What is autism?

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Abstract

Autism is a disorder that uniquely impacts core aspects of socialization. Since its first description by Kanner in 1943 a considerable body of knowledge has developed. It has traditionally been defined by a triad of impairments – in social interaction (autism), communication, and restricted interests & repetitive behaviors. Autism is an early onset disorder with a strong genetic basis and clear evidence of brain involvement. Progress has occurred in the areas of earlier diagnosis, provision of more effective management and in understanding the nature of the social brain difficulties in both autism and related conditions.

What is Autism?

Autistic disorder (also termed infantile autism, childhood autism, autism spectrum disorder, or Kanner's autism) was first described by Leo Kanner in 1943 as a condition marked by severe and sustained problems in social development (autism) along with unusual communication and a range of problems typically subsumed under the term resistance to change or insistence on sameness (1). The category of resistance to change includes restricted or stereotyped patterns of behavior and interest. The onset of the condition is in the first years of life, if not from birth. Although frequently associated with intellectual disability it appears that overall outcome has improved with earlier diagnosis and remediation. Although remarkably accurate in the descriptive sense, Kanner's initial report served to mislead early investigators; for example, his use of the word autism suggested some connection to schizophrenia and there was confusion on this point for some years.

Diagnosis

Kanner's initial description (1) emphasized an apparently congenital "inability to relate" to people and difficulties with change. Although he mentioned unusual communication problems, e.g., absence of spoken language or, in verbal individuals, idiosyncratic language, difficulties with pronoun use and social language, and echolalia, only subsequently have these been included as essential diagnostic features. First officially recognized as a diagnosis in DSM-III in 1980, the definition of autism has evolved over subsequent decades (2). For the last several decades (since 1994) the convergence of the American (DSM-IV) and WHO (ICD-10) definitions has witnessed an explosion of research. Both systems also recognized a number of other disorders within the overarching category to which autism was assigned (2). Recent changes in the American system in DSM-5 (3) have been somewhat controversial with an apparently more restrictive definition (2).
Early research on the condition was complicated by confusion with childhood-onset schizophrenia but several lines of work in the 1970s made it clear that autism was a distinctive disorder (4,5). Furthermore, it became clear that autism was a brain-based disorder with high risk for development of seizure disorder (6) and a very strong genetic basis (7,8).

Epidemiology

A number of epidemiological studies have been conducted. Overall the median rate of autistic disorder (strictly defined) is around 13 per 10,000 (9). The more broadly-defined autism spectrum disorder has been reported in around 1% of children in recent population-based studies (see paper on epidemiology of autism in this issue).
Issues of study design and sample selection impact identified rates, with smaller studies generally

reporting somewhat higher rates; although there has been a widespread impression that autism has increased in frequency, changes in diagnosis, increased awareness, and other factors make this unclear (9). There is a noteworthy gender difference with male predominance in autism (usually between 3 to 5 times as common in boys). Although the early impression of an association with social class has not been substantiated (10), cultural and ethnic issues have been rather uncommonly studied (11). There is a suggestion that under-diagnosis of the condition may be associated with lack of awareness or psychosocial adversity (12).

Natural History, Prognostic Factors, Outcomes

With earlier diagnosis and intervention there is some suggestion that long-term outcome has gradually improved (13), although not every child improves dramatically (14). Prognostic factors within the child include (at around age 5) the presence of verbal language and overall nonverbal abilities within the average range (13).
Before age 3 the diagnosis may be somewhat difficult to establish (15) but after that time the diagnosis is reasonably stable and generally life-long (16). By school age social interest may increase along with behavioral difficulties (17). As first noted by Kanner, in adolescence some individuals make gains while a smaller number lose skills (18). Adults are now more frequently able to be self-sufficient, with more attending college and post-secondary school programs (19), although it must be noted that research on adults with autism, particularly older adults, is sparse (20).

Etiology

Although changes in the syndrome occur over time, the major unifying theme across development is the persistence of social difficulties. Genetic and other neurobiological factors have been strongly implicated in pathogenesis and syndrome expression. The genetics of autism appear to be complex with multiple genes involved, many impacting some aspect of neural functioning (8).

