Autism Interest Group:

Differential Diagnosis of Autism Spectrum Disorders

or, Refining “Autism Awareness”

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3 Referrals to Child Development Clinic, Chosen at Random:

- “Autism? Unable to stay on task; can’t concentrate on one subject; quick to anger; hyperactive; social delays”
- “Autism? Only says a few words; flaps his hands, rubs his arms; limited eye contact; abnormal social development; stuffs his mouth with food.”
- “Autism? Very bright; excels in reading and writing; some social difficulties; has some quirky behaviors.”

» All 3 referrals could have ASDs.
» Then again, none may end up qualifying for ASD diagnosis
» In current era, of autism awareness, there seems to be some confusion that a referral for ASD diagnostic evaluation does not necessarily mean that a child has ASD
One important outcome of increased autism awareness is increased autism screening

- Purpose of screening is to cast a wide net, to make sure that people who might have a given condition receive diagnostic testing, and, if indicated, treatment.
- Screens, by definition, have high sensitivity and low specificity—in other words, a good screen should pick up 100% of people with the condition of concern, but should also pick up a large number of people who DO NOT have the condition.
- Screening is just the first step—a positive screen must be followed up by diagnostic testing.
Children who struggle with social interactions should be referred for further diagnostic assessment.

- But there are many causes of social difficulties.
Reality:

- Tourette’s/Tic Disorders
- ASD
- Anxiety
- ID, Aka “MR”
- RAD
- ADHD
Apparent Popular Perception:

ASD

Tourette's Syndrome
Tic Disorder

Autism Spectrum Disorder
Differential Diagnosis of ASD:

And the challenge of communicating these alternative diagnoses in this era of Autism Awareness:

- Intellectual Disability—now, more stigmatized than ASD; drop off in ID dx mirrors ASD’s rise.
- Global developmental delay—too vague for a lot of parents; feels like a lack of a dx
- ADHD—many parents assume that Rx will be forced.
- Mental health conditions, including depression, anxiety, adjustment d/o, PTSD, and Reactive Attachment Disorder—highly stigmatized!
- “Normal”, but quirky, temperamental, or “poor fit”—can be very challenging to communicate these days; parents may feel that you don’t believe them or are being dismissive of their concerns.
Other considerations when giving the “bad news” that it’s not ASD

• Myth that ASD is an especially treatable disability—we professionals are partly to blame for that, with emphasis that early dx of ASD is especially important

• Many parents assume (often incorrectly) that ASD is ticket to great gobs of services
  – Educational (may route child to more restrictive environment than necessary)
  – Medical (theoretically true, since passage of ABA law, but currently, there is no payment mechanism in place for ABA, and, actually, denials of MH services for ASD are common)
  – DD Services—ASD can help access these services, but is not the only way to access these services
  – SSI-- ASD can help access these funds but is not the only way.
A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, and manifest by all 3 of the following:

- 1. Deficits in social-emotional reciprocity
- 2. Deficits in nonverbal communicative behaviors used for social interaction
- 3. Deficits in developing and maintaining relationships
B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least two of the following:

- 1. Stereotyped or repetitive speech, motor movements, or use of objects
- 2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change
- 3. Highly restricted, fixated interests that are abnormal in intensity or focus
- 4. Hyper-or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment;
DSM V Diagnostic Criteria, cont’d

• C. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)

• D. Symptoms together limit and impair everyday functioning

• Note: Asperger Disorder and PDD-NOS have been folded into ASD and are no longer discrete diagnoses
So, what’s involved in a “gold-standard” ASD assessment?
Diagnostic Measures

- Autism Diagnostic Interview, based on DSM-V:

- Autism Diagnostic Observation Schedule: Structured diagnostic observations of child (ADOS)
Other Components of Diagnostic Assessment for ASD

- Cognitive assessment
- Adaptive behavior functioning
- Language and speech assessment
- Assessment of sensory processing and fine motor functioning
- Assessment of family history, social history, family dynamics
Some cases, to illustrate differential diagnoses:
“Austin”, background info:

- 6 year-old boy, has been diagnosed with “Asperger’s” by local psychiatrist and has educational eligibility for ASD services.
- Concerns include: “speech delay, little interaction with peers, and temper tantrums.”
- He expresses needs and wants, but can’t really converse.
- He parallel plays, but doesn’t engage in much actual play with peers.
- Mom notes that he is obsessed with dinosaurs, is quite brilliant about them, and can identify all their names.
- Mom notes that Austin seems to be a lot like a neighbor who also has been diagnosed with “Asperger’s”. 
“Austin”, assessment:

