

AUTISM IN INFANCY AND EARLY CHILDHOOD

Fred Volkmar, Kasia Chawarska, and Ami Klin

Child Study Center, Yale University School of Medicine, New Haven, Connecticut 06520;
email: fred.volkmar@yale.edu, kasia.chawarska@yale.edu, ami.klin@yale.edu

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■ **Abstract** Although initially described as an inborn disorder of affective contact, information on autism as it exists in infants has been limited. Delays in diagnosis, lack of information about the condition, and reliance on retrospective research strategies have been problematic. An awareness of the increased risk for siblings is now allowing the development of new, prospective approaches. Consistent with Kanner's original hypothesis, the available information strongly suggests a fundamental difficulty in the earliest social processes, which, in turn, impacts many other areas of development. New approaches to screening have lowered the age of initial diagnosis; this presents new challenges for early intervention. Directions for future research are highlighted.

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INTRODUCTION

Although first described by Kanner in 1943 as an inborn disorder of "affective contact," information on autism in infants and very young children is limited. Delays in diagnosis were common and a diagnosis often was not made until age

4 (Siegel et al. 1988). This has changed recently as public awareness of autism and the importance of early diagnosis have increased (Nat'l. Res. Council. 2001). A decade ago, information about the early development of autism was based on parent reports (Cohen et al. 1986) or retrospective review of home movies or videotapes (Osterling & Dawson 1994); over the past decade a few longitudinal studies of young children with autism have begun to appear (Lord 1995). As part of its 10-year plan for autism research, the National Institute of Mental Health has set the ambitious goal of actually reducing the frequency of autism in school-aged children through early diagnosis and intervention—a goal that would have seemed unthinkable a decade ago. In this chapter, we summarize what is known about autism as it exists in the first years of life, including the onset of the condition and behavioral and developmental features as well as approaches to screening. We review some aspects of assessment and intervention and conclude with a discussion of the limitations of current knowledge and areas important for the future.

DEVELOPMENT OF AUTISM AS A DIAGNOSTIC CONCEPT

Kanner's original paper (1943) emphasized that children with autism were born without the usual predisposition to be social. He grounded his description developmentally by citing the early emergence of social interest, an interest that we now know is present from birth (Chawarska & Volkmar 2004). Although his description was remarkably accurate in some aspects, it was modified based on clinical experience and research. For example, he initially believed children with autism had normal intelligence, but it is now clear that although markedly uneven profiles of ability are observed, most individuals with strictly defined autism function, overall, in the intellectually deficient range. Kanner's impression that children with autism did not exhibit other medical conditions was modified because a high risk for developing seizures became apparent as cases were followed (Volkmar & Nelson 1990). It also became clear that autism has a very strong genetic basis, with siblings having a 20- to 50-fold increase in their risk for developing autism (Rutter et al. 1997, Volkmar et al. 2004b).

The original definition of autism has evolved over time. Kanner emphasized two features: the "autism" and a group of unusual behaviors he subsumed under the terms "insistence of sameness" or "resistance to change"; the latter include unusual movements and mannerisms as well as a literal difficulty in dealing with novelty. Early work on autism was impeded by controversies over the validity of the condition, but by 1980 autism was officially recognized and included in a new class of disorder—Pervasive Developmental Disorder (PDD). Over the years other, apparently related, diagnostic concepts were proposed. Probably the most common and, somewhat paradoxically, the least studied of these conditions is Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS)—a category used for children with some features of autism (Volkmar et al. 2004a). The validity of these conditions apart from autism remains the topic of much interest and research; the primary focus of this review is on strictly defined autism.

A series of epidemiological studies have been conducted and generally suggest rates of autism between 1 in 500 and 1 in 1000 children; children with difficulties falling within the broader PDD category are probably three or four times as common (Fombonne 1999). Of the approximately 30 epidemiological studies, recent ones have tended to report higher rates of the disorder, although changes in diagnosis, increased awareness, and other factors complicate our understanding of this increase (Fombonne 1999, Wing & Potter 2002). Rates of autism in boys are three to four times higher than in girls (Fombonne 1999, Lord et al. 2002). In addition to the high risk for developing seizures and the strong genetic basis of the condition, a host of other research findings strongly implicates neurobiological factors in pathogenesis of the condition (Volkmar et al. 2004b).

