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OPINION

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Reconceptualizing Autism: Moving Beyond the Behavioral to Address Cause, Cure and Prevention

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Abstract: Since the publication of Leo Kanner's seminal paper in 1943, there has been essentially no definitive light shed on the cause, prevention or cure of autism. It is our contention that the reason lies, at least in part, with the original psychiatric conceptualization of the condition and the subsequent acceptance of this framework by health professionals ever since. We suggest an urgent revision of autism as a disease state such that its operationalization in major diagnostic systems such as the Diagnostic and Statistical Manual of Mental Disorders and International Classification of Diseases recognizes the biological variables known to be associated with autism.

Keywords: autism, psychiatric conceptualization

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Introduction

Since the publication of Leo Kanner's seminal paper in 1943,¹ research into autism has resulted in the publication of over 12,000 peer-reviewed papers, yet, there has been essentially no definitive light shed on cause, prevention or cure. The question that must be asked is, "Why?" It is our contention that the reason lies, at least in part, with the original psychiatric conceptualization of the condition by Kanner and the subsequent acceptance of this framework by health professionals ever since.

As autism was first "discovered" by a psychiatrist (Kanner) and is classified in the Diagnostic and Statistical Manual of Mental Disorders (DSM), historically, research into the condition has primarily been the domain of psychologists and psychiatrists (especially developmental). These professions typically view autism as a developmental or behavioral disorder and therefore field trials designed to establish the diagnostic definition of autism, as well as other research within the broad field of autism, tends to focus on behavioral or developmental aspects of the condition. The obvious limitation of this approach is the evident internally reinforcing model whereby autism is conceptualized as a behavioral or developmental disorder and therefore research and field trials that follow are designed to examine variables of that nature almost exclusively, thus confirming the original assumptions (e.g.²).

The majority of autism research tends to focus on assessment/diagnostic measures, behavioral/educational interventions, and incidence/prevalence rates (for a review see³). Where biological/physiological variables are considered, it has been predominantly in the area of neurological functioning (e.g.⁴). In contrast to the enduring conceptualization of autism as a behavioural or developmental disorder (or even a psychiatric or neurodevelopmental disorder), emerging evidence over the last decade has clearly identified a range of biomedical irregularities that are consistently present in individuals with autism, suggesting that there is a biological/medical component to autism that needs to be integrated into our understanding and formal conceptualizations if we are ever to gain a genuine and complete picture of the condition.

It is crucial to understand the origins of a condition such as autism as, once a disease state is defined and

labeled, the way in which a condition is perceived and treated by clinicians, researchers, medical professionals and policy makers is not easily altered. Therefore, if the process of properly recognizing and naming a condition is not rigorously conducted, via thorough medical examination and differential diagnosis, the end result can be instrumentally destructive to the fundamental goal of medical research to identify the cause of a condition and the subsequent determination of prevention and cure.

Autism as a Psychiatric Disorder: The Historical Context

In his seminal paper proposing the discovery of a new condition that would come to be known as autism, Kanner,¹ in a 19,000 word detailed case series analysis of 11 children, did not once mention considering a biological explanation for the "fascinating peculiarities" he observed, nor indicate that a single biological hypothesis was pursued. Kanner's bias to a psychiatric explanation for the patient presentations he was seeing is also reflected in his journal of choice, "Nervous child".

In his paper, the cases are presented in a narrative form with almost no references to medical examinations or evaluations and, instead, a relentless description of child and parental behavioral characteristics is provided, with the author clearly trying to draw behavioral/psychological parallels between the child and parents: "This much is certain, that there is a great deal of obsessiveness in the family background.¹ (p. 250)" In this perspective lies the genesis of the "refrigerator mother" theory of autism whereby autism was presumed for several decades to be caused by emotionally frigid mothers. Although the theory is attributed to another psychiatrist, Bruno Bettelheim, the theory was clearly supported by Kanner who in a 1960 interview described parents of autistic children as "just happening to defrost long enough to produce a child."⁵

In medical practice, the appropriate action when presented with a difficult case is to do the most thorough exploration as possible as to the patient's physical state (e.g. physical examination, blood, urine, imaging studies) and, if the diagnosis is still not clear from that point, to develop a list of possible diseases which might best explain the patient presentation.



Following, one then continues to think critically and conduct further testing as necessary in order to go about excluding disorders from the list of possibilities. This well known standard medical practice appears completely absent from Kanner's professional practice. Instead of conducting the aforementioned process, Kanner instead wrote a 19,000 word case description and suggested that he had discovered a new psychiatric condition. In an attempt to understand the approach of Kanner, it is useful to understand a little of the man and his background.

Kanner was an Austrian-born psychiatrist influenced by the dominant psychiatric paradigm of the time; psychoanalysis. Hence, his personal perspective appeared to be brought to bear on the original 11 children; "We must, then, assume that these children have come into the world with innate inability to form the usual, biologically provided affective contact with people, just as other children come into the world with innate physical or intellectual handicaps.¹ (p. 250)" In other words, Kanner assumed

that the children were abnormal *at birth*. This is an extraordinary assumption, completely unsupported by any science of the time, and is curiously incongruent with his support for the refrigerator mother theory of autism which places the primary cause of autism as occurring during the first months and years *after* birth.

Kanner, an avid poet with a strong love of the arts, and "with no more training in pediatrics or child psychiatry than he had received as a medical student,"⁶ announced to the world a new psychiatric disorder without ever explaining how the symptom profiles he had observed may not already be better explained by medical or psychiatric conditions known at the time. In a startling coincidence, at around the same time, Hans Asperger, another Austrian physician with a penchant for poetry, published the first definition of Asperger's syndrome in 1944.⁷ In a paper describing just four boys, he identified a pattern of behavior and abilities that he called "autistic psychopathy." His special interest was "psychically abnormal" children.

