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Autism Spectrum Disorder as an Initial Diagnosis in Adults

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The two reports currently accepted as the original descriptions of autism spectrum are Leo Kanner's 1943 article, *Autistic Disturbances of Affective Contact* (1), and Hans Asperger's thesis, *Autistic Psyhopathy in Childhood* (2), presented in 1944 at the University of Vienna. The article by Kanner described 11 patients, all exhibiting the core features of what would later be called Autism. The children were characterized by "a profound aloneness", limited and unique interests, and a tendency to repetitive behaviors. They disliked change and insisted on sameness. Three children did not talk, while in others speech was either limited or had a monologue-like quality with little contribution to communication. The 4 children described by Asperger were verbally expressive, although their speech tended to be inappropriate, with little attention to social cues. Language development thus formed the initial basis for differentiating Kanner's Autism from Asperger's syndrome. Kanner was also Viennese, although he had seen his patients in the USA, where he had emigrated earlier. His article was in English, and it initially drew more attention compared to Asperger's work. Nevertheless, his emphasis on differentiating this syndrome from childhood-onset schizophrenia was overlooked, as the prevailing psychoanalysis-oriented practice focused on psychodynamic formulation rather than diagnosis. Autism as a separate disorder was therefore not officially recognized until the publication of the DSM-III in 1980. Michael Rutter is the child psychiatrist who primarily contributed to this modification. It was also in the eighties when Asperger's autistic psychopathy was introduced and named after him by Lorna Wing. This was followed by the publication of his work in English, translated by Uta Frith (3). Wing & Gould's Camberwell study on symptomatology (4) and Frith's studies on theory of mind are the other cornerstones in the autism literature (5).

Today's formal classifications, DSM-5 and ICD-11, comprise the single diagnostic category of Autism Spectrum Disorder (ASD). This diagnosis has replaced the subtypes Autistic Disorder (classical autism), Asperger's Disorder (Asperger's syndrome), and other Pervasive Developmental Disorders, stipulating a simpler differentiation based on language skills and intellectual functioning. However, the diagnostic constructs defined earlier include a much wider variety: Atypical Autism is among the most widely known subtypes, defining those cases who do not fulfill all diagnostic criteria, where a full assessment is precluded by mental retardation or other comorbid conditions. High Functioning Autism refers to cases without any intellectual impairment, i. e., those with an IQ above 70. Broader Autism Phenotype is a general term for the subclinical symptoms in patients' relatives.

Such wide variety in diagnostic descriptions is unusual, given that the first case descriptions were published only 80 years ago, a relatively short period for history of medicine. Reduction to a single category has therefore been a matter of debate. In fact, many clinicians and researchers remain skeptical, questioning the validity of a unitary disease: Is it really possible to account for the large clinical variation with a single diagnosis?

Validity will probably remain as a problematic issue in psychiatry, particularly for disorders with a fluctuating course and varied clinical manifestations that are readily influenced by cultural norms and practicing conditions. It must be noted, however, that higher validity is not the only objective in modifying diagnostic criteria in formal classification systems. In the case of ASD, adoption of a single category does not simply reflect authors' consensus on higher validity. Practical consequences were also taken into account, including reliability and ease of diagnosis, initial detection in adulthood, and maintaining availability of treatment options to individuals after they have reached adulthood.

During the last few decades a "brain disease" model was promoted and readily embraced for many psychiatric disorders. This creates a potential for error in research methodology and clinical practice, especially for markedly heterogeneous disorders like schizophrenia, where a holistic approach is indispensable. Where does ASD stand in this context? Could the unitary definition reinforce the brain disease model and pave the way for further reductionism?

My opinion is that ASD is relatively less problematic in this aspect. Despite the fact that cross-sectional symptomatology in adults is highly varied with many indirect manifestations and shaped to a great extent by the patient's individual experience, the internal consistency of symptom dimensions is higher than in many other disorders. This is a good indicator, if not proof, of validity. Furthermore, disorders in the autism spectrum have historically lent themselves more readily to dimensional, quantitative assessment. The very nature of the symptoms has necessitated in the diagnostic process understanding of personal experience rather than straightforward observation. This has probably protected the autism spectrum from a forced medical conceptualization as one single brain disease.

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Industry influence on research orientation and high hopes for a medical cure remained moderate and did not take precedence over psychosocial and cognitive interventions. The current name of the disorder reveals heterogeneity and is therefore less misleading. It is probably safe to hope that management will not be limited to pharmacological treatment protocols.

