Diagnostic Uncertainty

Q. Dr. Ruedrich, clinicians in the field of MR/DD have struggled with the difficulty of applying our standard diagnostic criteria to patients with mental retardation and developmental disabilities (MR/DD). Because patients with developmental delay and verbal impairments may not be approachable with the standard psychiatric diagnostic interview, we must rely on caregiver information, symptoms and signs that do not require verbal report, and our observational skills. With your years of experience specializing in the treatment of this population, has the diagnostic task become easier?

A. I am not sure the task is easier or if the clinician becomes more comfortable with uncertainty. As we know, the current DSM-IV is not well oriented toward people with MR/DD.\textsuperscript{3,17} It relies very strongly on verbal information, or self-report, of the patient's symptoms.\textsuperscript{9,14-16} Thus, when working with populations capable of limited self-report, clinicians commonly extrapolate the criteria to their specialized population. This is necessary when working with young children, impaired geriatric populations, and most individuals with MR/DD. Generally, clinicians are effective in treating these populations, and a number of authors have offered either modifications of the DSM criteria using derived diagnostic systems, or specialized rating scales and structured clinical interviews.

Q. Could you speak about these systems?

A. The DSM derived systems include the many published accounts of modified criteria, using either “MR” Diagnostic Equivalents, or “Behavioral Equivalents.” For example, if a patient cannot articulate feelings of depression, caregiver report of, for example, sad facial expression and lack of interest in activities could be substituted for those required DSM criteria, leading to meeting the required five of nine criteria for a diagnosis of depression.\textsuperscript{4,9,10,14-16} Other authors have developed rating scales and structured clinical interviews especially suited to individuals with developmental disability.\textsuperscript{1,7,8,11}

To my knowledge, however, there is little research supporting the notion that changing a criterion here or there validates or invalidates the diagnostic impression. However, I do believe that such schemas make the clinician more comfortable: these techniques are a piece of “data” that can help the clinician feel that he or she is maintaining diagnostic consistency across a clinical practice.

For example, aggression is commonly interpreted as a sign of depression, anxiety or a psychotic illness in a patient with MR/DD, but aggression does not appear in any of these diagnostic categories according to DSM-IV. We believe that certain forms or types of aggression may represent a manifestation of those disorders in patients with MR/DD, but we need to do that in an internally consistent manner.

Q. Aggression, which may be the most common mental health referral problem to psychiatry clinicians, may be a symptom of many illnesses, or no illness at all. We know that persons with MR/DD may live in environments that are ill suited to individual needs and aggression may be a simple expression of frustration or a coping mechanism. How does the clinician, then, use aggression, or other behaviors not found in DSM criteria, as symptoms of a psychiatric disorder?

A. This typical situation involves many unanswered questions. For example, I saw a patient yesterday who has moderate mental retardation, a choreiform movement disorder, is mute, and is being treated for a presumed mixed
mood disorders with a mood stabilizer and had been doing well. His caregivers reported that his self-injury had not recently responded to treatment. When we started to detail his episodes, there appeared to be a striking pattern. This gentleman likes to sit in a rocking chair and will rock until the chair moves against the wall. Then he becomes self-injurious until someone moves him away from the wall. Thus, self-injury was serving as a communication mechanism to obtain the assistance he needed from staff. This one “behavior” in the context presented does not serve well as a marker symptom of any psychiatric disorder. If not explored, such reports from caregivers would suggest that his treatment is ineffective; treatment would be changed and he may have become acutely ill again.

Q. How would you proceed to diagnose a person with MR/DD who does not present with a full syndrome picture of a psychiatric disorder and meets only some criteria for any disorder, but does have worrisome or dangerous behaviors such as SIB or aggression?

A. The first and most important step is to not view any symptoms or sign cluster as being based in the developmental disability itself. We all have been warned about diagnostic overshadowing, or seeing only the disability as being responsible for aberrant behavior rather than a psychiatric disorder.12 Second, the clinician must look for any possible environmental stressors or lack of fit in supports. Having done this, the next step is to make the best DSM-IV fit available as a provisional diagnosis.

Q. When you speak of the “best fit available,” would this process ever apply to a patient of normal intelligence?

A. The process is similar. There is a significant clinical subjectivity to the DSM criteria-based diagnostic method itself, and all clinicians have to use their total clinical experience. No one should apply these criteria in “cook-book” fashion with real world patients. This should be true for all of medicine, but in the psychiatric field we are unnecessarily defensive about our diagnostic uncertainty. Let me give an example I use when teaching medical students about psychiatry.