Over the past decade a growing body of work has focused on brain-related aspects of social dysfunction, including lack of salience (i.e., relevance) of things like perception of biological motion or attraction to faces (21-23). Postmortem studies have revealed abnormalities in specific brain regions as well as in the cytoarchitechture of the brain (24). Although initially limited to lesion studies, developments in genetics have greatly advanced potential animal models of the condition (25). Although the focus of much interest by the lay press, evidence for environmental factors in the etiology of autism is rather limited (26).

Differential Diagnosis and Assessment

Autism can present major challenges for evaluation and treatment, given the marked range of abilities both across and within individuals; typically services from multiple service providers are needed. Autism should be differentiated from other developmental disorders such as language disorder or intellectual disability and from sensory deficits or extreme deprivation syndromes (27). In autism nonverbal skills are often more preserved than verbal skills. In language disorders it is typical for social interest and motivation to remain even in the face of communication problems. In intellectual disability social skills are usually not dramatically different from cognitive abilities, although the presence of stereotyped mannerisms in association with severe intellectual impairment is a source of confusion. Although DSM-5 has largely discarded potential subtypes of the broader spectrum of autism-related conditions, research interest will continue in the potential autism variants (2).

Various tests have been developed for purposes of screening (28) and for diagnosis (29). Providing an integrated, comprehensive view of the individual should be the goal of assessment (30).

Treatment

A large body of work now supports the use of behavioral and educational interventions (30) and a growing body of work has a strong research evidence base (2). Essentially goals of treatment aim to minimize the negative effects of autism on learning and development and to maximize the potential for more normative developmental processes (14). Some drug treatments may be helpful but, to date, these do not address core social problems (31).

Parents frequently engage in the use of alternative and complementary treatments that, by definition, lack a research basis. Parents should be helped to understand the importance of use of evidence-based treatments (30).

GP Comment.

What I learned from this paper?

1. The paper allowed me to refresh my knowledge of the prevalence/incidence and aetiology of the condition.

2. Early diagnosis being the key to improved outcomes, it is essential to have an index of suspicion if children fit the triad of symptomatology.

3. Educational and behavioural interventions rather than medication leads to improved outcomes.

4. Are we as modern clinicians over-diagnosing and over-medicating?

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References

1. Kanner L. Autistic disturbances of affective contact. Nervous Child. 1943;2:217-50.
2. Volkmar FR, Reichow B, Westphal A, Mandell DS. Autism and the Autism Spectrum: Diagnostic Concepts. In: Volkmar F, Paul R, Pelphrey K, Rogers S, editors. Handbook of Autism and Pervasive Developmental Disorders. 4th ed. Hoboken, NJ: John Wiley.
3. American Psychiatric Association. Diagnostic and Statistical Manual. 5th ed. Arlington, VA: APA Press; 2013.
4. Kolvin I. Studies in childhood psychoses. I. Diagnostic criteria and classification British Journal of Psychiatry. 1971;118:381-4
5. Rutter M. Childhood schizophrenia reconsidered. Journal of Autism & Childhood Schizophrenia1972 Oct. 1972;2(4):315-37.
6. Volkmar FR, Nelson DS. Seizure disorders in autism. Journal of the American Academy of Child & Adolescent Psychiatry. 1990;29(1):127-9.
7. Folstein S, Rutter M. Genetic influences and infantile autism. Nature. 1977;265(5596):726-8.
8. Rutter M, Thapar A. Genetics of Autism Spectrum Disorder. In: Volkmar FR, Paul R, Pelphrey K, Roger S, editors. Handbook of Autism. Hoboken, NJ: John Wiley.
9. Hill PA, Zuckerman K, Fombonne E. Epidemiology of Autism Spectrum Disorder. In: Volkmar F, Paul R, Pelphrey K, Rogers S, editors. Handbook of Autism and Pervasive Developmental Disorders. 4th ed. Hoboken, NJ: John Wiley.
10. Wing L. Childhood autism and social class: a question of selection? British Journal of Psychiatry. 1980;137:410-7.
11. Grinker RR. Unstrange minds. New York, NY: Basic; 2007.
12. Mandell DS, Ittenbach RR, Levy SE, Pinto-Martin JA. Disparities in diagnoses received prior to a diagnosis of autism spectrum disorder. Journal of Autism & Developmental Disorders. 2006.

