3]

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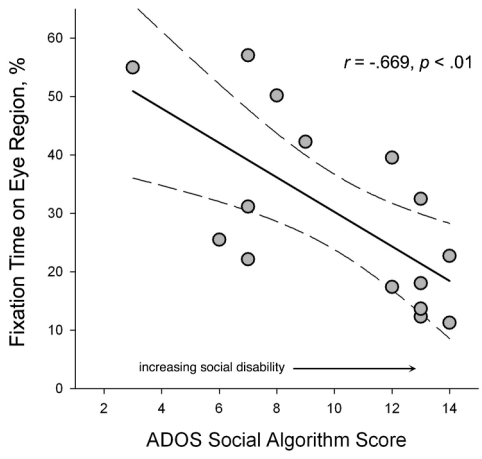
## Predictors of Outcome

*Jones, Carr, & Klin (2008; Arch Gen Psychiatry)*




Less fixation time on eyes predicts more severe social disability.



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## Making the Diagnosis of ASD Today

- Meets DSM 5 TR Criteria.
- Coping behaviors assessed.
- Co-morbid behaviors and disorders assessed.
- Corroborating data obtained about child and adulthood.
- Intellectual, achievement and neuropsychological data collected if warranted.

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## Making the Diagnosis of ASD in the Future

There has been a significant uptick in recent research leveraging artificial intelligence (AI) to develop diagnostic algorithms for Autism Spectrum Disorder (ASD). These studies demonstrate the potential of AI to revolutionize early detection, reduce diagnostic delays, and assist clinicians with accurate classification.

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## Machine Learning (ML)

Machine learning is a subfield of artificial intelligence (AI) that enables computers to learn from data and make decisions or predictions without being explicitly programmed. Algorithms in ML build models based on input data, identify patterns, and improve their performance over time.

There are three main types:

1. **Supervised learning** (with labeled data),
2. **Unsupervised learning** (finding patterns in unlabeled data),
3. **Reinforcement learning** (learning through feedback and reward signals).

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One notable 2023 study focuses on early ASD screening in children using machine learning (ML) models. The researchers evaluated various algorithms for their ability to identify early signs of ASD, particularly where traditional diagnostic delays are common. Their results showed strong performance in ML models, suggesting they could complement existing diagnostic pathways.

Alkahtani, H., & Aldhyani, T. H. H. (2023). *Early screening of autism spectrum disorder diagnoses of children using artificial intelligence*. Journal of Data Research.

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## Support Vector Machines (SVMs)

Support Vector Machines (SVMs) are a type of supervised machine learning algorithm used for classification and regression tasks. They work by finding the hyperplane that best separates data into different classes. The goal is to maximize the margin between the classes—the distance between the closest points (support vectors) of each class to the hyperplane.

SVMs are powerful for smaller, structured datasets and perform well in high-dimensional spaces but typically require feature engineering and don't scale as easily as deep learning models.

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Another 2023 paper applied AI strategies like support vector machines (SVM) and deep learning (DL) on data from EEG and MRI scans to diagnose ASD. The integration of these imaging modalities with AI allowed for more objective and data-driven assessments.

Sundas, A., Badotra, S., Rani, S., & Gyaang, R. (2023). *Evaluation of autism spectrum disorder based on the healthcare by using artificial intelligence strategies*. Computational and Mathematical Methods in Medicine.

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## Deep Learning (DL)

Deep learning is a specialized subset of machine learning that uses **artificial neural networks** with many layers (hence "deep"). It is particularly effective for large-scale, complex data like images, audio, and natural language. Deep learning models automatically extract and learn hierarchical features from data, often outperforming traditional ML algorithms in fields like computer vision, speech recognition, and NLP.

Examples include:

1. Convolutional Neural Networks (CNNs) for image processing
2. Recurrent Neural Networks (RNNs) and Transformers for language modeling

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A comprehensive review in 2023 detailed various AI applications for ASD triage, diagnosis, and prioritization. It emphasized how AI could streamline clinical workflows and support decision-making in under-resourced healthcare settings.