AUTISM IN THE FIRST YEAR OF LIFE

Age and Type of Onset

Parents report a range in the age at which they are first concerned about their child's development, with about 90% recognizing abnormality by 24 months (De Giacomo & Fombonne 1998). While speech delays or worries about hearing are common, other early concerns may be that the child is "too good" or is highly irritable (Stone & Lemanek 1990). Unfortunately, the specificity of these problems to autism remains unclear. Most of the available work is based on parent report. Prospective data are needed, although the interpretation of such data is complicated given the fast pace of change and the various meanings of the presence (or absence) of a behavior at specific time points (Lord & Risi 2000). For example, preintentional use of gestures such as reach-and-grasp motions in pursuit of an object is seen before age 9 months, but it usually becomes quickly synchronized to eye contact and indicates emerging intentional communication (Bates et al. 1979). After, but not before, the one-year developmental level a lack of conventional gestures such as nodding or pointing would be a source of concern. To complicate the task even further, early symptoms change over time (Kanner 1968, Lord 1995). Finally, effects of situation, novelty, task demands, and unusual patterns of environmental responsivity may result in considerable variability in the child's presentation, particularly in infants and young children (Natl. Res. Council. 2001).

The phenomenon of reported regression is another potential problem. Various studies (Kobayashi & Murata 1998, Rogers & DiLalla 1990, Tuchman & Rapin 1997, Volkmar et al. 1985) have documented parental reports of regression in 20% to 40% of cases. Unfortunately, various terms—which overlap to some degree—are used, e.g., regressive autism, setback autism, and developmental stagnation. These terms do, however, speak to some aspects of the underlying phenomenon, i.e., in some children there may be a gradual or more rapid loss of language and/or social skills (Kobayashi & Murata 1998, Rogers & DiLalla 1990, Tuchman & Rapin 1997). In other cases, the problem seems to be one not so much of loss as of a failure to make progress, e.g., the child seems to say one or two words but then

language does not progress (Sipersein & Volkmar 2004). Finally, in some rare cases the child progresses normally for several years (usually three to four) with normal language, cognitive, and self-care skills, and then gradually or abruptly loses these abilities and begins to exhibit more classic features of autism. The term childhood disintegrative disorder is used in such instances and available data suggest an even worse outcome than for more typical autism (Volkmar & Rutter 1995).

The relationships, if any, between these various subtypes and their validity as clinical phenomena remain controversial. For example, Osterling et al. (2002) found minimal differences between regressive and nonregressive autism, whereas Rogers & DiLalla (1990) have noted poorer outcome in the regression group. The latter group may also be more likely to exhibit dysmorphic features (Lainhart et al. 2002). Thus, consistent with Kanner's original (1943) impression, autism does appear to be a very early onset disorder but there are a small number of cases where regression is reported. It is possible such cases represent a specific subtype or potentially different diagnostic group, but the issue remains unresolved.

Clinical Presentation in the First Year of Life

Kanner's original (1943) report emphasized unusual social development. Subsequent research has refined his initial impression in various ways, e.g., infants with autism may have limited eye contact and diminished overall social responsiveness (Maestro et al. 2002, Sparling 1991). They also may be less likely to engage in motor or vocal imitation and to exhibit problems in arousal or unusual sensory responses (Dawson et al. 2000). The lack of comparative or normative data often presents difficulties for interpreting these findings, although in one study (Klin et al. 1992) preschool children were not exhibiting social behavior that normally would be expected prior to age 1 year. For example, the children with autism failed to assume anticipatory postures, reach for familiar persons, show interest in children other than peers, and engage in simple social interaction games.

In addition to parent report data, the retrospective analysis of home movies or videotapes provides another potential research resource. Such research has generally confirmed the early emergence of developmental differences in infants with autism (Adrien et al. 1992, Maestro et al. 1999, Osterling & Dawson 1994, Osterling et al. 2002). For example, Maestro et al. (2002) examined early videotapes of infants later diagnosed with autism as compared to age-matched typically developing infants; the infants who went on to be diagnosed with autism showed less visual attention to social stimuli, they smiled less frequently, vocalized less, and they engaged less in object exploration. However, the infants did not differ in terms of repetitive behaviors. In a study of 8- to 10-month-old infants, Werner et al. (2000) reported that those subsequently diagnosed with autism were less likely to respond to their own name, a result similar to that of Osterling & Dawson (1994) in their study of 12-month-olds. The use of a typically developing comparison

group limits some of these findings, i.e., differences observed may reflect general developmental delay rather than the effects of autism *per se*.

A more stringent approach includes the use of a developmentally delayed comparison group. Using a range of behaviors, Osterling et al. (2002) compared 12-month-old infants with autism to infants with mental retardation only: Significant differences were found in orienting to name and looking at people, using gestures, looking at objects held by people, and performing repetitive actions.

Although these data are limited in various respects, early signs of autism appear to include—as Kanner would have predicted—a lack of social interest in the first months of life, with reduced levels of social engagement and social-communicative interchanges; differences in the nonsocial areas are much less striking. By ages 6 to 12 months, differences become more pronounced in the communicative area, including a general lack of orientation toward 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 (Bates et al. 1979). 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.

