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AUTISTIC SPECTRUM DISORDERS: CAUSAL MECHANISMS AND RECENT FINDINGS ON ATTENTION AND EMOTION

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This paper provides an overview of selective research on autism. Autism forms part of a spectrum of related developmental disorders that vary in severity. Both their prevalence and severity argue for concerted efforts aimed at improving our understanding and treatment of the many individuals affected. We begin by outlining an important discovery that implicates an early prenatal insult to the developing brain stem in at least some people with autism (hereafter, the thalidomide discovery*; Miller & Stromland, 1993). Several lines of evidence consistent with this claim are summarized. We then turn to recent research on early developing mechanisms of attention and emotion in autism. Evidence to be reviewed points to impairment in the disengage function of visual attention, and data are provided on the relationship between disengagement and the regulation of emotional states. Research on emotion focuses on the hypothesis, derived from the thalidomide discovery, that there may be a physical/anatomical basis to the lack of facial expressiveness in autism. We end by discussing the implications of this work for future research and for supporting children and adults with autism.*

Autism is the most extreme form of a spectrum of related disorders (known as the

pervasive developmental disorders; PDDs). These disorders are defined behaviourally, by impairments in communication and socialization and by marked inflexibility in behaviour (APA, 1994). Perhaps most importantly, there is a striking failure to engage in reciprocal social communication, whether through using the eyes, facial expression, gestures or language. Regardless of measured intelligence, which can vary from severely mentally handicapped to superior, people with autism lack what is referred to as social or practical intelligence (Sternberg & Grigorenko, 2000). They are socially naïve, they have difficulty *reading* others, and, despite a desire for social contact, they lack social know-how. There is, in addition, a more general deficit in every day problem-solving skills.

Autism occurs with a frequency of 1-2 per 1,000 (e.g., Bryson, Smith & Clark, 1988), although prevalence of the entire spectrum of autistic disorders is substantially higher. Recent estimates indicate that 5-6 per 1,000 has some form of autism (including autistic disorder, Asperger syndrome and atypical autism or PDD-NOS; Bryson, 1997; Chakrabarti & Fombonne, 2001; Gillberg & Wing, 1999). These estimates are considerably higher than those for Down syndrome (1/1,000; Alberman, Mutton, Nicholson & Bowbrow, 1995), fetal alcohol syndrome (0.5-2.0/1,000; May & Gossage, 2001) or fragile-X (0.25-0.9/1,000 for males and 0.2-0.6/1,000 for females; Webb, 1991), thus placing autism as the most frequent of the severe disorders of development. Indeed, considerable debate exists about whether the prevalence of autism is increasing, or whether changes in diagnostic practices (i.e., a broadening of the diagnostic category to include less severe variants of autism) account for the overwhelming increase in the number of children coming to clinical attention. In either case, both the frequency and severity of autism argue for concerted efforts aimed at improving our understanding and treatment of the many individuals affected.

This paper has two main purposes: first, to outline important advances in our understanding of causal and neuropathological mechanisms in autism, and second, to describe recent findings on attention and emotion in autism. Research to be discussed implicates an early prenatal insult to the developing brain stem in at least some people with autism. After briefly outlining evidence for this claim, we turn to empirical findings that point to impairment in early developing mechanisms of attention and emotion in autism. Discussion focuses on the problem of orienting or moving attention in visual space, on the relationship between visual-spatial attention and the regulation of emotion and on the expression of facial emotion. Finally, we consider the implications of this work for future research and for supporting children and adults with autism.

Causal and Neuropathological Mechanisms

To date, the causes of autism remain largely unknown. Recently, however, a very important discovery was made (hereafter, the *thalidomide discovery*). Briefly, thanks to the thoughtful work of two ophthalmologists (Miller & Stromland, 1993; Stromland, Nordin, Miller, Akerstrom & Gillberg, 1994), a relationship has been established between prenatal exposure to thalidomide, a particular set of physical anomalies and autism: risk for autism is dramatically increased in individuals whose thalidomide exposure resulted

in ear anomalies, a lack of lateral eye movement and facial paralysis. Indeed, 33 percent with this set of anomalies developed autism, a rate well beyond that expected by chance. Two of these anomalies, a lack of lateral eye movement and facial paralysis, result from neurological impairment to cranial nerves VI and VII, and are diagnostic of Moebius syndrome (Kumar, 1990). This pattern of physical and neurological anomalies, including the overlap with Moebius syndrome, implicates an early prenatal insult to the brain stem (Rodier, Ingram, Tisdale, Nelson & Romano, 1996). That is, in at least some people with autism development has been disrupted during the initial stages of brain development (i.e., the period of neural tube closure or thereabouts).