• As I entered the room, Austin turned to me, came over, and held out an allosaurus toward me, and said “Wook! T-Wex!”
• “Very good!” I said, “and what’s this one?”, holding out an apatosaurus.
• “Dat one’s a wong-neck!” he replied eagerly.
• During the rest of the interview, Austin played happily, first with toy trucks, which he used appropriately; next, with the toy kitchen, offering me “food” he had made.
• Austin enjoyed the examination, and then enjoyed using the instruments to examine me.
• On team assessment, cognitive, adaptive, language, and fine/visual motor skills are all noted to be impaired. ADOS is non-clinical.
“Austin”, impressions:

- Austin is diagnosed with Intellectual Disability
- Note: Austin has “persistent deficits in social communication and social interaction” but these deficits ARE accounted for by general developmental delays.
- Sociability and communicative intent are actually relative strengths for Austin.
- Educational and mental health interventions for Austin should be planned with the understanding that social skills are a relative strength!
“Cody”, background info:

• 9 year old boy with extreme mood swings, high degree of anger and frustration, low self-esteem
• At grade level, but underperforming, since he doesn’t complete assignments. Short attention span and hyperactive in class.
• Gets “stuck”—if he feels he has been wronged, he can’t move on—this can ruin a whole day.
• Has trouble keeping friends. Is intense and “smothers”. Violent talk at school.
• Soils himself on regular basis, and hides underwear.
• School has enrolled Cody in IEP with ASD eligibility
• Cody is referred to therapy—therapist makes ASD diagnosis, and then drops Cody from services, as “MH agencies can’t treat ASD”.

“Cody”, assessments:

- Behavioral questionnaires – parents and teachers endorse ADHD symptoms and impairment.
- ADOS completely negative.
- Normal cognition, language
- Delayed academics, fine motor skills
- Generally disorganized, distractible, hyperactive, and impulsive during the visit.
“Cody”, Impressions:

• Social impairment is secondary to ADHD and also encopresis (soiling)
• Though he doesn’t have ASD, social skills class may well still be appropriate, depending on goals.
• Behavioral and medical treatment of ADHD and encopresis recommended.
• Focus on family therapy, rather than on individualized social skills counseling, recommended
• Note: Cody can recite rules of social interaction in clinic, but has difficulty generalizing these skills to life, due to his impulsivity
“Maxine”, background info:

- 4 year-old girl, with history of prenatal alcohol exposure, abuse and neglect for first 3 years, DHS involvement.
- Mom has now been sober for 1 year, and Maxine is now back with mom.
- Mom’s current concerns: aggression, tantrums, defiance, self-injury.
- History of feces smearing and eating, which has improved; ongoing toilet resistance.
- At pre-school, participates in structured activities, but shows little interest in peers.
- “Fixates” about “wanting to going to the park to play.”
- Mom acknowledges FASD and PTSD as possibilities, but states that she thinks Maxine has autism.
“Maxine”, assessment:

- Turns to me, makes eye contact, smiles
- Spontaneously starts conversation about toy barn
- Brings toy animals to me and pretends to make them make noises
- Pretends to examine me with stethoscope, otoscope, hits me a little too hard with the reflex hammer, and pronounces with a sly smile that I am “very sick, very sick!”
- ADOS—non-clinical
- Cognitive, language, fine-motor skills: average
“Maxine”, impressions:

• Clearly, NOT consistent with ASD
• Not consistent with classic FASD, either, though subtle effects of FAE cannot be ruled out.
• Clearly, the primary behavior problems are attributable to history of abuse/neglect/disrupted attachment.
• While making the diagnosis is straightforward, conveying this information sensitively is very challenging:
  – ASD dx used to implicate “refrigerator moms”
  – Parent-blaming has (thankfully) fallen out of fashion in our understanding of the etiology of ASD.
  – Problem is that ASD diagnosis is now a sought-after absolution for parents who DO need to acknowledge the role of environmental factors in causing maladaptive behavior in order to fix that behavior.
“Krystal”, Background Info:

- 9 year-old girl, speaks only to mother and 3 other people: aunt, friend, and friend’s mother.
- Performing below grade-level; doesn’t ask questions so doesn’t get needed help.
- Plays with other children at recess, but never talks.
- Play-dates with one friend, but never talks to this friend.
- No c/o early language dev’t. At age 4, gradually began to taper down the list of people she would speak to.
- Around age 4, grandmother died suddenly.
- School provides speech therapy, but she refuses to speak during therapy. Has been receiving counseling, with no improvement. Therapist suspects ASD.
“Krystal”: Exam/Observations:

