# The Basics of Autism Spectrum Disorder for Speech-Language Pathologists


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# About the Speaker

- **Speech-Language Pathologist** - Graduated with M.S. in Speech-Language Pathology from The University of Texas at El Paso in 2007)

- **PhD, Psychology** - Canterbury Christ Church University in the United Kingdom in 2023, also a **faculty member** in the Allied Health Professions in the school of Speech and Language Therapy.

- Established **True Potential** PLLC in 2008 Tucson Arizona, which to date serves young children diagnosed with Autistic Disorder and their families through educational and speech and language therapy services. Also established the non-governmental organization **Explora tu Potencial** in 2013, which is  still dedicated to developing widely available, effective. affordable and sustainable specialized care for individuals with ASD and their families in Ciudad Juárez, México.

- Designed and directed multiple **small- and large-scale educator training programs** for **private and public schools** in Mexico.

# Disclosures

- Financial — Daniel is a presenter of online CE courses by True Potential PLLC for which he receives royalties.

- Nonfinancial — Daniel has no relevant non-financial relationships to disclose.

# Learning Outcomes

As a result of this course, participants will be able to:

1. Describe how differences in **key brain structures and functions** are **associated with the symptoms of ASD.**

2. Broadly summarize our **historical understanding and definition of ASD**.

3. List the **latest and specific criteria** required to meet a **diagnosis of ASD**.

4. List ways in which **ASD** can be seen **across life stages** from early life to adulthood.

5. List some of the **standardized and non-standardized measures** you can use to evaluate communication skills in individuals diagnosed with ASD across different life stages.

# Course Agenda

**SECTION I**
Neurobiology of Autism Spectrum Disorder (ASD)

**SECTION II**
A Brief History of ASD

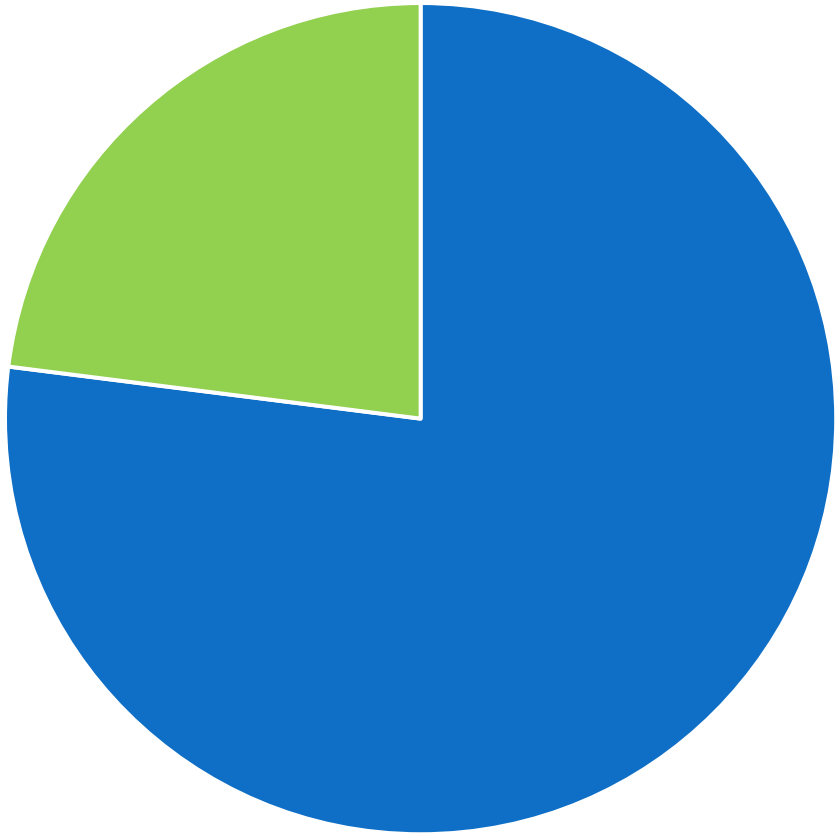
**SECTION III**
Current Definition /Diagnostic Criteria of ASD (DSM-5-TR)

