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Autism Research at the NICHD



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Autism Questions and Answers for Health Care Professionals

Autism is a complex, biological, developmental disability that causes problems with social interaction and communication. It is usually associated with restrictive or repetitive interests and behaviors throughout life.

In the past, little was known about this condition. It was thought to be rare, institutionalization was the main form of treatment, and it was said to be the result of “cold mothers.” Researchers have since discovered that all of these characteristics are untrue.

For a health care professional, the challenge of how to handle a patient with autism is especially difficult for a variety of reasons. This publication highlights questions commonly asked by health care professionals about autism and provides answers based on the latest autism research.

Many of the answers provided here resulted from studies conducted by the Network on the Neurobiology and Genetics of Autism: Collaborative Programs of Excellence in Autism (CPEA), a worldwide research network co-sponsored by the National Institute of Child Health and Human Development (NICHD) and the National Institute on Deafness and Other Communication Disorders (NIDCD), parts of the National Institutes of Health (NIH). The CPEA Network conducts research on the genetics and neurobiology of autism, through partnerships among more than 25 universities in the U.S., Canada, Britain, and France. As the largest single autism-specific research endeavor to date, the CPEA is trying to understand autism and its many facets. Other individual NICHD- and NIH-supported scientists are also contributing new knowledge to this cause.

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Information provided in this document about autism symptoms and diagnosis also comes from *The Screening and Diagnosis of Autism Spectrum Disorders*, an article authored by a multidisciplinary Consensus Panel assembled at the NIH in 1998. This panel, composed of seven NIH Institutes, nine professional organizations, and three autism parents' groups, reviewed over 2,500 scientific publications on screening for and diagnosing autism. The resulting article was published in the *Journal of Autism and Developmental Disabilities* in 1999 (Volume 29, Number 6, pages 439-484).

Subsequently, many medical academies, professional organizations, and parent and advocacy groups in autism, led by the American Academy of Neurology and the Child Neurology Society, met to build on the Panel's base, developing a practice parameter and providing clinical guidance for screening and diagnosing autism. The practice parameter was published in *Neurology* in 2000, and in *Pediatrics* in 2001. Other medical and professional journals are expected to publish the parameter in the future.

For a complete listing of the groups included in the Consensus Panel, or for more information about the CPEAs or autism research at the NICHD, please refer to the **Q: Where can I go for more information about autism?** section at the end of this document. The citations for the articles mentioned above are also available in this section.

Q: What is the prevalence of autism?

A: Although the exact prevalence of autism is not yet known, estimates range from one-in-500, to one-in-1,000 cases in the United States. Current statistics show that autism occurs in all racial, ethnic, and social groups, and that boys are three-to-four times more likely to be affected.

Initial studies done in the 1960s indicated that autism had a prevalence rate of four-to-five cases in 10,000, which is why the condition was thought to be rare. However, current prevalence estimates are not consistent with prevalence rates for a rare disease. Further, recent U.S. and international data point to a steep increase in prevalence of the condition.

However, many epidemiologists believe that recent changes in diagnostic criteria and in the conditions that are classified as autism spectrum disorders could account for some of the increased prevalence.

Q: What causes autism?

A: A variety of factors are thought to be involved with causing autism. Recent studies suggest a genetic element, possibly a predisposition, is involved in causing autism very early in a child's *in utero* growth. Other causes may include infectious, neurological, metabolic, and environmental factors.

Some of the researchers in the CPEA are focusing their efforts on possible genetic causes of autism. In 2000, scientists in the CPEA Network released the results of two studies that found genes were involved in autism. Additional papers were published in 2001 by CPEA researchers and other NIH-funded

scientists as part of an international consortium on genetics research. These results lead researchers to believe that some people could have a genetic predisposition that makes them more likely to develop autism. The CPEA Network and other NICHD-supported researchers are also looking into other factors that could be involved in autism, in addition to genetics. For a complete bibliography of autism-related publications from CPEA researchers and other NICHD- and NIH-funded scientists, visit the NICHD autism Web site at <http://www.nichd.nih.gov/autism/bib.cfm>.

Even though the research community is uncertain of the exact cause of autism, it is clear that the **quality of parenting is *not* a cause or a contributing factor to autism.**

Autism is a devastating disease for patients and families; any suggestion that the family caused the condition only worsens the situation.

