

Angelman Syndrome, a Parent's Brochure for Parents

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Your child has been diagnosed with Angelman Syndrome. If your pediatrician, neurologist or geneticist has information for you about AS, it is probably bad news. You may be told that your child is profoundly mentally retarded and will never achieve any level of independence or "quality of life." Your heart sinks.

If your child is a toddler or older, the diagnosis may come as a relief. You probably know already that something is "not right," and finding out what that is can help you help your child. You have had time to get to know your child, and the diagnosis doesn't change what you already know about your child. There may be comfort in learning that AS does not generally endanger health or shorten lives, and it is good to know what to watch for in terms of health issues like scoliosis and seizures.

If your child is an infant, however, you may feel a great deal of despair on hearing this diagnosis. Because AS is little-known among most medical professionals, the information you are given may be seriously out of date, based on studies done on people who were institutionalized as infants, who did not have the benefits of therapeutic interventions available today. You may be told-or led to believe-that your child will be profoundly mentally retarded, that he will never achieve any level of independence. It is important for you to know that better, more up-to-date information is available, information which may help you deal with your fears and disappointment. You will probably even want to share information with your child's doctor. Parents of children with AS are often in the position of "educating" medical professionals about the syndrome.

Or you may be one of the many parents who have a child with a disability but without a diagnosis. Not all children with AS have definite diagnoses, either; their chromosomal studies show no abnormalities, but their behaviors-even their appearance-allows a "clinical" diagnosis of AS to be made. In the beginning, Dr. Harry Angelman diagnosed children with AS (whom he originally called "Puppet Children") based only on clinical evidence.

Why is a diagnosis important? Many pediatricians even discourage parents who want to have more studies done to try to identify exactly what is wrong with their child. They may feel that this is a form of denial, that the parents believe they will find that their child has some "curable" condition. This may be true, of course, but a diagnosis of AS can be of vital importance to the person with AS and to her family and caregivers.

With no diagnosis, we can not plan for the future. We know that our children with AS will have a normal life expectancy, that there need be no loss of function, that our children keep learning into at least early adulthood. It is important because we know that our children will

need care for their entire lives, that they will never be completely independent. A diagnosis of AS helps us know what sorts of therapies and other interventions our children will benefit from most, and helps us avoid wasting time and effort on those which may not be at all helpful. With a diagnosis of AS, we know we will need to have an ongoing relationship with a neurologist, and that seizures are always a possibility.

Without a diagnosis, no matter what the disorder, parents tend to blame themselves and each other, and even other people involved such as the midwife, for what is wrong with their child. We know this is unfair and unreasonable, but we can't help fretting about "what if." A diagnosis of AS lets us know that the disability was nothing we or anyone else caused, nothing we could have prevented, nothing we did wrong. And it can guide genetic counseling for future pregnancies.

Knowing that a child has AS can help the parents find needed emotional and financial supports as well. In most countries, having a diagnosis helps social agencies find resources needed by families. Knowing the name of our children's disability can guide us to support groups of parents, where we find emotional and practical advice. Parent groups arrange for conferences around the world, where scientists and other parents offer help and information to one another.

This booklet is written by an experienced mother of a child who has Angelman Syndrome. I hope that it will be helpful to you. I have learned so very much from talking to other parents, either in person, on the phone, or on the Angelman syndrome electronic mailing list, and from attending conferences sponsored by my local AS group, the Angelman Syndrome Foundation (USA) and the IASO, that I want to share with you, especially if you are new to AS. I hope that this booklet will be of help and comfort to you.

Any material you may have been given by your physician or found for yourself probably covers the broad range of possibilities that exist in the child with AS. It is important for you to understand, and believe, that there is no child who has all these characteristics; and none of the characteristics applies to every child with AS.

Your child is an individual. He or she shares a medical diagnosis which will have a great impact on your child's life and the life of your family, but just as every person who has diabetes is not exactly like every other person who has diabetes, your child will not be exactly like any other child with AS. There are similarities; just as all diabetics have to control their blood sugar, all children with AS have to find ways to function in their own lives.

