Introduction

Autism is a disorder originating in early childhood that has extremely debilitating effects on social and communicative development.1-5 Because the causes of autism have not yet been identified, it cannot be prevented, nor do we have a cure or even particularly effective treatments. The symptoms of autism are quite heterogeneous but must involve deficits in social relatedness and communication, as well as the presence of restricted interests and repetitive behaviours.6

Subject

The investigation of autism is important because of its very serious consequences for life adjustment. Moreover, because autism involves deficits in social abilities that are manifested very early in typical development, the disorder serves as a model for understanding the prerequisites for communicative and emotional interactions.

Problems
Individuals with autism invariably have difficulties in social engagement and the acquisition of communicative skills. However, the ways in which these difficulties are manifested vary widely from person to person. Many individuals with autism do not develop functional language skills\textsuperscript{7,8} and show deficiencies in non-verbal communication.\textsuperscript{9} Those who do acquire language have problems in communicating because effective communication requires the ability to share perspectives with others, a major deficit in autism.\textsuperscript{10} The rate of mental retardation (MR) in autism has long been cited as approximately 75%, and a current review paper summarizing surveys with IQ data gave a median of 70%. However, recent studies have reported lower estimates, largely because of the inclusion of individuals with Pervasive Developmental Disabilities (PDD) and Asperger’s syndrome, who typically have lower rates of MR.\textsuperscript{11}

**Research Contexts**

Because autism was discovered relatively recently, there has been a great deal of change in the contexts in which autism is researched. Only in the last five to 10 years has there been sufficient consensus over the core symptoms of autism to enable reliable and valid diagnostic instruments to be created, a major research improvement. The demonstration by epidemiological studies of a greater prevalence than previously thought has also altered the research context.

**Key Research Questions**

One key research question concerns the psychological and physiological manifestations of autism and the factors responsible for these characteristics. Psychological theories of autism focus on problems in social and symbolic understanding,\textsuperscript{12,13} executive functions including attention shifting,\textsuperscript{14,15} and central coherence.\textsuperscript{16} Physiological manifestations are assessed with measures of head circumference, structural and functional imaging techniques (this can include magnetic resonance imaging [MRI], positron emission tomography [PET], and magneto encephalography [MEG]), electroencephalography, evoked potentials, and post-mortem anatomical studies of brain tissue. Most studies compare psychological and physiological characteristics of individuals with autism with the characteristics of non-autistic individuals of similar age, gender and frequently, developmental or language level. More recent studies have addressed the genetic basis for these characteristics as well as for the diagnosis itself.

A second research aim is to develop early identification measures for the syndrome of autism. The diagnosis of autism is now generally not made before a child reaches 30 months of age.
because of instability in diagnoses made before that age period. Early diagnosis studies use three approaches: the coding of behaviours from birthday party videotapes of children who are later diagnosed with autism;\textsuperscript{17} the assessment of two- to three-year-old children who have behaviours characteristic of autism; and the longitudinal follow-up of siblings of children with autism, who are at higher risk than siblings of non-autistic children.\textsuperscript{18}

Investigations of the effectiveness of interventions with children with autism have been increasing over the last few years. This is fortunate because, until this time, there have been very few randomized control studies of interventions with children with autism.\textsuperscript{19,20} Current studies improve on previous investigations in that the aims of the interventions are often based on theoretical considerations or empirical evidence of areas of deficits or environmental mediators of developmental progress. In the past, since less was known about autism specifically, interventions were less tailored to the problems of children with autism or characteristics of the children or their environments that are associated with improved development.

**Recent Research Results**

There is research evidence supporting most of the psychological theories of autism, but no single theory that explains all the symptoms and developmental aspects of the disorder. Many studies replicate the identification of specific and unique deficits in social and symbolic domains that are manifested as early as the second year of life. However, it is possible that these deficits arise from earlier problems in attention regulation or in social orientation and engagement. Moreover, deficits in social communication and play in early childhood do not explain the presence of repetitive behaviours and obsessive concerns or problems in decision-making that mar the functioning of more highly able individuals with autism. Theories of executive function and central coherence deficits explain these latter problems more adequately, but are limited in their application to problems with social communication and play.

While psychological studies have been quite successful in terms of replication, this has been less true for findings of differences in brain structure and/or function. Perhaps the strongest evidence is for a neurodevelopmental disorder in brain growth, in that the most common physical abnormality found in children with autism is large head size.

Genetic transmission is the most broadly supported causal mechanism. The higher rate of autism in monozygotic as opposed to dizygotic twins, as well as the increased sibling recurrence risk and
the presence of the broad phenotype in family members support this theory. A number of regions on different chromosomes have been identified in various studies and a variety of candidate genes are being investigated. Unfortunately, there has been only limited replication to date. Recently, the description of specific subgroups with similar physical or behavioural profiles (endophenotypes) has been shown to increase the power to detect linkage for autism susceptibility gene regions. Research on environmental teratogens (stimuli that disrupt normal development by damaging cells, altering chromosomes structure, or acting as abnormal inducers) that might contribute to autism is just beginning.

Conclusions

Autism is a developmental disorder of unknown origin that severely compromises the social development of children whose behaviour meets the diagnostic criteria. Most individuals with autism are sufficiently socially isolated and intellectually impaired that they have difficulties with employment and do not marry or have children. Research is needed that addresses the causes of the disorder as well as the causes of variations in its manifestations. Such research is likely to lead to more effective interventions that are created with two aims: 1) where possible, to treat and even prevent the disorder; and 2) to optimize the developmental progress and level of functioning of individuals with the disorder.

Implications for the Policy and Services Perspective

There is a great need for therapeutic and educational services for individuals with autism. School systems and social services are largely unable to provide the resources that are required to educate and treat the problems faced by people with autism at all age levels. At the same time, families are over-burdened by the challenges involved in rearing and supporting family members with autism. Currently, there is an emphasis on early detection so that interventions can be implemented that prevent the development of problems secondary to the social deprivation often elicited by the children’s social difficulties. At the same time, we need many more programs designed to help older individuals with autism to adapt to their life circumstances.

References


