ARTICLE

Autistic Disorder

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History

The word autismus was coined by the psychiatrist Eugen Bleuler in 1912 as he was defining symptoms of schizophrenia. The word autism first took its modern sense in 1943. Leo Kanner reported on 11 child patients with striking behavioural similarities and introduced the label 'early infantile autism' in 1943. This lead in the early 1950's and 1960's to think that autism somehow be a continuum with schizophrenia as Kanner used Bleuler's term 'autism'. Some of Kanner's initial observation about the disorder were not associated with low intelligence quotient (IQ) levels, presence of 'splinter skills' and 'savant skills', have well educated and successful parents. During 1970's a neurobiological basis of the disorder was suggested following the high rates of seizure disorder associated with autism. Michael Rutter synthesized Kanner's work and for the first time these were recognized as a separate class of disorders and quoted in the third edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM III) as the pervasive developmental disorder (PDD) in 1978. The role of behaviour therapy and the use of controlled learning environments emerged as the primary treatments during 1980's and 1990's. The current cornerstone of autism therapy is 'behaviour therapy'.

Epidemiology

First epidemiological study of autism was conducted by Victor Lotter in 1966 and reported a prevalence of 4.5/10,000. A median prevalence is of 8.7/10,000. Fombonne in 2005 suggested a prevalence of 13/10,000. Both clinical and epidemiological studies report higher rates in boys with an average ratio of 3.5-4:1. Ratios varies with IQ levels and studies report a M:F ratio upto 6:1 in individuals without mental retardation whereas ratio of 1.5:1 with mental retardation. Contradictory to Kanner's view autism is clearly seen in all social class and in all countries.

Aetiology

Psychological theories:

1. Kanner's 'emotional theory' that viewed the need of intensive psychotherapy for both mother and child.

2. 'Weak central coherence' hypothesis.

3. 'Executive dysfunction' hypothesis.

4. Most current and influential cognitive hypothesis is the 'theory of mind' hypothesis.

5. 'Enactive mind' hypothesis.

Genetics:

Family and twin studies demonstrate that genes play a greater role in autism then any other neuropsychiatric disorders. 60% of monozygotic twins share a diagnosis of autism. 20-80% of first degree relative of autistic individual are affected compared to general population. Recent studies says that no single gene accounts for autism. Specific findings regarding neuroglins, shank 3, contractin associated protein 2, neurexin 1 and the growing understanding of the biology of fragile X mental retardation 1 (FMR1) gene have provided the first concrete insight into molecular and cellular pathology underlying autism, pointing to the critical role of synaptic formation and function. 20-30% of boys with fragile X syndrome meet the diagnostic criteria for autistic spectrum disorder. 1-8% of boys of idiopathic autism have been found to carry the fragile X mutation. Tuberous sclerosis, an autosomal dominant disease associated with a range of phenotype like mental retardation (MR) and seizures, is prevalent in about 0.4-2.8% cases of autism.

Environment:

In 1967 Bernard Rimland suggested that autism was caused by mercury and heavy metal toxicity to which some children have a genetic sensitivity. He recommended treating autistic children with a gluten-free, casein-free diet and mercury chelation therapy (removal of mercury from the system). Other environmental factors that can possibly play an important role in causing autism include viral or bacterial infections, vaccines and thalidomide.

Immune theory:

Maternal antibodies directed against foetus may be produce in-utero. Lymphocytes of some autistic children react with maternal antibodies which raises the possibility that embryonic and extra-embryonic tissues may be damaged during gestation. However bulk of evidence dose not support the causative association.

Neurochemistry

The most consistent finding has been that over 25% of autistic children and adolescents are

hyperserotonemic. However, after 29 years of investigation, the mechanism of hyperserotonemia has not been determined. Hyperserotonemia has been found to be familial. A hyperdopaminergic functioning of brain might explain the overactivity and stereotype movements seen in autism and it is consistant that the use of dopamine receptor antagonist is effective in reducing these symptoms.

Elevated plasma norepinephrine has also been a replicated finding and cerebrospinal fluid (CSF) opiate activity has been found to be elevated in few studies. Research in neurochemistry must continue to work in concert with other subspecialties to form a more comprehensive and theory-based approach to the neurobiological correlates of autistic disorder.

Anatomical correlates

Most data collected on the basis of postmorten findings and neuroimaging studies. No clear cut current data is specific enough to point any one theory in clear favour over another. Principle areas of involvement includes limbic system, circuit within the temporal and frontal lobes.

Post mortem studies: Decrease numbers of cerebeller purkinje cells, granular cells. Glial cell deficient indicates scarring. Contrastly volumetric enlargement of cerebellum compared to controls. Involvement of limbic system showing decrease neuronal size, decrease aborization, increase neuronal packing density over amygdala, hippocampus, septum, anterior cingulate, and mammilary body.

Imaging studies: Amygdala enlargement in autism with decrease total neuronal numbers. Functional magnetic resonance imaging (FMRI) has shown hypoactive amygdala circuits with occipito-temporal cortex, subcortical areas. FMRI show low level activities in medial frontal and ventral temporal on social task. Underactivation of fusiform gyrus on ventral surface of temporal lobe during face perception. Corpus callosum, the major fiber pathway between two hemisphere, is reduced in autism.

