SPECIAL ARTICLE

Autism in infants: an update

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Although autism is a disorder of very early onset, knowledge on how it is first expressed in infancy has, until recently, remained limited. In recent years new strategies of research, including prospective studies, have substantially increased our knowledge regarding autism in infants. Research findings have suggested the very early emergence of significant differences in social information processes. In addition to having important implications for research, these findings also offer new opportunities for screening and early identification and, hopefully, for improved outcome.

Key words: Autism, infancy, diagnosis, social development

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In his original report on autism, Leo Kanner (1) indicated his belief that the disorder was congenital in nature. Subsequent work has confirmed that, while a small proportion of children seem to develop the disorder after some months of normal development (2), symptoms emerge within the first two years of life in a vast majority of cases (3). Indeed, many of the skills not expressed by older individuals with autism are readily mastered by typically developing infants (4). Early onset would also be consistent with the impressive body of work supporting a genetic basis for the condition (5).

Somewhat paradoxically, our knowledge on autism as it is first expressed remains limited. This is unfortunate for several reasons. First, it makes it more difficult to disentangle the complicated impact that autism has on various lines of development, i.e., to focus on particular processes and sequences of development which go awry. In addition, there is now a strong suggestion that early diagnosis and intervention may significantly improve outcome (6). It is possible that, if a diagnosis of autism could be made very early in life, intervention could begin much sooner. In this paper we review current knowledge of autism in the first months of life, with a focus on diagnostic and developmental issues, and discuss areas important for advancing future research.

CLINICAL PRESENTATION IN INFANCY

Various problems have, until recently, limited our knowledge of autism as it is first expressed in infancy. Of the first 11 patients described by Kanner (1), only one was below age 3 years, and over half were above 4 years of age. Until recently, it was relatively common for the diagnosis to be made around age 4, even when parents were worried for several years before (7). As a result, much of the available research on early development was based on parental retrospection or, less commonly, reviews of home videotapes (8,9). The first longitudinal studies of young children referred for a possible differential diagnosis raised interesting questions about expression of autism in the first years of life (10). As more information on early development of children with autism has become available, it has generally confirmed Kanner's original emphasis on disturbed social development as the major clinical hallmark of the condition. Infants with autism appear to have limited eye contact, social attention and responsiveness (11), are less likely to engage in vocal or motor imitation (12), and may have problems with regulation of arousal and unusual responses to sensory stimuli (12).

Until relatively recently, the most frequent source of information on early development was that provided by retrospective parents' reports, which are complicated to interpret for a number of reasons. It is clear that many parents are seriously concerned about the child's development in the first year of life, and almost 90% are so concerned by age 2 (13). Common concerns include observations of social deviance or oddity, worries that the child may be deaf, and that the child's language fails to progress. Prospective data, for instance derived from high-risk populations, are clearly needed (14), although, when such data are available, it will be important to consider them in light of the rapid pace of change in infancy (i.e., the meaning and significance of particular behaviors may have important developmental correlations). For example, an early preoccupation with visual stimulation (staring at fans) may gradually transform itself into the repetitive stereotyped behaviors more frequently observed between ages 2 and 3 years. Similarly, contextual and situational variables become much more important in evaluating infant behaviors, e.g., the effects of arousal or novelty may impact the behaviors of interests more dramatically.

As noted above, one important complication in the study of infants with autism is the observation that in some cases (about 20%) early development is reported to be generally within normal limits. A clear consensus on the significance of regression in autism has not yet emerged, although Rogers (15) emphasized that subtle delays may precede more obvious skills loss. The advent of prospective studies of high-risk samples of younger siblings of children with autism should help to validate the phenomenon, and establish whether children with regression do or do not con-

stitute a specific subgroup that requires further, but separate, study.

An important approach to overcoming the lack of direct access to infants with autism is retrospective analysis of videotapes. This approach has its limitations, but a series of such studies have now generally confirmed the early emergence of developmental differences in infants later found to have autism. For example, a review of videos of infants up to 6 months suggested that those later diagnosed with autism exhibited less social visual attention, smiled and vocalized less commonly, and engaged in less object exploration (11); interestingly, abnormalities were not observed in terms of repetitive behaviors. Slightly older infants (8 to 12 months of age) have been noted to be less likely to respond to their own names (9,16). However, while failure to respond to name might be an indicator that a 12 month old child would benefit from a further evaluation for a possible diagnosis of autism, passing the "name calling" test does not mean that the child is not at risk of developing the disorder. Osterling et al (17) found that 12 month old infants with autism, compared to infants with mental retardation, differed with respect to orientation to name, while comparison to a nondelayed group showed differences in other areas, including repetitive behaviors, use of gestures, and looking at objects held by people.