**SECTION IV**
Identification and Evaluation of Communication Impairments in ASD

# Section I

## Neurobiology of ASD

# Genetic Variants Related to ASD



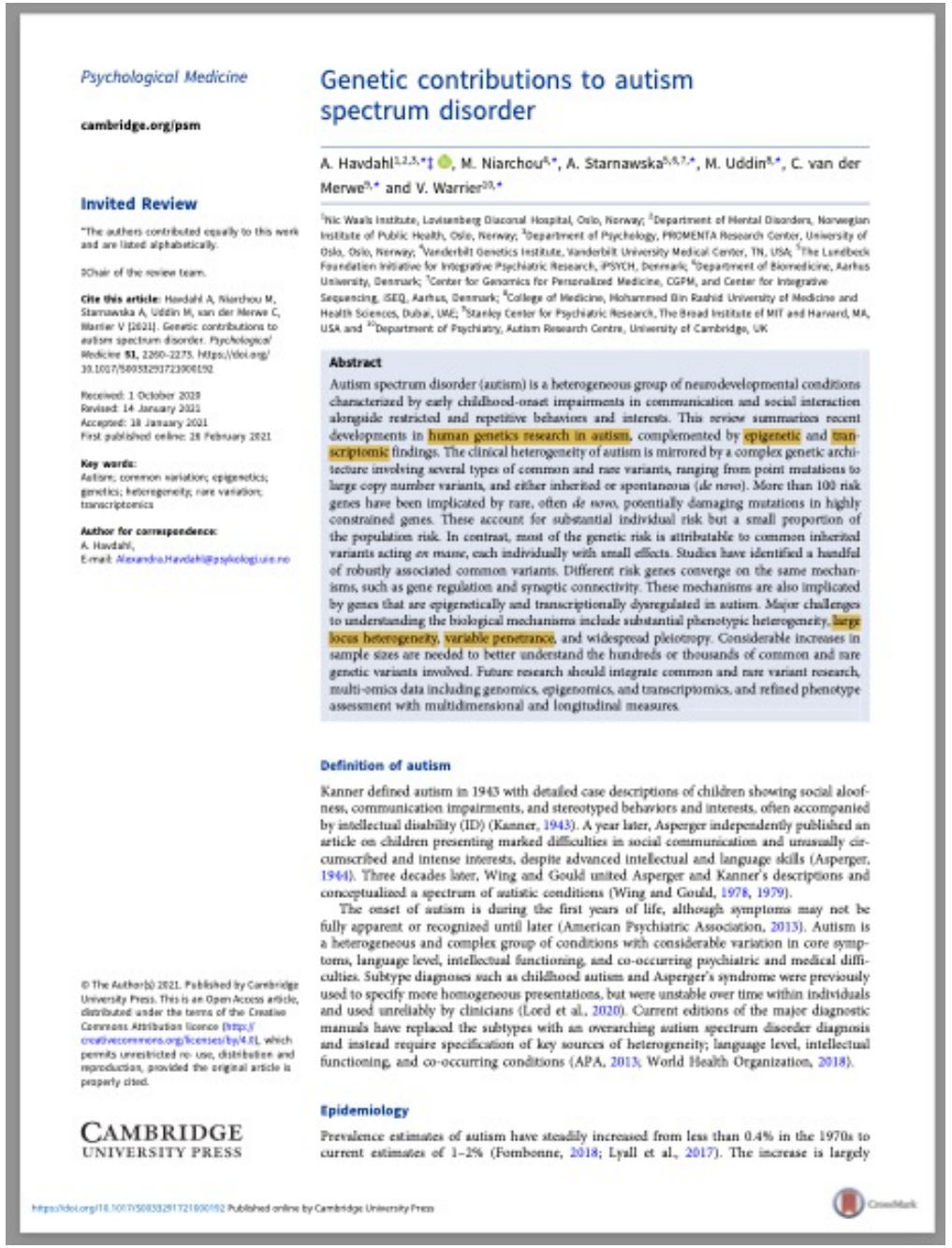
Inherited

Spontaneous/Environmental

# The Origin of ASD

"Most researchers agree that the development of ASD is not necessarily determined by a single gene, but rather is the result of a combination of mutations in many genes, with a **certain degree of heritability**. However, **ASD is not a genetic disorder**, especially in the early stages of development, and many potential risk factors, such as **poor maternal pregnancy and childbirth**, may trigger mutations that produce ASD symptoms."

