Guest Editorial

Modern Views of Autism

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

Conceptual clarification and the availability of new diagno-
sitic tools made it possible to research the causes of autism. The
first British twin study, published in the late 1970s, docu-
mented substantial differences between monozygotic and
dizygotic concordance rates and pointed to genetic contribu-
tions 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 dis-
cordant for autism often displayed subtle developmental abnor-
malities involving language, social interactions, and patterns
of behaviours and interests. This suggested the family trans-
mission 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 conceptualiza-
tion toward a more dimensional view of autism spectrum dis-
orders. 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 chro-
mosomes (especially chromosomes 2 and 7) as likely to har-
bour 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 defini-
tion 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 con-
trolled 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 prepara-
tions may raise the risk of autism (15). Other investigations
are currently exploring further the contribution of environ-
mental 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 trans-
late into practical help for some time. Therefore, it is impor-
tant 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 bene-
fits from intensive early interventions that often share the
same behavioural techniques, although treatment programs
may be packaged in diverse ways with respect to their particu-
lar ingredients and mode of delivery. Recently released expert
recommendations, based on state-of-the-art knowledge, pro-
pose a minimum of 25 hours weekly of intensive educational
interventions for preschool children with autism (17). In Can-
da and elsewhere, many young children with a PDD unfortu-
nately 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 incorpo-
rating both professionals and parents have formed every-
where. One example is the Canadian Autism Intervention
Research Network (CAIRN), which aims to improve services
for Canadian children suffering from autism (Web site:

Research into the causes and treatment of autism spectrum
disorders is fast expanding. In 1997, with a budget of $42 mil-
lion, 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 (STAAR T) centers. These centers 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 (ADDM) network. Five other states have just received funding to perform population surveillance and conduct etiologic research as part of the Centers for Excellence for Autism and Developmental Disabilities Research and Epidemiology (CADDRE) initiative (http://www.cdc.gov/ncbddd/dd/aic/states/default.htm#admm). 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.

References


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