"Absent or little speech" may be the most distressing AS characteristic for parents of newly-diagnosed children. While it is true that spoken language is rare in children with AS (although not as rare as was once believed), it is important to know that every child with AS communicates. Further, receptive speech ability (that is, understanding what is said) is much higher than expressive ability in children with AS. Many children understand a great deal of what is said; all children understand some of what is said. It is easy for parents to fail to talk enough to a child with AS; the lack of verbal feedback can be discouraging, and we sometimes do not do all the "baby talk" we would otherwise do. It is good for your child, even as a baby, to hear spoken-and sung-language, and I think it's good for you, too.

The children with AS communicate in a variety of ways. Some children use sign language, modified to their motor capabilities; some do speak fairly intelligibly but use supplemental means such as picture boards. Some children are able to point out written words. Many

children use electronic “talkers.” Nearly all children with AS use gestures, either with their own hands or with someone else’s!

It will be important to work closely with your child’s speech and language therapist to find the best ways for your child to communicate. It helps if the therapist is open to trying all kinds of communication. Sometimes therapists will express concern that using more than one type of communication will be confusing to the child with AS. Most of our children, however, already use more than one way to communicate, if we are able to understand it.

Because many children with AS have weak trunk muscles, they may miss developmental milestones that rely on gross motor abilities. At an early age, your child may be unable to bring himself to sitting and may fall to the side when he is put into a sitting position. He may slide forward out of his chair without some sort of restraining device. Many parents have found that hippotherapy (therapeutic horseback riding) is helpful to strengthen these muscles. Your child’s physical/ physio therapist can probably recommend exercises to help as well.

Some children with AS do not walk. Most children are mobile, however, and a great many do walk independently. Your child may need an assistive device, such as molded leg braces or a walker, or they may walk on their own. Walking usually is delayed in children with AS, often developing after the third birthday. If your child is going to be able to walk, she will walk when she is ready. Other than praise and encouragement, providing opportunities to walk, and working with your child’s therapists if assistive devices are needed, there is not much parents can do to speed up this process.

If your child is ambulatory, it is important to keep him/her walking. This may be difficult, especially when longer distances are involved—it’s easier on everyone to put the child in a stroller or wheelchair. If your child is allowed to sit and ride when they could be walking, they are in danger of developing contractures at the hips and knees, which may make it difficult for them to walk at all. Keeping your child active may help with hyperactivity and sleep problems as well.

Many children with AS are hyperactive. Various medications and behavioral interventions can help with this problem; there is no one “best” treatment for behaviors common in children with AS. Children with AS respond well to praise and other forms of positive reinforcement. It is helpful to work with a behavior specialist if one is available to you. Most important, however, is your understanding that this maddening behavior is not something your child is doing to make you miserable. It is easy for us parents to forget that our children must work very hard to restrain their impulsive behavior, and so we do not praise them for doing things that we would consider matter-of-course in other children. It is impossible to overdo praising our children.

Seizures are a frightening prospect for parents who have learned that their child has AS. Some children, however, never have seizures. Some children have a serious problem with seizures, and many different medications must be tried to find the right “mix” of ingredients and dosage. Some children with seizures respond well to the ketogenic diet. If your child has seizures, it is helpful to have an open, comfortable relationship with your neurologist so that you can discuss trying different approaches to controlling the seizures. Many children with AS fall between being seizure-free and having many seizures. They may take medication daily to prevent seizures; they may have the occasional seizure brought on by illness or stress, but the seizures are not a major factor in their lives.

Dr. Terry Hutchison, who is an expert on epilepsy in AS, describes the varieties of seizures our children may have. Many parents do not recognize seizures, and it is recommended that all parents take a look at Dr. Hutchison's "primer" on seizures (http://www.angelman.org/seizures_in_children_with_as.htm). He also addresses the various medications used for seizure control. This will be useful information to share with your child's neurologist.

Many children with AS have trouble sleeping through the night. Some children may resist falling asleep. More commonly, children with AS waken at 2 or 4 a.m. and may not go back to sleep. In addition to wreaking havoc with the household, sleep deprivation is a common trigger for seizures in children who may otherwise be seizure-free. Some successful strategies parents use include confining the child at night so that the rest of the household can continue to (try to!) sleep-there are commercially available enclosed beds where the child can be awake alone, and safe. Some parents have built their own versions of these beds, which is an option if neither the local health services nor your insurance will help pay for a commercially made bed.