In this exercise, I ask students to list the signs and symptoms of acute appendicitis; there are about ten. We choose acute appendicitis because this is a diagnosis that most people feel is well delineated, common, and fairly reliably diagnosed. I ask them to number how many signs and symptoms he or she would require before going with a diagnosis of appendicitis and proceeding to operate. The numbers usually range from two to nine. This indicates very wide diagnostic subjectivity. I then ask them to assume the patient met all ten criteria, the appendix is removed, and on pathological examination of the appendix, there is no evidence of appendicitis. Instead, the patient had any one of many other disorders that can mimic the symptoms and signs of acute appendicitis. We know that surgeons aim to have 10 to 15% of scheduled appendectomies return as “normal,” and that is considered good practice. Psychiatry is the same. We have criteria, but there is no absolute certainty. The main difference is that today we cannot pathologically confirm our clinical diagnosis in a living patient. As a result, we become unnecessarily defensive, in that we cannot say, for example, that we correctly diagnosed 90% of cases of depression. Additionally, we have no laboratory tests that can aid assessment among our diagnostic criteria. We have to rely on largely subjective data.

The majority of the DSM criteria call for a subjective report by the patient. We often have to jettison that in diagnosing people with communicative language difficulties, or MR/DD below moderate levels, who may not be able to self-report at all.

Another important point is that many DSM disorders do not require objective distress on the part of the patient. The mental disorder may require distress and/or dysfunction. For example, a person may have a clinical depression even though he or she does not have subjective feeling of being depressed. A person who is manic feels great: everyone they interact with does not, and the patient may ruin his or her life and financial situation. So, the clinician must know the patient, and feelings about the patient, even though the patient’s self-report may be less than accurate.

Presently, it has become unfashionable in our field to admit to diagnostic uncertainty. In psychiatry, a decade or two ago, it was more acceptable to say that one’s diagnostic impression was provisional. Lately, either our own pride, or managed care requirements for documentation make us present our work in a way that conveys
more confidence in our diagnoses than we should be stating. The risk of this approach is that we sometimes believe ourselves, forget our diagnoses should begin as clinical impressions, and immediately treat the person as if he or she definitely has the condition. In reality, we have often based our diagnosis on caregiver observations and limited data. If we do not regard our diagnoses as provisional, it is less likely that we will entertain any subsequent diagnostic hypotheses that might be true. We should maintain that all our diagnoses are provisional and constantly reformulate our diagnostic thinking.

For example, I become concerned when I hear people say with certainty that a person with profound MR is depressed or psychotic. I saw a fellow yesterday for an initial visit who had a previous diagnosis of an anxiety disorder. His caregivers said he was anxious but could not say why they thought so. I asked about activity, expression, perseverative behaviors, and we could not come up with any subjective or objectively observed sign or symptom that was consistent with anxiety disorder. So, I tried to reformulate his diagnosis by taking a comprehensive history, and conducting a full mental status examination, looking for another DSM disorder that might better represent his symptoms. I settled on possibly Attention Deficit/Hyperactivity Disorder (ADHD), or Impulse Control Disorder, but I have the attitude that these are provisional. He may still have another psychiatric condition, a medical condition we cannot identify, or even a neuroleptic induced akathisia.

**Q.** So, in this case, you made a “presumptive” diagnosis. We often do such diagnostic labeling, and follow with a list of “rule-outs.” The patient may have a bipolar disorder, but we must also rule out depression, anxiety, psychosis, etc. But, the difficulty is that we can only “rule out” some medical conditions, such as hypothyroidism. Two decades ago, everyone in the field used to refer to their “working diagnosis,” and this probably best represented our work. We admitted to following an ongoing process where we were constantly reexamining our patients. Certainly, DSM has improved our practice by striving for diagnostic criteria. But DSM is also limited in delineating the “trajectory” of illness. All illnesses have a course; the symptoms of psychiatric diagnoses exacerbate and remit. But DSM-IV tends to take a cross-sectional snapshot. It is not longitudinal, giving the rich life course information that is so important to diagnosis. Do you agree?

**A.** Yes. With patients of normal intelligence, it is somewhat easier because we also expect them to give us their life course history. But with all patients, we know them when we follow them longitudinally. We know that with or without treatment, their symptoms are very different after months or years of follow up. For example, I have a patient of normal intelligence who in her earlier years, had a very serious eating disorder, and during those years, she may or may not have also had a depressive disorder. Now, she presents with a devasting and recurrent major depression, treated with many different combinations of medications and ECT. She still has some signs of an eating disorder, although she does not meet the criteria. Does she still have an Eating Disorder? When did one disorder begin and the other start? Were they there together? Is it in remission? All of these questions the clinicians must ask, in constant reformulation. Because we have no laboratory tests and no pathological confirmation, we have to have constant diagnostic reconsideration.

**Q.** Many clinicians feel that their diagnoses are supported when the patient responds to a medication. For example, if a patient’s symptoms are better when treated with Depakote®, he or she must have a bipolar disorder. Do you feel this logic is ever justified?

**A.** This use of post-hoc treatment analysis is common in psychiatry, but often not justified, particularly in persons with MR/DD who may be receiving symptomatic treatment for symptoms that are not obviously part of an identified psychiatric disorder. For example, Depakote® has mood stabilizing properties that may help ameliorate symptoms of bipolar disorder as well as depression and anxiety disorders. It also has anecdotal efficacy (non-specifically), for aggression and self-injury. Further, its side effects of sedation may also provide symptom relief. In no way does “response” to Depakote® prove the person had a bipolar disorder. This “post-hoc treatment analysis has led to an “epidemic” of bipolar disorder being diagnosed in many
psychiatric patients, persons with MR/DD, and children who have ADHD.

