Atypicality, intelligence, and age: a conceptual model of autistic spectrum disorder

Annotation

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Statement of problem

The conditions subsumed under the term 'autistic spectrum disorder' (ASD) vary in severity at the time of initial presentation; they also vary in expression over time. Unfortunately, the most widely adopted reference standard for the diagnosis of ASD, the *Diagnostic and Statistical Manual of Mental Disorders, 4th edition* (DSM-IV),[1] does not quantify severity of expression of ASD at the time of initial diagnosis, nor can it be used to document longitudinal changes in symptom expression, other than to state in a 'yes/no' fashion that an individual might no longer meet criteria for diagnosis. In clinical practice, measures intended to quantify the severity of ASD, such as the Childhood Autism Rating Scale,[2] the Gilliam Autism Rating Scale,[3] or the Autism Diagnostic Observation Schedule (ADOS),[4] are typically given once, rather than serially (unlike IQ or academic achievement testing, which are commonly given every 2 or 3 years, if not annually). Finally, the diagnosis of ASD is generally made without reference to the affected individual's level of general intelligence. Most scales for assessing ASD do not take the participant's chronological age or level of general development into account, although there are exceptions.[5,6] These clinical practices constrain our thinking about ASD. Here I present a schema for thinking about ASD along three dimensions: (1) severity of atypical symptoms, (2) level of general cognitive ability or comorbid mental retardation*, and (3) age. This schema is not meant as a diagnostic instrument in and of itself. Rather, it is intended as a frame of reference within which to locate the scores of existing instruments, and as a way of conceptualizing ASD. I suggest that this frame of reference will lead to improved clinical care for individuals of all ages with ASD, and might point the way toward further researchable questions in areas pertaining to the etiology, educational management, and epidemiology of autism and related disorders.

Severity of expression of ASD

Many scales have been devised for establishing the diagnosis of ASD and/or quantifying the severity of expression of atypical features.[2–13] The terms 'atypical' or 'atypicality' refer to developmental and behavioral features that would never be encountered in normally developing children, or in children with uncomplicated developmental delay. Virtually all of these rating scales enumerate or quantify atypical features in three or four major areas: social relatedness, use of language, repetitious behaviors, and (optionally) sensory issues (Table I). Children with severely atypical features are generally considered to have fully expressed autism of the 'classical' or 'Kanner' type. Children with moderately atypical features are generally considered to have pervasive developmental disorder (PDD). Children with mildly atypical features, plus hyperverbal but pragmatically impaired speech, obsession with a narrow range of arcane topics, and fine motor clumsiness, are commonly classified as having Asperger syndrome (AS).

It is generally accepted that Kanner-type autism, PDD, and AS represent, if not on points on a continuum, then at least overlapping clinical sets. There is a general consensus regarding the core features of ASD. Impaired eye contact, delayed echolalia, sniffing of non-food objects, and intense preoccupation with lining up objects, are encountered almost exclusively within the setting of ASD. The precise boundaries for ASD, however, and the optimal way to group symptoms for purposes of patient care or research remain subject to debate.[14] For example, fine and gross motor awkwardness, although commonly seen in ASD, are not unique to ASD; whether they should be regarded as core features is uncertain.

Association of ASD and IQ

ASD of any degree of severity can be seen in association with any degree of general intelligence. This has led to the evolution of overlapping and often imprecise terms, such as 'high-functioning autism' (HFA). We have depicted the relationship between IQ and severity of ASD graphically, as a way of clarifying the nomenclature and as a way of bringing to the fore the explicit or implicit assumptions built in to various diagnostic terms (Fig. 1). In this graph, all of the clinical features listed in Table I have been compressed into the *x*-axis, with decreasing atypicality running from left to right. The *y*-axis represents general intelligence, with an IQ of 70 (the cut-off for mental retardation) at the origin. Thus, within this two-dimensional model HFA falls into the upper left-hand quadrant (Fig 1. quadrant II): children with severely atypical features, but IQ within the normal range. AS

*UK usage: learning disability.
NB: List of abbreviations on last page of annotation.

maps to the upper right-hand quadrant (Fig 1. quadrant I), indicative of mildly atypical features and average or better general intelligence. In contrast to HFA and AS, the term PDD carries no associated implications regarding IQ, and therefore occupies a large area in the center of the graph, slightly to the right of the origin, extending above as well as below the *x*-axis. It should be apparent from Figure 1 that there is a potential overlap zone among HFA, 'high-functioning PDD' (i.e. PDD with normal general intelligence) and AS. Thus, the model constitutes a useful way of resolving disagreements as to whether a specific child has HFA 'or' PDD, 'or' AS, when in fact all three diagnoses lie in the same region of the plane.

