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Special Article

Present Conceptualization of Early Childhood Neurodevelopmental Disabilities

Michael I. Shevell, MD, CM, FRCP

Neurodevelopmental disabilities are collectively a common problem in child health that frequently prompts neurologic assessment and intervention. They are a group of heterogeneous conditions that share a disturbance in the acquisition of basic developmental skills in a chronologically appropriate manner. Lacking uniform diagnostic means of ascertainment, their recognition depends on fulfilling present consensus opinion regarding the various subtypes now recognized. Distinctive subtypes of neurodevelopmental disabilities can be accurately diagnosed according to present consensus conceptualization. Dual diagnosis of neurodevelopmental disabilities in the same child is possible, given present opinion. It can be expected that these conceptualizations will be dynamic and guide ongoing research efforts that will elucidate basic mechanisms, effective therapeutic interventions, and evaluate outcomes.

Keywords: neurodevelopment disabilities; development; cerebral palsy; mental retardation; autism

The neurodevelopmental disabilities of early childhood are a group of chronic heterogeneous disorders that share as an essential core feature a recognized disturbance, either qualitative or quantitative in nature (or both), in the acquisition and expression of early developmental skills.1 This disturbance may be defined as being quantitatively (ie, greater than 2 standard deviations below the mean) significant or qualitatively apparent (ie, lack of milestone acquisition in comparison to chronologic peers).2 Traditionally, these developmental skills have been clustered into various domains: motor (fine and gross), speech and language, personal-social, cognition, and activities of daily living (eg, dressing, self-hygiene, toileting).3 These domains are not completely distinctive nor mutually exclusive; oral motor skills are a precondition for speech expression, fine motor and language skills are often a precondition for an accurate assessment of intrinsic cognitive ability.

Collectively, neurodevelopmental disabilities are a common problem in child health and thus a frequent reason for specialty medical (ie, pediatrics, developmental pediatrics, pediatric neurology, child psychiatry) and allied health (ie, occupational therapy, physical therapy, speech-language pathology, audiology, psychology, nursing, social work) assessment and intervention.4 Lifelong morbidity is often attached to the diagnosis of a neurodevelopmental disability with attendant additional medical, rehabilitation, educational, vocational, and supportive needs that may challenge the individual, family, and society. This challenge is especially evident currently given moral, legal, and service trends as framed by the current World Health Organization model of health and functioning that promotes integration and the maximizing of individual participation despite impairments that impart activity limitations.5

Neurodevelopmental disabilities have traditionally been divided into subtypes. These subtypes function as “terms of convenience” or “symptom complexes” that through consensus conceptualization and usage convey in a short-hand way recognizable, distinctive, and distinguishable entities.6 Accurate diagnosis of these entities provides a template for (a) medical and rehabilitation evaluation, (b) comprehending pathogenesis, (c) determining service needs requirements, (d) informing family counseling, and (e) shaping future expectations and realities. Early childhood neurodevelopmental disabilities in common medical use currently include cerebral palsy, global

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developmental delay, mental retardation/intellectual disability, developmental language impairment, and autistic spectrum disorder.

Early childhood neurodevelopmental disabilities subtypes share marked within class heterogeneity. This heterogeneity applies to individual presentation, evolution, causation (ie, etiology), interventions required, challenges faced, and outcomes achieved (however measured or assessed). This heterogeneity, together with a lack of objective uniform diagnostic tests and apparent degrees of overlapping co-occurrence between the neurodevelopmental disabilities, challenges our consistent conceptualization and accurate recognition of these subtypes.

This review will attempt to provide a summary of what is presently consensus opinion regarding the conceptualization of early childhood neurodevelopmental disabilities subtypes, so as to facilitate the early recognition of these disorders. Early consistent recognition will facilitate the timely and appropriate allocation of relevant resources that seeks to optimize eventual outcome for these children and their families.

Cerebral Palsy

At a frequency of 1.5 to 2.5/1000 live births based on established population registries, cerebral palsy represents the most common cause of physical impairment in children. It is a relatively “ancient” concept in child neurology, having been described in the 19th century by various German (Hirsch [1842], Heine [1860]), English (Little [1843]), Canadian (Osler [1886-1889]), American (Sachs and Peterson [1890]), and Austro-Hungarian (Freud [1893]) physicians. However, an attempt to formally define and classify cerebral palsy lay dormant until the 1950s.