13. Howlin P. Outcomes in Adults with Autism Spectrum Disorders. In: Volkmar F, Rogers S, Paul R, Pelphrey K, editors. Handbook of Autism, 4th edition. Hoboken NJ: Wiley; 2013.
14. National Research Council. Educating Young Children with Autism. Washington, D.C.: National Academy Press; 2001. 307 p.
15. Lord C. Follow-up of two-year-olds referred for possible autism. Journal of Child Psychology and Psychiatry. 1996;36(8):1065-76.
16. Chawarska K, Klin A, Volkmar FR. Autism spectrum disorders in infants and toddlers: Diagnosis, assessment, and treatment. New York, NY: Guilford Press; US; 2008. p. 348.
17. Loveland KA, Tunali-Kotoski B. The School-Age Child with an Autistic Spectrum Disorder. In: Volkmar FR, Klin A, Paul R, Cohen DJ, editors. Handbook of Autism and Pervasive Developmental Disorders. 1. 3rd ed. Hoboken, NJ: Wiley; 2005. p. 247-87.
18. Kanner L. Follow-up study of eleven autistic children originally reported in Journal of Autism & Childhood Schizophrenia 1971, Apr. 1971;1(2):119-45.
19. Volkmar F, Wiesner L. A Practical Guide to Autism. Hoboken, NJ: John Wiley; 2009.
20. Piven J, Rabins P, Autism-in-Older Adults Working G. Autism spectrum disorders in older adults: toward defining a research agenda. Journal of the American Geriatrics Society. 2011;59(11):2151-5.
21. McPartland JC, Wu J, Bailey CA, Mayes LC, Schultz RT, Klin A. Atypical neural specialization for social percepts in autism spectrum disorder. Social Neuroscience. 2011;6(5-6):436-51.
22. Schultz RT, Gauthier I, Klin A, Fulbright RK, Anderson AW, Volkmar F, et al. Abnormal ventral temporal cortical activity during face discrimination. Archives of General Psychiatry. 2000;57(4):331-40.
23. Vander Wyk BC, Voos A, Pelphrey KA. Action representation in the superior temporal sulcus in children and adults: an fMRI study. Developmental Cognitive Neuroscience. 2012;2(4):409-16.
24. Casanova MF, El-Baz A, Vanbogaert E, Narahari P, Switala A. A topographic study of minicolumnar core width by lamina comparison between autistic subjects and controls: possible minicolumnar disruption due to an anatomical element in-common to multiple laminae. Brain Pathol. 2010;20(2):451-8.
25. Lewis MH, Tanimura Y, Lee LW, Bodfish JW. Animal models of restricted repetitive behavior in autism. Behavioural Brain Research. 2007;176(1):66-74.
26. Wing L, Potter D. Epidemiology of Autism. In: Goldstein S, Naglieri JA, Ozonoff S, editors. Assessment of Autism Spectrum Disorders. New York, NY: Guilford; 2008. p. 18-54.
27. McClure I. Developing and Implementing Practice Guidelines. In: Volkmar F, Paul R, Pelphrey K, Rogers S, editors. Handbook of Autism and Pervasive Developmetnal Disorders. 4th ed. Hoboken, NJ: John Wiley.
28. Ibanez LV, Stone W, Coonrood EE. Screening. In: Volkmar F, Paul R, Pelphrey K, Rogers S, editors. Handbook of Autism 4th ed. Hoboken NJ: John Wiley.
29. Lord C. Diagnostic Instruments. In: Volkmar F, Paul R, Pelphrey K, Rogers s, editors. Handbook of Autism. 4th ed. Hoboken, NJ: John Wiley.
30. Volkmar FR, Wiesner LA. A practical guide to autism: What every parent, family member, and teacher needs to know. Hoboken, NJ: John Wiley & Sons Inc; US; 2009.
31. Volkmar FR, Woodbury-Smith M, King BH, McCracken J, State M. Practice Paramters for the assessment and treatment of children and adolescents with autism and pervasive developmental disorders. Journal of the American Academy of Child & Adolescent Psychiatry. J Am Acad Child Adolesc Psychiatry 1999 Dec; 38 (12 Suppl) 32S-54S.