AUTISM: AGES 1 TO 3 YEARS

The quantity and quality of available research rises dramatically with respect to toddlers. In part this reflects the fact that it is around this time that parents are more likely to seek evaluation (De Giacomo & Fombonne 1998). In addition to grossly apparent developmental delays (e.g., failure to speak), unusual behaviors may prompt concern and may begin to include stereotyped motor mannerisms (e.g., hand flapping) or idiosyncratic uses of materials (e.g., spinning things) (Wimpory et al. 2000).

Parental report has been used to compare children with autism to those with both typical development and developmental delay. By age 30 months, differences from typical peers are readily apparent, and social deviance, problems in communication, and unusual responses to the nonsocial environment are striking (Ornitz et al. 1977). Parent report data have identified various behaviors that differentiate young children with autism from those with developmental delay. These include both person-to-person behaviors (anticipatory postures, turn taking, intensity of eye contact) and behaviors in which an object is the focus of joint interest (joint attentional skills such as pointing to materials, following a point of another person, or giving objects) (Wimpory et al. 2000). Limited affective engagement and unusual sensory behaviors are also observed (Hoshino et al. 1982).

By this developmental period, it becomes feasible to employ contemporaneous parent report and direct observation. For example, in a prospective questionnaire study, Dahlgren & Gillberg (1989) reported that a number of items discriminated groups of children with autism (below age 3) from those with typical development or developmental delay. Difficulties in the area of social development, unusual gaze, abnormal play, and lack of responsivity to speech were observed. Cox et al. (1999) followed a group of 20-month-old infants prospectively; the infants were high-, medium-, or low-risk for autism as identified through a screening using the Checklist for Autism in Toddlers (CHAT) (Baron-Cohen et al. 1992). At 20 months, behaviors that discriminated the children included range of facial expression, use of conventional gesture, and pointing to indicate interest. A follow-up study was conducted at 42 months, and additional items were noted to differentiate the group, e.g., desire for shared enjoyment, imaginative play, offering comfort, and nodding; none of the repetitive, stereotyped behaviors differentiated the groups. Lord (1995) followed 2-year-olds evaluated for a possible diagnosis of autism. A standard set of assessments was employed, including the Autism Diagnostic Interview (ADI) (Le Couteur et al. 1989), with items specifically selected as relevant to children under 2 years of age. After 12 to 15 months, the children were re-evaluated using the same measures. Items that differentiated the groups again included social activities (seeking shared enjoyment, social reciprocity, use of another person as a tool, interest in other children), and communicative tasks (attending to voice, pointing, using and understanding gesture). Two behaviors, directing attention and attention to voice, correctly identified 82% of children. By age 3, finger mannerisms, attention to voice, pointing, and use of the other person's body were, in addition, able to correctly classify all subjects.

These results suggest significant changes between the second and third birthday, with higher levels of more "typical" autistic behaviors present by age 3; conversely, some of the behaviors suggestive of autism at age 2 substantially decreased by age 3 in the developmentally delayed comparison group (Lord 1996, Lord & Pickles 1996).

This growing body of research illustrates important areas where findings converge, but it also suffers from a number of significant limitations. The available data suggest that for many infants, differences may begin to become apparent in the first months of life; however, these can be subtle and current data have important limitations. For example, in retrospective studies, parents may be more accurate in reporting negative, rather than positive, symptoms (Stone et al. 1994) and may have difficulties in noting some of the deficits in play and joint attention that seem more frequent between the second and third birthday (Charman et al. 2001). Videotaped materials provide important advantages, but they suffer from the potential disadvantages intrinsic in selective taping, e.g., lack of evidence for areas of difference in what parents chose to record. Comparisons across studies can be difficult due to differences in method, comparison group, sample selection, and so forth. As diagnostic methods for infants become more robust and as prospective studies are undertaken, it may be possible to more adequately address these concerns.

DIAGNOSTIC ISSUES

A growing body of work has emphasized the importance of early case detection and intervention for improving long-term outcome (Natl. Res. Council. 2001). Although early signs of autism are apparent in many cases before age 12 months, diagnosis of infants and very young children is quite complex. Several approaches have been utilized (see Lord & Corsello 2004 and Volkmar et al. 2004b for reviews).