Q: Is there a link between autism and vaccines?

A: To date there has been no conclusive, scientific evidence that any part of a vaccine, nor any combination of vaccines causes autism. There is also no conclusive data that any type of preservative (i.e., thimerisol) used during the manufacture of vaccines plays any role in causing autism. In 2001, the Institute of Medicine (IOM) and the American Academy of Pediatrics (AAP) released findings from their separate reviews of the available evidence on a possible link between vaccines and autism. Both groups found, independently of one another, that existing evidence does not support such a connection (Immunization

Safety Review Committee 2001; Halsey et al 2001).

Because there is no conclusive scientific evidence of a link between autism and vaccines, the National Immunization Program at the Centers for Disease Control and Prevention (CDC), along with the AAP and the American Academy of Family Physicians, suggest that physicians follow the recommended childhood immunization schedule that is published every year (MMWR 1998; Halsey et al 2001).

Physicians are advised to take careful family histories of all their patients to bring to light any factors that might influence their recommendations about the timing of vaccinations.

The CPEA Network, funded by the NICHD and the NIDCD, with additional funding from the CDC, is working to study autism and its relation to the measles/mumps/rubella (MMR) vaccine. CPEA researchers will compare vaccination records of groups of people with autism, to those who do not have autism, to see if the onset of autism symptoms was associated with getting the MMR vaccine or other vaccines. Lab tests in this study will also look for any signs of persistent infections that could be related to the MMR vaccine.

The NICHD fact sheet titled *Autism Research at the NICHD—Autism and the MMR Vaccine* has more information about this and other studies related to vaccines and autism. This publication and other fact sheets about autism are available on the NICHD Autism Web site, at www.nichd.nih.gov, or from the NICHD Clearinghouse at 1-800-370-2943.

Q: What is the clinical phenotype of autism?

A: Because of the similarities and differences among people with different forms of autism, health care professionals now view autism as having a broader clinical phenotype than was once thought. The expanded phenotype goes beyond the standard definition for autism, to include, as the DSM-IV states, a *range of impairments* rather than the *absolute presence or absence* of a certain behavior or symptom (DSM 1994). The DSM-IV uses the terms “pervasive developmental disorder (PDD)” and “autism spectrum disorder (ASD)” to describe five variations of autistic behavior; the International Classification of Disease (ICD), published by the World Health Organization (WHO), has eight variations of PDD.

Q: What disorders does PDD or ASD include?

A: According to the DSM-IV, the umbrella PDD or ASD category includes:

- Autistic disorder (sometimes called “classic” autism)
- Asperger syndrome
- Childhood disintegrative disorder
- Rett syndrome
- PDD Not Otherwise Specified (NOS) or atypical autism

Depending on his or her specific symptoms, a person with autism or ASD can be in any one of these categories.

In 1999, NICHD-supported researchers identified the gene responsible for Rett Syndrome, one of the conditions included in the ASD category. Rett Syndrome occurs only in girls and causes them to develop autism-like symptoms after seemingly normal development. This discovery could lead to improved detection, prevention, and treatment of Rett Syndrome.

Advances in detecting, preventing, and treating Rett Syndrome may shed light on ways to understand and treat ASDs, including those aspects of ASD that may involve regression.

Q: What is my role as a health care professional in caring for a child with autism?

A: Because diagnosis of autism is difficult, only a health care professional who specializes in the treatment of children with autism should perform a formal evaluation and diagnosis for autism. However, health care professionals who are not specialists can assist in getting a person with autism the help he or she needs.

Autism is a treatable condition. People with autism benefit from early intervention and show multiple outcomes to treatments. Health care professionals who are not autism specialists can provide the vital “first step” in the process of diagnosis—screening. By recognizing the symptoms of autism, you can refer children who exhibit these symptoms to the appropriate specialist(s) for further evaluation. By increasing your own knowledge of the common symptoms of autism, and sharing that

information with your colleagues, your staff, the families in your care, and others in the community, you can help ensure that those who need help get it.

Q: What are the symptoms of autism?

