

Complications of a Medically Complicated Child

There is a phenomenon in psychology that states that active observers—people who are involved in an action—have a great need to predict and control a situation. This couldn't hold more true for me, the mother of a child who is "medically complicated." My 20-month-old son is the actor, and I am the active observer. My son doesn't realize how unusual his life is, but I know that running from a meeting to a hospital appointment one or more times each week is not usual for many parents. I yearn to control this chaotic frenzy, but until Sam's health problems are under control, our lives will not be, either.

"Is life always like this?" our social worker asked us. She had been assigned to us by the state because our son is receiving physical and occupational therapies as part of the early intervention program. Having a social worker is perceived to be "a good thing," but it is one more appointment I need to arrange in an already crowded schedule. She also suggested we attend a support group for parents of medically complicated children. While a support group may be helpful, I don't really feel that I have the time to attend.

It is assumed that our son Sam has a connective tissue disorder. My husband and I had always thought that there was something not quite right about Sam: He cried a lot during his early months; he made no attempt to move or roll over; and he seemed weak. His thyroid screen was positive, but upon repeating it, it showed normal results. He was born with two large hematomas on his head, but the x-rays indicated that there were no abnormalities present. Sam developed a case of thrush that lasted for 5 months. After a normal immunologic evaluation, it finally cleared up without intervention. When he was 6 months old, I discovered he had several pubic hairs, but the endocrine tests that followed also showed normal results. We tried to laugh and say, "There is always something going on with Sam."

When Sam was 9 months old, we became very concerned with how his feet turned up and toward the ankle, and how his legs turned out in a bowlegged appearance. He was also developmentally delayed. At a well-visit, our pediatrician also noticed these problems. We were referred to an orthopedic surgeon, who assumed the problem was due to unusual positioning in utero, and referred Sam to physical therapy. Physical therapy helped a little, but it became clear that a 2-month fix was not going to help our son in the long term.

Later, our pediatrician caught a glimpse of Sam's left eye turning outward. Assuming this was strabismus, she referred us to a pediatric ophthalmologist for further examination. It was on 16 July 2002 that we found out that Sam had bilateral ectopia lentis. Sam's lenses in both eyes were not located behind the pupil and iris as they should have been, but were displaced outward and upward. As a

result, Sam was focusing through the curved part of the lenses, and he was highly myopic. One reason that Sam had not wanted to move very much became clear: He couldn't see beyond what was literally in front of his face. While this information was disturbing enough, we learned that dislocated lenses were often associated with genetic syndromes, such as the Marfan syndrome, homocystinuria, and the Weill–Marchesani syndrome. The latter two were unfamiliar to me, but I knew a woman whose brother had died of the Marfan syndrome, and that was all I could focus on for several days.

Such began the life of being a parent of a medically complicated child. We learned that we needed to become our son's biggest advocate in a confusing health care system. We learned all we could about dislocated lenses and associated syndromes through the Internet, through colleagues, and through our pediatrician, who, in this HMO world, gave more of her time to us than is imaginable. When we saw a vitreoretinologist who wanted to remove Sam's lenses, and we instinctively felt that this was not right, we contacted the National Institutes of Health and Johns Hopkins University and learned that if Sam did have the Marfan syndrome, removing the lenses was not the proper intervention. We immediately canceled the surgery, and we then found a leading pediatric ophthalmologist who specialized in genetic conditions. Our pediatrician readily agreed to refer us to this specialist; we were fit into his busy schedule within a week, and we learned that the first strategy to follow was providing Sam with corrective lenses. Days after this first visit, Sam received his glasses. He wasn't happy about wearing them at first, but within minutes, he looked around and said "Ooohhh." He was 12 months old, and he was seeing the world clearly for the first time.