- On exam, she is cooperative, makes eye contact, exhibits typical social referencing and joint focus of attention, but doesn’t vocalize.
- During the interview, she spends 45 minutes, completely focused on her self-portrait, which she draws meticulously, and never gets to the other 2 assignments:
“Krystal”, Impressions:

• Selective mutism (form of social anxiety).
• SLP agreed; other school personnel did not, but continued to insist on ASD eligibility.
• I rec’d continued counseling and also Rx’d SSRI.
• A few weeks into Rx she spoke to a waitress.
• > month into treatment, she spoke to her best friend for the first time.
“Ray”, Background Info:

• 14 year-old boy: “Does he have Asperger’s or is he just a butt-hole?”, mother asked me, in front of her son.
• Difficulty making friends; annoyed with others easily.
• Number of annoying habits, including throat-clearing, coughing, making “fart noises”, bouncing, tapping, head-rolling, and fidgets.
• Teased about these behaviors and would like to stop.
• Ray has been diagnosed in the past with ADHD and treated with stimulant, which caused exacerbation of tics, weight-loss, and diminished energy. Several other stimulants were tried, with similar effects.
• Ray has begun to hoard things; was dx’d with OCD.
• Aggression towards sister and cat escalated; the family was beginning to consider residential treatment.
• A new psychiatrist dx’d Asperger Disorder, prescribed anti-psychotic medication, with some improvement in tics and attitude, but also significant weight-gain and sedation.
“Ray”, assessment:

- Exam: Pleasant, cooperative, normal non-verbals and prosody.
- Little ticcing seen in office, some fidgetiness
- ADOS—non-clinical
- Normal cognitive and language skills
- Obese
“Ray”, Impressions:

- Tourette syndrome, with secondary social impairments.
- The key is that Ray is bothered by these habits.
- Also, it is important to note that while Tourette’s is defined by the tics, it has associated features, including the temper flares, OCD, and ADHD symptoms.
- I explained the diagnosis—mother visibly softened with news.
- Ref’d to counseling. Rec’d tic accommodations. Rec’d sports/ exercise. Rec’d d/c-ing anti-psychotic medication.
- On follow-ups over the next year, he is doing very well, academically and socially, off of all medication.
“Melvin”: Background Info:

- 14 year-old boy with previous diagnosis of high-functioning autism, first diagnosed at 3 years of age
- His mother wants to know, does he still have high-functioning autism or Asperger Disorder?
- Now is in high school, getting straight A’s; lettered in cross-country and choir, on the chess club, has many friends.
- Plays piano concerts; sells tickets -- half the proceeds for college and donates the other half to charity, incl. CDRC!
- Intense interests in piano, choir, chess, and the Trailblazers, but these interests aren’t intrusive.
- One repetitive activity: repeatedly throwing a ball at a wall, daily, for an hour after school as his “down time”.
- He dated a girl for awhile. After a few dates, she tried to kiss him and he said he wasn’t ready. She, taken aback, “What, are you autistic?” Melvin said no, but afterwards was wracked with guilt for having lied. Mother wonders if Melvin will be able to be intimate in the future.
"Melvin", Assessment:

- Exam: Very pleasant, but with flat affect, stiff mannerisms, unnatural and unrelenting gaze at times—eye contact appears "learned", which it is!
- Pedantic speech with flat prosody.
- On the ADOS, he scored solidly in the range of autism in Communication and Reciprocal Social Interaction.
- On cognitive assessment, he has superior scores in verbal and non-verbal domains; academics and vocabulary also in superior range.
“Melvin”: Conclusions

• Autism, like any developmental disability, is not just a list of symptoms -- the person must have disability resulting from that impairment.

• DSM-V. Section D. “Symptoms together limit and impair everyday functioning”

• If you make a charitable donation to the CDRC, you can have whatever diagnosis you pay for…! 
Summary

• Autism is not as rare as we used to think
• Autism is not as common as some people seem to think
• There are other (often more common) explanations for social difficulties, communication difficulties, and/or repetitive behaviors and movements, in addition to autism
• Autism Awareness is, of course, very important!
• And--diagnostic accuracy matters, too, to drive appropriate interventions, so diagnosticians must take care not to over-call ASD.
Thank you

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