A: The symptoms of autism vary in both occurrence and severity, which makes diagnosis difficult. According to the DSM-IV, the *general* symptoms for autistic disorder (hereafter referred to as autism) include the following:

- A total of six (or more) items from a list of qualitative impairments in the following categories: **1) Social interactions** (at least two from this group), **2) Communication** (at least one from this groups), and **3) Restrictive repetitive and stereotypic patterns of behavior, interests, and activities** (at least one from this group).
- Delays or abnormal functioning in (1) social interaction, (2) language used as social communication, or (3) symbolic or imaginative play, with onset before age three.
- Symptoms that cannot be described as Rett syndrome or Childhood Disintegrative Disorder.

Please refer to the DSM-IV for a complete listing and explanation of these criteria.

Q: When is the usual onset of symptoms?

A: The symptoms of autism are usually measurable by 18 months of age, although parents and experts in autism treatment can usually detect symptoms before this time. Formal diagnoses can be made by age two, but are usually made between ages two and three, when the child shows delays in language development.

Recent studies show that at least 20 percent of children with autism experienced a “regression,” as reported by their parents. These children have a mostly normal development followed by a loss of social and communication skills. There is little information, however, about trends in such regression, such as age of onset, severity, or possible triggers.

To learn more about regression in autism, the CPEA Network, with additional funding from the CDC, is conducting research on autism, regression, and the MMR vaccine. CPEA scientists will compare vaccination records of three groups of people: those who had autism at birth, those who regressed into autism after seemingly normal development, and those who do not have autism. Through this comparison, the scientists hope to determine if regressive autism might be a definable subgroup of ASD, and if the MMR vaccine plays any role in the onset of symptoms in that subgroup.

Q: Are there any indications that require immediate evaluation for autism?

A: A child should *definitely be evaluated for autism immediately*, if he or she has:

- No babbling by 12 months
- No gesturing (pointing, waving bye-bye, etc.) by 12 months
- No single words by 16 months
- No two-word (not just echolalic) phrases by 24 months
- *Any* loss of *any* language or social skills at *any* age

Q: What other parental concerns should prompt a health care provider to evaluate a child for autism?

A: There are a number of symptoms reported by parents that should be **red flags** for pediatricians. You should evaluate a child for autism, if the parents report that the child:

- Does not respond to his/her name
- Cannot tell what he/she wants
- Has language delay(s)
- Doesn't follow directions
- Appears deaf at times
- Seems to hear sometimes, but not others
- Doesn't point or wave bye-bye
- Used to say a few words, now doesn't
- Has tantrums
- Has odd movement patterns
- Is hyperactive, uncooperative, or oppositional
- Doesn't know how to play with toys
- Gets stuck on things over and over
- Doesn't smile socially
- Seems to prefer to play alone
- Gets things for him/herself
- Is very independent
- Does things "early"
- Has poor eye contact
- Is in his/her "own world"
- Tunes people out
- Is not interested in other children
- Walks on his or her toes
- Has unusual attachments to toys or other unusual objects (i.e., always holding a certain toy or saving or holding string)
- Lines things up

Q: Do parents typically overreact when they think their child has a problem?

A: Parents are the best observers of their children’s day-to-day growth and development because they spend so much time with the children. In most cases, parents who suspect that their child has autism will present it to their child’s doctor at a well-child visit as either a suspected speech or language delay or a problem with social development (with or without a speech-language problem). Most pediatricians take parents’ concerns about speech or language delay seriously and follow up with evaluation using standard screening methods. Concerns about social development should also be taken very seriously. In the *Screening and Diagnosis of Autism Spectrum Disorders* (Filipek et al 1999), the Consensus Panel indicates,

Any concerns regarding problems with social development should always be taken seriously, as seriously as an older child’s complaint of back or chest pain. Unlike “stomachaches” and “headaches” which are common, self-limiting, and can often be treated symptomatically without a diagnostic workup, a complaint about of back or chest pain is rare and deserves investigation. Similarly, parents rarely complain of social delays or problems, so any and all such concerns should be immediately investigated. It is even more significant when parents voice additional concerns in the communication and behavior areas as well as in socialization. (452)

Children with autism and their families reap many benefits from early intervention. Treating parental concerns seriously and respectfully is important to the child, to the family, and to your relationship with them as a health care provider.

Q: How can I determine whether a parental concern actually constitutes a social or behavioral development problem?

