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Guest Editorial

Modern Views of Autism

Eric Fombonne, MD, FRCPsych¹

In a seminal paper describing the first 11 cases of autism, Kanner pointed to the innate disturbance of affective contact occurring in the infancy of these children and to unusual personality traits in their parents (1). These observations could have indicated genetic mechanisms underlying the syndrome; however, the predominance of psychoanalytical theories and the particular focus on maternal deprivation in post-World War II child psychiatry led to misconceptions of autism as an infant's response to early disturbances of the mother-child relationship (termed the "refrigerator mother"). The word "autism" was first used by Bleuler to index a cardinal sign of schizophrenia, and its use to describe the syndrome unfortunately led to 30 years of controversy about the validity of autism as a distinct syndrome vis-à-vis adult psychoses. The subsequent period of confused terminology (for example, "infantile schizophrenia," "early childhood psychosis," and "sybiotic psychosis") largely reflected untested psychoanalytical models and prevailed up to the late 1960s.

Then, systematic empirical investigations progressively helped to establish the validity of autism as a separate syndrome. First, the syndrome's strong association with seizures and mental retardation was recognized, as were unusual psychometric profiles differing from those seen among individuals with mental retardation but without autism, and it became obvious that autism is a disorder of brain development. Second, the first adult outcome studies showed that autism is a lifelong handicap and that, despite important gains

made by some individuals, long-term (especially social) deficits persist, even in the minority of subjects (10%) who achieve the best outcomes (2). Third, comparative studies showed that autism is separate from childhood schizophrenia and differs from it in response to treatment, in family history, in longitudinal outcomes, and in a range of associated clinical features (for example, association with mental retardation, sex ratio, and core symptoms) (3). Similar studies also helped to differentiate autism from language disorders and other developmental disorders. Autism appeared to involve communication impairments rather than language impairments only. Moreover, concurrent social and behavioural deficits could not be explained solely on the basis of language and communication problems. Fourth, controlled studies of parenting styles and behaviours in families with children suffering from autism indicated that parents of such children were no different from other parents (4). It also became increasingly evident that autism differed from syndromes seen in children raised in institutions or suffering from maternal deprivation (that is, attachment disorders). Unlike these other syndromes, autism was observed to be remarkably stable across rearing contexts, and no evidence ever emerged to document benefits from the sad practice of removing children from their families to provide them with "compensatory" therapeutic milieu experiences (5). Finally, in the early 1970s, experimental studies showed that developmental gains could be achieved in children with autism when they were educated with active, rather than passive, techniques; when the home and classroom environments were structured to capitalize on their strengths and compensate their deficits; when the educational environment offered a high teacher-pupil ratio and addressed the multiple deficits across developmental areas with individualized educational plans; and when parents acted as cotherapists to promote the generalization of learning in

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these children (6–8). The Treatment and Education of Autistic and Communication Handicapped Children (TEACCH) program was developed by Schopler in the early 1970s. This approach to autism became particularly influential worldwide and progressively replaced obsolete, unstructured, psychoanalytically based approaches to the treatment of autism (6).

Conceptual clarification and the availability of new diagnostic tools made it possible to research the causes of autism. The first British twin study, published in the late 1970s, documented substantial differences between monozygotic and dizygotic concordance rates and pointed to genetic contributions to autism. Current heritability estimates for autism are above 90%, which makes autism a strongly genetic disorder. In addition, it was observed that monozygotic cotwins discordant for autism often displayed subtle developmental abnormalities involving language, social interactions, and patterns of behaviours and interests. This suggested the family transmission not only of narrowly defined autism but also of a broader phenotype comprising developmental abnormalities conceptually similar to those seen in autism but associated with milder impairments. This broad phenotype of autism was subsequently validated in various family studies and is still the object of much research. The concept of a lesser variant of autism, together with the recognition of autism in children with normal intelligence (that is, high-functioning autism and Asperger disorder), progressively shifted our conceptualization toward a more dimensional view of autism spectrum disorders. In the mid-1990s, strong evidence for genetic factors in autism, the development of reliable and valid diagnostic tools, and technological progress in molecular genetics led to molecular genetic studies of autism. Because there are few consistently identified biological abnormalities in autism, and consequently few candidate genes, most investigators have relied on affected-relative pairs designs. Several groups are now actively working to identify genes involved in liability to autism. The first molecular genetic findings were published in 1998 (9); since then, published results from several molecular genetic investigations point to particular areas on some chromosomes (especially chromosomes 2 and 7) as likely to harbour autism susceptibility genes. World laboratories are currently working night and day on these investigations.