Categorical Approaches

Categorical approaches to autism are exemplified in systems such as the Diagnostic and Statistical Manual (DSM; Am. Psychiatric Assoc. 1994) and International Statistical Classification of Diseases and Related Health Problems (ICD; WHO 1992). These have the advantages of official recognition and a long history of facilitating research and clinical work; in many ways, they also serve as the conceptual basis for dimensional approaches (Volkmar et al. 2004a). In his original paper, Kanner (1943) emphasized two essential features—the autism as well as the entire set of behaviors he subsumed under the term “insistence on sameness.” Early progress in the field was delayed because of a lack of explicit definition and confusion about continuities with other disorders (particularly childhood schizophrenia). During the 1970s, a consensus emerged on the validity of autism, and diagnostic guidelines that were more formal were proposed. Rutter (1978) emphasized the early onset of the disorder and the characteristic problems in social development and communication (not just due to associated mental retardation), along with the presence of unusual behaviors of the type Kanner had conceptualized as “insistence on sameness.” His approach was highly influential in the definition that appeared in DSM-III (Am. Psychiatric Assoc. 1980), where autism was first granted official recognition as a category of disorder.

Over the years, various changes and modifications have been made in the DSM and ICD definitions of autism. The original DSM-III definition focused on the “infantile” aspects of infantile autism, e.g., delineating the presumed initial presentation of the disorder; developmental issues in syndrome change were not satisfactorily addressed. This concern led to major changes in the revision of DSM-III, wherein developmental issues were addressed in the guidelines for the disorder although with the price of over-diagnosis of the condition in more intellectually disabled individuals (Volkmar et al. 1994). The current system, DSM-IV (Am. Psychiatric Assoc. 1994), was developed based on an international study done in collaboration with the developers of ICD-10; these two systems are now essentially the same. The current definition of autism is historically continuous with Kanner’s original work and with Rutter’s subsequent modifications. In this system, autism is defined on the basis of problems in the areas of social development, communication and play, and restricted and stereotyped interests (with social factors being given slightly more weight); by definition autism must have its onset by age 3 years. This system has a reasonable balance of sensitivity and

specificity over the IQ range (Volkmar et al. 1994). In addition to autism, it includes a number of other specifically defined disorders, including Asperger's disorder, Rett's disorder, and childhood disintegrative disorder as well as the "subthreshold" PDD-NOS. The data used to develop this definition included a substantial cohort of 3- to 5-year-olds, but few children under age 3. By ages 3 to 5 the definition appears to work reasonably well, although for infants and young children the data on its sensitivity and specificity are much more limited. As the work on early development suggests, two kinds of problems are particularly likely. Some children at age 2 exhibit the social and communication-play problems typical of autism but do not yet exhibit the unusual stereotyped movements or other behaviors in the third category of disturbance; many of these children go on to do so before their third birthday. Less frequently, children below age 3 may appear to exhibit all of the features required for a diagnosis of autism but then lose these features as they mature (Lord 1995). Furthermore, Stone and colleagues (1999) note that some of the DSM-IV criteria clearly are not as applicable to very young children, e.g., criteria involving peer relationships and conversational skills. At least one attempt has been made to provide an alternative categorical diagnosis system for autism and related disorder specifically for infants and young children (Nat'l. Cent. Clinical Infant Prog. 1994), although this system suffers from a number of problems and lacks substantive empirical basis.

A somewhat different approach to the issue of categorical diagnosis is employed in instruments like the Autism Diagnostic Interview-Revised (ADI-R) (Lord et al. 1994) and the Autism Diagnostic Observation Schedule (ADOS) (Lord et al. 2000)—a semistructured interview with parents and direct child assessment, respectively. Both instruments were developed to be keyed to categorical diagnostic criteria and emerged as the most widely used diagnostic instruments for research purposes (see Lord & Corsello 2004). The ADI-R works well for children over 4 years of age but much less so for younger children (Cox et al. 1999, Lord 1995, Stone et al. 1999). Lord (1995) noted that among 2-year-olds this instrument over-diagnosed among the more mentally handicapped and conversely under-diagnosed among the more intellectually able (sensitivity and specificity both approximately 0.50). Cox and colleagues (1999) reported a similar result in a study of 20-month-olds. Some modification in the scoring rules increased the sensitivity of the instrument, but low specificity remained problematic. By age 42 months, sensitivity was improved but the tendency to over-diagnose autism in children with severe cognitive delay remained (Cox et al. 1999).

In contrast to the ADI-R, the ADOS employs observation of and interaction with the child. DiLavore et al. (1995) used an early version of this instrument to compare young children with autism (age range 38 to 51 months) to children with developmental delay (ages 2 to 3 years) and noted several areas of differences between the groups, e.g., use of social smile, sharing pleasure, eye contact, giving to others, response to name, use of gesture, differentiation of parent from examiner, and levels of stereotyped behaviors. These results are similar to those reported by Stone et al. (1994).