Single photon emission computed tomography (SPECT) demonstrates decreased brain function in the following areas - frontal lobes (Area M, para saggital section of areas 8 and 9 of Broadmann, area 10), temporal lobes (area 38, anterior temporal, and 22). There is also hypoperfusion in occipital lobes, visual association areas, both posterior parietal areas and cerebellar vermis and mesial aspects of both cerebellar lobes. Finally there is decrease of perfusion in area 24, anterior cingulate in the left hemisphere.

Childhood autism

A PDD defined by the presence of abnormal and/or impaired development that is manifested before the age of three years, and by the characteristic type of abnormal functioning in all three areas of social interaction, communication, and restricted, repetitive behaviour. The disorder occurs in boys three to four times more often than in girls.

Clinical features

1. Social deficit

Childern with autism seems to dwell in a separate world with little interest in parents and siblings.

During first year of life:

i) Impairment in reciprocal social interactions, failure to cuddle, failure to raise arm in anticipation of being picked up, lack of imitation of speech and gesture, failure to point to or show objects to others, poor eye contact.

ii) Lack of social interest, some children may stiffen in protest when cuddled by parents, may not seek parental attention, lack of social interest make them 'easy babies'.

iii) Lack of interest in domestic imitation, do not engage in common games like imitating their mother or father, inability to model adult behaviour and master them by role play, autistic children fail to develop social play with other children and often prefer solitary activities.

2. Impaired communicaton

Children with autism have impaired verbal and nonverbal communication. 30-40% of children remain without spoken language. Delay in acquisition of language are most frequent presenting complain of parents. Remarkably different pattern of speech e.g. immediate and delayed echolalia. Speech is described as monotonous, mechanical or robot-like, pronoun reversal. Lack of nonverbal communication like eye to face gaze, absence of facial expression, humour and sarcasm are a source of confusion.

3. Restricted repertoire of activities and interest

Children with autism have difficulty tolerating change and variation in routine. Peculiar kind of object attachment - hard objects then soft. Repetitive stereotype movements - toe walking, body rocking, head banging.

Associated features include cognitive impairment, abnormal response to sensory stimulus, sleep and eating problems, mood and affective disorder, self injury and aggression.

Course and prognosis

Autism is a life long disability with most individual needing significant family and community support. With early intervention long term outcome improves. 15-20% can achieve independence and self sufficiency in adulthood. During adolescence autistic children may exhibit behavioural detoriation with onset of seizure. Intervention should aim to facilitate acquisition of important adaptive skills for maximizing the potential for independence.

Positive prognostic factors are communicative speech by the age of five years, higher nonverbal intellectual levels, verbal IQ greater than 50, overall cognitive ability in normal range.

Diagnostic asssment

A diagnosis of autistic disorder is based on the clinical history, neuropsychiatric interview and observational assessment.

History of present illness focuses on characteristics typical of autism: Extent of sociability, level of language development, play, presence of stereotypes.

Past history focuses on: Mother's pregnancy, birth history, child's early development milestones, history of infection and accidents that may involve the brain.

Family history: Information about other members with diagnosis of autism, other developmental disorders, history of mood disorders in family.

Physical examination: Evidence of specific disorders associated with autism like tuberous sclerosis, fragile X and other neurological signs.

Mental status examination: Attempts should be made to engage the child in imaginative play, verbal conversation; attention should be focused on gaze avoidance, initiating social communication, sustaining conversations, problem with joint attention, and stereotype movements must be assesed.

Tools

Childhood Autism Rating Scale (CARS): A 15 item scale. Classify into mild, moderate, severe degree of impairment. Helps in deriving a descriptive summary of the extent of autistic behaviour. Should be used in conjunction with history, descriptive behaviour at home, school or community setting.

Indian Scale for Assessment of Autism (ISAA): National Institute for the Mentally Handicapped (NIMH) has developed a scale for identification and rating the severity of autism in India. This scale has 40 statements, divided under six domains - social relationship and reciprocity; emotional responsiveness; speech, language and communication; behaviour patterns; sensory aspects and cognitive component.

Electroencephalography (EEG): The incidence of EEG abnormality in autism in the absence of clinical seizure disordres ranges from 10-83% but depends on the number of recordings. Abnormality in the form of diffuse and focal spikes, paroxysmal spikes and wave pattern, multifocal spike activity and mixed discharge. Incidence of EEG abnormality is significantly higher in mentally retarded autistic individual.

Another abnormality associated with autism is the auditory P300 which represents the brain's processing of sensory stimulus which reflect abnormality in higher auditory processing and neural pathways.

Neuroimaging: Neuroimaging techniques are not routinely used in the absence of any specific indication.

Differential diagnosis: Other PDDs, childhood schizophrenia, mental retardation with behavioural problems, mixed receptive-expressive language disorders, acquired aphasia with convulsions, congenital deafness, psychosocial deprivation.

Management

Aim is to promote learning, language acquisition, communication and self help skills, reduce disruptive behaviour.

Nonpharmacological: Behavioural therapy, educational therapy, psychotherapy, family therapy.

Pharmacological: Major tranquilizers, selective serotonin reuptake inhbitors (SSRIs), naltrexone, clonidine.

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