Joudar, S. S., Albahri, A. S., Hamid, R. A., & Zahid, I. A. (2023). *Artificial intelligence-based approaches for improving the diagnosis, triage, and prioritization of autism spectrum disorder: A systematic review of current trends and challenges*. Artificial Intelligence Review

60

Further advancing this trend, another 2022 study proposed a hybrid model that combines multiple medical tests and sociodemographic data for ASD classification. The integration of diverse datasets led to more accurate diagnostic outputs.

Alqaysi, M. E., & Albahri, A. S. (2022). *Diagnosis-based hybridization of multimedical tests and sociodemographic characteristics of autism spectrum disorder using artificial intelligence and machine learning*. Computational and Mathematical Methods in Medicine.

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Collectively, these studies signal that AI is not only capable of assisting in ASD diagnosis but could also redefine best practices for early intervention and individualized care.

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# Collective Insights: Recent Autism Research (2023–2025)

- Earlier biological detection using **genetic, EEG, and neuroimaging biomarkers**
- Increasing use of **AI and multimodal sensing technologies**
- Emphasis on **developmental brain trajectory modeling**
- Recognition of **sensory, emotional, and comorbid conditions**
- Expansion of **personalized and non-pharmacological interventions**
- Integration of **computational neuroscience frameworks**
- Growing exploration of **gut–brain axis and microbiome effects**
- Greater focus on **transdiagnostic and overlapping genetic mechanisms**

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## Hypothetical Case Study: AI-Assisted Asd Diagnosis

### Patient Profile

Name: Liam, Age: 3 years

Referred by a pediatrician due to delayed speech and social disengagement.

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## Step 1: Data Collection

- Liam's parents complete a detailed online behavioral questionnaire based on standardized tools (e.g., ADOS, ASRS). Meanwhile, his medical records, developmental history, and demographic information are uploaded securely.
- Additionally, Liam undergoes:

  **EEG and MRI scans** to capture neurological patterns.

  **Video recordings** of behavior during play and interaction using a smartphone app.

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## Step 2: AI Preprocessing

- The AI diagnostic system preprocesses this multimodal data:
- **Natural language processing (NLP)** analyzes questionnaire responses for ASD-indicative language patterns.
- **Computer vision algorithms** assess facial expressions, eye gaze, and repetitive behaviors from video.
- **Deep learning models** interpret EEG/MRI scans to detect neurodevelopmental biomarkers associated with ASD.

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# Step 3: Diagnostic Prediction

- Using a trained ensemble model (e.g., combining Random Forest and CNNs), the AI provides the following.
- **Risk probability score**: 87% likelihood of ASD
- **Diagnostic category**: Moderate ASD (based on DSM-5 criteria translated into a rules-based model)
- **Confidence intervals** and visual explanations (e.g., heatmaps showing critical moments in videos)

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**Random Forest**
**Ensemble Learning**
**Main Idea:**
A Random Forest is a collection of decision trees. Each tree is trained on a random subset of the data (with replacement), and the final prediction is made by averaging the outputs (for regression) or through majority voting (for classification) from all trees.
**Strengths:**
Handles both classification and regression tasks effectively.
Resistant to overfitting (due to averaging across many trees).
Works on tabular data (e.g., spreadsheets with features like age, income, etc.).

**Convolutional Neural Networks**
**Deep Learning**
**Main Idea:**
CNNs are a class of neural networks that utilize convolutions (a type of filter) to detect spatial hierarchies in data automatically. They excel at processing images, videos, and spatial data, recognizing features such as edges, textures, and shapes.
**Strengths:**
Excellent at handling high-dimensional image data.
Automatically extracts features (no manual engineering required).
Employed for tasks like image classification, object detection, and facial recognition.

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## Step 4: Clinical Review

- A neuropsychologist reviews the AI's findings alongside their own assessment. The AI tool provides:
- A **summary report** highlighting flagged behaviors and brain activity patterns
- Recommendations for follow-up (e.g., speech therapy, ABA therapy)
- The clinician confirms the diagnosis and tailors an early intervention plan.

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## Step 5: Continuous Monitoring

The app schedules periodic reassessments and collects behavioral data through parent check-ins and additional video uploads, allowing the AI to update predictions and therapy outcomes.