By ages 6 to 12 months, differences become more pronounced in the area of communication, including a general lack of orientation towards verbalization in general and to their own name in particular. Infants with autism are less interested in people at a time when most infants begin to more fully integrate object exploration with social interaction and become more clearly intentional. On the other hand, some behaviors frequently reported by parents have not so clearly emerged as areas of difference using videotaped analyses (e.g., difficulties in arousal regulation). Such problems may be less specific to autism; alternatively, the failure to find differences may have more to do with the nature of the available videotaped materials.

Research on the development of toddlers (ages 1 to 3) with autism is greater in quantity than that on infants, and is relevant to work on infancy in that it highlights areas which may serve as precursors of behaviors later observed. In general, the data indicate that differences from typical peers are readily observable to parents and others by at least age 30 months. The behaviors that differentiate children with autism from those with developmental delay include both person-to-person behaviors (anticipatory postures, turn taking, intensity of eye contact) and behaviors which involve some aspects of joint attention (e.g., pointing to materials, following the point of another person or giving objects). In addition, limited affective responses and unusual sensory and motor behaviors are more frequently observed (3). Other areas of difficulty include abnormal play and limited response to speech (18,19). In general, the results (based on both parent report and observation/assessment) indicate that between the second and third birthday higher levels of more "typical" autistic behaviors are present, so that a diagnosis can be made with greater certainty by that time (20).

DIAGNOSTIC ISSUES

Since Kanner's first description of autism (1), the diagnostic concept has undergone modification based on research and clinical work, while retaining important historical and conceptual continuities with Kanner's description (21). Kanner emphasized the centrality of the social difficulties as well as the presence of a set of unusual behaviors he subsumed under the term "insistence of sameness" or "resistance to change". These behaviors included unusual movements and mannerisms as well as problems in dealing with change and novelty. This approach was generally retained in the DSM-IV, where the final definition of autism was based on a large multinational field trial (13) including information on nearly 1000 cases seen at over 20 sites around the world. Of these cases, over 300 were less than 5 year of age (although most were aged between 3 and 5). The final diagnostic approach provided reasonable coverage over the range of syndrome expression in autism and was applicable from early childhood (i.e., around age 3) through adulthood. At the time the DSM-IV was formulated, there was much less concern about the diagnosis of autism in infants and it appeared that the approach derived worked satisfactorily by age 3. Examination of some of the DSM-IV field trial data for children under age 5 did reveal a few items with stronger developmental correlates. In general, such items were discarded since they would not be applicable to the entire range of syndrome expression. For example, attachment to unusual objects has low sensitivity (0.50) but high specificity (0.90), so that, when it is observed, it has high predictive power for autism but only in this younger age group.

The applicability of current (DSM-IV) approaches to the diagnosis of autism in infants and very young children has been questioned on several grounds: for instance, some of the criteria are not clearly applicable to infants, reducing the potential pool of available criterion items (22).

Another approach has focused on the development of screeners and checklists. Such instruments provide an important alternative to the more detailed DSM type diagnostic approach, but entail a somewhat different set of concerns or constraints: for instance, overdiagnosis may be much less of a concern than underdiagnosis; time constraints and issues of efficiency are important as is ease of use and the degree of training required for use (23). Level 1 screeners are intended to identify children likely to have disability from typical peers, while Level 2 screeners focus more specifically on differentiating children at risk for autism from those with other difficulties. The latter are typically intended for more specialized settings. Screeners to date have not focused on children under 1 year.

DIRECTIONS FOR THE FUTURE AND IMPLICATIONS FOR CLINICAL SERVICES

Recent work employing new approaches to the study of social attention has begun to emerge and has important potential for contributing to both screening and early diagnosis. These findings build on the observation that older individuals with autism exhibit highly unusual patterns of attending to people (24). Similar results have now been obtained with toddlers (25). These results are consistent with the general lack of salience of social motivation and interest (26). Another approach has used automatic attention cuing (27) to further clarify differences in early gaze processing. The use of strategies based on these techniques may offer important new approaches to early screening that are more physiologically based.

As diagnostic and screening methods improve, more and more infants will be referred for assessment and treatment. It remains unclear what services will best be employed in treatment. The recent US National Research Council Report (6) has summarized work relevant to somewhat older, i.e. preschool, children, while there is uncertainty about models of service provision that will be most appropriately provided to infants. However, the potential for focusing on remediation in the earliest months of children's lives does provide the possibility for substantially improved outcomes.

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