- Chen et al., 2024 p. 8



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**Invited Review**

"The authors contributed equally to this work and are listed alphabetically.

‡Chair of the review team.

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# Genetic contributions to autism spectrum disorder

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**Abstract**

Autism spectrum disorder (autism) is a heterogeneous group of neurodevelopmental conditions characterized by early childhood-onset impairments in communication and social interaction alongside restricted and repetitive behaviors and interests. This review summarizes recent developments in human genetics research in autism, complemented by epigenetic and transcriptomic findings. The clinical heterogeneity of autism is mirrored by a complex genetic architecture involving several types of common and rare variants, ranging from point mutations to large copy number variants, and either inherited or spontaneous (*de novo*). More than 100 risk genes have been implicated by rare, often *de novo*, potentially damaging mutations in highly constrained genes. These account for substantial individual risk but a small proportion of the population risk. In contrast, most of the genetic risk is attributable to common inherited variants acting *en masse*, each individually with small effects. Studies have identified a handful of robustly associated common variants. Different risk genes converge on the same mechanisms, such as gene regulation and synaptic connectivity. These mechanisms are also implicated by genes that are epigenetically and transcriptionally dysregulated in autism. Major challenges to understanding the biological mechanisms include substantial phenotypic heterogeneity, large locus heterogeneity, variable penetrance, and widespread pleiotropy. Considerable increases in sample sizes are needed to better understand the hundreds or thousands of common and rare genetic variants involved. Future research should integrate common and rare variant research, multi-omics data including genomics, epigenomics, and transcriptomics, and refined phenotype assessment with multidimensional and longitudinal measures.

## Definition of autism

Kanner defined autism in 1943 with detailed case descriptions of children showing social aloofness, communication impairments, and stereotyped behaviors and interests, often accompanied by intellectual disability (ID) (Kanner, 1943). A year later, Asperger independently published an article on children presenting marked difficulties in social communication and unusually circumscribed and intense interests, despite advanced intellectual and language skills (Asperger, 1944). Three decades later, Wing and Gould united Asperger and Kanner's descriptions and conceptualized a spectrum of autistic conditions (Wing and Gould, 1978, 1979).

The onset of autism is during the first years of life, although symptoms may not be fully apparent or recognized until later (American Psychiatric Association, 2013). Autism is a heterogeneous and complex group of conditions with considerable variation in core symptoms, language level, intellectual functioning, and co-occurring psychiatric and medical difficulties. Subtype diagnoses such as childhood autism and Asperger's syndrome were previously used to specify more homogeneous presentations, but were unstable over time within individuals and used unreliably by clinicians (Lord et al., 2020). Current editions of the major diagnostic manuals have replaced the subtypes with an overarching autism spectrum disorder diagnosis and instead require specification of key sources of heterogeneity; language level, intellectual functioning, and co-occurring conditions (APA, 2013; World Health Organization, 2018).

## Epidemiology

Prevalence estimates of autism have steadily increased from less than 0.4% in the 1970s to current estimates of 1–2% (Fombonne, 2018; Lyall et al., 2017). The increase is largely



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# Pre and Postnatal Risk Factors

1. Poor **maternal health**
2. Complications in **childbirth**
3. Exposure to **contaminants** before and after birth

# Environmental Risk Factors

- Heavy metals including **aluminum, cadmium**, **chromium**, **copper**, **lead**, **arsenic, manganese** and **mercury** have found in significantly higher concentrations in individuals diagnosed with ASD (Aschner et al., 2024; Akyuzlu et al., 2014; Ding et al., 2023; Seneff et al., 2012).

