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Utilizing a transdiagnostic neuroscience-informed approach to differentiate the components of a complex clinical presentation: A case report

Alicia B. Vanden Bussche^{a,b,c}, Nancy A. Haug^{a,b}, Tali Manber Ball^b, Claudia B. Padula^{b,c},
Andrea N. Goldstein-Pierarski^{b,c}, Leanne M. Williams^{b,c,*}

^a Palo Alto University, 1791 Arastradero Rd., Palo Alto, CA 94304, USA

^b Department of Psychiatry and Behavioral Sciences, Stanford University School of Medicine, 401 Quarry Rd., Palo Alto, CA 94304, USA

^c Veterans Affairs Palo Alto Health Care System, 3801 Miranda Ave., Palo Alto, CA 94304, USA

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ABSTRACT

Background: Recent research recognizes considerable overlap in the clinical presentation of psychiatric disorders such as Autism Spectrum Disorder, Attention Deficit Hyperactivity Disorder and Social Anxiety Disorder. The diagnostic approach collects symptoms to reflect a single underlying psychopathological process. The Research Domain Criteria (RDoC) emphasizes psychopathology as arising from combinations of abnormalities in core underlying constructs that can be measured at many levels of analysis, from biological to behavioral. Patients who present with clinical heterogeneity may benefit from transdiagnostic case conceptualization that integrates detailed symptom information across multiple measurements spanning multiple domains of functioning based in the RDoC framework.

Case presentation: We report on one case that was included in a research study focused on advancing knowledge towards a transdiagnostic, brain-based model of anxiety and depression. The 20-year-old male patient presented at a community mental health clinic for inattention, low mood, sleep problems and anxious symptoms. The patient also presented with primary problems in negative valence systems (anxiety, avoidance, and bias towards negative information), cognitive systems (fluctuating cognitive ability over time, poor concentration and ability to focus), and social processing systems (deficits in social communication skills). Conceptualizing this case through a transdiagnostic lens augmented the patient's treatment plan by including a more integrative approach. Treatment included social skills training, progressive relaxation exercises, and basic psychoeducation in emotional expression and independent living skills.

Conclusion: This case illustrates the utility of a transdiagnostic approach, particularly when a traditional diagnostic model generates conflicting evidence and/or multiple comorbidities. RDoC provides a framework for integrating abnormalities across multiple dimensions. Furthermore, it lays the foundation for future integration of brain-behavior relationships into case conceptualization and personalized treatment approaches.

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Abbreviations: ASD, Autism Spectrum Disorder; ADHD, Attention Deficit Hyperactivity Disorder; RDoC, Research Domain Criteria; SAD, Social Anxiety Disorder; MINI, M.I.N.I. International Neuropsychiatric Interview; DSM-5, diagnostic and statistical manual of the American psychiatric association 5th edition.

* Corresponding author at: Department of Psychiatry and Behavioral Sciences, Stanford University School of Medicine, 401 Quarry Rd., Palo Alto, CA 94304, USA.

E-mail addresses: avandenb@stanford.edu (A.B. Vanden Bussche), nhaug@stanford.edu (N.A. Haug), tmball@stanford.edu (T.M. Ball), claudiapadula@gmail.com (C.B. Padula), agoldpie@stanford.edu (A.N. Goldstein-Pierarski), leawilliams@stanford.edu (L.M. Williams).

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Background

Presenting psychopathology often does not fit neatly into our current diagnostic boundaries. An implicit assumption of the current diagnostic system is that the collection of particular symptoms reflects a single underlying psychopathological process. Complementing this approach, the Research Domain Criteria (RDoC) considers that complex clinical presentations arise from combinations of abnormalities in core underlying dimensions that can manifest across multiple levels of analysis, from biological to

behavioral. From this RDoC lens, an adequate description of psychopathology relies on more than categorical collections of symptoms and provides direct links to a brain-based understanding of psychopathology.

The Research Domain Criteria (RDoC) framework, an initiative within the National Institute of Mental Health, aims to better understand basic dimensions of functioning underlying the full range of human behavior from normal to abnormal [1,2]. The RDoC framework consists of functional constructs (i.e. concepts representing a specified functional dimensions of behavior), and proposes behavioral paradigms, brain circuits, genes, and molecules that can be used to measure them [1]. Constructs are grouped into domains of functioning, which reflect current knowledge surrounding major systems of cognition, emotion, motivation, and social behavior. Currently, there are five domains in the RDoC matrix: 1) negative valence systems; 2) positive valence systems; 3) cognitive systems; 4) systems for social processes; 5) arousal [1,2].

In the present case report, we integrated clinical, neuropsychological, and neurocognitive behavioral measures for a complex case of a young man presenting with symptoms of Autism Spectrum Disorder (ASD), Social Anxiety Disorder (SAD), and Attention Deficit/Hyperactivity Disorder (ADHD). We aimed to generate an integrated, transdiagnostic case conceptualization and use this conceptualization to develop a treatment plan.