The new diagnostic criteria include another modification: Actual observation or caregiver reporting of onset before 3 years of age is not a prerequisite for diagnosis. It is stated that symptoms may not be detected until later because of minimal social demands and support from parents or caregivers in early years. This is an accurate observation with major implications for clinical practice. Symptoms that escaped detection earlier can now be diagnosed in adults, therefore the disorder must be included in the differential diagnosis in adult psychiatry.

Nevertheless, ASD is still rare as an initial diagnosis in adults (6). What could be the reason for that?

One explanation is that the established prototypes of ASD were shaped by the earliest standard descriptions and are limited to severe cases and dramatic presentations. Furthermore, severe ASD is commonly accompanied by additional symptom dimensions, some of them severe enough to be diagnosed as comorbid disorders. The widespread prototype in the lay as well as professional community is muddled by additional features like intellectual disability and abnormalities in posture and coordination, none of which is necessarily inherent in ASD *per se*. Likewise, patients with Asperger's syndrome are expected to display obvious weirdness. Those assumptions obscure high-functioning cases (those without intellectual disability), whose symptoms are attenuated with higher adaptive capacity and social support. In fact, all neurodevelopmental disorders are characteristically open to environmental influence, both positive and negative, and the clinical picture at any time is the product of a complex nature-nurture interaction.

Another reason could be the relative prominence of other diagnoses at cross-sectional assessment. Those could be indirect manifestations of ASD's core features or comorbid disorders. Crises, brief psychotic disorders, depression, and anxiety may mask ASD. While they may be severe enough to warrant an additional diagnosis, in some cases they lead to an overdiagnosis of chronic and severe mental disorders. It is not surprising therefore that many adults with an initial diagnosis of ASD have been in treatment with other, usually several diagnoses. Foremost among them is schizophrenia, a common and readily diagnosed disorder that has somehow remained immune to skepticism regarding reliability and validity. The age-old relationship between ASD and schizophrenia is beyond the scope of this article, however it deserves further exploration, given their history as well as the overlap between their manifestations and both genetic and environmental etiology. Other diagnoses that commonly accompany or mask ASD are mood disorders, anxiety disorders, obsessive-compulsive disorder spectrum, and personality disorders. Detection of ASD in those cases has important implications in both research and clinical practice.

The scientific relevance stems from the fact that accurate and precise phenotyping is crucial for complex disorders/traits, for which molecular genetics has recently gained momentum. Epidemiology may also be problematic, as indicated by the wide range of reported incidence and prevalence. The credibility of research in large samples might be misleading for us, authors and reviewers, as we tend to overlook the limitations brought about by reliance on the initial diagnoses in the case files, some of which are based on superficial assessments or simple screening.

Clinical implications of an ASD diagnosis are also relevant in that they include modifications in management, whether the previous diagnoses are viewed as comorbid disorders, complications, or indirect manifestations of a single diagnosis (7). Detection of previously undiagnosed ASD often reveals the need for prioritizing psychosocial interventions and family support. Syndromal (secondary) cases might receive upon reassessment previously missed diagnoses that caused the disorder. If the underlying syndrome is one with a known pattern of inheritance, families can be provided with genetic counselling and testing. Patients who have been in treatment for chronic schizophrenia might need a review of the dose and planned duration of antipsychotic treatment. Ongoing psychotherapy for anxiety or depression could be bolstered with added information.

Heritability of ASD is above 80%. Association with several types of chromosomal variation and findings from neuroimaging studies provide robust evidence to the role of biological factors in initiating this disorder. Genetic determinants include multiple single nucleotide variants, apparently associated with the disorder with small potential effects; rare but highly penetrant copy number variants, either *de novo* or inherited; and well-known syndromes that are known to be direct causes of ASD (5). Taken together, however, chromosomal variants fail to explain the high heritability. This supports the clinical observations regarding the role of individual experience and environmental factors in the etiology and course of ASD. The phenotype, which manifests mainly in social relationships is complex, prone to life-long alteration, and readily modified by gene-environment interactions.

Lorna Wing's aphorism emphasized the uniqueness of each case: "If you have seen one child with autism, you have seen *one* child with autism."

The neuropsychiatric nature of ASD is not necessarily an indication for limiting management to a purely medical model. On the contrary, it appears that highly individualized management is indispensable, tailored to the person's developmental history, current psychosocial conditions, subjective experience, and cognitive capacity.

Although it is appealing to target definite explanations of etiology or potentially curative medical treatments, currently unmet needs in ASD include psychosocial interventions and collaboration between professions. A holistic approach should include support for families, special education, general medical assessment, and genetic counselling. We need to work together with other medical branches, psychologists, education experts, and social workers. It is our duty to inform health authorities and funding agencies in their decisions about health policies and funding of research and clinical services.

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