This AI-assisted approach doesn't replace the clinician but **augments diagnostic accuracy and speed**, especially in under-resourced or rural areas where developmental specialists may be scarce.

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## Characteristic Cognitive Impairments to Evaluate in ASD

- The ability to attribute mental states to oneself and others.
- The ability to display emotional reaction appropriate to another person's mental state (joint attention of emotion).
- The ability to plan and attend to relevant details in the environment.
- The ability to understand the communicative content of gaze.
- The ability to work cooperatively with others (joint attention of behavior).
- The ability to understand, comprehend, analyze, synthesize, evaluate and differentiate in particular social information in the environment.

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## CAAB STRUCTURE AND SUBTESTS

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**Social Attention:**
- Joint Attention
- Visual Absurdities

**Social- Emotional Knowledge:**
- Blurred Faces Task
- Perspective Taking
- Emotion Charades
- Faux Pas Task

**Social Communication & Interaction:**
- Conversational Probes
- Social Sequencing
- Understanding Body Posture
- Empathy

**Social Problem Solving:**
- Figurative Language
- Flexibility
- Central Coherence Visual Puzzles
- Social Attribution Task
- Social Causality

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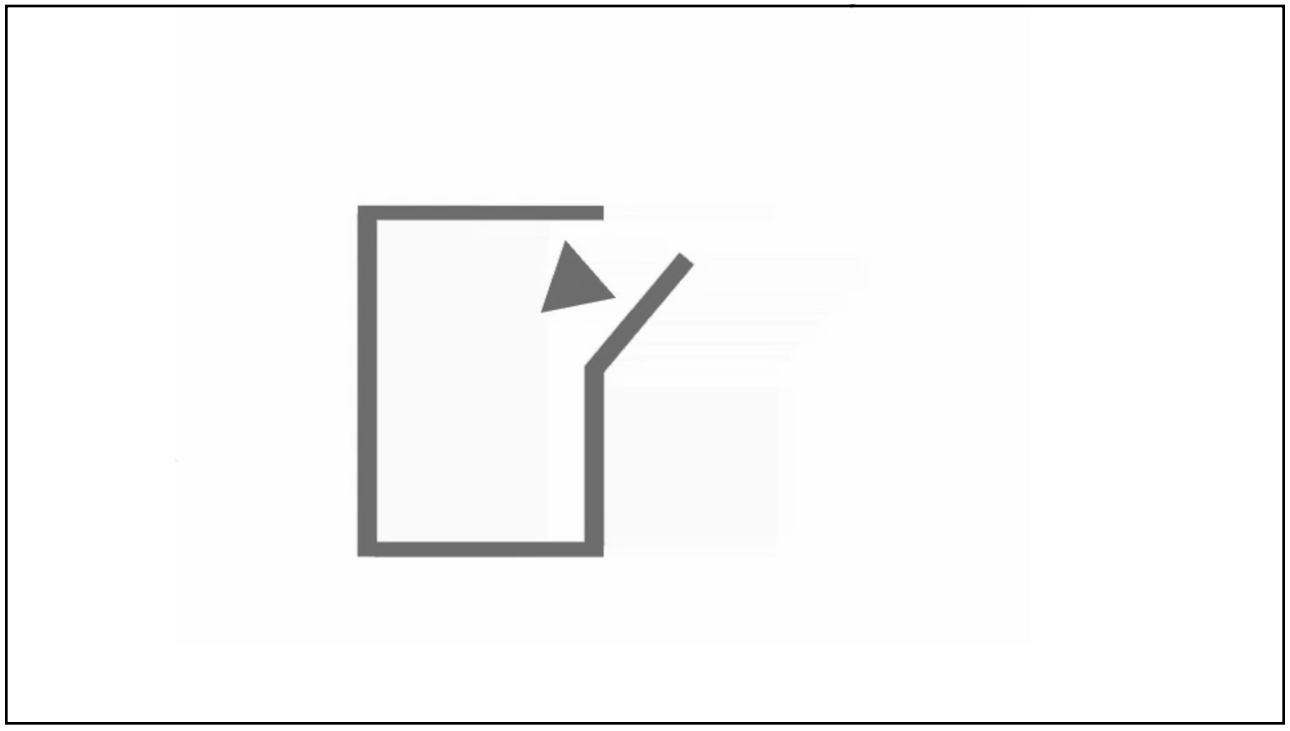

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**What Secretary Kennedy's Plan Misses**

Kennedy's plan reduces autism research to a simplistic search for a "cause," as though autism were a singular condition with a singular explanation. It fails to incorporate the foundational principle of developmental psychopathology: that behavior emerges from complex, evolving interactions among genes, brains, and environments. The plan lacks scientific rigor and risks diverting resources from practical, evidence-based strategies by relying on retrospective chart reviews and neglecting longitudinal evidence.

