On the Etiology of Autism

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Abstract

Autism, categorized as a Pervasive Development Disorder (PDD), is characterized by impaired social interaction, impaired communication, and repetitive behavior. The cause of the disorder has been unclear since its first description by Kanner in 1943. Historically, the prevailing explanation was psychogenic in nature, namely that the mother's emotional "frigidity" is to blame (giving rise to the term "refrigerator mother"). This reasoning resulted in the suffering and guilt of many caregivers who faced such an accusation. Through the 1970s, this theory was largely assumed to be correct, but most modern clinicians believe that the etiology is mostly genetic and neurological in nature. Recent research was thought to put an end to this debate on environmental factors, yet present-day proponents of the psychogenic model still assert that poor parenting explains the disorder.

Introduction

Autism has come to the forefront of public opinion. Concerns about vaccinations have become national legal battles as parents and researchers alike search for answers on the cause of this devastating disorder. The increasing public awareness of one of the rarest disorders (Klin, 2006, S5) has propelled a large number of investigators to pursue this question in hopes of being able to intervene and correct the course of development for afflicted individuals. Even so, the etiology of autism remains elusive, despite a half-century of research.

Although not usually placed in the same category as unsolved problems in advanced theoretical physics or computer science, the Autism Spectrum Disorders (ASD) have proven to be among the most vexing questions under scientific investigation (Kennedy & Norman, 2005). Definitively proving the origin of this disorder would clearly be an immense achievement, yet it seems unlikely that such a result is imminent. However, recent progress has opened new avenues not known previously, lending hope to the many families who know the hardships of being more than an observer to this psychological curiosity.

Of course, like many unsolved problems, many causes have been proposed since the first description of autism. Without a solid grounding on the facts, however, it is difficult to judge such explanations. This puzzle contains a number of oddly-shaped and otherwise unrelated pieces that are, in some way, shape, or form, expected to come together to form a unified whole.

Behaviorally, autistic persons are well known for being characterized by impaired social interaction, communication, and repetitive, unproductive behavior. Other representative signs include apparent deafness, apparent mental retardation in 60-70% of the population (Klin, 2006, S4), and a 25% incidence of epileptic seizures (Volkmar, 2007, 185). Beyond these classic symptoms, however, harder to explain factors are present, including biting (sometimes self inflicted) as well as profound eating difficulties (Klin, 2006; Oltmanns, Neale, & Davison, 1995; Schopler & Mesibov, 1985).

In terms of heredity, autism has been described as congenital since its first classification (Kanner, 1943), though more recently ASD individuals have been shown to have normal psychological development before seemingly taking a 180 degree turn to enter an autistic state (Chawarska, Klin, & Volkmar, 2008, 15). Parents tend to be intellectual (Kanner, 1943), but not enough to be predictive. Having siblings is uncommon — many parents cannot withstand the thought of bearing another ASD child — but in the statistically rare case of a second-born autistic child, they often present a similar (if not identical) disorder. Finally, the disorder is observed more often in males (Oltmanns et al., 1995; Wing, 1966). As might be expected, the significance of these combined factors remains unclear even today (Chawarska et al., 2008).

The prognosis of such persons is often bleak. Most will be reliant on caregivers until the end of their life, although some do show marked improve-

ment (Volkmar, 2007, 56). Clearly, it is desirable to improve the quality of life for affected persons, including both the individual presenting autism as well as their family. Such a focus has not always been the case, as older approaches of diagnosing and treating this disorder differed greatly.

Historical Perspectives

Most renditions of the history of autism begin with Kanner's original classification of "infantile autism" as a disorder separate from other childhood psychological illnesses. However, digging deeper reveals that a French physician in the 1700s named Itard wrote about a boy that contemporary researchers have classified as autistic (Schopler & Mesibov, 1985, 6). Besides showing that autism existed before modern technology, this and other reports are often forgotten; this first encounter is often eclipsed by Kanner's description.

Fombonne points to Kanner's 1943 paper as the first appreciable step in the diagnosis criteria for autism (2003, 503). In a time dominated by Frued's academic disciples, Kanner's writings were atheoretical and fact-driven (Volkmar, 2007, 2). His contemporaries had often confused the symptoms with other closely related disorders, using psychodynamic categorizations such as "infantile schizophrenia" and "early childhood psychosis" for what Kanner showed to be a distinct condition (Fombonne, 2003, 503).

Psychodynamic Explanations

Even after Kanner's pioneering work, psychodynamic explanations still found their way into his newly-christened disorder. At the outset, the early pattern of using adult terms such as "schizophrenia" and "psychosis" carried on, reflecting the untested and unfounded psychoanalytical theories which dominated even into the late 1960s (Fombonne, 2003, 503).