A: Be specific when asking questions about a child’s development. The more specific the question, the more exact the response. For instance,

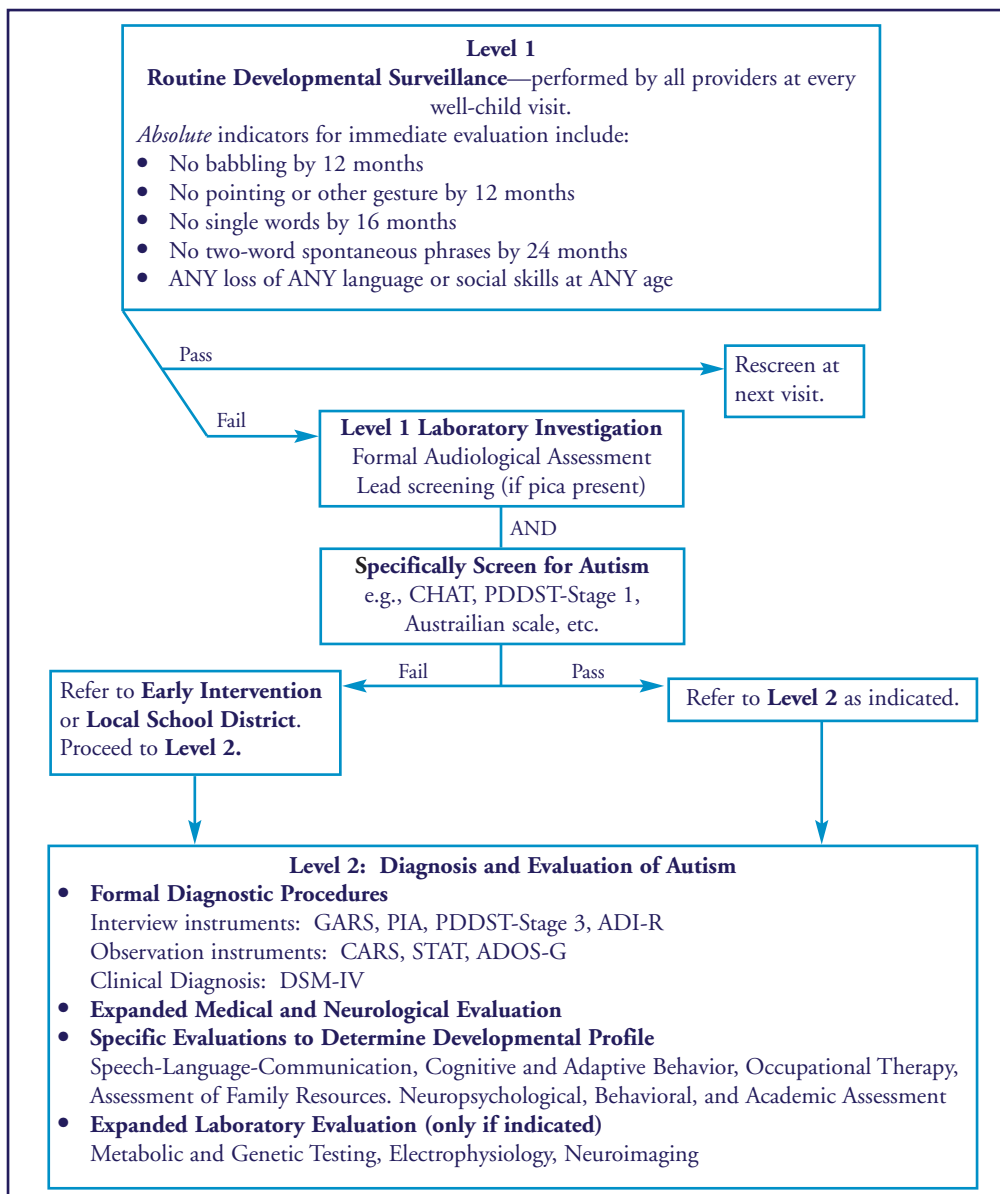
- *Does your child...*Smile in response to a smile from others?
- *Does your child...*Engage in reciprocal, back-and-forth play?
- *Does your child...*Have anything odd in his/her speech?
- *Does your child...*Avoid or have limited pretend play?

Consult the list of “Red Flags” on the previous page to identify additional questions and question topics.

Q: What is the typical process for diagnosing a child with autism?

A: A general flow chart describing the suggested screening and diagnosis process is shown below. For a more complete description, please consult the American Academy of Neurology/Child Neurology Society’s Practice Parameter (Filipek et al 2000).

