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BRIEF REPORT

Brief Report: Intersection of Sets of Symptoms Between Congenital Blindness and ASD: Proposing of Differential Criteria

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Abstract
To propose novel differential criteria of the DSM-5 for diagnosing transient Autistic-like behaviors in children with congenital blindness as a secondary condition. Most references indicate a significantly higher prevalence of autism in children with congenital blindness compared to sighted children. These behavioral symptoms may be transient Autism-like behaviors that should be diagnosed as a secondary condition. Differential criteria are proposed: gaining more adaptive responses to effective interventions; presenting more efficient adaptation to environmental changes; gaining improved use of language in a more typical manner; acquiring more mature interactions with family as well as with others and, proving more positive prognosis due to spontaneous maturity and life experiences. Decreasing false-positives and true-negatives in the assessment process and diagnosis of primary vs. secondary ASD and comorbid conditions. Developing novel assessment tools to distinguish between ASD and autism-like behaviors in the intersection area. Future revision of DSM publication may reconsider these proposed changes in diagnostic criteria.

Keywords DSM-5 · ASD criteria · Congenital blindness

Introduction

In DSM-5® (American Psychiatric Association, 2000, 2013), there are four main categories of behavioral symptoms for the diagnosis of ASD (Autism Spectrum Disorder), while each category includes several subcategories of behavioral symptoms: (A) Persistent deficits in social communication and social interactions across contexts, not accounted for by general developmental delays, and manifest by 3 of 3 symptoms; (B) Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least 2 of 4 symptoms; (C) Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities), and (D) Symptoms together limit and impair everyday functioning. In addition, the age criterion in DSM-5 stated: "symptoms must be present in the early developmental period but may not become fully manifest until social demands exceed limited capacities or may be masked by learned strategies in later life". The DSM-5 has rule-outs to distinguish between ASD and other developmental conditions such as intellectual developmental disorder or global developmental delay, but it does not delineate clear rule-outs to distinguish between ASD and congenital blindness. Thus, the primary aim of this manuscript is to propose a novel diagnosis: Transient Autism-like behaviors in children with congenital blindness.

The presence of Autism in children with blindness was systematically investigated throughout the last decades worldwide (Perez-Pereira, 2005; Molinaro et al., 2020). Previous quantitative studies (Absoud et al., 2011; Ek et al., 1998, 2005; Fraiberg, 1977; Hobson & Lee, 2010; Mukaddes et al., 2007; Parr et al., 2010; Rogers & Newhart-Larson, 1989) have investigated the prevalence of Autism in children with blindness. These studies all together found that 859 early blinded children yield a 48% prevalence of ASD. This prevalence is even higher if the blindness was congenital and complete, irrespective of its etiology. This significantly high prevalence is 31 times higher than the 1.5% prevalence among sighted children (Baio et al., 2014). This staggeringly wide gap in prevalence should be reevaluated in terms of diagnostic criteria. In other words, this gap indicates a separate diagnosis other than Autism, presumably "autism-like behaviors in congenitally blind children" and presents an

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intersection between ASD symptoms (as a primary diagnosis) and transient symptoms seen in children with congenital blindness. A literature review by Jure et al. (2015) revealed that autism, as a primary condition, was diagnosed in children with Retinopathy due to Prematurity (Ek et al., 1998), in children with bilateral optic nerve hypoplasia (Ek et al., 2005) and in children with Leber's Congenital Amaurosis (Hobson & Lee, 2010; Rogers & Newhart-Larson, 1989). A follow up reassessment by Hobson and Lee (2010) of nine children in their adolescence resulted in formal cancelation of the previously diagnosed ASD due to maturity and gradual recovery of symptoms in 8 out of the 9 adolescents. These findings present some evidence that strict symptoms and criteria of the DSM-5 for ASD can lead to incorrect diagnosis in some complex, under-diagnosed cases such as congenitally blind children. However, these finding are based on few studies with small number of participants.

Occupational therapists, and other practitioners and educators, working with children with congenital and acquired visual impairments notice that children with congenital blindness or severe early visual loss may present behaviors and psycho-motor responses that evoke an immediate declaration of "autistic behaviors" (Jure et al., 2015). Moreover, these children show basic impairments in interpersonal interactions and in social mutuality and communication strategies, but mostly transient and develops through time, maturity and daily experiences in the various educational settings and communities. The significant role of educators and therapists and other professionals in developing theses children's skills and competencies should not be forgotten.

Consequently, occupational therapists administer various assessments to investigate the type and severity of such behavioral disturbances, such as the Childhood Autism Rating Scale (C.A.R.S) (Schopler et al., 1986), in addition to assessing the behavioral responses towards environmental stimuli such as the Sensory Profile (SP) (Dunn, 1999). However, there is a foundational lack of a valid and objective methodology adapted for the visually impaired population, thus diagnosis of ASD in children with VI is often based on subjective clinical impression, with inconclusive prevalence data (Molinaro et al., 2020).