This claim has since been explored in an innovative program of research spearheaded by Patricia Rodier (Rodier, 2000; Rodier et al., 1996). Among the several lines of existing support, an autopsy of a child with autism revealed a dramatic reduction in neuron numbers in the region of the brain stem where the 7th cranial (motor) nerve develops (which incidentally lays adjacent to the sensory neurons, disruption of which may underlie the unusual sensory responses in autism). Development of an animal model of autism resulted in parallel findings: a relative decrease in the number of motor neurons was found following prenatal exposure to a drug (valproate acid) during the same period of prenatal development implicated in the thalidomide cases. Similarly, data from an entire population with autism indicate that an anomaly involving the ear (posterior rotation, evident in 45% of cases) distinguished autism from both typical development and non-autistic developmental disorders (Rodier, Bryson & Welsh, 1997). Finally, preliminary evidence implicates developmental or Hox genes that control development of the brain stem in the etiology of at least some cases with autism (i.e., those from multiple incidence families; Ingram, Stodgell, Hyman, Figlewicz, Weitkamp & Rodier, 2000). The working hypothesis is that genetic and/or environmental (e.g., toxins such as thalidomide or valproate acid) factors increase risk for autism, and that the particular factors or combination thereof will vary across families.

Visual-Spatial Attention

Broadly speaking, there are two forms of attention—focused/sustained attention, as required in recognizing objects or patterns, and visual-spatial attention, or the ability to move attention fluidly through visual space. Focused/sustained attention has been researched extensively, in part because it has been implicated in attention deficit disorder (with or without hyperactivity; ADHD/ADD): children with ADD/ADHD have difficulty focusing and sustaining attention, as required, for example, in detecting an object or figure embedded among several others (Campbell, 1973). In contrast, people with autism are actually superior to typical controls at detecting embedded figures (Shah & Frith, 1983; also see O'Riordan, Plaisted, Driver & Baron-Cohen, 2001, for evidence of superior feature detection). Such findings are consistent with evidence of overly focused attention in autism (Rincover & Ducharme, 1987), and with anecdotal reports of good pattern recognition (e.g., strong matching skills and hyper-sensitivity to disruptions or changes in pattern).

Visual-spatial attention is a more basic form of attention that develops earlier and is

mediated by different neural structures. It is defined by three operations--the ability to disengage attention from an object in one location, and to shift and re-engage attention on a new object in a different location. This form of attention, and specifically the disengage operation, is normally operative by at 3-4 months of age (Johnson, Posner & Rothbart, 1991; Hood & Atkinson, 1993; McConnell & Bryson, submitted). Prior to this, typically developing infants are able to engage attention, but their attention has been described as *obligatory* or *sticky*: they have difficulty disengaging from, and may remain stuck on, one of two competing stimuli. Visual-spatial attention is of particular interest because of its role not only in cognitive development but also in the regulation of emotional states (Rothbart, Ziaie & O'Boyle, 1992). Probably the most basic way we deal with emotionally upsetting events or thoughts is by disengaging attention or distracting ourselves from the source of the upset.

Over the past several years, we have been studying visual-spatial attention in autism. Our operating assumption is that impairment in the disengage operation might underlie such diverse phenomenon as not orienting to name, repeating the same motor movement or speech sounds, being preoccupied with certain stimuli or thoughts, and having difficulty moving from one activity to another. In our first set of experiments, preliminary evidence was provided for difficulties disengaging and/or shifting attention, which were particularly marked when moving attention from the right to the left side of space (Wainwright-Sharp & Bryson, 1993; 1996). However, we were unable to distinguish whether the problem was one of disengagement, shifting, or both. More recently, we have been using a simple visual orienting paradigm that provides independent measures of the two operations. Briefly, the children are seated in front of three computer screens positioned side-by-side. Once the child orients to a central fixation stimulus, a second stimulus appears in one of the two lateral screens, and the time taken to begin an eye movement to the lateral stimulus is measured. The critical manipulation is whether, upon presentation of the lateral stimulus the central stimulus remains on (requiring that the child disengage and shift) or is turned off (requiring a shift alone).