Some parents get into bed with their child when she wakes, or take the child into the parent's bed. Some parents have successfully trained their children to play quietly in their rooms at night. Most parents wisely take measures to ensure the safety of the children at night, locking or blocking bedroom doors, installing alarms, etc. It can be very dangerous-even deadly-for our children to be awake alone at night and able to roam inside (and outside) the house or apartment. Keeping your child safe is not child abuse. Allowing him to wander alone at night is.

As with hyperactivity, it is important for parents to understand that sleep difficulties are not something our kids do "on purpose." Studies have shown that children with AS have an abnormal pattern of secreting melatonin, the hormone that helps us fall asleep and stay asleep. Some children are helped to sleep by receiving a melatonin supplement before bedtime; some are not. Some children who are helped are not helped all the time. An important paper by Dr. Joseph Wagstaff about the use of melatonin in children with AS is at <http://asclepius.com/iaso/wagstaffsleep.html>. You may want to show this to your child's doctor. Medications such as antihistamines, which are commonly used in other children, may have dangerous effects in children with AS, particularly in lowering seizure thresholds. Talk to your physician before trying any of these, even over the counter medications.

Probably the most difficult part of caring for a child with AS is taking care of yourself. If your child does not sleep well, or long enough, you may find yourself too tired to be a good parent. If there is another adult in the household, it is helpful to plan "night duty" so that everyone gets at least some sleep. You may want to try giving your child medication to help induce sleep, although experience has shown that normally prescribed "sleep" medications do not work well with our children. This must be addressed on an individual basis. What works for one child may not work for another.

It is also helpful, I have found, to lower your standards for housekeeping. Even hiring someone to come in weekly to clean is not very useful because the mess reappears almost immediately. Better to strip down the rooms your child with AS uses most, so that keeping the rest of the house tidy is possible. If you can, it helps to buy extra sets of underwear, socks and shirts for everyone in the household, so that laundry can be put off for a day or so if necessary. Beth Sturr (<http://www.bethsturr.com>) offers much practical advice for everyday problems.

When it comes to personal care, a few children with AS are virtually independent in feeding, washing, dressing and toileting themselves. Some children, however, do not learn to use the toilet and must wear diapers (nappies) day and night, and many fall in between the two extremes. Your child will probably be toilet trained to some degree, although later than you would like. Your child will at least be able to cooperate in dressing and other aspects of personal care. Many parents have been successful with "schedule training," where the child is placed on the toilet at specific intervals during the day, waiting for success. School is a good place for schedule training to begin. Your child's teacher should be able to guide you in this matter.

Education and social services are two areas where parents, while dependent on others to deliver the services, have a good deal of power in determining what the services will be. You may have to spend time and energy to obtain what you feel your child needs, but the expenditures will be worth it. Wherever you live, you will probably find yourself spending much more time and energy on your child's educational needs than you would like to. It is unfair, to say the least-we are already working hard, probably on too little sleep-but once you establish yourself as a presence at your child's school, your child will benefit in untold ways.

It is useful to educate yourself as much as you can about AS. There are excellent links at the IASO website that can guide your search, and Frank van Hof has an excellent annotated directory of websites that will help you learn. Once you become an "expert," your work with teachers, therapists, neurologists and others will be much easier and more efficient.

It is my belief-shared by many experienced parents-that other parents are the best source of practical information for parents of children with AS. I hope you will want to join the Angelman syndrome electronic mailing list www.asclepius.com/iaso/asmaillists.html, where parents and caregivers, even some therapists and teachers, chat informally about daily life and how to cope. Important scientific information is also shared on the list. We are an international group.

From my exposure to other families and children with AS, I have come to believe that our children have greater potential to learn and grow than is commonly accorded them by medical and educational professionals.

I have been overwhelmed by the caring that exists among the AS families I know, and by the dedication of parents and caregivers to making life better for all our children.

What I know for sure about Angelman Syndrome:

Your child will be exceptionally loving and sweet natured. Your child will drive you crazy at least twice a day. Your child needs you to expect the best from him and for him. In return, your child will help you become a better person in ways you never dreamed.