Psychodynamic theories stunted the growth of the field, causing many researchers to overlook otherwise salient aspects of the problem. One pronounced example is the phenomenon of pronoun reversal, a linguistic aspect of the disorder. (For an example of this, consider the child having the intention of communicating hunger. Rather than using the utterance "I am hungry," he or she would use "you are hungry," reversing the pronouns.) The psychodynamic adherents interpreted this as evidence of the child's "inability to separate himself or herself from others." However, as has been shown in studies of Typically Developing (TD) children's language, such instances are forms of echolalia presented out of context, a process that typically disappears at a young age. Freudian thinking construed this problem to be that of a normally functioning person trapped underneath an "autistic shell" (Schopler & Mesibov, 1985, 5). Interestingly, Kanner saw this disorder as covering up intellectual potential, showing a similar view (Volkmar, 2007, 2). Furthermore, echolalia was viewed by the psychodynamicists as "suggesting that children understood a great deal more than they let on and were somehow holding back what they know," a ridiculous notion to modern theorists (Schopler & Mesibov, 1985, 5).

"Refrigerator Mothers"

Although the Freudian observations may have been taken as genetic even then, but 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" (Fombonne, 2003, 503). This being combined with the popular press' promotion of psychodynamic interpretations in the 1950s and 60s (Schopler & Mesibov, 1985, 5) led to the "'refrigerator mothers' hypothesis". At this time, the common understanding was that the etiology of autism was rooted in "parents who were emotionally unresponsive to their children" (Klin, 2006, S4).

This concept was especially distressing to mothers of autistic children who grew to view themselves as the singular cause for their child's disorder. Reportedly, Kanner also believed in such an assessment, but remarked that it was not supported by the data (Tustin, 1981; Wing, 1966, 31). Even today, this thinking lingers on as parents often find themselves in denial of the possibility of autism when it becomes evident, believing such a harsh etiological account will apply to them as well (Chawarska et al., 2008, 301).

Even if the cause is not "frigitity", the problems that autism causes in family functioning is easily apparent, sometimes being called a "family disability." Often, one parent becomes a full-time caretaker for the child and the other focuses on their career (Chawarska et al., 2008, 317-8). It is not surprising, therefore, that the divorce rate in families with an autistic child is high. These parents have understandably begun demanding answers in hopes of ending their frustration.

While the "refrigerator mother" explanation has since been abandoned in the United States, it is still able to be found in Europe and Latin America. Starting in the 1960s, growing evidence began to suggest autism was a congenital brain disorder, "found in all countries, social economic and ethnic/racial groups in which researchers sought it"

(Klin, 2006, S4).

Legacy

Surprisingly, the psychodynamic explanation of the disorder is less than archaic, even in the United States (Schopler & Mesibov, 1985, 6). Through the 1980s, authors such as Tustin likens the disorder to a decision between organic and psychogenic explanations, the characteristic behaviors being an "autoimmune reaction to the self" and a "mismangaged psychological birth" (1981, 10-14). As seems to be traditional in Freudian thinking, conjecture enters most of the writings, coming in terms of "psychosomatic cores" that are used without any scientific proof of existence (Tustin, 1981, 14).

As a further result of this clash of theories, many parents of autistic children have become misinformed of what treatment options are available to them. As such, ASD children are often the target of controversial treatments, "interventions that are popular despite an absence of scientific or theoretical support." Unfortunately, autism's proclivity against explanation has been its own downfall, as much speculation is made about possible causes and remedies. All too often these are claimed to be miracle cures when in fact they are nothing but (Chawarska et al., 2008, 243).

Autism did not enter into the Diagnostic and Statistical Manual of Mental Disorders (DSM) until 1980, and even then it was colored by previous interpretations. Although a goal of the DSM III was to be atheoretical, its description retained many of its previous qualities from being classified as "childhood schizophrenia." However, some great strides were made, as a new classification called Pervasive Development Disorders was made to contain autism in this edition of the DSM (Chawarska et al., 2008, 3). As the DSM has improved, both the fourth edition (DSM-IV) and the ICD-10 have gained more accurate definitions (Volkmar, 2007, ix).

Modern Approaches

Recent history has been marked by a large growth in data concerning autistically developing children. Particularly, the 1990s led to a new goal: early diagnosis and intervention. It is thought that the ability to collect more data, especially that from the first year of life, will lead to a better understanding of the disorder. This has largely come to pass in the form of eye tracking techniques. Previously, researchers could only see the damage after it had been done; now they have the chance to track and study when and how the disorder develops, opening the door to many useful longitudinal studies. Today, the consensus (subtracting Freud's ever-present legacy) is that autism is a "behavioral syndrome caused by one or more factors acting on the central nervous system" (Chawarska et al., 2008, 277).

