Diagnosing Autism Spectrum Disorder: Who will get a DSM-5 diagnosis?

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Key Points

- There are concerns that the proposed DSM-5 criteria will not identify the more able individuals currently diagnosed with high-functioning autism and Asperger syndrome.
- Only one study has reported both good sensitivity and specificity in the same sample.
- No study has yet considered how changes at the ‘sub-domain’ level might affect overall sensitivity and specificity, and few have included individuals of different ages and ability levels.
- The current research designed and compared different DSM-5 algorithm versions using items from the Diagnostic Interview for Social and Communication Disorders (DISCO).
- Our results clearly demonstrate that algorithm design is an essential consideration when designing new diagnostic tools to meet the proposed DSM-5 criteria.
- Moreover, good sensitivity was reported across age and ability level, suggesting that the algorithms do not miss higher functioning individuals.

CRITERIA FOR DSM-5 ASD:

- Social-emotional reciprocity
- Developing and maintaining relationships
- Non-verbal communication
- Impaired Social Communication
- Restricted and fixedated patterns of behaviours
- Repetitive and stereotyped speech
- Excessive adherence to routines
- Fixed and rigid adherence to routines
- Hyper or hypo-reactivity to sensory input

Algorithm design

- The DISCO is based on the concept of a spectrum of autistic disorders that predated the earliest ICD and DSM criteria for autism and is therefore not constrained by existing international diagnostic classifications.
  - Items were selected from the 320 DISCO items and mapped onto the DSM-5 criteria.
  - The assignment of items was confirmed by three independent clinicians.
  - An algorithm is a set of items from the DISCO that are combined by the DSM-5 “rules” i.e. must meet all three A sub-domains, to provide a diagnostic algorithm.
  - Three versions of the DSM-5 algorithm differed in number of DISCO items required at the sub-domain level:
    - Initial: thresholds were set to one item as proposed by DSM-5
    - Youden J – optimal thresholds for sensitivity and specificity were chosen using the point at which each sub-domain ROC curve maximally deviated from the chance line using the Youden J statistic: (maximum=(sensitivity + specificity)-1).
    - Modified: this algorithm also used sensitivity and specificity values calculated from ROC curves; the threshold was selected that maximised specificity while maintaining the highest level of sensitivity.

Relaxing the criteria?

- Previous work has “relaxed” the DSM-5 criteria in order to improve sensitivity. In the current study only the sensitivity of the Youden J algorithm was improved by relaxing the A rule (2/3) (sample 1=.94; sample 2=.71), but this is still less sensitive than the “unrelaxed” Initial (1.94) and Modified algorithms (1.85).
- As the purpose of DSM-5 is to guide clinicians and should represent the current conceptualisation of ASD, we argue that relaxing of criteria is less optimal than the use of an algorithm that matches directly the pattern of behaviours specified by these guidelines (e.g. the Modified Algorithm).

Testing the three DISCO DSM-5 algorithms

- Sample 1: 36 ASD; 17 intellectual disability; 14 language impairment
- Sample 2: 52 ASD; 26 intellectual disability
- The current study is one of the first to develop and validate a new DSM-5 algorithm from a single standardised diagnostic tool.
- The Initial Algorithm had the highest level of sensitivity (identified most individuals with ASD) but lowest specificity (identified too many of the control group).
- The Youden J Algorithm had the highest level of specificity but lowest sensitivity.
- The Modified Algorithm had comparable sensitivity to the Initial Algorithm and comparable specificity to the Youden J Algorithm using McNemar’s test (p=.01).

DSM-5 across age and IQ

- The algorithms did not vary across age group or high and low ability using chi-square tests. The initial and modified DISCO DSM-5 algorithms suggest individuals across a broad range of age and abilities will receive a DSM-5 diagnosis.
- Why is the DISCO good at capturing DSM-5 across age and IQ?
  - Endorsement for the majority of items (72%) was consistent across age and ability.
  - A small set of items were observed in above 90% of cases in the high and low ability groups and in both children and adults.
  - Sharing interests and enjoyment; friendships; awareness of others’ feelings
  - In addition, a small minority of algorithm items were more relevant for higher functioning individuals:
    - Reciprocal communication, interrupting conversations, anger toward parents, long-winded and pedantic speech, maintenance of sameness in routines, repetitive themes, insistence on perfection, collecting facts on specific subjects, repetitive activities related to special skills
    - And a small number of items more relevant for older individuals:
      - Anger toward parents, imaginative activities, long-winded and pedantic speech, tone of voice, repetitive themes, collecting facts on specific subjects
    - The combination of these global, as well as more specific, items may have contributed to the inclusivity of the DISCO DSM-5 algorithms.

References and Contact

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