ENCYCLOPEDIA
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[Archived] Autism and Its Impact on Child Development

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Introduction

Autism and related pervasive developmental disorders are more common than previously recognized, affecting up to one in 100 individuals.[1] Although symptoms and accompanying functional impairments can improve with development and as a result of specific interventions, the condition is lifelong and results in considerable morbidity and cost to the individual, his or her family and society.

Subject

The term “autism spectrum disorders” is now commonly used to describe a range of neurodevelopmental conditions that differ in underlying etiology and vary in manifestation.[2] What they share in common with Kanner’s description of classically “autistic” children is a primary impairment in social relatedness and reciprocity.[3] Once considered particular characteristics of rare individuals in the population, these impairments are now more commonly understood as a broad spectrum of individual differences that are widely distributed in the general population.[4]

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Problems

The primary impairment in social reciprocity, peer relationships and emotional engagement is accompanied in different individuals by varying degrees of language and communication impairment, mental retardation and patterns of non-functional restricted, repetitive and stereotyped behaviours. Sensory abnormalities, including either hyposensitivity or hypersensitivity, and unusual interests in some sensations are common. A lack of imaginative play indicates an underlying difficulty with generation of ideas that is highly relevant in the development of understanding, and thinking about, other people and other situations. All of these characteristics can be seen in varying degrees of severity.

Both the DSM-IV[5] and the ICD-10[6] classification systems include diagnostic categories for individuals who show some but not the full complement of impairments necessary to meet criteria for autism: Asperger's syndrome, atypical autism and pervasive developmental disorder ("unspecified" in ICD-10; "not otherwise specified" in DSM-IV). This group is a mix of those with greater severity in one area than others, mild impairments in several areas or late onset (after three years of age).

Research Context

While it has been established that autism is an organic condition and that the condition is highly heritable, in only a minority of cases can a specific neurologic or genetic cause be identified.[2] As a result, considerable research activity is currently focused on identifying the genetic and neuro-pathological basis. However, as the autism phenotype may be arrived at by a number of pathogenic routes that overlap somewhere in brain development and function, as yet unidentified biological or genetic markers will not necessarily be present in all cases and case definition will continue to be reliant on the behavioural picture alone. Targets for behavioural research include establishing the neuropsychological processes that are impaired, earlier identification and establishment of accurate and reliable diagnosis in toddlers, and testing the efficacy and effectiveness of various intervention programs and approaches.

Key Research Questions and Recent Research Findings

Although etiology can be established in a few cases (e.g. children with fragile-X syndrome or tuberous sclerosis), there is evidence for complex polygenic inheritance. However, while attempts to identify susceptibility genes by association studies have produced several candidate

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genes on different chromosomes, no gene has yet been identified.[7] The recurrence risk for subsequently born siblings is approximately 5-10%, although milder impairments of social communication skills or language are found in as many as 20% of relatives. Familial susceptibility to the “broader phenotype” of autism has implications for genetic counselling. Autism is more common in boys than girls (4:1), but no explanation for this discrepancy has been substantiated.[8] Between 15% and 30% of children with autism experience a period of stasis or regression, most commonly in speech and social behaviour, usually between 12 and 20 months of age, although the causes of this regression are not well understood.[9]

There is agreement that social processing (of faces, emotions, mentalizing skills) is impaired and there is evidence that the brain systems that subserve such cognitive functions are structurally and functionally disrupted. However, the developmental cause of such impairments may be earlier disruptions in the development of brain circuits that underlie the social reward and social orienting systems.[10,11]

There has been progress in the earlier identification and diagnosis of cases, in part via efforts to develop prospective screening instruments[12] and the prospective study of “high- risk” samples, such as younger siblings of already diagnosed children.[13] However, this presents clinical challenges: establishing the reliability of early diagnosis, modification of treatment approaches for toddlers, use of assessment tools with younger children and the ability to indicate prognosis.[14]

There is some evidence for effectiveness of intensive applied behavioural analysis approaches to early intervention, but there are also limitations with respect to outcomes and the generalization of cue-dependent behaviours.[15] There is also evidence for benefits from social and communication-based approaches[16] and approaches that provide visual cues and structure that many preschoolers with autism find difficult to generate themselves[17]. Important elements of intervention programs for preschool children with autism include a focus on the development of pragmatic and functional communication skills (whether verbal or non-verbal), joint engagement and joint social activities, promotion of emotional engagement and regulation, and helping parents to manage behavioural tantrums and maladaptive routines.[15] For the most treatment-resistant children with the poorest prognosis (e.g. those with no verbal communication by mid-school years, accompanied by extreme social aloofness and mental retardation), there is a need to determine whether augmentative communication approaches might help adaptation.

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Depending on family resources and access to support and services, the impact on the family can be considerable, especially at times of important transition (diagnosis, school entry, school transfer, entry to adulthood). Research into the effectiveness and acceptability of support services for families and for adults with autism is scant. One emerging trend is the identification of comorbid psychopathology (e.g. anxiety, OCD) in adolescents and adults, most notable in individuals with average IQs, that can lead to additional challenging behaviour.

Conclusions

Our understanding of autism, once considered a rare and almost always severe childhood disorder, has undergone a revolution in the past 20 years. It is not a rare disorder. Its manifestations can vary widely and it can present in high- as well as in low-IQ individuals. Together with increasing evidence of the positive benefits of early intervention, these changes in conceptualization and application of the diagnosis mean that our notions of outcome and likely progress are also undergoing a revolution. Alongside advances in genetic and neuro-scientific research, a reconceptualization of autism has led scientists to ask fundamental questions regarding social behaviour and communication that have relevance for populations of children in general and not just those relatively rare children who have impairments in these abilities sufficient to meet diagnostic criteria for autism.

Policy Implications

The costs of autism to individuals, families and society are considerable. Internationally, there is an impetus to improve early identification and treatment in order to minimize the impact and to ameliorate the negative secondary sequelae of late diagnosis and ineffective treatments. Community health practitioners and kindergarten staff require training in the identification and management of autism. Basic research into the etiology and underlying psychological impairments that characterize autism are required alongside more applied research into early identification, effective interventions and support for families. At a broader societal level, the recognition that aspects of autism are connected more generally to individual differences in social behaviour (for example, between males and females[18]) challenges the notion of autism as a distinct and necessarily “impaired” way of processing and understanding the social world. This calls for greater societal acceptance of differences in social engagement and social behaviour.

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