Collective attempts yielded an initial, and ultimately lasting, conceptualization of cerebral palsy as “a permanent but not unchanging disorder of movement and posture, appearing in the early years of life and due to a non-progressive disorder of the brain, the results of interference during its development.” Thus, cerebral palsy was envisioned at its essential invariable core as a motor disorder of early onset arising in the brain as a result of a fixed unchanging lesion, which has a lifelong impact, the clinically evident nature of which may change over time. Subsequently, for the purposes of clarity, transient motor disorders and those motor disorders that were the product of a progressive neurodegenerative process were excluded from a diagnosis of cerebral palsy. As a corollary, a classification scheme based on the qualitative (ie, spastic, dyskinetic, ataxic-hypotonic, mixed) nature and topographic (ie, quadriplegic, diplegic, hemiplegic) distribution of the motor abnormalities observed on formal neurologic examination was put forward by Minear. This classification scheme, without formal validation regarding reliability or consistency, was widely uptaken and remains the standard clinical mechanism for the description of cerebral palsy that provides insight into the localization of brain injury, its etiology (and presumed pathogenesis), and expected neuroimaging findings.

A second collective attempt in the early 1990s refined the original definition by explicitly emphasizing the heterogeneity of cerebral palsy and its potential causation as either being acquired or congenital, the latter the by-product of a defect in brain formation. This attempt yielded the following consensus of cerebral palsy as “an umbrella term covering a group of non-progressive, but often changing, motor impairment syndromes secondary to lesions or anomalies of the brain arising in the early stage of development.” A later effort by Badawi and colleagues explicitly listed a long list of chromosomal, genetic, and syndromal diagnoses that have been traditionally either included or excluded from consideration as “cerebral palsy.” Reflecting an emerging interest in function, gross motor functional capabilities, especially pertaining to the key function of ambulation, were used to develop an alternative approach to cerebral palsy classification. The Gross Motor Function Classification System effectively captures meaningful differences in gross motor skills among children with cerebral palsy. It provides considerable insight into severity, service needs, and motor prognosis and has been rigorously tested for reliability and validity and is psychometrically robust. Not surprisingly, substantial correlation between the complementary classification schemes (ie, neurologic subtype and gross motor skills) has been demonstrated.

That cerebral palsy is often more than simply a motor impairment syndrome has long been recognized. Indeed, for many children and their families, the major burden of disability and care that limits participation and resulting quality of life lies outside the motor domain. Comorbidity may be a secondary consequence of the unmitting motor impairment (ie, scoliosis, musculoskeletal deformities) or the result of additional manifestations of the underlying brain anomaly or lesion (ie, epilepsy, cognitive impairment, speech-language difficulty, dysphagia that limits oral feeding, behavioral issues, sensory limitations). Thus, the most recent consensus definition for cerebral palsy expands on the essential core of motor impairment to include explicit recognition of both functional limitations and comorbidities: “cerebral palsy describes a group of permanent disorders of movement and posture, causing activity limitations, that are attributed to non-progressive disturbances that occurred in the developing fetal or infant brain. The motor disorders of cerebral palsy are often accompanied by disturbances of sensation, perception, cognition, communication and behaviour, by epilepsy, and by musculo-skeletal problems.”
Global Developmental Delay and Mental Retardation/Intellectual Disability

The conceptualization of global developmental delay as a distinct diagnostic entity is a relatively recent phenomenon. It reflects both our emerging emphasis on development and function across a broad spectrum of early childhood skills and the actual framework within which families seek assessment, evaluation, and intervention for their children (ie, Why is my child not yet walking? Why is my child not yet talking?21). It also is a by-product of inherent conceptual and methodological difficulties in defining and reliably measuring purely cognitive skills in the young child (ie, a child under 5 years of age).23 Recognizing global developmental delay requires, as an essential precondition, an awareness that developmental trajectories are invariably individualized possessing wide variation within what can be accepted as “normal” without a clear dividing boundary line between “normal” and “abnormal.”23 Also at times, development is not smooth nor consistent but rather is “saltatory” in character featuring periods of rapid skill acquisition followed by periods of skill practice and consolidation.24 Global developmental delay refers to the symptom complex in the young child, where there is an evidence of a delay in skill acquisition in 2 or more developmental domains.25 Operationally, it was defined in the American Academy of Neurology/Child Neurology Society Practice Parameter evaluation guideline as a significant delay in 2 or more developmental domains, with “significant” meaning performance 2 or more standard deviations below the mean on objective norm-reference chronologically appropriate standardized evaluation measures relevant to the domain tested (eg, motor-Alberta Infant Motor Scale, Peabody Developmental Motor Scale, Bruininks-Oseretsky Test of Motor Proficiency, Language-Peabody Picture Vocabulary Test, Expressive One Word Vocabulary Test, Clinical Linguistic Auditory Milestone Scale, Clinical Evaluation of Language Fundamentals; behavior/adaptation-Vineland Adaptive Behavior Scales, Pediatric Evaluation of Disability Inventory, Wee Functional Independence Measure, multiple domains-Batelle Developmental Inventory).22 Although delay in 2 domains usually implies delay across all domains, apparent subtypes of global developmental delay related to cognitive skills have been demonstrated.26