Dimensional Approaches to Diagnosis and Checklists

Dimensional diagnostic approaches offer many potential advantages, particularly for infants and young children. Their use offers better approaches in dealing with developmental change and is not necessarily incompatible with categorical approaches (see Lord & Corsello 2004). These instruments also have some intrinsic limitations. The focus on highly deviant behavior creates some challenges for these instruments, e.g., items may be more difficult to develop, and behaviors of interest may be low in frequency or difficult to rate reliably. Other problems include the way the instrument addresses (if at all) the issue of history and change, the nature of the intended informant (parent report, teacher report), and the format (interview versus direct observation). Low-frequency but highly significant diagnostic behaviors may not be observed during direct assessment; on the other hand, employing parent or teacher report raises other issues in terms of reliability, potential effects of informant knowledge and sophistication, and so forth. One approach might be to combine multiple informants or sources of information, although this creates additional difficulties (Offord et al. 1996). In terms of item or scale development, differences may exist in the extent to which the absence of specific skills is emphasized versus the presence of highly unusual behaviors. For example, are the widely recognized difficulties in eye contact in early social engagement better conceptualized as a lack of normal social engagement or as the presence of high abnormal patterns of functioning? For some items/scales, there may be strong correlates with age or cognitive development (Tadevosyan-Leyfer et al. 2003). To complicate the situation further, the presence of deviant behaviors may be differentially correlated with the absence of more developmentally appropriate behaviors. Attention to both delay and deviance is required. In observational approaches, the absence of a specific behavior may be much more difficult to interpret than its presence (Lord & Corsello 2004).

Despite these concerns, a number of dimensional rating scales and checklists have been developed. These instruments differ in many ways, with intended uses ranging from diagnosis and diagnostic assessment to population screening. These approaches have had a significant role in research as well as in clinical work, although there is—with some noteworthy exceptions centering particularly around screening—a dearth of information on their use in infants and very young children. For example, the Childhood Autism Rating Scale (CARS) (Schopler et al. 1980) has been widely employed in the assessment of older children; it rates the child along a series of dimensions of severity of autism. In Lord's (1995) study, CARS and the ADI-R were in general agreement with each other and with the diagnoses of clinicians. However, both CARS and the ADI-R were less accurate for children 2 years of age or younger, with CARS over-diagnosing the condition.

In contrast to instruments like the ADI-R and CARS, another line of work has centered on development of brief rating scales/checklists that might be used more for screening than for definitive diagnosis. The development of such instruments is particularly important for prospective studies (e.g., of newborn siblings at risk for

autism) and for epidemiological studies. Development of screening instruments entails consideration of a somewhat different set of concerns and constraints (see Aylward et al. 1997, Coonrod & Stone 2004). Level 1 screeners are intended to identify children at risk for more general disability; such instruments are frequently used in general medical (pediatric) practice and are usually concerned with the potential identification of a broad range of developmental problems. In contrast, level 2 screeners focus more specifically on differentiating children at risk for autism from those with other difficulties, such as overall cognitive delay or language impairment. Level 2 screeners are more detailed and focused and more frequently used in specialized settings.

The Checklist for Autism in Toddlers is a Level 1 screener that combines parent report and observation of the child at 18 months (Baron-Cohen et al. 1992, 1996, 2000), and samples a small number of items thought to suggest autism. Results from the original study (from which children with severe delays were excluded) suggested high levels of specificity but low sensitivity: subsequent modifications have been undertaken (Baird et al. 2000, Scambler et al. 2001).

Robins and colleagues (2001) attempted to address some of the concerns about the original CHAT in the modified CHAT (M-CHAT); it employs 23 items in a screener designed for 2-year-olds. In their report, the instrument appeared to have reasonably good sensitivity and specificity. Predictors of autism included items related to joint attention, social engagement, and communication, consistent with earlier reports of symptoms typical of autism in 2-year-olds (Baron-Cohen et al. 1996, Lord 1995). This instrument is still in development; rates of false negative and false positive cases remain to be determined.

Second stage screening tools have also been developed. The Pervasive Developmental Disorders Screening Test (PDDST) is a parent report measure for children under age 6 years with different versions for different levels of screening (Siegel & Hayer 1999). Early results appeared promising but they have not yet been published in a peer-reviewed journal. The Screening Tool for Autism in Two-Year-Olds (STAT) (Stone et al. 2000) is designed for children from ages 2 to 3 years. It includes 12 pass/fail items that address aspects of communication, social engagement, and play; administration is in a play-like setting and takes about 20 minutes. Early results were encouraging but again the instrument needs to be evaluated in larger, and community-based, samples.