- Relationships between **phthalates** and ASD, and other disorders, have been documented through multiple studies (Jeddi et al., 2016). xs

# Environmental Risk Factors

Significant relationships have been found between exposure to herbicides like **glyphosate** and pesticides like **organophosphorus** and **organochlorine** (Chen et al., 2024; Pu et al., 2020).

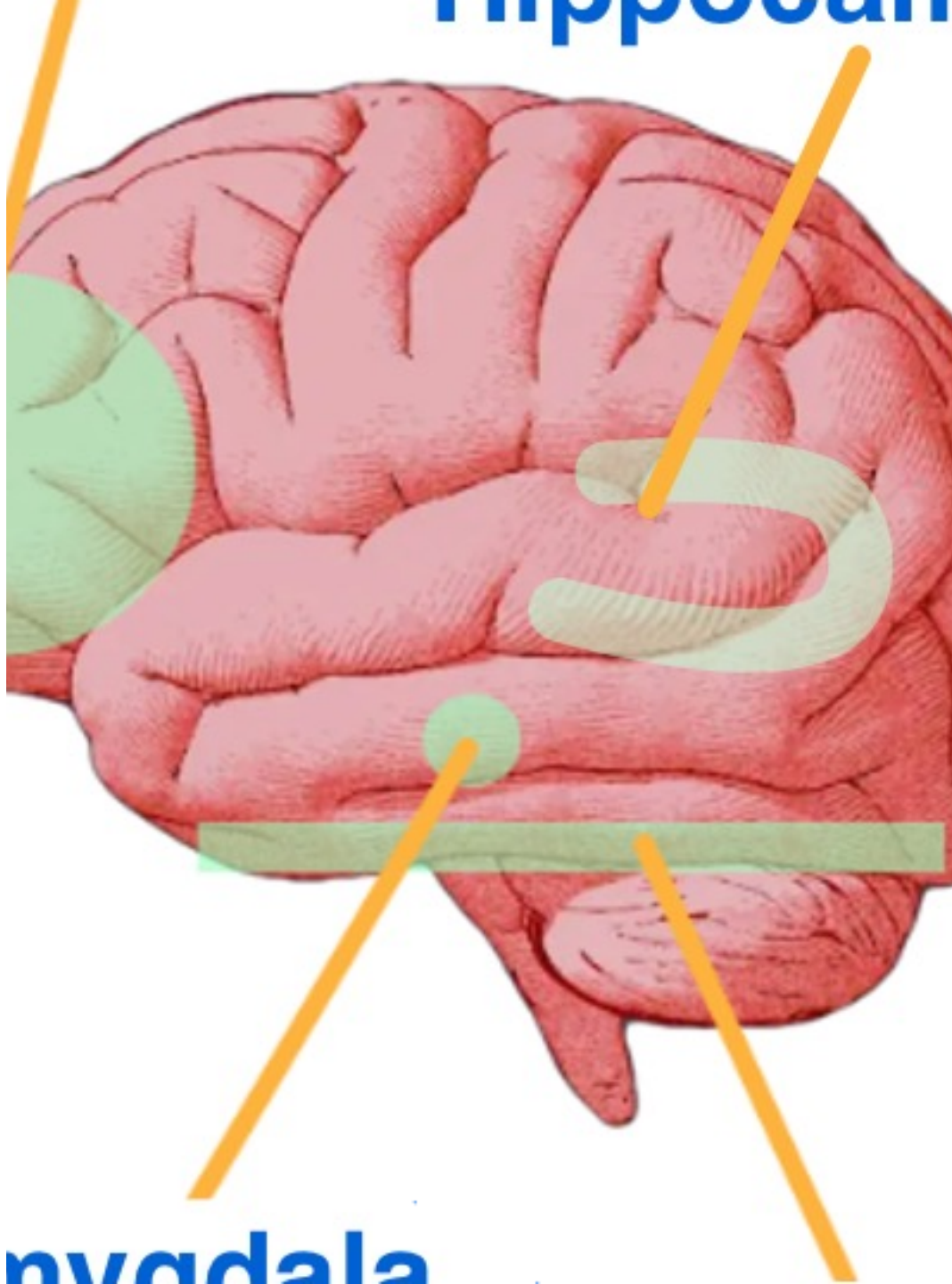
# Resulting Differences in Neurological Functioning