Our main finding is that relative to matched typical children and children with Down Syndrome, children with autism have marked difficulty disengaging visual attention (Landry & Bryson, in press; also see evidence for more subtle difficulties executing fast shifts of attention). Indeed, on at least 20 percent of trials they remain stuck on one of two competing stimuli for the entire 8-second trial duration. These findings for 4- to 7-year-old children with autism parallel those reported for typically-developing 2-month-olds (Hood & Atkinson, 1993; Johnson et al., 1991; McConnell & Bryson, submitted), thus underscoring the significance of the impairment. Moreover, we find no relationship between difficulty disengaging and either nonverbal intelligence or receptive language level. The problem appears to exist in virtually all children with autism, including those of average or above average measured intelligence. We have since replicated and extended these findings with a larger and more diverse group with autism (n=32; age range=3-12 years), showing that the disengage problem exists in the vertical as well the horizontal plane (Rombough & Bryson, submitted). We also provide evidence for a right-downward orienting bias: children with autism have particular difficulty disengaging and

shifting attention to the left and upwards in space. These findings are consistent with reports of *tunnel vision*, or overly focused attention in autism (Rincover & Ducharme, 1987). Support is also provided for our claim that people with autism suffer a form of developmental spatial neglect, that is, they lack awareness of information in large areas of space (Bryson, Wainwright-Sharp & Smith, 1990).

One additional finding bears emphasizing. In the course of coding eye movement data from the visual orienting task, one of us (Landry, 1998) observed that the children with autism were showing self-regulatory behaviours typical of infants. Following the lead of Rothbart et al. (1992), these were categorized into approach and avoidance or distress behaviours. Analysis of the data revealed a strong relationship between difficulty disengaging and the presence of distress behaviours. Children with autism showed a preponderance of distress behaviours such as rapid and shallow breathing, excessive mouthing and gaze avoidance. In contrast, children with Down syndrome, who had no difficulty disengaging, showed a preponderance of approach behaviours such as smiling and leaning forward towards the stimuli. In typical infants, development of the disengage operation is associated with infants being more soothe able (Johnson et al., 1991), and with increased smiling and less distress to limitations (McConnell & Bryson submitted), as reported by parents. Support is thus provided for the claim that the disengage function of visual attention serves to regulate emotional states (Rothbart et al., 1992). Direct experience would suggest that in individuals with autism the challenge is to distract them from upsetting events or thoughts before their emotional reactions become extreme, and to teach them strategies for detecting and regulating their own emotional states.

Facial Emotion

A lack of facial expressiveness is well documented in autism (e.g., Snow, Hertzig & Shapiro, 1987; Yirmiya, Kasari, Sigman & Mundy, 1989). This has been viewed as part of the nonverbal communicative problem, and is generally attributed to impairment in the development of mentalistic processes such as the capacity for shared affect, inter-subjectivity or inter-personal relatedness (Hobson, 1993). While we do not challenge the veracity of such interpretations, we do note that they are descriptive rather than explanatory. The overlap between Moebius syndrome and autism suggests a physical/anatomical basis for the lack of facial expressiveness in autism. Recall that Moebius syndrome is defined by 6^{th} and 7^{th} cranial nerve dysfunction, resulting in a lack of lateral eye movement and facial paralysis; the children are unable to smile or express other facial emotion, and their faces lack the communicative intent normally expressed in back-and-forth eye movements. One outstanding possibility is that diminished and/or faulty innervation of the face may contribute to the lack of facial emotion in autism.

We explored this possibility by conducting a microanalysis of the facial muscle movements of young children with autism (Czapinski & Bryson, in press). The children were engaged in semi-structured play, which, through the use of high-interest toys and activities, was designed to optimize the expression of positive emotion. The entire session (about 30 minutes, following a warm-up period) was videotaped, and facial muscle movements were coded from videotapes using a standard, well-researched method (the

Maximally Discriminative Facial Movement Coding System; Izard, 1979). In this system, individual muscle movements from the three facial regions (mouth, eyes and brow) are coded independently, and codes are provided for translating patterns of movements into different emotions. However, our focus was on the muscle movements themselves (vs. the emotion represented alone).