The Sensory Profile (Dunn, 1999) reveals the type and severity of the sensory modulation disorder in these individuals. Usually, this population presents sensory seeking behaviors in preferred domains, while simultaneously presenting sensory avoiding behaviors in other domains. For instance, seeking tactile sensation by the overwhelming use of manual functions and hand reach behaviors aiming to investigate the surrounding environment (both human and nonhuman), while avoiding many types of food textures which gradually, but systematically developing picky-eating behaviors. In many cases, in a somewhat contradictory manner, young children with congenital blindness avoid touching some objects that are perceived as "threatening" or "fearful" (e.g., unfamiliar pet or unfamiliar textures) and, thus, provoking anxious behavioral responses. In these cases, the young children react anxiously and respond in an uncommon psycho-motor manner (e.g., rubbing fingers, shaking arms, pressing own eyes by fingers, rocking upper body trunk, gazing on light source, etc.). All these characteristic behaviors and responses significantly affect the children's communication and social-interactional skills due to engagement in preferred and non-preferred stimuli but, these "Autism-like" behaviors are usually transient and become absent with development, maturity, experience and learning. Moreover, the loss of vision since birth and the restricted experiences within the over-protected life environments naturally and expectedly lead to a disturbed assimilation of perception, knowledge and skill even though these children have potentially intact cognition and executive functions (Choodbary et al., 2020). Although children with congenital blindness develop efficient executive functions in adolescence and adulthood, their experiences and learning strategies are profoundly different than sighted children or children with functional residual vision. The learning process is episodic, partial, limited and restricted by task-related factors or by environmental factors, repetitive in nature, and predominantly cannot be integrated with visual stimuli nor with eye-contact, gaze and visual tracking skills. High proportion of congenitally blind children has transient dysfunctions in language acquisition in general, and in syntax, grammatical and pragmatic use of language in a mutual manner due to the loss of direct eye contact with the others and in an accurate orientation to the others' exact location and distance from the speaking child. Comparably, individuals with autism often have significantly more sustained difficulties in processing non-literal and pragmatic elements of speech (Andrews & Wyver, 2005; Bowler, 2007). These impairments in language components and pragmatic use in children with congenital blindness profoundly affects their social interactions and interpersonal relations, including the lack of initiation of conversation and sustaining the communication coherently due to the loss of relevant information from the surrounding environment.

In summary, disturbed, uncommon and unintentionally deviated behaviors among infants with congenital blindness may become spontaneously acquired and assimilated in distorted psycho-social schemas and rigid behaviors (Fazzi et al., 2019). These stereotypical behaviors may further become affected by perceptual disturbances due to the loss of vision and the insufficient sensory integrative processing and the development of self-image and self-representation (Fraiberg, 1977). Furthermore, the human natural tendency of the parents to protect their children from surrounding hazards and risk factors may further impact the children's learning experiences and affective responses in a more restricting way. Parents prevent, halt or shift their child's actions during play activities and

personal activities of daily living leading to unfinished activities and engagements. As a result, the child learns and assimilates the habituation repetitive, episodic and insufficient engagement in routines, interactions and social participation even in adolescents with visual impairments (Pinquart & Pfeiffer, 2011).

Based on the previous findings and evidences, the differential criteria for the determination of autism-like behaviors as a secondary condition, compared to children with primary ASD, are proposed as follows: gaining more adaptive responses to effective developmental interventions in follow up assessments; presenting more efficient adaptation to environmental changes across life-contexts; gaining improved use of language in a more normative and correct manner through time; acquiring more mature interactions with family as well as with other unfamiliar individuals and, proving more positive prognosis due to spontaneous maturity and life experiences. As a consequent implication clinicians, educators and researchers are required to prove evidence for distinguishing between the diagnosis of transient Autism-like behaviors in children with congenital blindness as a secondary condition and the ASD as a primary diagnosis. Professionals are also required to prove the existence of sustained ASD symptoms in some children with congenital blindness despite of professional long-term interventions as an indication of comorbid conditions (primary ASD in congenitally blind children). On the long-term level, future DSM versions may take into consideration these proposed changes of the diagnostic criteria based on novel research evidence and on new findings retrieved from newly developed assessment tools and diagnostic methodology. This process may take a decade, but it may result in decreasing false-positives and true negatives in the diagnosis of children with primary ASD and children with transient, secondary ASD-like symptoms in children with congenital blindness. Moreover, children with congenital blindness who may suffer from dual diagnosis (i.e., ASD and congenital blindness) will be also diagnosed in a more accurate and case-specific methodology.

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