Review and learn in detail specific brain structures responsible for:

- **Speech and Language**
- **Other Social Communication Skills**

# Some Key Brain Areas

- Hippocampus
- Prefrontal Cortex
- Amygdala
- Fusiform Gyrus



# The Hippocampus

Cognitive Mapping

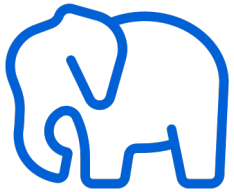
Affordance Perception

Model-Based Planning

# The Prefrontal Cortex

Decision Making

Personality and Behavior Regulation



Working Memory

Attention and Concentration

# The Amygdala

Key Emotional Processor

Fight or Flight Response

Emotional Memory

Social Interactions

Decision Making

# The Fusiform Gyrus

Face Recognition

Reading

Connectivity

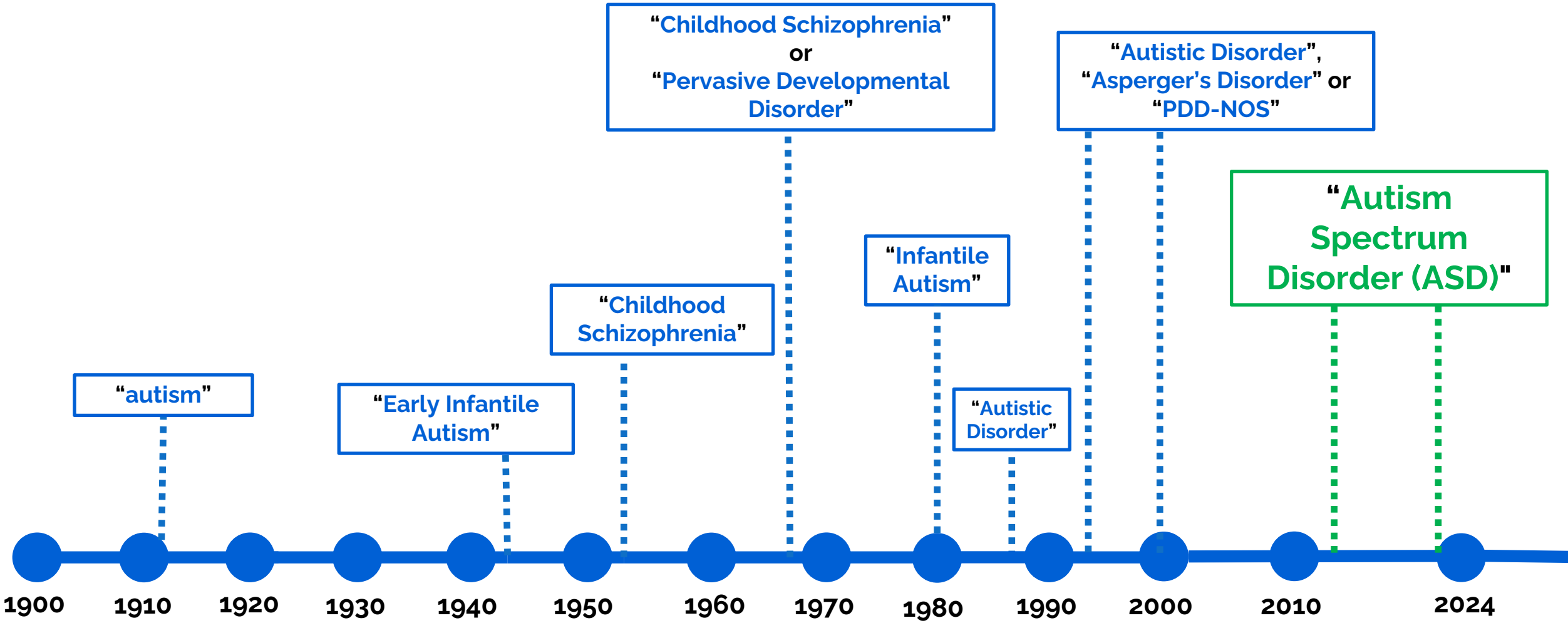
# Stay informed

- Keep up with the **most current research** on ASD.
- Understand how ASD affects **individuals and those around them**.
- Understand the **wide-ranging** impacts of ASD.