Algorithm for Diagnosis of Autism



Q: Are there any screening or diagnostic tools I can use to help identify children who might need additional evaluation?

A: Yes. The Consensus Panel that wrote The Screening and Diagnosis of Autism Spectrum Disorders recommends several developmental screening tools:

- *The Ages and Stages Questionnaire*
- *The BRIGANCE® Screens*
- *The Child Development Inventories*
- *The Parents' Evaluation of Developmental Status*

In addition, there are a number of screening tools that are specific to autism, including:

- *Pervasive Developmental Disorders Screening Test (PDDST)*
- *Checklist for Autism in Toddlers (CHAT)*
- *Australian Scale for Asperger's Syndrome*

The Panel also indicates that the *Denver II* (formerly the *Denver Developmental Screening Test-Revised*) is **not recommended** as a developmental screening tool for autism.

Health care professionals should perform developmental screening at every well-baby and well-child visit, through the preschool years. If a child shows any of the characteristics listed earlier as *definite* indications for further evaluation, the child and his or her family should see a specialist in child development or

another appropriate health care professional to begin testing for autism.

For a complete list and description of these diagnostic tools, consult the American Academy of Neurology/Child Neurology Society's Practice Parameter (Filipek et al 2000).

Q: What do I do once a child in my care is diagnosed with autism?

A: According to Public Law 105-17: Individuals with Disabilities Act-IDEA (1997), the child's primary care provider is required to give the family a referral to an early intervention service. Services vary by state. The family with a child under age three should be referred to the community zero-to-three service system. The local school district must provide free and appropriate services for a family with a child three or older. You should also make sure the child and his or her family follow-up the diagnosis with appropriate care and treatment.

Q: Is there a cure for autism?

A: To date, there is no cure for autism, and children do not outgrow the symptoms. However, there are a number of treatments available that can help people with autism and their families lead more normal lives.

Individualized, intensive behavioral interventions, preferably beginning before age two-and-a-half or three, have provided the most dramatic and lasting improvements, in some cases resulting in normal to near-normal

functioning. Any delay in diagnosis and referral to services can diminish the benefits of intensive, early interventions. In addition, there are pharmacological treatments that do not cure autism, but may relieve some of the symptoms.

Q: Are there treatments for autism?

A: Many families of children and adults with autism are finding new hope from a variety of treatments for autism. These treatments include (but are not limited to):

- **Individualized Education Programs (IEPs)** are an effective way to prevent problem behaviors typically related to autism. IEPs involve a variety of interventions, including some of those mentioned below, and are designed to help a child or adult with autism to overcome his or her specific problems. Children with autism seem to respond very well to IEPs that are properly designed and systematically implemented.
- **Comprehensive Treatment Programs** encompass a number of different theories about treating autism. These programs range from specific methods of learning, to applied behavior analysis, to reaching certain developmental goals. In general, children need to be in this type of program for 15-40 hours a week, for two years or more, to change their behaviors and experience benefits.
- **Applied Behavior Analysis (ABA)** generally focuses on reducing specific problem behaviors and teaching new skills. Recently, ABA programs have broadened their scope to

include interventions for use before or between episodes of problem behaviors, in addition to interventions that are useful during or after these episodes. By showing children or adults with autism how to handle things like a change in schedule, furniture that has been moved, and meeting new people, ABA can reduce the chances that these situations will trigger problem behaviors.

- **Positive Behavioral Interventions and Support (PBS)** is an approach that tries to increase positive behaviors, decrease problem behavior, and improve the child or adult’s lifestyle. The PBS method looks at the interactions between people with autism, their environments, their behavior, and their learning processes to try and develop the best lifestyle for them.
- **Pharmacological treatments** can also be effective in improving the behavior or abilities of a person with autism. In general, these medications are called “psychoactive” because the drugs affect the brain of a person with autism. Medication is often used to deal with a specific behavior, like reducing self-injurious behavior or aggression, which may allow the person with autism to focus on other things, like learning.

When considering treatment options, it is important that patients with autism be evaluated for co-morbid, potentially treatable conditions, such as seizures, allergies, gastrointestinal problems, or sleep disorders. Treatment of these co-morbid features may not cure autism, but can lead to improvements in quality-of-life for both patients and their families.

References

Diagnostic and Statistical Manual of Mental Disorders, 4th edition. (1994) Washington DC: American Psychiatric Association.