Autism Spectrum Disorder is broadly described as markedly abnormal or impaired development in social interaction and communication, and is characterized by a restricted and stereotyped repertoire of activities and interests. Manifestations of autism vary, depending on age and level of development [3]. Children who are high-functioning (i.e., IQ within normal limits) and have milder presentations of ASD are less likely to be diagnosed until older childhood or beyond [4]. Diagnosis of ASD in this group can be complicated by clinical heterogeneity and comorbidity with other psychiatric disorders such as ADHD and SAD [5,6]. Similar to ASD, ADHD often involves difficulties with social interaction, communication, and restricted interests [7], while SAD can include social withdrawal, freezing or failing to speak in social situations.

Due to symptom overlap, these diagnostic categories may be a poor indication of the underlying pathology. In contrast, the negative valence systems, cognitive systems, and systems for social processes are key constructs that span ASD, ADHD, and SAD diagnoses. Thus, patients who have overlapping clinical presentations may benefit from an RDoC-informed, transdiagnostic assessment.

Case presentation

Presenting concerns

The patient is a 20-year-old, unemployed male from a high socioeconomic background living at home with his parents. The patient was brought to a community mental health clinic for psychological treatment by his parents, who were concerned about his mental health. He sought treatment to assist with distractibility, procrastination, and fidgeting behavior that he could not control. He described having angry outbursts and impulsive tendencies. He reported struggling with the above symptoms since 10 years of age. The patient endorsed significant social anxiety and social skill deficits that interfered with engagement in social interactions. He also reported low mood and frequent urination at night causing him discomfort and sleep disturbance.

Developmental history

According to a psychosocial history and collateral reports, he was born approximately three weeks late and his delivery was dif-

ficult. He exhibited hydrocephalus and did not cry or move around. Developmental milestones were achieved at expected age levels and language milestones were achieved earlier than expected. As objectively reported by his family, he had a remarkable memory and a “fast brain.” However, he experienced a number of sensory and motor coordination issues that started during early childhood. He was disturbed by loud noises, heard sounds unrecognized by others, was bothered by clothing tags and sometimes chewed on erasers. He also had difficulty with balance and problems with his ankles rolling. He did not have friendships or significant close relationships, other than family members.

Psychosocial history

The patient struggled academically and socially while away at college, which precipitated his leaving the university after two years. He had difficulty making friends and oftentimes worried that other individuals were ignoring him. Although he received adequate grades during his freshman year, he could not keep up with classes and described feeling as if his teachers were going too fast during lectures. He reported an inability to focus and complete schoolwork. The patient explained that nervousness about tests and grades tended to make his situation worse. After returning home, he had difficulty driving an automobile due to checking multiple directions before turning and fear of getting into an accident. He subsequently stopped driving altogether, leading to significant reliance on his parents.

Mental status exam and cognitive screening

The patient was dressed casually and had appropriate hygiene. His behavior was odd, including hopping while walking and repeating the clinician’s questions. He was fidgety and frequently stared at the floor. He had prolonged latency when speaking and thought processes were inhibited. It was unclear to the examiner if the patient was internally preoccupied or unable to focus due to attention deficits. Mood was apathetic, withdrawn, and slightly irritable. Affect was blunted and guarded. Judgment was fair, as evidenced by expressed interest in seeking help. Insight was fair, due to recognition of behaviors causing substantial impairment. His full scale IQ was in the average range, and he exhibited mild cognitive impairment on a cognitive screening measure.

Diagnostic assessment

After six sessions of psychological treatment at the community health clinic, the patient’s therapist diagnosed Social Anxiety Disorder, Attention-Deficit/Hyperactivity Disorder, Predominantly Inattentive Type, and Unspecified Neurodevelopmental Disorder. SAD was diagnosed based on the patient’s excessive fear of scrutiny in social situations, strong fears that he may become panicked in public and social avoidance. Comorbid ADHD was based on the patient’s inability to sustain attention, tendency to become easily distracted, procrastination, disorganization in his work habits, and difficulty paying attention to others; symptom onset occurred prior to age 11. Unspecified Neurodevelopmental Disorder was diagnosed based on the patient’s additional impairments in social functioning that were not better accounted for by ADHD, ASD, or SAD. It was noted that his symptoms might have organic etiology related to hydrocephalus at birth or other neurological conditions; thus further evaluation and medical work-up was recommended. In addition, the therapist indicated that Internet Gaming Disorder should be ruled out, based on the patient’s preoccupation with games on the internet, withdrawal symptoms when he did not play internet games, needing to spend more time playing Internet games in order to feel satisfied, and continuing to play games on

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