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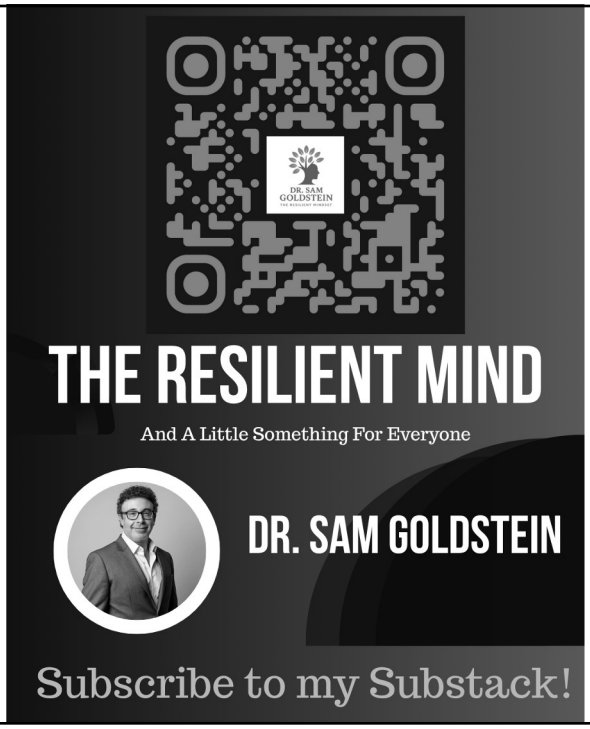


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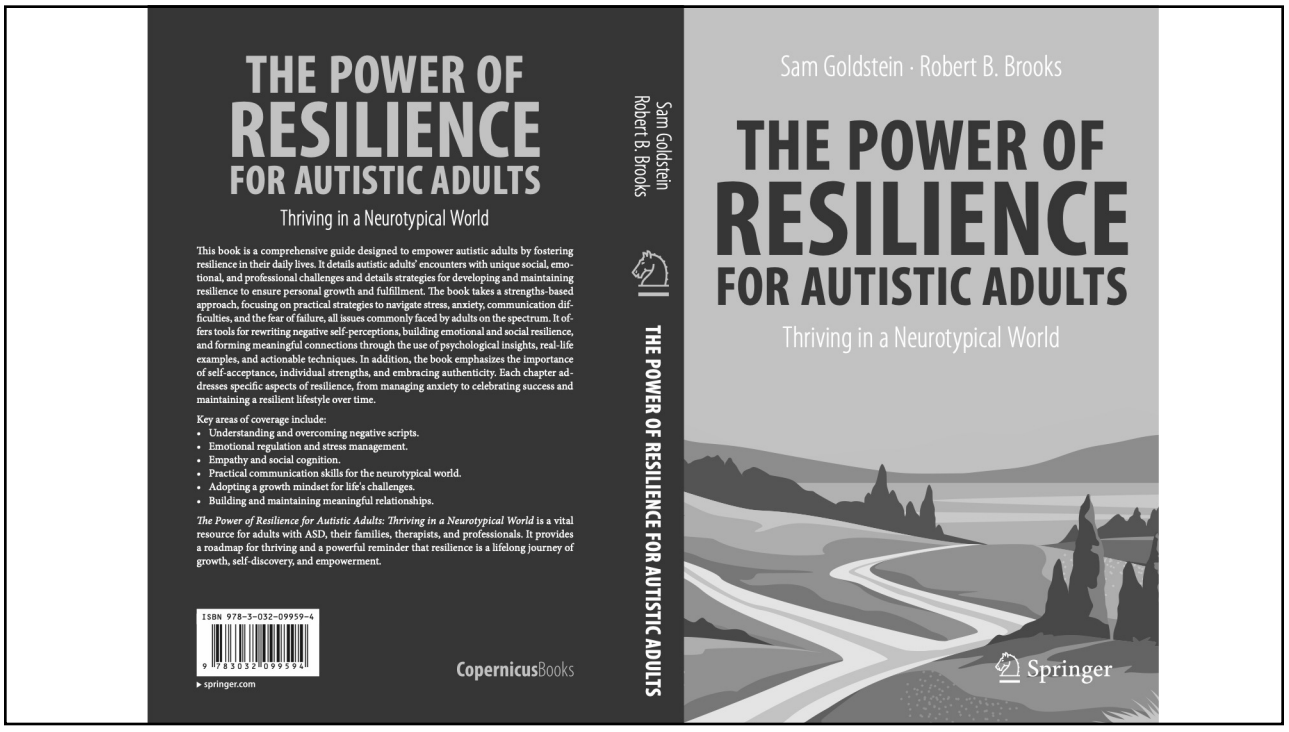