Filipek PA, Accardo PJ, Ashwal S, Baranek GT, Cook, Jr EH, Dawson G, Gordon B, Gravel JS, Johnson CP, Kallen RJ, Levy SE, Minshew NJ, Ozonoff S, Prizant BM, Rapin I, Rogers SJ, Stone WL, Teplin SW, Tuchman RF, and Volkmar FR. (1999). The screening and diagnosis of autism spectrum disorders. *Journal of Autism and Developmental Disabilities*, 29(6):439-484. This article provides more detailed explanations, descriptions, and rationales for most of the recommendations presented in this document.

The Consensus Panel responsible for this article included the following Institutes and organizations: American Academy of Audiology; American Academy of Neurology; American Academy of Child and Adolescent Psychiatry; American Academy of Pediatrics; American Academy of Mental Retardation; American Association on Mental Retardation; American Psychiatric Association; American Psychological Association; American Psychological Society; American Speech-Language-Hearing Association; Child Neurology Society; Autism Society of America; Autism Society of America Foundation; Autism Research Institute; Autism National Committee; Cure Autism Now; National Alliance for Autism Research; and FRAXA (Fragile X Research Foundation), with liaisons from the NICHD, the NIDCD, the National Institute of Mental Health, and the National Institute of Neurological Disorders and Stroke. For a more complete understanding of the screening tools and methods presented here, consult the full text of the article.

Filipek PA, Accardo PJ, Ashwal S, Baranek GT, Cook, Jr EH, Dawson G, Gordon B, Gravel JS, Johnson CP, Kallen RJ, Levy SE, Minshew NJ, Ozonoff S, Prizant BM, Rapin I, Rogers SJ, Stone WL, Teplin SW, Tuchman RF, and Volkmar FR. (2000). Practice parameter: screening and Diagnosis of autism; Report of the Quality Standards Subcommittee of the American Academy of Neurology and the Child Neurology Society. *Neurology*, 55:468-479.

This practice parameter also provides detailed explanations, descriptions, and rationales for much of the information presented in this document. You may also want to consult your own professional organization for more information on its practice parameter.

Halsey N, Hyman S, and The Conference Writing Panel. (2001). Measles-mumps-rubella vaccine and autistic spectrum disorders. *Pediatrics*, 107(5): e84-107

Immunization Safety Review Committee, Board on Health Promotion and Disease Prevention, Institute of Medicine. (2001). *Immunization safety review: measles-mumps-rubella vaccine and autism*. Stratton K, Gable A, Shetty P, and McCormick M (Eds.). Washington DC: National Academy Press.

MMWR. (1998). Measles, mumps, and rubella vaccine use and strategies for elimination of measles, rubella, and congenital rubella syndrome and control mumps: recommendations of the advisory committee on immunization practices. *MMWR*, 47(RR8).

Q: Where can I go for more information about autism?

A: The NICHD Clearinghouse provides information on autism and autism research, and on other topics related to the health of children, adults, and families. The information specialists at the NICHD Clearinghouse are available at:

Mail: PO Box 3006, Rockville, MD 20847

Phone: 1-800-370-2943

Fax: 301-984-1473

Email: NICHDClearinghouse@mail.nih.gov

The NICHD Autism Web Page, www.nichd.nih.gov/autism, offers information on NICHD autism research, including the CPEA Network, current grants and funding mechanisms, ongoing clinical trials, and the NIH Autism Coordinating Committee. A list of currently funded grants and project abstracts, as well as a complete bibliography of publications from CPEA researchers and other NICHD- and NIH-supported scientists is also available through this Web site. With a variety of information on topics related to autism, the NICHD Autism Web site is a good place to start your search for information.

Q: Are there other autism information resources I can consult?

A: The National Library of Medicine (NLM) offers the following features:

- MEDLINEplus, serves as a gateway to all the NIH resources about autism, including the NICHD autism Web page and Web sites of other Institutes that are researching different aspects of autism. It also provides links to recent news articles about autism, as well as to non-government organizations with autism as their focus. To access MEDLINEplus, go to www.nlm.nih.gov/medlineplus, and do a search for “autism.”
- MEDLINE, allows you to search hundreds of medical journals, millions of medical abstracts, and the NLM Medical Subject Headings;
- Loansome Doc, allows you to request printed copies of journal articles for a fee.