Change in symptoms over time

The clinical features summarized in Table I can be thought of as indices of severity of expression of ASD at a fixed point in time. However, these same features can also be thought of as representing a fairly predictable sequence, or pathway, that children follow as their symptoms gradually evolve over time. This observation goes back to Kanner's original publication in 1943, which was itself a 5-year longitudinal report,[15] and has been replicated numerous times since.[16–27] Because these observations predate most of the presently available therapies for ASD, they strongly support the notion of a natural history for ASD. This is not to negate the benefits of therapy; rather, it underscores the importance of considering how much of a child's improvement over time might be due to the natural evolution of the condition, rather than to specific interventions.[28] The long-term prognosis for any given child with ASD is governed by the joint impact of the severity of expression of ASD and the level of general intelligence. To capture these observations, it is necessary to introduce a third dimension – time (or age) – to the conceptual model (Fig. 2). In Figure 2 the features of ASD from Table I remain embedded in the *x*-axis, and intelligence continues to be represented on the *y*-axis. Time (or age) is represented on the *z*-axis. As any given child progresses through time, his or her symptom expression will change. To some degree, the evolution of a child's clinical features over time is dictated by his or her starting location in the *x–y* plane; for example, children in quadrant I (average or better general intelligence, plus moderate to mild atypicality) will fare better, as a rule, than children in quadrant III (severe atypicality plus subnormal general intelligence).

Discussion

This three-dimensional model represents a useful way of approaching several sets of issues, includeing diagnosis and prognosis, intervention strategies, and etiology/ epidemiology.

Table I: Degree of expression of atypical features

Clinical domain	*Decreasing atypicality →*		
	Severe	*Moderate*	*Mild*
Impaired relating to others	No eye contact No physical affection Cannot be engaged in imitative tasks	Intermittent eye contact Seeks affection 'on own terms' May invade personal space of others (not true affection) Engageable in imitative tasks, although with difficulty	Good eye contact Shows interest in others, but often does not know how to join in Easily engaged in imitative activities Rigid: has difficulty if perceives that rules have been broken
Delayed and deviant language	Nonverbal No response to voice: may 'act deaf' No use of gestures as a means of compensating for absence of spoken language May use 'hand-over-hand' to guide caregiver to desired objects	Echolalia, delayed echolalia Odd inflection May use some stock phrases in an attempt to communicate Makes use of visual communication modalities (symbol cards; sign language)	Speaks fluently, but lacks understanding of verbal nuance, inference, or humor Difficulty with 'theory of mind' language tasks (fibbing; framing topic for partner; conversational repair)
Repetitious behaviors	Cognitive Extreme distress if routines are changed or when required to transition from one task to another Behavioral/motoric Frequent self-stimulatory, stereotypical movements (flapping, spinning, toe-walking, finger twiddling) Fascination with odd objects (tags, wheels, fans, etc.)	Cognitive Same, but with diminishing level of distress; may be able to accept verbal preparation for changes in routine Behavioral/motoric Motor stereotypies infrequent; may re-emerge when excited Complex repetitious play activities (lining up objects, memorizing numbers, letters, etc.)	Cognitive May demonstrate conscious awareness of preference for routines; easier to self-modulate Behavioral/motoric Motor stereotypies absent Play remains repetitious but repetitive quality is more subtle; preoccupation with arcane topics (e.g. bus schedules, solar system)
Sensory phenomena	Intense aversion or attraction to specific classes of stimuli Auditory: covers ears Visual: visual self-stimulation (lights/ patterns); looks at objects from odd angles Tactile: rubbing, licking, mouthing, deep pressure Olfactory: sniffing Extreme food selectivity 'Increased pain threshold' Fears: heightened/blunted	Same, but diminishing intensity	Same, but diminishing intensity

