Understanding Autism

Parents and Pediatricians in Historical Perspective

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Both primary care providers and subspecialists in pediatrics encounter families who are actively involved in the diagnosis and treatment of their children. Parents of children with an autism spectrum disorder in particular are often aware of scientific issues, and their expertise and desire for a medical cure for autism sometimes put them at odds with the medical team. We investigated the role of parents and advocates in autism research and treatment over the last 50 years. Our review of scientific publications and archival sources documents how parents and advocacy groups have done the following: (1) organized research funding; (2) constructed clinical research networks; (3) suggested new avenues for research; (4) popularized empirically based therapies; and (5) anticipated paradigmatic shifts in the understanding of autism. We believe that this historical account will help pediatricians and researchers recognize that families can contribute to expert understanding of complex medical conditions such as autism and that the existence of partnerships with families of children with autism is a critical component of future research and treatment programs.

Arch Pediatr Adolesc Med. 2007;161:392-398

Clay’s parents brought their 4-year old son to the developmental assessment team with hopes of helping their child with autism; they left disappointed and divided. Clay had received a variety of medical treatments, including hyperbaric oxygen, oral fluconazole, and chelation, prior to the team’s evaluation. He was also on a diet that eliminated gluten- and casein-containing foods and was receiving a variety of nutritional supplements, including high doses of vitamin B6 and magnesium. Clay’s mother believed that he had improved at least temporarily with each of these interventions; his father was more skeptical. Both parents hoped that the medical team would recommend treatment with secretin. When the developmental assessment team described the results of recent randomized controlled trials demonstrating no benefit from secretin compared with placebo, Clay’s mother pressed the team. Did the authors run separate analyses on the subset of children with gastrointestinal symptoms? Did the studies have enough statistical power to rule out the possibility of benefit from secretin, and what outcome measures were used? Although she had no formal medical or scientific training, Clay’s mother lodged several other criticisms of the studies and became increasingly frustrated that the team did not seem to want to help her son. For their part, the team felt that their professional competence was being challenged, and several team members worried that in pursuit of alternative treatments, Clay’s family was failing to focus on educational interventions that could improve Clay’s social and academic functioning. After a 2-day team evaluation and a 90-minute family conference, no one was happy. Clay’s mother found no support for her quest to help her son, Clay’s father had renewed doubts about the wisdom of expensive and intrusive treatments, and the team wondered why their expertise and hard work brought only frustration to everyone involved.

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This case scenario is a composite drawn from the experiences of a university clinical service, but such conflict is not unique to developmental assessment teams. Both primary care providers and subspecialists in pediatrics frequently encounter families who are well informed and actively involved in the diagnosis and treatment of their children. In general, pediatricians have led the medical profession in learning to respect patient and family autonomy; indeed, family-centered pediatric practice is endorsed explicitly by the American Academy of Pediatrics. Nearly all pediatricians have sometimes felt angry or bewildered, however, by families and patients who challenge their authority as the professional medical provider. Clay’s case stands out because of the degree to which Clay’s parents had mastered the scientific process and how their expertise and desire for a medical cure for autism put them at odds with the medical team.

Such conflicts arise in many areas of medicine, especially in the case of childhood disorders, but autism is a particularly salient example because of the condition’s past history and current notoriety. Autism has become one of the most difficult and controversial child health issues because of the remarkable increase in observed prevalence, controversies over etiology, and the search for specific medical interventions. Although there is evidence for the effectiveness of a variety of behavioral and educational interventions, alternative therapies for autism spectrum disorders are particularly diverse and widely applied as families seek medical treatments for an otherwise chronic condition. Pediatricians, meanwhile, are faced with the problem of continuing to provide understanding support for families who may want to investigate treatments that are unfamiliar or insufficiently supported by evidence and controlled trials.

Classic autism was first described in the medical literature by Leo Kanner in 1943, and through the 1950s the condition remained in the domain of psychiatry. Bruno Bettelheim and many of his colleagues in psychology interpreted autistic behavior in classic Freudian terms as a syndrome of ego damage resulting from unconscious parental rejection. By the 1960s, medical professionals began to seriously consider nonpsychological frameworks for understanding autism, and in 1964, Bernard Rimland described infantile autism as a neurological disorder with a strong genetic component. The first study showing a high degree of concordance in a series of monozygotic twins was published in 1977, and by the mid 1990s, most researchers agreed that autism was a highly heritable genetic neurological disorder. The recent apparent rise in the number of cases of autism suggests an environmental component of the etiology to many parents, although epidemiologists and psychologists have generally viewed these shifts as a result of improved ascertainment and as a consequence of greater physician and public awareness. They also observe that growing prevalence reflects an increasingly broad definition of autistic disorder over the last 20 years as well as the inclusion of Asperger syndrome and pervasive developmental disorder not otherwise specified in the current concept of autism as a spectrum of disorders.