In contrast to global developmental delay, mental retardation (originally known unfortunately as “feeble mindedness”) has long-standing as a medical diagnosis. It was originally conceptualized within the framework of a single measurable objective trait (ie, the intelligent quotient or IQ) that provided a mechanism for stratifying severity.27 The last half century has witnessed 6 collective efforts at arriving at a consensus definition that ultimately has reflected a recognition that mental retardation goes beyond a static measure of intelligence to reflect the dynamic holistic interactions between an individual with limited intellectual and adaptive skills and a personal-specific contextual environment.28,29 The most recent consensus definition for mental retardation was put forward in 2002 as “a disability characterized by significant limitations both in intellectual functioning and in adaptive behavior as expressed in conceptual, social and practical adaptive skills.”30 This definition envisioned adaptive behavior as including those skills that an individual needs to acquire to adequately function within his or her environment. The deficits of mental retardation within the World Health Organization model lead to impairment (ie, performance incapacity), activity limitations, and participation restrictions (ie, the opportunity to function).5 As a neurodevelopmental disability, mental retardation is evident at an early age and lifelong in its time course requiring systems of support throughout the lifespan (eg, familial, educational, vocational).30

Recent developments have witnessed the emergence of the term “intellectual disability” to replace that of mental retardation.31 These terms are synonymous in their meaning. Indeed, the American Association of Mental Retardation has changed its name to the American Association of Intellectual and Developmental Disability to underline a preference for the term “intellectual disability” as it better captures our emphasis on behavior and contextual factors reflecting more accurately the changes in our construct of disability.

As presently construed, the diagnosis of both global developmental delay and mental retardation/intellectual disability requires an awareness of situational contextual factors and a sensitivity to social-cultural and linguistic modifiers.23 Accurate and reliable diagnosis requires observation over time, rather than a single “one-shot” assessment.32 While most times an initial diagnosis is based on clinical judgment, experience interdisciplinary assessment using multiple objective measures is preferable for accuracy.

Clearly, there is a complementarity between the diagnosis of global developmental delay and mental retardation/intellectual disability.23 Although nonsynonymous, they are chronologically framed by what can be assessed and measured in children below and above the age of 5 years. Although the term “delay” in global developmental delay suggests potential catch-up with maturation, longitudinal studies have not shown this to be true.33 Indeed, many older children and adults with mental retardation/intellectual disability were originally labeled as having global developmental delay as young children and many children first labeled with global developmental delay will go on with time to manifest mental retardation/intellectual disability later. Thus, global developmental delay may be considered a chronologically limited placeholder and its diagnosis provides medical and rehabilitation guidance.
with respect to etiologic evaluation, rehabilitation service needs, family counseling, and prognostication.23

Developmental Language Impairment

Arriving at a definition for developmental language impairment, one of the more common subtypes of early childhood neurodevelopmental delay34 has been hampered by a lack of consensus, a multiplicity of variously used nomenclature, and a tendency to extend the terminology of acquired adult aphasias to the pediatric population.35 Analogous, indeed synonymous, terms with developmental language impairment used over the years have included developmental dysphasia, developmental aphasia, developmental language disorders, and specific language impairment.34