As psychopharmacology develops and we have more specific neurotransmitter probes, we can more accurately use post-treatment formulation. When we had “dirty” drugs (i.e., less specific in their actions), it was more difficult to decide what part of the patient’s symptomatology responded.

Q. What would be an example of a “clean” drug?

A. The SSRI’s (selective serotonin reuptake inhibitors, e.g., Prozac®), which also have little sedation and few side effects, are a good example. If a patient responds well to an SSRI, we can probably assume that he or she has a disorder of that neurotransmitter symptom.

Q. We know that SSRIs can treat generalized anxiety disorder, panic disorder, and depression; so can we say even our “cleanest” drugs have multiple effects?

A. Or, as likely, we may be artificially separating depression from anxiety, on a clinical basis, when there may not be that separation in the person. It may be that our models will be adjusted, and that patients may have etiologically overlapping conditions.

There is talk of changes in the schema for DSM-V. For example, groups interested in MR/DD want a more etiologically based system. To diagnose MR at present, we have a label on Axis II, mild, moderate, severe and profound. One proposal for DSM-V would identify an individual as having developmental disability due to an etiologic diagnosis. For example, a person could have “developmental disability due to Down syndrome.” Then, modifiers would add other information such as “needing extensive supports in home living,” along the lines of the Multidimensional/Intensity of Support model from the American Association on Mental Retardation definition. This would allow for more relevant assessment, and other important psychiatric information could be incorporated, such as the new information that is accumulating about behavioral phenotypes. Examples would be the eating disorders associated with Prader-Willi syndrome, or compulsions associated with Autistic Disorder.

Q. Will this address our most fundamental diagnostic uncertainties?

A. Not entirely. Clinical uncertainty is common in medicine. For example, a patient may present to their primary care physician with a sore throat, or “pharyngitis.” This may be due to streptococcal pharyngitis (“strep throat”) or many other conditions. But if no specific etiology is identified, the internist is quite comfortable diagnosing “pharyngitis due to unknown etiology.” DSM tried to improve our diagnostic rigor by establishing criteria that were very reliable—meaning they could be objectively measured, and in field trials clinicians using them would come to the same diagnostic conclusions. However, the validity, or whether these diagnoses were actually true or not, has still to be determined. As I said, so far we still have no pathological confirmation of our clinical impression.

Q. What would you recommend to clinicians to improve their practice?

A. One important thing we can do as teachers is to share our clinical uncertainty with our students—the mark of a good clinician is to not make a firm diagnosis at the initial visit or following a first contact. The best clinicians seek other information, assess the environmental fit, get to know the person over time and follow the longitudinal course, evaluate for other explanations like drug toxicity, medical conditions, side effects of other medications or psychotropic medications, and constantly reevaluate the initial clinical impression. This is true for all of psychiatry, but very much so for all patients with MR/DD, as well as children and organically or otherwise-impaired geriatric patients.

I think we can also say that clinicians should be more confident about diagnoses in MR/DD than we were 20 years ago. We should be proud of and embrace our diagnostic uncertainty because we are demonstrating we are better clinicians if we do, maintaining an open mind, examining diagnostic hypothesis, and reanalyzing our data. This includes interviewing multiples caregivers, and obtaining as much history as possible, and sometimes sending patients to a colleague for a second opinion. Although we can clearly say that...
clinicians working with persons with MR/DD can be more confident about our diagnoses than we could be 20 years ago, we have advanced far enough that we can sometimes say the emperor is naked and be comfortable with it.

Q. To conclude, is diagnostic uncertainty a good thing?

A. Yes.

REFERENCES


CORRESPONDENCE: Stephen L. Ruedrich, M.D., Department of Psychiatry, MetroHealth Medical Center, 2500 MetroHealth Drive, Cleveland, OH 44109, e-mail: sruedrich@metrohealth.org.
ANNOUNCEMENTS

March 28-30, 2001
5th Conference of the European Union of Supported Employment
Support for All
For more information contact:
Mike Evans
Employment Disability Unit
Dundee City Council
Dunsinane Avenue
Dundee DD1 3QN Scotland
Phone: +44(0) 1382 828180 • Fax: +44(0) 1382 828148
e-mail: mike.evans@dundeecity.gov.uk • suse.jobs@bigfoot.com
Web Page: www.suse.org.uk

April 30-May 4, 2001
YAI/National Institute for People with Disabilities
22nd Annual International Conference
Crowne Plaza Manhattan Hotel
New York, NY
For more information contact:
Aimee Matza
Phone: 1.212.273.6193 • Fax: 1.212.629.4113 • e-mail: amatza@yai.org

May 29-June 2, 2001
American Association on Mental Retardation
125th Annual Meeting
Pioneering New Directions in the 21st Century
Adams Mark Hotel
Denver, CO
For more information contact:
Phone: 1.800.424.3688 • Web Page: www.AAMR.org