Other Level 2 screeners include the Autism Behavior Checklist (ABC) (Krug et al. 1980) and the Gilliam Autism Rating Scale (GARS) (Gilliam 1995). The ABC is a 57-item checklist with items differentially weighted depending on the strength of their association with autism in a large sample of individuals (from age 18 months to 35 years). Although it is quickly administered and widely used, there are some concerns about its psychometric properties, and its usefulness in infants and very young children remains unclear. The GARS is a 56-item behavior checklist designed to screen for autism in individuals from ages 3 to 22; it can be rapidly completed with little training. Its utility in infants and young children

remains to be established; there is some concern about its sensitivity with preschool children (South et al. 2002).

Clinical Diagnosis

To date the most robust method to diagnose autism in infants and very young children remains the use of experienced clinicians (Adrien et al. 1992, Cox et al. 1999, Lord 1995, Stone et al. 1999). The clinical diagnosis of autism is highly stable, with 72% to 87% of cases retaining the diagnosis at follow-up (see Klin et al. 2004 for discussion). It presumably is the case that experienced clinicians draw upon a much richer range of knowledge than that embodied in diagnostic guidelines or criteria (Klin et al. 2000). At present clinical diagnosis continues to be the "gold standard" in diagnosis of autism spectrum disorders in infants, toddlers, and preschoolers (Lord 1995, Stone et al. 1999). Development of better (categorical and dimensional) approaches specific to infants and very young children is an important research priority and one that is essential in understanding the boundaries of the disorder in relation to broader autism spectrum conditions.

DEVELOPMENTAL FEATURES OF AUTISM IN THE FIRST TWO YEARS OF LIFE

Social Functioning

Social difficulties are the single most powerful predictor of diagnosis for older individuals with autism (Siegel et al. 1989, Volkmar et al. 1994); this likely is true for infants as well. Preschool children with autism often fail to demonstrate social skills typically present in the first months of life (Klin et al. 1992). Eye contact is limited as is overall social engagement and responsivity (Dawson et al. 2000, Sparling 1991). Difficulties in the area of joint attention are striking (Mundy et al. 1990, Sigman & Ruskin 1999); such behaviors are central in development of communicative and social-cognitive abilities (Tomasello 1995). Over time, both initiation and response to bids for joint attention do increase in children with autism (Leekam et al. 2000), but are severely compromised in natural contexts (Klin et al. 2002a,b). The pattern of acquisition of these skills in autism may be atypical (Carpenter et al. 2002, Klin et al. 2003).

For typically developing children, imitation and play are important for symbolic (Piaget 1952a,b; Vygotsky 1990) and social-cognitive development (Trevvarthen & Aitken 2001)¹. Older children with autism consistently have difficulties in imitation

¹It is important to distinguish between true imitation, which entails an aspect of intentionality as well as the translation of observed movements of others into motor output from echopraxia, and simple mimicry, which occurs without an appreciation of the person's goal (Tomasello 1995).

(Hobson & Lee 1999, Loveland et al. 1994, Smith & Bryson 1994), and these difficulties are present by at least the second year of life (Charman et al. 1997, Dawson et al. 1998, Roeyers et al. 1998). Levels of imitation positively correlate with language levels (Sigman & Ungerer 1984).

Play activities in the typical child progress from simple object exploration to functional object use to pretend play. Although the evidence on this issue is limited, in the first months of life strong differences are not observed between infants with autism and typical or delayed peers (Baranek 1999, Maestro et al. 2002, Osterling et al. 2002). But by 9 to 12 months, abnormalities are evident (Baranek 1999) and become progressively more deviant due to higher levels of perseveration (Osterling et al. 2002). Although early functional play routines may be observed (Charman & Baron-Cohen 1997), by around age 2 years differences from typically developing peers are striking: play is less purposeful, less symbolic, and less developmentally complex (McDonough et al. 1997, Mundy et al. 1986, Sigman & Ruskin 1999, Stone et al. 1990).

By the end of the first year of life, typical infants have developed strong patterns of attachment; suck behaviors maintain the proximity of the child to the caregiver while also facilitating exploration (Rutter 1995). Children with autism do form attachment to parents (Capps et al. 1994, Rogers et al. 1993, Shapiro et al. 1987, Sigman & Mundy 1989, Sigman & Ungerer 1984) and differentially respond to familiar and unfamiliar individuals (Landry & Loveland 1989, Sigman & Ungerer 1984). However, the quality of attachment behaviors may be unusual (Rogers et al. 1993). Among younger children with autism, attachments to unusual objects are relatively common (Volkmar et al. 1994), but the significance of this is unclear.

Communicative Development

Concerns about the child's speech and communicative development are among the most frequent initial presenting complaints (De Giacomo & Frombonne 1998). Even before they begin to produce spoken language, patterns of sound production in children with autism are abnormal (Wetherby et al. 1989) as is vocal quality (Sheinkopf et al. 2000), a likely precursor of the noteworthy deficits in intonation and vocal quality seen later (Shriberg et al. 2001).