# Section II

## A Brief History of Autism Spectrum Disorder

# A Brief History of ASD



# 1 in 36

## 2.78%

**of children diagnosed with ASD in the United States**

**2024**

# Section III

## Current Definition / Diagnostic criteria of ASD (DSM-5-TR)

# Terminology

- Autism Spectrum Disorder or (ASD) according to definition in **DSM-5-TR** (APA, 2022)

- Corresponding ICD-11 code is **6A02** (WHO, 2018)

## Criteria A

“**Persistent deficits**" in **three areas** of social communication including:

1. **Reciprocity**
2. Comprehension and use of **nonverbal communication**
3. Difficulties establishing and maintaining **social relationships**

# Criteria B

**At least two** of four stereotypical behaviour patterns:

1. **Stereotyped or repetitive** movements
2. Insistence on **sameness**
3. Highly restricted, **fixated interests**
4. **Hyper/hypo reactivity** to specific stimuli

# Criteria C, D and E

The observed symptoms :

C. Occurred **early in development**

D. “Cause clinically **significant impairment** in social, occupational, or other important areas of current functioning,” (APA, 2022)

E. Are **not explained by other categories of disability** such as intellectual disability and global developmental delay.

# Additional Specifications:

Diagnosis must specify if:

- There are accompanying **intellectual and language impairments**.
- The condition is associated with a known **medical** or **genetic** condition or **environmental** factor
- The condition is associated with another **neurodevelopmental**, **mental**, or **behavioral** problem.

# Section IV

## Identification and Evaluation of Communication Impairments in ASD

# ASD in Early Infancy (0-36 months)

## Early Motor Impairments

- Delayed developmental milestones **(C)**
- Slow or stagnant development of gross and fine motor skills **(C)**
- Prone to accidents **(C)**

# Early Signs of Social Communication Impairments

- Lack of response to name **(A2)**
- Slow vocabulary development **(A1)**
- Delayed speech or symbolic communication **(A1)**
- Limited reciprocal interactions **(A3)**
- Limited attention to people's faces **(A2)**
- Difficulty establishing nonverbal communication **(A2)**

# Repetitive and Restricted Behaviors and Sensory Sensitivities

- Repetitive movements **(B1)**
- Strong adherence to routines **(B2)**
- Intense interests in specific objects **(B3)**
- Over- or under-reactivity to sensory input **(B4)**
- Unusual sensory interests **(B4)**

# ASD and Early Childhood (3-6 years)

# Social Communication Challenges

- Difficulty with peer interactions **(A1)**
- Limited use of gestures and facial expressions **(A2)**
- Delayed or unusual speech development **(A1)**

# Repetitive and Restricted Behaviors

- Repetitive play patterns **(B1)**
- Insistence on sameness **(B2)**
- Intense focus on specific interests **(B3)**

## Sensory Sensitivities

- Over- or under-reactivity to sensory stimuli **(B4)**

- Unusual sensory interests **(B4)**

# Impact on Learning and Development

- Difficulty with imaginative play **(A3)**
- Challenges in following instructions **(A2)**
- Emotional regulation difficulties **(A3, D)**

# Speech and Language Evaluation Tools for Early Infants and Young Children

## Standardized Assessment Tools for Early Infants and Young Children

- Social Communication Questionnaire (SCQ; Rutter et al., 2003)
- MacArthur-Bates Communicative Development Inventories (CDI; Fenson et al., 1994)
- Preschool Language Scale, Fifth Edition (PLS-5; Zimmerman et al., 2011)
- Mullen Scales of Early Learning (MSEL; Mullen, 1995)

# Non-Standardized Assessment Tools for Infants and Young Children

- Language Sample Analysis
- Interviews and Observations with parents and primary caregivers
- Developmental Observation Checklist System

# ASD and the School Age (6-12 years)

# Early Developmental History

- Delayed language development **(C)**
- Co-morbidities **(D)**
- Associated with known medical or genetic conditions **(E)**