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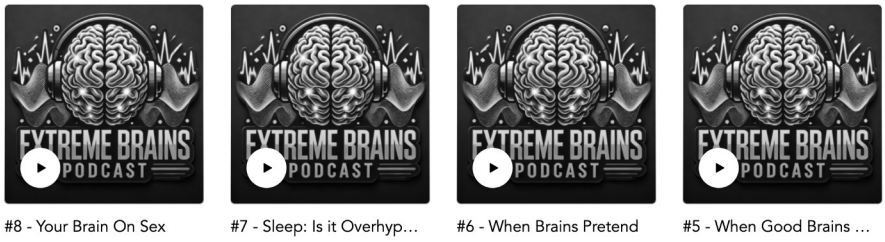
## Extreme Brains Podcast

EXTREME BRAINS is a lively, thought-provoking podcast in which Sam, David, and James—three friends with sharp wit and unique perspectives—gather around a microphone to critique current events, explore life's challenges, and unpack the absurdity of modern times. Whether dissecting the day's headlines, debating life's perplexing questions, or finding humor in the chaos, this podcast offers listeners an engaging mix of critical insight, camaraderie, and laughs.

Listen on:




#8 - Your Brain On Sex
#7 - Sleep: Is it Overhyp…
#6 - When Brains Pretend
#5 - When Good Brains …

https://extremebrainspodcast.podbean.com/

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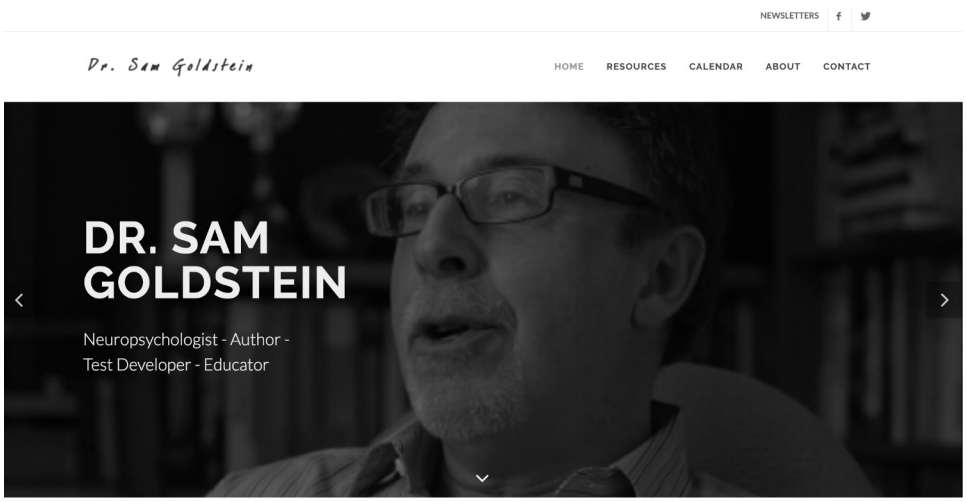
# www.samgoldstein.com

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# Questions?

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