We recently moved to a large, urban area, and this has corresponded to excellent medical care. Sam's diagnosis is not yet defined: He does have something, although no one knows what. Still, it is difficult for most of the physicians we see to realize the full spectrum of what we are dealing with. They view Sam through the lens of their own specialty: For example, two orthopedic surgeons think Sam has the Stickler syndrome; the otolaryngologist does not. The cardiologist and ophthalmologist think he has the Marfan syndrome. The physiatrist and neurologist think that he is doing well; the geneticist is concerned. Sam recently developed several inguinal hernias, thought to be common in people with connective tissue disorders. But Sam does not fit any one pattern clearly. Gene testing can be performed, but at \$2800 per test, the geneticists are under pressure from the health insurance company to do this only when there is clear clinical evidence that a particular syndrome may be indicated. After nearly 6 months of weekly visits to specialists, we have now been given permis-

sion for karyotyping. In May 2003, Sam was tested for the Marfan syndrome. Soon, we may know more, or no more.

Apart from a diagnosis, we need Sam to receive treatment to improve his functional status. As a psychologist, I can also see how important it is for Sam to achieve. I watch him make every effort to bend down and reach a fallen object; I can follow his eyes and see him planning his strategy to get across a room without letting go of a chair, a wall, or a table. Physicians X and Y both recommended that Sam start using corrective cable braces to turn his legs inward; however, Physician X stated that he should start using them once he starts to walk, as the cable braces will slow down his progress in walking; Physician Y stated that Sam should start using the cable braces now, and that it is more important for Sam to correct his muscle problems before he begins to walk. Who is right? Interpreting medical advice is among the challenges we face in parenting a medically complicated child.

An integrated health care system in which physicians of various specialties meet to discuss difficult cases may be one way in which medically complicated children can be best served. When Sam was diagnosed with ectopia lentis, the natural next step was for him to have an echocardiogram, given the association between dislocated lenses and the Marfan syndrome. At this appointment, we asked the cardiologist if Sam's difficulties with breathing and excessive sweating could be related to a heart problem. He dismissed this, told us that Sam's aortic root was large but not abnormally so and that no cardiac problems existed. Six months later, in our new city, Sam had a follow-up echocardiogram to recheck the aortic root size. It was then that a patent ductus arteriosus was diagnosed: This congenital heart defect may be responsible for his grunted breathing, excessive sweating, and poor weight gain. The difficulty in diagnosing this may have been due to fragmented health care. At the time of the second echocardiogram, Sam had seen many more specialty physicians, and perhaps having more input from other physicians led to a more thorough

exam in the latter case, which resulted in the detection of the patent ductus arteriosus.

Physicians struggle to determine Sam's diagnosis; therapists struggle to get Sam to reach for that ball, to turn those knees in, to take an unaided step; but we, as parents of a medically complicated child, struggle with much more. I coordinate Sam's medical records so that every physician knows what every other physician is thinking. Most physicians seem grateful for this. I try to arrange multiple procedures with multiple surgeons on the same day so that Sam will undergo anesthesia as little as possible. Many surgeons seem to want this to happen, but their scheduling staff is not always as accommodating. I consult with our daycare center to determine how Sam can best be served next year in a classroom where everyone is walking but he may not be. I meet with our daughter's teachers to discuss her behavioral problems, possible signs of the stress she feels. I struggle with keeping up with my work when I need to take off so much time to attend medical appointments. While I know that Sam is not any physician's only patient, I wish sometimes that they and the office staff would attempt to act like it. Recently, a pediatric surgeon asked me how I was coping with all of Sam's complications. It was the first time any physician had asked me how I felt. I wish it happened more often. But more important, I wish that physicians had a better system in which they could ask their colleagues of other specialties how *they* felt about a medically complicated child.

Rani Ghose, PhD
Bedford, MA 01730

Note: Names have been changed to protect the identities of the patient and the treating physicians.

Requests for Single Reprints: Rani Ghose, PhD; e-mail, rani_ghose@hotmail.com.

Ann Intern Med. 2003;139:301-302.