DIAGNOSIS AND PROGNOSIS

The DSM-IV bases the diagnosis of ASD on symptoms that are typically most evident in moderately to severely atypical preschool children. In contrast, our model enables the clinician to locate the patient within a three-dimensional diagnostic space, taking into account the effects of age, rate of cognitive development, and severity of expression of atypical features. Thus, the model goes several steps beyond the narrow question 'Does this child have ASD?', placing the child's clinical features into a broader and more developmentally detailed context. This will be particularly helpful when considering infants and toddlers, or adults with ASD, who might not manifest symptoms consistent with DSM-IV. (For example, in very young children deficits in social interaction and play might be more prominent than insistence upon maintenance of routines.[29]) Given two children with similar levels of atypicality, the functional outcome is better for the child with the higher IQ.[16,20,27,30–36] What is not clear is whether the more favorable outcome for this group is simply due to higher IQ in itself (as would be true for children without ASD) or whether it is also tied to a greater decrease in expression of autistic symptoms over time among children without mental retardation. As additional data are collected with more refined tools such as the ADOS[5] or the Diagnostic Interview for Social and Communication Disorders,[6] it will be useful to map these data onto this three-dimensional space, to gain a fuller understanding of the interactions among these three variables: atypicality, intelligence, and age.

INTERVENTION STRATEGIES

Our model provides a framework for thinking about the optimal intervention strategies for any given child, and for charting the child's progress over time. Children who are younger, more severely atypical, and/or suffering from mental retardation might be more appropriate candidates for intensive, one-on-one behavioral interventions, whereas children who are older, less atypical, and/or have normal intelligence might be more appropriately served through more socially based-group interventions. One of the biggest hurdles in the field of developmental pediatrics is the dearth of randomized, blinded, prospective studies of different interventions. This lack of controlled data arises in part because of ethical concerns and in part because the educational system does not proceed from the scientific model. Many educational practices are unsupported by controlled studies. For example, there are no controlled data to show how many hours per week a typically developing youngster needs in order to learn to read, much less any data bearing on questions such as 'How much special education, and what kind is "enough" for a child with ASD?' Given the difficulty of conducting randomized trials of specific educational interventions, the best substitute for evaluating the efficacy of different interventions would be a model that predicts the degree of improvement that would be expected regardless of therapy. Given a child's age and starting point in the two-dimensional plane represented by Figure 1, it would be useful to have the ability to predict the likely range of outcomes over time, irrespective of treatment modality. We are currently engaged in data collection to determine whether the two-way and three-way relationships among age, IQ, and severity of atypicality can be modeled statistically

ETIOLOGY/EPIDEMIOLOGY

ASD is etiologically heterogeneous.[37] Certain regions of the clinical space represented by Figure 2 are more likely to be occupied by patients with specific etiologies, or classes of etiologies. For example, boys with fragile X syndrome and autism are more likely to be found in quadrants III and IV, because the fragile X mutation usually results in mental retardation in affected males. It remains to be seen whether other genetic or environmental factors cluster in another regions of the diagnostic space represented in Figure 2. Testing patients who cluster in different portions of this three-dimensional space might shed light on specific causes of ASD. Our model also lends itself to thinking critically about secular trends in the prevalence of ASD, e.g. is the apparent increase in incidence of ASD due to

