SESSION 3: DIAGNOSIS AND IDENTIFICATION
Catherine Lord
July 2, 2015 // 11:00 am EST

Course Materials
These materials are intended to provide some background information and organizational structure to the Summer Institute session on diagnosis and identification. The materials for this session were designed to enhance trainees' learning experience from this session and to provide background information for trainees who have limited prior exposure to diagnostic instruments and core behavioral phenotypes in ASD. The design of this session assumes that trainees are familiar with the concepts covered in this document, including the glossary of terms and primary suggested readings, prior to the session. Additional resources and suggested readings are provided for trainees desiring a more in-depth consideration of these issues.

These materials were developed by Allison Meyer (graduate student at UNC; allison.meyer@unc.edu), Rebecca Grzadzinski (graduate student at Cornell/Columbia University; reg2016@med.cornell.edu), and Meagan Talbott (postdoctoral fellow at UC Davis MIND Institute; mtalbott@ucdavis.edu).

Register for this course and other sessions in this series at sfari.org/summer
Learning Objectives
The Summer Institute for Autism Research was established in direct response to requests from early career researchers (graduate students, postdocs, etc), who asked INSAR and SFARI for greater training opportunities in multidisciplinary topics. In designing the Summer Institute, the priorities were: (1) to provide a multidisciplinary training platform for young scientists from various backgrounds; (2) allow international participation; and (3) make it freely available. Thus, the inaugural Summer Institute covers broad topics (which are geared to researchers outside the respective topic areas), is offered over a free web platform, and allows researchers from around the world to connect with the presenter. The overarching goal of the Summer Institute is to expose junior scientists to topics they are not currently engaged in, with the hope that basic scientists and clinical scientists could learn from each other to ultimately advance the understanding of autism spectrum disorders.

The current session, Diagnosis and Identification, is lead by Dr. Cathy Lord. The overarching objective of this session is to provide the trainee with insight into the standard diagnostic procedures for Autism Spectrum Disorder (ASD) and the limitations of these standard procedures. Participation in the course will provide trainees with an understanding of:

1. The different aims one might have in attaining a standard diagnosis
2. The limits of current diagnostic measurement
3. Factors to consider when interpreting scores from existing diagnostic measures
Glossary of Key Terms
Below is a breakdown of some of the technical terminology used in this session.

1. **Diagnostic and Statistical Manual-Fifth Edition (DSM-V):** This manual describes the diagnostic criteria for neurodevelopmental, psychiatric and mental disorders including Autism Spectrum Disorder (ASD). Previous versions of the DSM included a category called Pervasive Developmental Disorders which encompassed Autistic Disorder, Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS), Asperger’s Syndrome, Rett’s Syndrome, and Childhood Disintegrative Disorder. Individuals with diagnoses of Autistic Disorder, PDD-NOS, or Asperger’s Syndrome based on DSM-IV were all considered part of the autism spectrum. Diagnosing ASD using DSM-V criteria requires an evaluation of an individual’s behavior in the following domains: 1) Social communication and social interaction and 2) restricted, repetitive patterns of behavior or interests. To make a diagnosis, these behaviors must occur in early in development, cause significant functional impairment, and not be fully explained by the individual’s general intellectual ability.

2. **Social-emotional reciprocity:** sharing of interests and emotions (affect), behaviors used to initiate and regulate social interactions (greetings, turn-taking, conversational skills), and responses to others’ social bids. Individuals with ASD may have impairments in this area that include various aspects of social-emotional functioning, such as difficulty maintaining a back-and-forth conversation (not responding to another person’s comments, talking at length on a topic of interest without referencing the other person’s interest or own thoughts).

3. **Nonverbal communication:** Communicating information and engaging in social interactions with others without using words or vocalizations. For example, nonverbal communication refers to communication through eye contact, gestures (e.g. pointing, showing the size of an object), body language, or facial expressions. Individuals with ASD may have impairments in this area of communication exhibited by atypical eye contact (e.g. looking away from a person while talking to them or overly focused eye contact), lack of or odd facial expressions, or limited use of gestures to communicate information.

4. **Repetitive behavior:** Repeated actions or behaviors outside of what is developmentally appropriate for an individual. Repetitive behavior can include repeated motor movements (e.g. flapping hands), repetitive language (e.g. repeating back words from a TV show or echoing another individual’s words), or repeated use of objects (e.g. spinning wheels on a toy truck, lining up objects).

5. **Insistence on sameness:** This symptom includes difficulty managing changes in routine and extreme distress when there are small changes. Individuals with ASD may also want certain situations (including social situations) to be exactly the same every time.

6. **Restricted interests:** Restricted Interests in individuals with ASD are very intense and/or abnormal in their scope. For example, and 5-year-old boy with ASD may have a restricted interest in dinosaurs which is developmentally appropriate for his age. However, his focus on this interest is very intense and he has difficulty playing with toys that are not dinosaurs or having conversations that are not related to dinosaurs. Restricted interests may also be unusual in their scope. For example, a young child with ASD may be very interested in antique electronics or drainpipes which is not an interest that is otherwise developmentally appropriate.
Other Relevant Key terms:
1. **Autism Diagnostic Observation Schedule (ADOS):** A semi-structured, observation-based assessment of social-communication and restricted/repetitive behaviors used to aid in the diagnosis of ASD. The ADOS consists for 5 different modules. The module for an assessments is chosen based on the individual’s language ability and age.

2. **Autism Diagnostic Interview-Revised (ADI-R):** A standard and comprehensive parent interview used to assess for behaviors associated with ASD currently and in the past.

3. **Screening measures:** A number of short, parent-report measures can be used during the diagnostic process. These measures are used alongside behavioral measures (e.g. an ADOS) and parent interview to help inform diagnostic decisions. Some of those screening measures include:
   a. Social Communication Questionnaire (SCQ)
   b. Social Responsiveness Scale (SRS)
   c. Child Behavior and Communication Checklist (CBCL)

4. **Item Response Theory:** Used in psychometrics in the design and analysis of assessments (e.g., tests, questionnaires) that focuses on the item-level as opposed to the test-level understanding.
Primary Suggested Readings:


*Participants who are unfamiliar with and/or would like to see examples of the behavioral constructs discussed here are encouraged to view the Autism Speaks Video Glossary, which provides video examples of each of these behaviors in both typically developing young children and those with ASD. [http://autismspeaks.player.abacast.com/asdvideoglossary-0.1/autismspeaks/login](http://autismspeaks.player.abacast.com/asdvideoglossary-0.1/autismspeaks/login)*

Further Suggested Reading:


Get Engaged

Continue the conversation and connect with peers currently working or interested in autism.

- Join SFARI’s private autism research discussion group on Facebook: http://on.fb.me/1yWVOoO
- Become an INSAR member: http://www.autism-insar.org/membership
- Contact the INSAR Student & Trainee Committee: studentcommittee@autism-insar.org