



Review

Additional considerations for the early detection and diagnosis of autism: Review of available instruments

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ABSTRACT

Once considered rare, autism is now viewed as a common and highly debilitating condition that has generated great interest among mental health professionals worldwide. The disorder is quite heterogeneous, can be very debilitating, and has a lifelong course. The general consensus among researchers, therefore, is that intervention should start at a very early age as a means of enhancing prognosis. The linchpin of early treatment is early detection and diagnosis. As a result of this recognized goal, psychiatrists and psychologists have been engaged in intensive efforts to develop methodologies and methods which will allow for the identification of autism at two years of age or younger. The current review focuses on research for early detection and diagnosis, relevant issues in identifying the disorder, and available assessment methods and instruments for making a diagnosis. Recommendations for future research are provided.

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The field of autism has become a major area of study in the fields of mental health and education worldwide (Adcock & Cuvo, 2009; Bhaumik et al., 2010; Brim, Townsend, DeQuinzio, & Poulson, 2009; Fernell & Gillberg, 2010; Smith & Matson, 2010). Once considered to be a rare condition, autism is now designated as one of the most common, and certainly one of the most debilitating, childhood disorders (Fombonne, 2008). Neurodevelopmental in origin, it is composed of social and communication deficits, as well as rituals and stereotypies. Autism, while treatable, has a lifelong course (Cederlund, Hagberg, & Gillberg, 2010; Johansson, Gillberg, & Råstam, 2010; Matson, Fodstad, & Rivet, 2009; Matson & Neal, 2009;

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Matson, Neal, Hess, Mahan, & Fodstad, 2010; Niklasson & Gillberg, 2010; Niklasson, Rasmussen, Óskarsdóttir, & Gillberg, 2009; Rose, Bramham, Young, Paliokostas, & Xenitidis, 2009). Additionally, the problems associated with autism are generally considered to be lifelong as well (Adcock & Cuvo, 2009; Dawson, Matson, & Cherry, 1998; Matson et al., 1996; Matson, Matson, & Rivet, 2007; Miniscalco & Sandberg, 2010). Nonetheless, there has been a response to behavioral and pharmacological interventions to varying degrees depending on comorbid intellectual disabilities and severity of core symptoms of autism (Al Anbar, Dardenne, Prado-Netto, Kaye, & Contejean, 2010; Chan et al., 2009; Devlin, Leader, & Healy, 2009; Matson, Mahan, & Matson, 2009; Petscher, Rey, & Bailey, 2009). As a result, a strong focus on early intervention has also resulted in an equally strong focus on early detection (Gutierrez et al., 2009; Hayward, Gale, & Eikeseth, 2009). This recognition is not specific to autism, however, as researchers studying a range of childhood disorders have come to the same conclusion, that early detection and treatment are critical (Birbeck & Drummond, 2005; Tremblay & Limbos, 2009).

For early assessment and diagnosis, it is also important to address a number of collateral problems. Some of these high co-occurring problems, such as immune factors, epilepsy, and intellectual disabilities are physiological (Matson & Neal, 2010; Matson & Shoemaker, 2010; Stigler, Sweeten, Posey, & McDougale, 2009). Additionally, various forms of psychopathology and challenging behaviors co-occur at high rates for persons with autism (Bakken et al., 2010; Davis et al., 2010; Farmer & Aman, 2010; Gillberg, 2010; Hagberg, Miniscalco, & Gillberg, 2010; Kroeger & Sorensen-Burnworth, 2009; LoVullo & Matson, 2010; MacNeil, Lopes, & Minnes, 2009; Matson & Dempsey, 2009; Matson, Dempsey, & Fodstad, 2010; Matson, Hess, & Boisjoli, 2010; Matson, Mahan, Hess, & Fodstad, 2010).

1. Early detection

Parents tend to notice autistic symptoms at a very young age. Most parents (76.2%) had concerns before age 3 and 83.3% thought their child had symptoms before age 2 in hindsight (Jónsdóttir, Saemundsen, Antonsdóttir, Sigurdardóttir, & Ólason, 2011). In another study 50% of parents reported concerns before 12 months of age (Kishore & Basu, 2011). And Planche (2010) posits that many symptoms which are present from birth may go undetected for many months. However, even after parents become concerned they seek professional help only after several months (Kishore & Basu, 2011; Planche, 2010). This has led some researchers to suggest that pediatricians be particularly vigilant at routine checkups with respect to developmental delays and core features of autism (Mandell et al., 2010). Early detection also varies with types of autism spectrum disorder, with autism being diagnosed 5 months earlier than Pervasive Developmental Disorder – Not Otherwise Specified or Asperger Syndrome. Presumably, this occurs because autism symptoms are more severe, and thus easier to identify, than those symptoms for Pervasive Developmental Disorder – Not Otherwise Specified or for Asperger Syndrome. Cultural factors may be at play also. Chakrabarti (2009) assessed a cohort of children diagnosed with autism and noted that mean age of symptom recognition did not correspond to a quick diagnosis. Rather, a 32-month delay between symptom recognition and diagnosis occurred. The author also makes the important point with respect to interdisciplinary collaboration. While a child psychiatrist or child clinical psychologist is likely to make the diagnosis, initial contact is almost always with the family pediatrician. Therefore, the pediatrician must know what to look for and to have good referral sources.

The diagnosis is further complicated by heterogeneity in symptoms and the collateral skills that are also affected. For example, Kalb, Law, Landa, and Law (2010) conducted observations between children with autistic regression, children with autism whose symptoms had plateaued (stopped gaining skills), and children who evinced typical development. The authors found that while children who regressed had better skill development than autistic children whose skills plateaued, long term the regression group had the poorest prognosis. Similarly, and with respect to collateral skills, children have also been assessed in conjunction with core features of autism. Ray-Subramanian, Huai, and Wesimer (2010) studied 125 toddlers with autism (23–39 months of age) using adaptive and developmental measures; the Vineland Adaptive Behavior Scale and Bayley Scales of Infant and Toddler Development. They note that developmental delays were evident as early as 2 years of age. These data replicated Matson and associates who had previously found similar results with children as young as 17 months of age (Matson, Hess, Sipes, & Horovitz, 2010; Matson, Mahan, Fodstad, Hess, & Neal, 2010; Matson, Mahan, Hess, et al., 2010; Matson, Mahan, Kozlowski, & Shoemaker, 2010). Furthermore, these symptoms appear to be stable once they appear (Chawarska, Klin, Paul, Macari, & Volkmar, 2009).

2. Methods of early identification

A number of procedures have been used for early identification of autism. Some methods, such as home videos and experiments aimed at determining toddler reactions to stimuli, have been helpful in alerting clinicians and researchers in establish content of diagnoses. Other methods, particularly those empirically derived and tested, have been used for early identification of children with autism and for early diagnosis.

2.1. Content of diagnoses

Home videos allow for direct observation of autistic children at a very young age. However, there is a general lack of standardized assessment in these home videos. Length of the video, number of people in the video, the setting in which the video is filmed, and tasks or activities the child was involved in at filming are among the many factors that are likely to vary.

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