Although many medical professionals are aware of this basic history of autism, few know about the critical role that families have played in advancing both the clinical and scientific understanding of autism as well as treatment options. Throughout the history of autism in the United States, parental observations and priorities have come into periodic conflict with professional opinions, whether those opinions concerned the adequacy of parents’ childrearing abilities or their belief that there is an epidemic of new cases of autism. Perhaps because of this conflict, parents have often been at the vanguard of critical changes in expert understanding of autism.

LEARNING FROM PARENTS

From the first description of autism, parents have been essential to advancing research. In his case reports, for example, Kanner credited the meticulous notes provided by parents as the primary source for his research. Foreshadowing decades of conflict to come, however, his acknowledgment also contained an accusation: “[t]he obsessiveness of the parents of the autistic children was a veritable boon to me with regard to the case histories. Few children have ever been observed by their parents with such minute precision.” In public settings, Kanner went on to describe the parents of children with autism as “just happening to defrost long enough to produce a child.” Building on Kanner’s observations, Bettelheim and his colleagues believed that providing psychotherapy for parents as well as their children was a logical step in treating autism. Parents suffered as a result. In a 1967 memoir read by generations of parents, Clara Claiborne Park observed that the “depersonalization” of the clinical setting might easily have caused otherwise warm and engaged parents to appear aloof. She wondered whether “refrigerator professionals create refrigerator parents.”

Some parents responded by challenging conventional medical wisdom. Rimland, for example, published in 1964 one of the first surveys of evidence that autism is a neurological disorder with a genetic component. Like many parents, the Rimlands had diagnosed their children themselves at a time when professionals were often unfamiliar with the disorder. Perhaps because of Rimland’s training as an experimental psychologist, neither he nor his wife had much patience with the psychogenic theory of autism. Rimland argued that the atypical social tendencies and intellectualism of the parents of children with autism might be evidence of mild autistic tendencies in parents—what researchers now refer to as the broader autism phenotype. Other parents helped educational professionals understand autism as a developmental disorder. For example, autism was categorized by the US Office of Special Education under the term severely emotionally disturbed through the 1970s. In changing the terminology in 1980, government officials credited “voluminous” evidence provided by parent groups on the devastating effects of categorizing autism as an emotional disorder.

In 1974, the editors of the Journal of Autism and Childhood Schizophrenia (later renamed the Journal of Autism and Developmental Disorders) also recognized the value of families’ perspectives in a section called “Parents Speak.” The articles are a telling record of parents’ genuine in-
terest in research findings and provide examples of how parents anticipated scholars. Three early articles in the series, for example, commented on topics that have recently caught the attention of medical scholars: autism in adolescence, possible biological factors in autism, and parental stress and resilience.

Parental observations about the onset and natural history of autism have also been vindicated by recently published studies. At least since the 1970s, a subset of parents has described how their children had a period of normal development before the onset of autism. In this regressive form of autism, parents reported that their children acquired words, maintained eye contact, and demonstrated joint attention only to lose these skills at some point in the second year of life. Only recently have researchers begun to focus on children with autism who regress. Indeed, a 2005 analysis of birthday videotapes confirmed parental claims, finding that some children did lose social and communication abilities between their first and second birthdays.

Parents also helped move research on the genetics of autism forward in the 1990s. The founders of the Cure Autism Now Foundation (CAN), Jon Shestack and Por- tia Iverson, understood that if autism were a complex and etiologically heterogeneous genetic disorder, a large sample of genetic material was necessary to understand the disorder. Because researchers did not seem to be sharing genetic samples, these parents contacted families and enrolled them in a new gene bank organized through CAN, the Autism Genetic Resource Exchange. To prevent needless reproduction of results, CAN made the publication and sharing of data conditions of use. The Cure Autism Now Foundation was able to amass data and samples from more than 400 families at an initial cost of more than $6 million in private donations. Fewer than 10 years later, the Autism Genetic Resource Exchange has become part of a larger consortium called the Autism Genome Project that itself was initially sponsored by the National Alliance for Autism Research. Samples from the Autism Genome Project are maintained at a repository sponsored by the National Institute of Mental Health, and all qualified investigators are granted equal access.