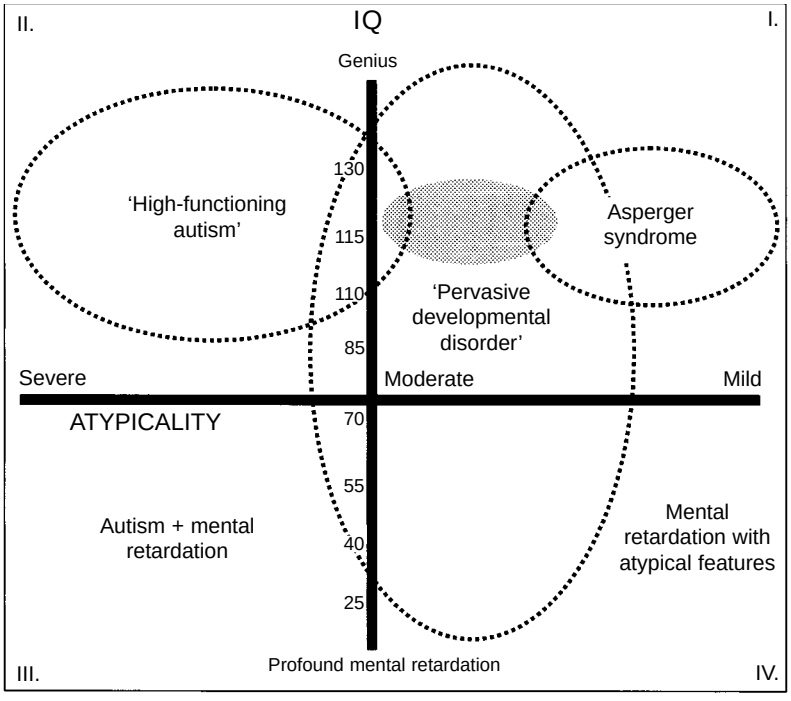

Figure 1: *Relationship between degree of atypicality and intelligence. Origin corresponds to moderate atypicality (x-axis) and an IQ of 70 (y-axis). Children with severely atypical features and an IQ in normal range (quadrant II) are sometimes referred to by the term 'high-functioning autism' (HFA). Children with moderately atypical features have pervasive developmental disorder (PDD). PDD can occur in presence of normal IQ (quadrant I) or in the presence of mental retardation (MR; quadrant IV). Children with mildly atypical features, normal general intelligence, hyperverbal behavior, narrow interests, and physical clumsiness have Asperger syndrome (AS) (quadrant I). There is a zone of potential overlap among children with HFA, PDD, and AS (stippled region); treatment is similar regardless of the diagnostic 'label'. Children with mental retardation might have fully expressed autism ('low functioning autism', quadrant III), or their primary developmental disability might be mental retardation, with a smattering of atypical features (quadrant IV).*

increased recognition of individuals in quadrant I or to an actual increase in incidence of new cases? Our model by itself cannot answer this question, but it invites meta-analysis of existing data as well as critical analysis of prospectively collected data, to determine whether there have been secular trends in the distribution of recognized cases of ASD by degree of atypicality, IQ, or age.

LIMITATIONS OF THIS MODEL

Model-building is a two-edged sword. The utility of any model lies partly in its ability to account for observed phenomena within a readily manipulable conceptual framework. All models therefore involve some degree of simplification. Condensing all the atypical features listed in Table I into one continuous axis is necessarily an oversimplification, in the same way that representing an individual's intelligence by one number ('full scale IQ') is an oversimplification. For example, our model does not explicitly address Wing's subtyping of children with autism as being 'aloof', 'passive', or 'active but odd',[38,39] although in some measure these subtypes correspond to our categories of 'severe', 'moderate', and 'mild' impairment within the social domain. We have intentionally omitted any mention of other developmental/ neuropsychiatric comorbidities, such as depression, Tourette syndrome, or attention deficit hyperactivity disorder. This is not to say that such comorbidities are uncommon or inconsequential. Rather, we have omitted these comorbidities from our model primarily to enable us to present parents with a readily comprehensible 'big picture' of the clinical manifestations of ASD. Our model is also silent on the question of etiology, because it is intended primarily to serve as a frame of reference for capturing the clinical expression of ASD (although we expect that certain etiologies will indeed map to certain portions of the three-dimensional clinical space depicted by our model). We accept these simplifications only to the extent that doing so affords parents, caregivers and researchers insights that would otherwise have been obscured by the details; however, we must not lose sight of the fact that any model represents a simplified view of reality. A second limitation in our model is our implicit assumption that 'general intelligence' and 'atypicality' vary independently of one another. We have depicted the *x*- and *y*-axes as orthogonal to one another, when this might not actually be so in nature. Third, at a practical level, it can be difficult to assess 'general intelligence' (our *y*-axis) independently of 'atypicality' (our *x*-axis). For example, adaptive skills are generally well correlated with measured IQ in non-autistic individuals. However, this relationship does not always hold in children with ASD, in whom adaptive skills might be substantially lower than would be expected on the basis of their level of intelligence.[40] Conversely, some skills seem to be highly modular and might stand well above the individual's day-to-day level of abilities. These are generally regarded as 'splinter skills', divorced from 'general intelligence' (hyperlexia, for example). Arriving at a concrete estimate for a given child's location on the *y*-axis at any given point in time might therefore be quite problematic, especially in the child who is 'untestable' by conventional means. Likewise, differentiating how much of a child's stereotypical behavior might be due to severe global cognitive delay, rather than atypicality, can be equally challenging. Thus, arriving at a precise location on the *x*-axis (degree of atypicality) in the presence of severe mental retardation can be equally challenging. In such circumstances the benefit to the parents of our model lies not so much in locating their child precisely within the model but simply in conveying to parents that there *is* such a model, so that they will be better able to compare their child with other children and with materials they will read about ASD. Over time, it is almost always possible to chart a child's course, even though at any given instant it might not be possible to locate the child's position precisely within our frame of reference. Furthermore, from a research standpoint, one is generally dealing with aggregate data on large numbers of individuals. Even though there will be an irreducible degree of ambiguity about the *x* and *y* coordinates for any given individual at a specific point in time, useful information can be gleaned from looking at group trends over time.