The development of nonverbal communicative abilities is intimately involved for typical children in the development of conventional communicative abilities and marks the beginning of intentional communication (Bates et al. 1979). However, very young children with autism communicate less frequently than matched developmentally delayed children (Stone et al. 1997, Wetherby et al. 1989). They are less likely to use contact and conventional gestures but are more likely to use highly unconventional gestures such as manipulating their conversational partner's hand to obtain objects (Stone et al. 1997).

Children with autism have difficulties with both the expression and production of affective responses, and the range, frequency, and integration of affective

displays are unusual (Ricks & Wing 1975, Snow et al. 1987, Yirmiya et al. 1989). Children with autism are less likely to look at an apparently distressed adult (Sigman et al. 1992) and they have difficulties imitating facial displays of emotion (Loveland et al. 1994).

Cognitive Development

Assessment of infants and very young children with autism present particular challenges (Klin et al. 2004) given the inherent limitations of instruments in this age group and the particular difficulties posed for assessment of children with autism. As developmental tests sample a broader range of skills, i.e., as the child becomes somewhat older, delays typically become more apparent in tasks that require language-based problem solving, symbolic thinking, or aspects of social interaction; tasks that are less verbal, e.g., visual matching, may be closer to age-expected levels (Klin et al. 2004). One consistent observation made first by Kanner (1943) concerns the emergence of noteworthy discrepancies across developmental areas. This developmental *decalage* has its origin in early childhood (Sigman & Ungerer 1981), and it not only persists over time but it often becomes even more striking (Ehlers et al. 1997, Freeman et al. 1988, Klin et al. 1995). Differences in sensorimotor development do not seem to be syndrome-specific (Cox et al. 1999, Dawson et al. 2002b, Morgan et al. 1989, Sigman & Ungerer 1984). Although studies of older children have demonstrated difficulties in executive functioning (Pennington & Ozonoff 1996), studies with younger children generally have not revealed syndrome-specific differences; in some instances preschool children with autism perform better than matched developmentally delayed controls on tasks (Dawson et al. 2002b, Griffith et al. 1999).

Attentional abnormalities in older children with autism are well documented. Difficulties in autism have to do with selective attention, with problems in attending to multiple aspects of stimuli and with auditory stimuli in particular (Burack et al. 1997). Infants later diagnosed with autism show that they attend less to people than to objects as compared to both typically developing and delayed controls (Baranek 1999, Maestro et al. 2002, Osterling et al. 2002); this is also true after the second year of life (Dawson et al. 1998). Selective social attention is particularly impaired, e.g., during free play 20-month-olds with autism were more likely to look at objects than at people than were either typical or delayed peers (Swettenham et al. 1998).

Recent work employing new approaches to the study of social attention has shown that older individuals with autism exhibit markedly deviant patterns of attention to people and, particularly, to faces as compared to developmentally matched controls (Klin et al. 2002a,b). Similar findings are emerging with very young children with autism (Klin et al. 2003). Figure 1 shows a toddler with autism viewing a videotape of a popular children's program: Rather than focusing on either the large purple character or the two children, the child's point of regard is on the intersection of several corners and the rainbow backdrop.

Various attempts have been made to account for these early differences in social-visual attention. The problem might have to do with avoiding complex

visual stimuli, e.g., faces (Swettenham et al. 1998), or with avoiding unpredictable and variable social stimuli (Dawson & Lewy 1989). A third hypothesis posits the fundamental problem as a lack of social motivation and salience (Dawson et al. 2002a, Klin 1991). In a study of automatic attentional cueing (by either eye movement or nonbiological movement) in 2-year-olds, the visual attention of the children with autism could be cued by directional changes in eye movement, although cue-specific differences also suggested different underlying strategies (Chawarska et al. 2003). Data that clarify the nature of these differences are critically needed.

ASSESSMENT AND INTERVENTION

Multiple aspects of the child's development must be assessed, usually necessitating the involvement of a range of professionals with attendant issues of coordination and communication. Additionally, diagnostic issues can be complex and sometimes are clarified only with time. Translation of assessment results into practical implications for intervention is critical, as is involvement of parents. Developmental assessment is a crucial aspect of this process (Klin et al. 2004); it helps to frame subsequent observations and can provide crucial information on intervention strategies. Speech-language-communication assessment is also vital. Delay and deviance in the area of communicative ability is universal in autism and is one of the most central areas for intervention; hence, speech-language-communication assessment is a core component of the diagnostic process (Wetherby & Prizant 2000). Assessment of adaptive skills is also critical; the inability to translate what the child can do in a structured testing situation into the real world is a universal problem and one with important implications for treatment (Carter et al. 1998, Sparrow et al. 1984). The final diagnostic process should aim at the integration of all available information, including historical information, the child's current developmental levels, behavioral strengths, and vulnerabilities. A growing number of assessment instruments for infants and young children are now available (see Klin et al. 2004 for a review), and, as noted previously, a number of instruments have been developed specifically to aid in the diagnosis of autism in young children.