The core essential feature of developmental language impairment is a recognizable impairment in language skills that is disproportionate when compared to achievement in the other nonlanguage developmental domains.36 This impairment has been variably defined either by that which is clinically recognizable with respect to either expressive or receptive (or commonly both) language skills or by a predetermined statistical cut-off on accepted standardized measures of preschool language (ie, Peabody Picture Vocabulary Test, Expressive One Word Picture Vocabulary Test, Clinical Linguistic Auditory Milestone Scale, Clinical Evaluation of Language Fundamentals).37 Although there may be deficits in nonlanguage skills, this should not cross a threshold of severity that when combined with the language deficits then merits a diagnosis of either a global developmental delay or mental retardation/intellectual disability.35 In addition, the presence of autistic features or significant bilateral hearing loss precludes the diagnosis of a developmental language impairment.35

Thus, the definition of developmental language impairment requires documentation of a discrepancy between language and nonverbal skills, especially that which has been deemed cognitive.38 Controversy exists regarding the actual measurement of these skills and the size of the discrepancy needed, both between language impairment and normal language proficiency and between language and nonlanguage developmental domains.39

The most widely used definition for developmental language impairment mandates nonverbal performance IQ within the normal range (ie, above 85 or no more than 1 standard deviation below the normal mean of 100) and a score on an accepted language measure that falls below 1.25 standard deviations below the mean.38,39 This corresponds to the 10th percentile of a normally distributed measure and coincides with the level at which a speech-language pathologist will reliably clinically identify a child as having a meaningful language impairment.40 Depending on the rigor of the definition used for diagnosis, clinical service provision, or research, some authorities will insist on a 1 standard deviation or more discrepancy between verbal and nonverbal performance.41

Recent studies have highlighted that children with developmental language impairment frequently have subtle impairments in motor domains.42 These impairments are frequently overlooked initially due to the magnitude of the child’s language deficits. This nonlanguage impairment may preclude proper language acquisition and thus their recognition and remediation provides a distinct therapeutic benefit to children affected. Longitudinal studies have also shown that over time, deficits in other domains, especially cognitive, become apparent in children with developmental language impairment.33,44 These deficits may not have been apparent earlier due to the extensive use of language to infer cognition or alternatively proper language skills are a precondition for later successful cognitive development. Ultimately, the cognitive limitations may render themselves apparent in the educational environment as a verbal-based learning disorder.45

Autistic Spectrum Disorders

The term Autistic Spectrum Disorder is frequently used “broadly and synonymously” with both “autism” and “pervasive developmental disorder.” The term “autism” was initially used by Bleuler early in the 20th century in the context of the mental illness schizophrenia to describe the “escape from reality” that was commonly observed in the psychotic behavior of individuals with this condition.46 In the initial clinical descriptions of children with an autistic spectrum disorder reported independently, and nearly coincidently, by Leo Kanner47 and Hans Asperger,48 the term “autism” or “autistic psychopathy” was used in an attempt to capture the clinically evident profound social disconnection of these children.

The first attempt to establish formal criteria for diagnosing autism was that of Kanner and Eisenberg in the 1950s.49 Omitted from their criteria was any mention of profound disturbances in language. Deficits in behavior (“repetitive, ritualistic behavior, which must be of an elaborate kind”) and social skills (“a profound lack of affective contact”) were highlighted. This conceptualization was modernized in an influential way by Rutter in 1977 when he attached “impaired language and communication skills” to being a core invariant feature of autism, which he also labeled interchangeably as either “infantile autism” or “childhood autism,” in addition to the previously highlighted impaired social relationships and an insistence on sameness.50

These core features were incorporated into the initial Diagnostic and Statistical Manual attempt to provide a definition in its third edition (1980) and have remained in
Table 1. DSM-IV Criteria for An Autistic Spectrum Disorder