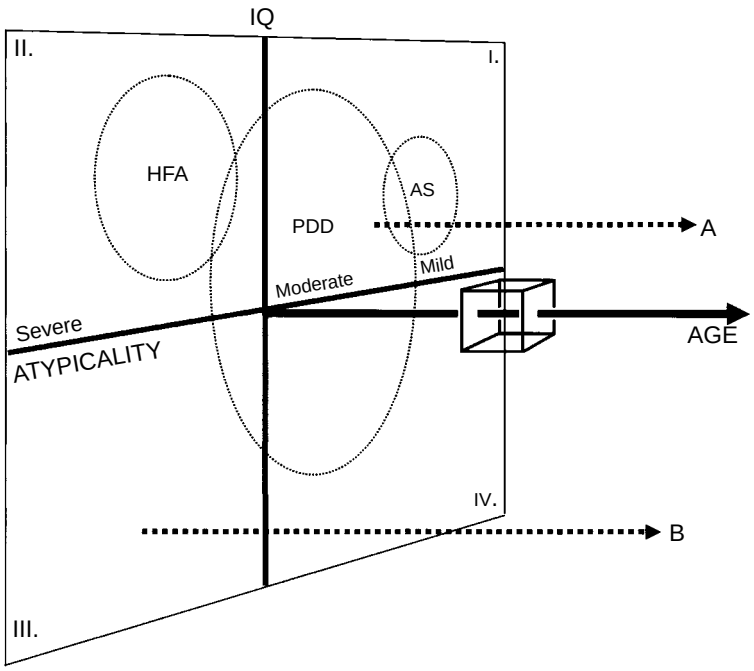

Figure 2: *Degree of atypicality, intelligence, and age. Same information is shown as in Table I and Figure 1, with the addition of time (age) as the third dimension (z-axis). Origin on z-axis represents birth. Point in starting plane where child is initially located is a major determinant of long-term outcome. Here, course for a child with pervasive developmental disorder (PDD) and normal IQ ('A') and a child with severe mental retardation plus severe autistic spectrum disorder (ASD) ('B') are represented schematically. Cube indicates diagnostic space schematically that forms the basis for symptoms of ASD listed in DSM-IV: preschool children with moderate to severely atypical development. This is only a small subset of the entire universe of individuals with ASD. AS, Asperger syndrome; HFA, 'high-functioning autism'.*

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List of abbreviations

AS	Asperger syndrome
ASD	Autistic spectrum disorder
DSM	Diagnostic and Statistical Manual of Mental Disorders, 4th edition
HFA	High-functioning autism
PDD	Pervasive developmental disorder