A large body of available evidence strongly supports the importance of intensive educational and behavioral interventions in children with autism. The recent National Research Council (2001) report has summarized the range of programs around the country with demonstrated efficacy in the treatment of children with autism. Although there are some differences in these programs, there are also many areas of similarity. Areas of difference include aspects of theoretical orientation, the degree to which it is the child or the adult who sets the learning/teaching agenda, and the degree to which the curriculum is guided by developmental principles. Areas of general agreement include the use of highly structured approaches to teaching, intensive involvement with the child, and a general focus on "learning to learn" challenges, i.e., basic abilities to participate in and benefit from instruction (Nat'l. Res. Coun. 2001).

Applied Behavior Analysis (ABA) has been the most widely studied treatment method, although there has been an increasing move to more eclectic models drawing from a range of other effective methods as well (Nat'l. Res. Counc. 2001, Smith 2000). Some attempts have been made to tailor treatments to presumed "core" deficits (Kasari 2002); differential response to treatment has now been included in some newer neuropsychological models of the condition (Mundy 2003). Some reasonably well-designed controlled treatment studies have now appeared (Drew et al. 2002, Eikeseth et al. 2002). However, even for well-established treatments like ABA, the results of recent randomized control trials (Smith et al. 2000) have not been as dramatic as those initially reported (Lovass & Smith 1988); it sometimes proves difficult for families to maintain the intensity recommended by some intervention programs. There is some suggestion that the age at which treatment begins may be an important factor (Harris & Handleman 2000). Available treatment studies have generally focused on preschool or school-age children, and few studies have directly addressed the issues of intervention in infancy. This problem will become more critical as early diagnosis improves (Volkmar et al. 2004).

Various factors appear to be central in successful intervention programs (Howlin 2000). Children with less classical autism may respond better than those with more strictly defined autism, and children with better cognitive abilities or higher levels of engagement may respond more positively (see Drew et al. 2002, Kasari et al. 2001, Koegel et al. 1999, Rogers 2000, Siller & Sigman 2002, Volkmar et al. 2004c, Whalen & Schreibman 2003, Wolery & Garfinkle 2002). For all treatments, generalization of skills learned across settings is critical (Hwang & Hughes 2000, Strain & Hoyson 2000). Unfortunately, even though earlier detection and intensive intervention have led to an overall improvement in outcome, some children fail to do well even with excellent intervention; the study of such children may help clarify how treatments can be more individually tailored to the child. Although there has been a growing interest in teaching approaches for social skills, much of this literature has focused on somewhat older children. In addition, much of the information on effective treatments has emerged through single-subject designs. There is a critical need for research that addresses issues of treatment mechanisms and moderators as well as individual differences in response to treatment(s) (Paul 2003).

SUMMARY AND FUTURE DIRECTIONS FOR RESEARCH

Although autism was first described 60 years ago as a disorder with onset in infancy, our knowledge about infants with the condition remains limited. Until recently most of the available information was derived from retrospective reports or videotapes. Knowledge of the strong genetic basis of autism, heightened media and public attention, and an awareness of the importance of early diagnosis and treatment have focused increased attention on autism in the first months of life. Research findings to date have supported and refined Kanner's original (1943)

hypothesis that the problem is indeed fundamentally a social one, with studies consistently revealing early deficits in joint attention, gaze and eye contact, and early emerging social interest and routines. These difficulties appear to then have important implications for emerging skills in other areas.

To date a major limitation of the research on infants with autism has been the reliance on retrospective approaches. The development of better screening methods and the study of high-risk populations, e.g., siblings of those with autism, now provides us with the opportunity to design prospective studies. The results of such studies will further help us refine current theories of both the psychological and neurobiological basis of autism and to address issues of the broader spectrum of autism-related disorders as well. Results will have important implications for understanding the role of social factors in other aspects of development and the potential for allowing us to develop even better behaviorally and biologically based approaches to screening and early diagnosis. A focus on specific processes is critically needed, e.g., how deficits in joint attention become entwined with other attentional, communicative, and cognitive processes (Volkmar et al. 2004b). Advances in assessment and methodology may help extend work from older and more able individuals to the first years, if not the first months, of life. The increasing numbers of infants and very young children identified as at risk will present important new challenges for service delivery and research on early intervention.

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