Table 1. Diagnostic and statistical manual edition 4 text revision for autistic disorder.

-
- A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):
1. Qualitative impairment in social interaction, as manifested by at least two of the following:
 - a. Marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 - b. Failure to develop peer relationships appropriate to developmental level
 - c. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. by a lack of showing, bringing, or pointing out objects of interest)
 - d. Lack of social or emotional reciprocity
 2. Qualitative impairments in communication as manifested by at least one of the following:
 - a. Delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
 - b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
 - c. Stereotyped and repetitive use of language or idiosyncratic language
 - d. Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
 3. Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
 - a. Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
 - b. Apparently inflexible adherence to specific, nonfunctional routines or rituals
 - c. Stereotyped and repetitive motor manners (e.g. hand or finger flapping or twisting, or complex whole-body movements)
 - d. Persistent preoccupation with parts of objects
- B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years:
(1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.
- C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.
-



In this, we see that Asperger reveals his psychiatric bias to be similar to Kanner's. In a further similarity, Asperger also appears to have failed to conduct any biological testing or make any meaningful attempt to determine whether the four boys had, in fact, an already known condition, rather than proposing that somehow, mysteriously, a new disease had landed at his doorstep exclusively in the guise of just four young boys (Asperger was unaware of Kanner's paper at this time).

So, the names Kanner and Asperger are inextricably linked to the autism spectrum disorders, and many herald the two men as the fathers of child psychiatry. Several important questions remain however. How likely is it that these two men, "discovered", by chance, a brand new disorder, never before seen in the human species, which appeared simultaneously on separate continents at around the same time? And what does it suggest that both men were Austrian in upbringing and heavily influenced by the psychoanalytic perspective of behaviour and human development, both failed to undertake testing of physiological variables or attempt medical explanations for the behaviour they were seeing, and both published articles of their "discovery" as a new psychiatric disorder?

Autism Today

Modern perspectives of autism are still clearly influenced by Kanner. This point is most clearly exemplified by the 'evolution' of autism in the DSM. Autism first appeared in the third edition of the DSM as 'Infantile Autism' in a new class of conditions titled Pervasive Developmental Disorders.⁸ Substantial revisions to the definition of autism were made in the DSM-III-R, with the criteria broadened to reflect a developmental orientation and the heterogeneity of autism.⁹ Subsequent editions of the DSM, both the fourth edition and the text revision of the fourth edition, have largely retained the same definition (see Table 1).^{10,11}

Asperger's disorder was first included in the fourth edition of the DSM (and continued in the text revision of this edition).^{10,11} As with autism, the disorder is contextualized as a behavioral/developmental condition, the primary difference with autism being that there is comparatively less disability. Table 2 presents the DSM-IV-TR definition of Asperger's disorder.

As can be clearly seen by the DSM definitions, autism spectrum disorders continue to be placed firmly in a developmental/behavioral context with the criteria

Table 2. Diagnostic and statistical manual edition 4 text revision for asperger's disorder.

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- A. Qualitative impairment in social interaction, as manifested by at least two of the following:
1. Marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 2. Failure to develop peer relationships appropriate to developmental level
 3. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. by a lack of showing, bringing, or pointing out objects of interest to other people)
 4. Lack of social or emotional reciprocity
- B. Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
1. Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
 2. Apparently inflexible adherence to specific, nonfunctional routines or rituals
 3. Stereotyped and repetitive motor mannerisms (e.g. hand or finger flapping or twisting, or complex whole-body movements)
 4. Persistent preoccupation with parts of objects
- C. The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.
- D. There is no clinically significant general delay in language (e.g. single words used by age 2 years, communicative phrases used by age 3 years).
- E. There is no clinically significant delay in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction), and curiosity about the environment in childhood.
- F. Criteria are not met for another specific Pervasive Developmental Disorder or Schizophrenia.
-



centered upon the core triad of behavioral symptoms: impaired socialization, communication and repetitive/obsessive behaviours. This exclusive diagnostic triad is likely to be seen as increasingly out of step with modern understandings of autism that recognize biomedical irregularities such as elevated oxidative stress,^{12,13} mitochondrial-respiratory disorders,^{14,15} generalized inflammation,¹⁶ neuroinflammation,^{17,18} gastrointestinal abnormalities,^{19,20} fatty acid deficiencies,^{21,22} and elevated urinary porphyrins.^{23–25}

Conclusion

Some 70 years have passed since autism was first identified, yet we are no closer to understanding what it is caused by, nor how the condition may be cured or prevented. We argue that this lack of progress is at least partially attributable to Kanner and the manner in which he conceptualized autism as a parentally-mediated psychopathology. Why do we need to be concerned about this? Simply put, autism is a devastating condition, lifelong in duration, with the majority of afflicted individuals requiring supported living arrangements. The majority of sufferers will never engage in meaningful employment, marry nor have children, and cannot engage in meaningful conversation. Autism affects not only the individual, but the family unit and community as a whole.

To avoid simply treading the same unfruitful path of the previous 70 years, we would suggest an urgent revision of autism as a disease state such that its operationalization in major diagnostic systems such as the DSM and International Classification of Diseases recognizes biological variables known to be associated with autism. The affect of this would be to facilitate a more multi-disciplinary and inclusive range of health disciplines to research the biological bases of autism. Improving our understanding of these bases is a fundamental way of addressing the touchstones of medical research into autism; cause, cure, prevention and treatment.

Disclosures

This manuscript has been read and approved by all authors. This paper is unique and is not under consideration by any other publication and has not been published elsewhere. The authors and peer

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