Our findings on expressed emotion replicate those reported earlier (e.g., Snow et al., 1987). Briefly, relative to both matched children with language disorder and a typically developing group, children with autism expressed less facial emotion, and were less likely to direct their emotion at others (vs., e.g., an object of interest; Czapinski, 2000). However, equally striking was the marked individual variation in facial muscle movements among the children with autism. Analysis of their discrete muscle movements revealed three main findings. First, there was significantly reduced movement in the mouth and eye (vs. the brow) regions in autism, although this varied across children (Czapinski & Bryson, in press). Some had particularly reduced movement in the mouth region, others in the eye region, and still others in both regions. The movements were less frequent, weaker and of briefer duration than those observed in either the language-disordered group or the typical children. Children with autism were further distinguished by a significant proportion of atypical muscle movements not observed in the other two groups, nor documented in Izard's system, thus underscoring their uniqueness. These included a preponderance of asymmetrical movements such as rising of one eyebrow, typically the right, as well as unusual combinations of facial muscle movements. Finally, we observed ptosis or droopy eyelids in a subset of children (at least 3/15). One child actually tilted his head backwards in order to see objects handed to him.

While we recognize that our findings are not definitive, taken together, they are consistent with the possibility that some disruption in the neuromuscular pathway contributes to the lack of facial expressiveness in autism. Diminished and/or faulty innervation of the face may, at a minimum, make it more difficult for children with autism not only to move individual facial muscles but also to form the particular combinations of muscle movements required to express different emotions. This might explain, for example, why the children are more able to do so when physically aroused (e.g., when tickled or on a swing). In any event, we do emphasize that a lack of facial expressiveness does not imply a lack of emotion. Indeed, this was brought home to us when coding data from the visual orienting task. Despite the lack of recognizable facial emotion, upon closer analysis it was clear that the children with autism were expressing a great deal of emotion, notably distress, but also positive emotion, although in ways characteristic of much younger children (i.e., typical 12-month-olds). We also emphasize that it is well established that we mirror the emotion expressed by others. The main implication for children with autism is that, by virtue of their lack of facial expressiveness, they are less likely to see and experience the facial emotion of others. It is thus critically important that we make concerted efforts to compensate for their non-expressiveness by ensuring that we are facially expressive in our interactions with them.

Summary and Research and Clinical Implications

Autism forms part of a spectrum of related disorders (including Asperger syndrome, and atypical autism or pervasive developmental disorder not otherwise specified) that vary in severity. Increasing recognition of the autistic spectrum has had profound implications for estimates of prevalence. Autism occurs with a frequency of 1-2 per 1,000, but prevalence of the entire spectrum is much higher, estimated at 5-6 per 1,000 (Bryson, 1997; Chakrabarti & Fombonne, 2001; Gillberg & Wing, 1999). Clinical services are currently overwhelmed with the large numbers of children coming to attention. Recent research implicates an early prenatal insult to the developing brain stem in at least some people with autism (Miller & Stromland, 1993; Rodier et al., 1996). There is also evidence of impairment in the ability to disengage visual attention, a function that normally develops by 3-4 months of age. One question we are currently pursuing is whether impaired disengagement might serve as an early marker of autism. It will also be important to identify the neural mechanisms that underlie impaired disengagement in autism, and to evaluate the efficacy of treatments, both behavioural and psychopharmacological, that might alleviate the disengage problem. Our experience suggests that certain forms of touch and motor behaviors such as pointing are good candidates for systematic study. By the same token, it would appear that active (i.e. motoric) engagement in an alternate activity is a particularly effective way of distracting individuals with autism. Finally, preliminary evidence implicates a physical/anatomical basis for the lack of facial expressiveness in autism. Specifically, some disruption in the facial neuromuscular pathway might contribute not only to the lack of facial emotion but also to the feeding and speech problems that frequently characterize autism. Future research might explore this hypothesis further by more directly evaluating the integrity of facial innervation in individuals with autism. It also remains possible that stimulation of the facial muscles might enhance the production of spontaneous facial expressions. It is to be hoped that advances in understanding autism will allow us to more effectively intervene and optimize the potential of the many individuals affected.

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