# Behavioral Patterns and Sensory Sensitivities

- Repetitive movements **(B1)**
- A strong adherence to routines and Intense interests **(B2, B3, B4)**
- Rigidity in thinking **(B2)**
- Hyper- or hypo-reactivity to sensory input **(B4)**
- Unusual sensory interests **(B4)**

## Signs of Social Communication Impairments in School Environments

- Struggles with conversational skills **(A1)**
- Difficulties with attention and joint attention skills**(A3)**
- Difficulty with social interactions . **(A2, C)**
- Difficulty making friends **(A3)**

# Speech and Language Evaluation Tools for School-Age Children

## Standardized Assessment Tools for School-Age Children

- Clinical Evaluation of Language Fundamentals, Fifth Edition (CELF-5; Wig et al., 2013)
- Peabody Picture Vocabulary Test, Fifth Edition (PPVT-5; Dunn, 2019)
- Test of Narrative Language, Second Edition (TNL-2; Gillam & Pearson, 2017)

## Criterion-Referenced and Non-Standardized Assessment Tools for School-Age Children

- Dynamic Assessment of Language Learning
- Educator and Parent Questionnaires
- Play-Based Assessments

# ASD During the Teen Age (13-17 years)

## Social Communication Challenges

- Limited back-and-forth conversations **(A1)**
- Difficulty understanding social cues **(A1, A2)**
- Challenges in making and maintaining friendships **(A3)**

# Repetitive and Restricted Behaviors

- Strong adherence to routines **(B2)**
- Intense interests or preoccupations **(B3)**

## Sensory Sensitivities

- Over- or under-reactivity to sensory input **(B4)**
- Stereotypical, repetitive and restrictive behaviors **(B1)**
- Seeking sensory input **(B4)**

# Impact on Academic and Daily Life

- Difficulty with executive functioning **(A1, A2, A3)**
- Social isolation **(D)**
- Heightened anxiety and stress **(D)**

# Speech and Language Evaluation Tools for Teenagers

# Standardized Assessment Tools for Teenagers

- Clinical Evaluation of Language Fundamentals, Fifth Edition (CELF-5; Wig et al., 2013)

- Test of Narrative Language, Second Edition (TNL-2; Gillam & Pearson, 2017)

- Social Language Development Test – Adolescent (SLDT-A; Bowers et al., , 2008)

## Criterion-Referenced and Non-Standardized Assessment Tools for Teenagers

Language Sample Analysis

Observations of Peer Interactions

Self, Educator and Parent Questionnaires

# ASD and Adulthood (18+ years)

# Social Communication Challenges

- Difficulty with social interactions **(A1)**
- Maintaining relationships **(A3)**
- Literal interpretation of language **(A1, A2)**

# Repetitive and Restricted Behaviors

- Repetitive routines and rituals **(B2)**
- Focused interests **(B3)**
- Repetitive movements **(B1)**

## Sensory Sensitivities

- Sensitivities to sensory stimuli **(B4)**
- Limited exposure to social and otherwise learning activities **(B4)**

# Impact on Daily Life and Employment

- Low employment rates **(D)**
- Difficulties with social communication in the workplace**(D)**
- Difficulty understanding and following workplace rules**(D)**

# Speech and Language Evaluation Tools for Adults

# Standardized Assessment Tools for Adults

- Assessment of Functional Living Skills (AFLS; Partington & Mueller, 2013)
- Communication Checklist - Adult (CC-A; Bishop, 2013)

# Criterion-Referenced and Non-Standardized Assessment Tools for Adults

Narrative Analysis

Workplace Communication Observations

Functional Communication Assessments

# Considerations for Assessment Design

**Select** your assessment tools **carefully**.

Stay up to date about the **physical and mental wellbeing** of the person and family members.

**Refer as needed** while providing adequate support.

# Considerations for Selection of Interventions

Select interventions grounded in **rigorous research**.

Learn to **adapt and individualize** interventions for each individual.

Use a **variety** of interventions depending on individual need.

# It was our pleasure!

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