Genetic

If factors impinging upon the central nervous system are thought to be the cause, one obvious avenue is a genetic explanation for its abnormal development. Surprisingly, a genetic role in the disorder was not thought to exist in 1976, while current geneticists have found it to be one of most heritable given evidence from twin studies (Volkmar, 2007, 157). Current estimates have put its heritability above 90%, making it very strongly genetic (Fombonne, 2003, 504). As such, there has been an explosion in attention: genetics has gone from being used with the word "if" to "where" (Volkmar, 2007, ix-x).

Of course, this makes it possible to create very reliable diagnoses; the ability to do so is limited only by the technology to implement them. This evidence combined with a valuable goal has led to the development of "reliable and valid diagnostic tools" and insights through the field of molecular genetics. Unfortunately, with this search for genetic etiology currently taking place, it is "obvious that research findings will not translate into practical help for some time," although the future is bright (Fombonne, 2003, 504).

Neurobiological

Biology, as we know, is largely the result of the expression of proteins from genetic factors. With this in mind, deciding to investigate neurobiological etiology is quite logical taking the consensus of autism's cause into account. As might be expected in such a difficult disorder, there are only a few consistent abnormalities expressed in autistic individuals, and thus very few genes that have been identified as taking part (Fombonne, 2003, 504). However, such abnormalities are not rare, being present in 75% of autistic subjects, although they differ from patient to patient. Much to neuroscientists' dismay, no "gross morphological abnormalities" have been found (Volkmar, 2007, 188)

With a large amount of research taking place, the amount of available data has grown immensely. However, this disorder seems like a frayed edge of a shirt: every time it is pulled, the more it unravels. The current research has given many intriguing insights and breathed new life into the field, yet still not much is known (Volkmar, 2007, xi). Even

so, growth is apparent, albeit slow. Take for example electroencephalographic (EEG) evidence: in 1966, causes in the CNS were very unclear (Wing, 1966, 34) whereas today EEG abnormalities are excellent evidence (Volkmar, 2007, 186). Furthermore, the burgeoning field of neurochemistry has identified several implicated neurotransmitters, such as serotonin (Volkmar, 2007, 195). Indeed, much has changed in 40 years.

While neurobiological evidence such as this has been found, it has not satisfied all the symptoms described in the introduction. A very limiting factor is the small sample sizes associated with autism, given its rarity. Furthermore, a review of the literature before the improved diagnostic criteria of the DSM-III-R has made accurate measurements difficult (Volkmar, 2007, 179).

Environmental

Again, the legacy of the psychodynamicists lives on as even recent research has gone back to investigating family influence on autistic behavior. Admittedly, the goal has changed much since it was last investigated, as currently the effects of autism on the entire family has become a major concern. The reasoning for opening this Pandora's box again has been said to be that schizophrenia is a similarly expressed disorder that has been shown to be brought about by such external forces, at least in part (Greenberg et al., 2006, 229-30). Other new environmental factors have been investigated, including (but not limited to) diet, heavy metals, infectious agents, pharmacological agents, and/or immunizations (Chawarska et al., 2008, 280-283).

Vaccines

Immunizations have been the subject of several recent court rulings and, much to the chagrin of some researchers, has become the poster child of autism to the American public. However, *no* single environmental risk factor has 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 is implicated. The mercury (thimerosal) used to preserve such vaccines has also been questioned, but all links have been disproved (Fombonne, 2003, 504). The logic behind such a connection was that factors in these immunizations could trigger out-of-control infections and lead to brain damage that could develop into autism (Chawarska et al., 2008, 256). Given the previous discussion of neurobiological factors, such evidence is on shaky ground at best, as it would likely be visible in examinations of such afflicted individuals. Additionally, as has been shown, autism predates the MMR vaccination (Chawarska et al., 2008, 282).

This presumed linkage has brought about serous public health concerns in that it has contributed to a reduction of vaccinations in several countries, including the United States and Japan. This has caused a number of deaths which could have otherwise been avoided. Surprisingly, a Japanese city made the decision to stop administering MMR vaccines. As expected, a study of this situation showed that autism rates did not decrease (Chawarska et al., 2008, 257). The evidence for vaccines being an etiology would seem to be entirely anecdotal in nature.

Conclusions

As has been shown, the field of autism research has grown and learned from a long history of research. Better, more reliable early detection methods have increased, as has awareness of the disorder in general. Even so, evidence such as the confusion on vaccinations and other "folk etiologies" for the disorder have raised the need to educate the public on more than just awareness of the symptoms of the disorder. While useful in terms of identifying individuals for treatment, the outcome has also interacted with lay opinion and made some otherwise-manageable problems worse.

Furthermore, the discussed psychodynamic explanations of autism have often been subject to having too much "read into" the measured behavior. However, modern research is not free of this affliction; many current research tracks have assumed that autism is a *social* developmental disorder without defining criteria for such a term. Indeed, there is a rich amount of remaining factors that need to be "teased apart" in order to satisfactorily explain this disorder, and as such, present and future researchers should be mindful of such implications.

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