More generally, parent groups have pushed for increases in federal funding for autism research. In 1997, the National Institute of Child Health and Human Development began a 5-year, $45 million program to establish an international network on the neurobiology and genetics of autism, the Collaborative Programs of Excellence in Autism. When the program was renewed in 2002, the member institutes committed another $60 million over 5 years. Parents also lobbied for Congress to pass the Children’s Health Act of 2000, which resulted in funding from the National Institutes of Health for individual researcher grants, the National Institutes of Health–funded Studies to Advance Autism Research and Treatment Centers Program, and provisions for epidemiological studies through the Centers for Disease Control and Prevention. The National Institutes of Health recently announced a new program of Autism Centers of Excellence involving up to $24 million in annual funding, and in December 2006, the Combating Autism Act, which authorizes significant amounts of future appropriations, was signed into law after energetic lobbying by parent groups.

Although it is difficult to say just how important the role of parent advocacy has been in securing this funding, parent groups have clearly been a guiding force in autism research. Grants for pilot studies from the National Alliance for Autism Research, for example, resulted in multimillion-dollar National Institutes of Health grants for a number of researchers. More importantly, both CAN and the National Alliance for Autism Research maintain grant review policies that distinguish them as parent organizations focused on treatment and cure in addition to basic research. Parents are guaranteed a say in the review process: CAN maintains a scientific review committee comprising scientific degree–holding parents of children with autism; this review committee ranks projects after an initial review by a scientific advisory group (written communication, Alycia Halladay, PhD, December 27, 2005). The National Alliance for Autism Research maintains a similar 2-tiered system (written communication, Therese Finazzo, January 6, 2006). The critical roles of these organizations in autism research—setting goals as well as distributing funds—seem only likely to grow. In 2005 following the diagnosis of their grandson, General Electric and NBC Universal executive Bob Wright and his wife Suzanne founded Autism Speaks with starting funds of $30 million. The Cure Autism Now Foundation and the National Alliance for Autism Research have both merged their operations with this larger organization. Meanwhile, the foundation of the “hedge fund titan” James Simons, who has an affected daughter, has begun to focus on targeted funding for genetics research.

Parents have often promoted research in the course of investigating promising interventions for their own children. Although some popular treatments may appear to be the result of a frantic search for solutions by desperate parents, it is important to remember that treatment-oriented approaches have also yielded reliable knowledge about autism. For example, the National Society for Autistic Children (later the Autism Society of America) was founded by Rimland and other parents in part to promote a then-new treatment possibility, applied behavior analysis, or the Lovaas technique. Although Ivar Lovaas and his colleagues began their work in the late 1960s, the widespread adoption of their techniques came in part because of the activism of parents who sought to obtain the best treatments for their children, often in opposition to those who argued that their children were untreatable. Rimland, for example, promoted applied behavior analysis in talks to parents around the country based on his observation of sessions at the University of California, Los Angeles. By 1987, Lovaas published the results of a controlled trial that, despite criticisms of his methods, remains one of the primary references for the efficacy of behavioral thera-
pies. Variations on these interventions remain the most widely used treatments for autism, although no direct comparisons of the efficacy of these methods are available. Significantly, the authors of almost all of the existing behavioral programs for children with autism have emphasized the centrality of parental participation, even when they disagreed with the discrete trial methods of Lovaas.

While some parent-run autism organizations have focused on genetics research and behavioral therapy, the Autism Research Institute and its Defeat Autism Now! (DAN!) conferences have pursued medical treatment. The Autism Research Institute was also founded by Rimland, who was initially interested in the use of high-dose vitamin B6 and magnesium as a treatment for autism; Defeat Autism Now! conferences are built around the premise that each child with autism will respond to an individualized regimen of nutritional supplementation, elimination diets, and detoxification therapies including chelation. These complementary and alternative treatments often concern medical professionals because there are few if any controlled studies of safety and efficacy. Proponents of alternative medicine respond that most studies are complicated by the variety of symptoms and severity in children with autism and by the possibility that improvement in some children is obscured by a lack of efficacy in others. Theodore Page has provided some theoretical support for this explanation in his review of the metabolic aspects of autism; he concludes that it is likely that “different metabolic, transport, signaling, and developmental defects cause a common defect in neural circuitry which is responsible for autism.”

In the absence of simple medical interventions for autism, it is not surprising that families would turn to alternative medicine to help their children, learning of new treatments from an informal network of families as well as from listservs, the Internet, and searches of the medical literature. One mother of a child with autism, Karyn Seroussi, described how she treated her son with an elimination diet after hearing from her mother-in-law that her husband had begun talking as a child only after milk was removed from his diet. Such anecdotal reports can be powerful evidence for families considering new therapies, especially when the risks seem relatively small.