Epidemiologic research has also accelerated in the last 15 years (10), and new surveys focusing on a broader case definition of pervasive developmental disorder (PDD), of which autism is only a single form, indicate that prevalence estimates are much higher than previously thought (60/10 000; see 11). Debate continues as to whether the evidence indicates that the incidence of autism has truly increased over time, once other explanations for increased prevalence estimates are controlled for (for example, when the broadened concept and diagnostic criteria, as well as improved case identification, are

considered) (12). Increased incidence would be consistent with environmental risk factors having an etiologic role, either acting alone or interacting with susceptibility genes. However, no single environmental risk factor has yet been shown to substantially increase the risk of autism, despite claims that the measles virus included in the measles, mumps, and rubella (MMR) immunization given to children at the beginning of their second year may be involved (13,14) or that the mercury (thimerosal) used to stabilize vaccine preparations may raise the risk of autism (15). Other investigations are currently exploring further the contribution of environmental risk exposures to the development of autism.

While the search for genetic and environmental causes is underway, it is obvious that research findings will not translate into practical help for some time. Therefore, it is important to maintain a strong research focus on devising and assessing interventions that promote normal development in children with autism to improve the long-term outcome for children suffering from this devastating disorder. In the late 1980s, researchers investigated the efficacy for very young children of early intensive educational programs based on applied behavioural analysis (ABA) principles. The results showed that substantial cognitive and language gains could be achieved and maintained at follow-up (16). Initial claims of a cure were most certainly exaggerated, but in the last decade, many other studies have shown similar developmental benefits from intensive early interventions that often share the same behavioural techniques, although treatment programs may be packaged in diverse ways with respect to their particular ingredients and mode of delivery. Recently released expert recommendations, based on state-of-the-art knowledge, propose a minimum of 25 hours weekly of intensive educational interventions for preschool children with autism (17). In Canada and elsewhere, many young children with a PDD unfortunately still receive much lower levels of service, in terms of both quantity and quality. Advocacy for subjects with autism has been required to influence social policies toward this developmental disorder, and efficient associations incorporating both professionals and parents have formed everywhere. One example is the Canadian Autism Intervention Research Network (CAIRN), which aims to improve services for Canadian children suffering from autism (Web site: <http://www.cairn-site.com>).

Research into the causes and treatment of autism spectrum disorders is fast expanding. In 1997, with a budget of \$42 million, the US National Institute of Child Health and Human Development (NICHD) started a 5-year international network of 10 Collaborative Programs of Excellence in Autism (CPEA) to unravel the disorder's mysteries (<http://www.nichd.nih.gov/autism/cpea.cfm>). The network resulted from a congressionally mandated conference that took place in April

1995 to identify gaps in our knowledge of autism and directions for future research (18). The CPEA network is now in its second cycle of funding. In the last year, another initiative from the National Institutes of Health (NIH) led to the funding (\$65 million over 5 years) of 8 new Studies to Advance Autism Research and Treatment (STAART) centres. These centres aim to understand underlying brain abnormalities and causes of autism and to improve prevention, early detection, diagnosis, and treatment. The Centers for Disease Control (CDC) has recently funded 7 US states to conduct epidemiologic surveys of autism as part of its Autism and Developmental Disabilities Monitoring (ADDMM) network. Five other states have just received funding to perform population surveillance and conduct etiologic research as part of the Centers of Excellence for Autism and Developmental Disabilities Research and Epidemiology (CADDRE) initiative (<http://www.cdc.gov/ncbddd/dd/aic/states/default.htm#addm>). Following the recommendations of a special review committee in the UK, the Medical Research Council and the Department of Health have earmarked research funds for autism research (http://www.mrc.ac.uk/index/public-interest/public-topical_issues/public-autism_main_section/public-autism_review.htm). Fortunately, the Canadian Institutes of Health Research (CIHR) have just funded 2 major training grants for autism research; these will significantly boost research capacity in Canada in the next decade.

Devoting an In Review series to autism is therefore timely. Autism is a lifelong condition and adult psychiatrists should be knowledgeable about its various presentations, its treatment, and its long-term outcome. The articles in this series offer state-of-the-art reviews of recent clinical and research developments in autism. Topics covered include diagnosis, comorbidity, outcome, epidemiology, genetics and

neuroimaging, early detection, intensive educational programming, and psychopharmacology. We are fortunate to have received contributions from highly distinguished autism experts. Space constraints prevented the inclusion of some areas (such as neuropsychology); however, interested readers will easily find good references on this topic.

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¹Canada Research Chair in Child Psychiatry, Professor of Psychiatry, McGill University, Montreal Children's Hospital, Montreal, Quebec.