A. A total of 6 (or more) items from 1, 2, and 3, with two from 1 and at least one each from 2 and 3:
1. Qualitative impairment in social interaction, manifested by at least 2 of the following:
   a. Marked impairment in the use of multiple nonverbal behaviors, such as eye-to-eye gaze, facial expression, body postures, and gestures, to regulate interaction
   b. Failure to develop peer relationships appropriate to developmental level
   c. Lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (eg, by lack of showing, bringing, or pointing out objects of interest)
   d. Lack of social or emotional reciprocity
2. Qualitative impairment in communication, as manifested by at least one of the following:
   a. Delay in or total lack of the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication, such as gesture or mime)
   b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
   c. Stereotyped and repetitive use of language or idiosyncratic language
   d. Lack of varied, spontaneous make-believe or social imitative play appropriate to developmental level
3. Restrictive repetitive and stereotypic patterns of behavior, interests, and activities, as manifested by at least one of the following:
   a. Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal in intensity or focus
   b. Apparently inflexible adherence to specific nonfunctional routines or rituals
   c. Stereotyped and repetitive motor mannerisms (eg, hand or finger flapping, twisting, or complex entire-body movements)
   d. Persistent preoccupation with parts of objects
B. Delays or abnormal functioning occurs in at least one of the following areas, with onset before age 3 years:
   (1) social interaction
   (2) language as used in social communication
   (3) symbolic or imaginative play

place since. Essentially, autistic spectrum disorders are defined by behavioral characteristics that attempt to capture a spectrum of both distribution-specific behaviors demonstrated and their severity that is independent of underlying biology or cognitive skills. Rutter’s core-affected domains remain and all must be affected to some degree: (a) social interaction, (b) language, communication and imagination, and (c) behavioral flexibility. Operational features of behavior consistent with specific abnormalities in each of these domains are provided (see Table 1). For proper diagnosis, an individual must possess at least 6 of the behavioral descriptors in total, with a minimum of 2 from the social interaction domain, 1 from the language/communication/imagination domain and 1 from the behavioral flexibility domain. In addition, the onset of the disorder must be evident before the age of 3 years. Given the multiplicity of behavioral descriptors and the parameters of diagnosis, considerable variability in phenotypes exists subsumed under the autistic spectrum disorder label. Some subtypes have been explicitly recognized: (a) one defined by biology (Rett syndrome), (b) another by clinical presentation (childhood disintegrative disorder), (c) another by the preservation of basic language skills (Asperger syndrome), and (d) by a threshold of severity in terms of the number of autistic behavioral descriptors observed (pervasive developmental disorder—not otherwise specified).

The diagnosis of an autistic spectrum disorder is currently facilitated in most diagnostic centers by the application of standardized questionnaires (Autism Diagnosis Interview–Revised) or observation schedules, such as the (a) Autism Diagnosis Observation Schedule, and (b) Childhood Autism Rating Scale, that provide a mechanism of objectivity to assist both in the categorical ascertainment of a diagnosis (ie, autistic spectrum disorder vs no autistic spectrum disorder) and a grading of severity (pervasive developmental disorder vs pervasive developmental disorder—not otherwise specified). Not unexpectedly, given considerable heterogeneity, diagnostic agreement between these measures has been less than perfect and parcellation of the autistic spectrum disorder entity into distinct behaviorally homogeneous subtypes remains as yet consistently attained.

Overlap and Dual Diagnoses

The consensus conceptualization of the neurodevelopmental disabilities has yielded entities that may coexist in the same individual. This observation is readily apparent in the clinical situation. The child with a cerebral palsy may also have a global developmental delay, mental retardation/intellectual disability, or an autistic spectrum disorder. Similarly, the globally developmentally delayed child may have autistic features insufficient for the certain diagnosis of an autistic spectrum disorder. The line between global developmental delay with autistic features and pervasive developmental disorder—not otherwise specified has not yet been distinguished with certainty. Finally, the child with an autistic spectrum disorder has variable cognitive skills, thus in many, mental retardation/intellectual disability can be formally diagnosed.
The coexisting of features characterized in the different neurodevelopmental disabilities should not preclude their independent diagnoses as explicit recognition provides the mechanism for systematic adequate appropriate standardized evaluation, service needs provision, programming access, family counseling, and prognostication.

Conclusion

Accurate recognition of a child’s neurodevelopmental disability is an essential precondition for providing appropriate care. The absence of objective laboratory-based markers together with the considerable implicit heterogeneity of these conditions challenges the clinician. Consensus definitions have emerged that reflect collective experience and wisdom that provides guidance in informing and shaping clinical decisions at various levels. These levels include etiologic evaluation, rehabilitation referrals, service needs provision, programming access, counseling, and prognostication. The absence of objective means of diagnosis implies that for the foreseeable future our conceptualizations will remain in a necessary and unavoidable state of flux. Periodic reassessments of our conceptualizations will be inevitable to incorporate new knowledge and insights that will hopefully advance our understanding and the provision of care for these children and their families.

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