Parents also turn to alternative medicine to address their children’s medical problems, ranging from allergies and immune problems to sleep disturbances, hyperactivity, and gastrointestinal difficulties, as much as the core symptoms of autism. A published compilation of testimonials from parents using biomedical treatments is filled with similar accounts: “Brian also suffered from multiple ear infections, acid reflux, and food intolerances,” “Matthew had always been very small for his age and had constant diarrhea,” “We battled not only thrush, but also constant, chronic congestion, eczema, and food sensitivities.” Parents often complain that pediatricians do not recognize common medical problems in their children because expressions of pain and discomfort are seen as merely part of the underlying neurological condition of autism, although some researchers have begun to consider the possibility of connections between physiological and behavioral symptoms.

In addition to voicing questions about the relative importance of autistic symptoms and what are seen as comorbid conditions, parents of children with autism have long argued that there are clinically distinct subsets of children within the category of classic autistic disorder. Indeed, it was parents’ insistence that their children had a variety of physical symptoms that led Andrew Wakefield, a gastroenterologist then at the Royal Free Hospital in London, England, to investigate a group of children with significant intestinal pathological abnormalities in addition to developmental regression. Wakefield’s contention that children with regressive autism and bowel symptoms reflected a “unique disease process” linked to the measles-mumps-rubella vaccine drew enormous attention from parents and the media. Wakefield’s work has been criticized for its study design, leading to the highly unusual “retraction of an interpretation” by nearly all of his coauthors. An expert panel of the Institute of Medicine recently confirmed that there is no link between the measles-mumps-rubella vaccine and autism. However, Wakefield’s description of a subset of children with a history of regression and a higher incidence of gastrointestinal symptoms has found some support in recent research, and a recent poster presentation by one group of researchers maintained that an association between the measles virus and bowel disease exists in some children with autism.

Parents have also expressed concerns about a relationship between the increasing number of vaccines in infancy and the increasing rates of autism diagnoses, particularly the potential of the ethylmercury-containing preservative thimerosal to act as a developmental neurotoxin. Physicians initially responded by postponing certain vaccines until thimerosal was removed; further research has not revealed any link to developmental disabilities, and a recent study found that the removal of thimerosal from vaccines has not been associated with any changes in autism prevalence. The same Institute of Medicine review committee that examined the evidence linking the measles-mumps-rubella vaccine to autism also confirmed the conclusion of a 2001 committee that although the hypothesis of an association between neuromuscular disorders and the use of thimerosal-containing vaccines was biologically plausible, it was not supported by epidemiological evidence. Parents counter by citing a number of suggestive laboratory studies, including one study finding that mice with autoimmune tendencies showed an increased susceptibility to thimerosal and another study suggesting that thimerosal may affect immune responses by altering the functioning of dendritic cells. Concerned parents have formed advocacy groups devoted to the autism-mercury hypothesis, while public health officials voice concerns about declining vaccination rates. This particularly contentious issue, as well as the unproven use of chelation therapies to treat possible “mercury toxicity” in children, may act to divert attention from the important observations of parents and obscure their potential contribution to understanding autism.
ANTAGONISM, PARTNERSHIP, OR COLLABORATION?

In the first half of the 20th century, parents of children with developmental disabilities gathered in small local groups to compare notes and wonder about their children’s future. In the 1930s and 1940s, these parent organizations began working together and started to change society’s views toward children with disabilities. They argued that there was no shame in disability and that local schools and governments had an obligation to support families who cared for their children at home. By 1932, the largest parent group, the National Association for Retarded Children, had 119 local chapters. A variety of professionals such as teachers in schools with special education programs also provided a voice for children with disabilities. Together, these grassroots organizations helped build the disability rights movement that dramatically transformed American laws, culture, and organizations. The Individuals With Disabilities Education Act, the Americans with Disabilities Act, and a variety of court decisions have worked to ensure that persons with disabilities have full access to work, schools, and recreational activities.

During the last 2 decades, advocacy groups have developed new relationships with researchers and medical practitioners. The wide availability of information on the Internet, combined with an increasing skepticism toward medical authority, has led to the formation of advocacy groups with an unprecedented interest in taking part in research on particular disorders and to very specific ideas about both the disorder and the ways that research should be carried out. Patients and their caregivers also have acquired a historical perspective: scholars in disability studies have demonstrated the ways that systematic exclusion from economic opportunities has been as important as medical factors in creating perceptions of persons with disabilities. Similarly, parents of children with autism are acutely aware of the history of psychogenic theories and are legitimately concerned about the ability of pediatricians and developmental specialists to dismiss parental claims in favor of an interpretation based on prevailing theories.

Beginning with human immunodeficiency virus and AIDS treatment activists in the last 2 decades, patient groups have gone beyond supporting research through funding to actively question the ways that clinical trials, for instance, are designed. This new generation of advocates often brings a formidable mastery of the vocabulary and methods of research to bear on their critiques. Medical anthropologists have called the emerging forms of patient activism devoted to specific disorders “biosociality” or “biological citizenship.” They emphasize the idea that now as never before, social groups are organized around shared experiences of illness and disability and these organizations act as interest groups that bring pressures to bear on legislators, courts, and the medical research community by virtue of the fact that researchers depend on patient populations for access to research subjects and materials.

Patient advocacy groups are not always easy partners with researchers, but they offer opportunities for understanding disorders in ways that are difficult to envision within the confines of clinical work. Patients and their caregivers have daily experiences with illness; they witness its alterations over periods of weeks and months and are often in a privileged position when it comes to detailed reporting on the efficacy of treatments. In the case of a behaviorally diagnosed disorder such as autism, this daily intimacy provides important resources: parental reports can enable subtyping across medical groups, such as children with regressive autism or gastrointestinal disturbances. Indeed, the recent interest of the National Institutes of Health and other national research organizations in community-based, participatory research acknowledges the value of including patients and advocates in all aspects of medical research.

CONCLUSIONS

Parents of children with disorders on the autism spectrum often suspect that they are regarded as “problem parents” because of their insistence on alternative disease models for autism. Many, like Clara Claiborne Park and Karyn Seroussi, express the sense that their status as parents invalidates their observations about their children in the opinions of medical practitioners. Meanwhile, pediatricians are concerned about the willingness of parents to fully disclose the range of interventions that they are using with their children, and many have voiced concerns that unproven treatments might be used in favor of behavioral and educational interventions. While case scenarios such as the one at the beginning of this article are not uncommon, it is critical to understand that parents of children with autism generally share the values of medical professionals. Both parents and pediatricians are seeking amelioration of symptoms and the best possible quality of life for children, and nearly all agree on the potential value of medical and behavioral interventions.

In this article, we have attempted to note the many instances when parents have contributed to the expert understanding of autism: as acute observers of their children, noting nuances of behavior and treatment response that might be invisible to pediatricians; as researchers, identifying new avenues of clinical and basic science; and as advocates, disseminating knowledge and generating resources for further research. It is important that physicians be clear with parents about the strengths and weaknesses of both standard medical knowledge and alternative treatments. The history of parental involvement with research also suggests that each parent of a child with autism is a valuable source of information and localized expertise, not only about their own child but about the disorder of autism in general.

Most importantly, parents and caregivers will be crucial in making any treatment effective on the level of the individual child. It is unlikely that autism will respond to a single decisive intervention; it is far more likely that both pediatricians and parents will be operating in the realm of partial and incremental improvements for quite some time. We hope that this article helps professionals to understand the historical contributions of families of
children with autism and to thus foster the collaborative relationship that is critical to improving the care of persons with autism.

Accepted for Publication: November 16, 2006.

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Author Contributions: Study concept and design: Silverman and Brosco. Acquisition of data: Silverman. Analysis and interpretation of data: Silverman and Brosco. Drafting of the manuscript: Silverman. Critical revision of the manuscript for important intellectual content: Silverman and Brosco. Obtained funding: Silverman. Administrative, technical, and material support: Silverman.

Financial Disclosure: None reported.

Funding/Support: This work was supported by a Jacob K. Javits Fellowship from the US Department of Education (Dr Silverman), a Mellon Postdoctoral Fellowship (Dr Silverman), and grant 033954 from the Robert Wood Johnson Foundation General Scholar Program (Dr Brosco).

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Announcement

Trial Registration Required. In concert with the International Committee of Medical Journal Editors (ICMJE), Archives of Pediatrics and Adolescent Medicine will require, as a condition of consideration for publication, registration of all trials in a public trials registry (such as http://ClinicalTrials.gov). Trials must be registered at or before the onset of patient enrollment. This policy applies to any clinical trial starting enrollment after July 1, 2005. For trials that began enrollment before this date, registration will be required by September 13, 2005, before considering the trial for publication. The trial registration number should be supplied at the time of submission.

For details about this new policy, and for information on how the ICMJE defines a clinical trial, see the editorials by DeAngelis et al in the September 8, 2004 (2004;292:1363-1364) and June 15, 2005 (2005;293:2927-2929) issues of JAMA. Also see the Instructions to Authors on our Web site: www.archpediatrics.com.