

Health Care Guidelines The Down Syndrome Health Care Guidelines follow and individual's development from birth through adulthood and provide information about potential health concerns at each stage. They are compiled by the Down Syndrome Medical Interest Group, a national affiliation of health care providers who specialize in caring for individuals with Down syndrome. The guidelines help define the standards of quality care for individuals with Down syndrome. In addition to specific recommendations for screening tests, they include information about the kinds of medical conditions that individuals with Down syndrome are at risk for and suggestions for early intervention, diet and exercise, and other issues across the lifespan. T

he late William I. Cohen, M.D., specialized in developmental and behavioral pediatrics in the Child Development Unit of the Children's Hospital of Pittsburgh. He was director of the Down Syndrome Center of Western Pennsylvania and an associate professor of pediatrics and psychiatry at the University of Pittsburgh School of Medicine. Dr. Cohen also co-founded the Down Syndrome Medical Interest Group and was medical editor of

Down Syndrome Quarterly

. He edited the "Health Care Guidelines for Individuals with Down Syndrome: 1999 Revision," published in

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to watch a presentation by Dr. Cohen on health concerns of people with Down syndrome

William I. Cohen, M.D. on the Down Syndrome Health Care Guidelines □ ***The Health Care Guidelines for individuals with Down syndrome were written for medical and health care professionals, as well as educators, but they also are designed as a resource for parents and other caregivers. Why is this document such an important tool?***

The importance of the document is to support parents in getting the screening tests done that we believe are important to keep children and adults with Down syndrome healthy. Pediatricians may have between three and five children with Down syndrome in their practice, and although they may be knowledgeable about the current recommendations, I don't expect them to be able to keep up in the same way as parents, who really become experts about the needs of their

children. And even though we write about and present these issues in a variety of forums - at scientific meetings and continuing medical-education conferences - the issue may not be as compelling for physicians as it is for parents.

The "Health Care Guidelines" are supposed to help define for parents what is needed, so they can communicate with their primary-care physician and say, "This is what is recommended. This is what we need to do." It is a helpful tool for physicians, too, because it defines the medical vulnerabilities and the necessary screenings.

We designed the guidelines to come into physicians' offices through the professional circuit, but also through families. If I send out something to all the physicians in our area, it may or may not get filed in a place that's going to be useful. But when Mrs. Smith comes in with Billy and has the document in her hand, it goes into Billy's chart where it needs to be.

What is the history of the guidelines? Why were they developed and how long have they been in use?

The first person to begin to define the needs of individuals with Down syndrome in terms of health and screening was Dr. Mary Coleman. This goes back to the 1970s, when she began to define the medical vulnerabilities and describe what tests should be done on a regular basis. They were subsequently published in Down Syndrome Papers and Abstracts for Professionals, which was basically a newsletter. They were revised, generally, every couple of years. Dr. Nancy Roizen was editor for a number of revisions. In 1992, together with the Ohio-Western Pennsylvania Down Syndrome Network, we undertook the revision. Subsequently, the Down Syndrome Medical Interest Group, a group of health professionals, took over the editorship in 1996.

It's interesting to note that you can go around the world and find that national Down syndrome organizations have very similar documents.

The American Academy of Pediatrics, under the auspices of the Committee on Genetics, also issues health-supervision guidelines for Down syndrome and other genetic disorders. The most recent revision, in 1999, by the Down Syndrome Medical Interest Group, was coordinated with the American Academy of Pediatrics, so they now take on the weight of practice guidelines.

That was a very important step.

The Down Syndrome Medical Interest Group will continue to be consultants to the Committee on Genetics when they undertake their next revision.

We are now preparing that revision. Individuals with particular expertise or interest are asked to review the current recommendations and to see what the evidence suggests in terms of changes or additions. Usually the changes are small and accretional as opposed to global and far-reaching.

In addition to specific recommendations for screening tests, there is information about the kinds of medical conditions that individuals with Down syndrome are vulnerable or susceptible to. One of the conditions of particular importance is obstructive sleep apnea. Sleep problems are quite common, and although there is not a screening mechanism per se, the guidelines talk about the importance of asking questions about that and to consider obstructive sleep apnea as a potential contributor to a variety of learning or behavioral problems.

The guidelines offer a scientific discussion of the problem as background, above and beyond the recommendations for periodic screening.

What advice can you give to new or expectant parents about selecting a pediatrician and other health care professionals to work with their child with Down syndrome?

That's a very interesting question, and it comes up very often when we see patients in the Down Syndrome Center. The notion that many folks would have is that they need to find someone who's very knowledgeable; that it really is going to help them to have somebody who is an expert on Down syndrome. In my experience, some of the people who believe they may be experts on Down syndrome may not be current. They may be enamored of their expertise and fail to listen to parents about what may be different with their child.

A mother once came to me with a new baby and said, "My doctor is a family physician, and he said that he doesn't know anything about Down syndrome." I anticipated that she had reacted to

this as if it was a problem, but she went on to say, "And so he told me that any information I could bring him would be so appreciated." He, indeed, is the kind of physician you want-one who is open to partnering with a family. The knowledge that someone has is necessary, but not sufficient. In fact, not having the knowledge is less of a problem than believing you have it.

What we need when we have a child with special health care needs is someone who will collaborate-who is open to listening to parents and seeing them as partners. We need to know how to identify someone with that style. One of the best ways to find a pediatrician is to ask families of other children with Down syndrome in the area. Going to parent support groups can be very helpful.

The guidelines - even in their summarized form - are very complex and scientific. What is the best way for parents and other caregivers to educate themselves about the many issues they address?

This may be a matter of formatting; it may have become too dense. We've already talked about a "Headline News" version for the next version, instead of "The MacNeil Lehrer Report." On the other hand, parents don't want to get half-information. They would rather have it all and then sift through it. When we see new babies, one parent may want all the information and will enjoy getting as much background they can, whereas another family member may be overwhelmed. We like to try and let people know that they can choose how much to read or not read. It's very rare that you get a baby with an owner's manual.

And there's good news and bad news in the guidelines. We talk about things that can happen, but rarely do occur. Dr. Allen Crocker from Boston Children's Hospital has talked about "ghosts," which terrify families. These are things that occur very, very rarely but do happen with a slightly greater incidence in children with Down syndrome. Still, they linger out there even though the likelihood is low.

We encourage parents to do what's comfortable for them. The important thing is that the information is there for them and they can choose to use it as they want. To say, "Well, we're going to dumb it down" would be insulting to the parents. There are a lot of sophisticated parents out there. It's fascinating how knowledgeable the family of a child around six weeks old can be about this condition. I point this out to the residents, but then I say to them, "Well, of course, all they have to learn is one disorder and you have to learn the whole textbook in pediatrics." But it is pretty impressive because the need to know is so great.

I am trying to foster in pediatric residents the idea that these parents are going to be their allies. They should learn to collaborate with them and listen to what they have to say, and also learn to prioritize based on what the family's issues are.

The guidelines have very specific recommendations for five age groups: newborn, infancy, childhood, adolescence and adulthood. Is it realistic to expect a health care professional to maintain such a high degree of diligence?

I don't think they're onerous. We're talking about hearing screening every six months to about three years, vision screening annually, thyroid function testing annually, celiac screening at about two years of age, neck x-ray between three and five. And then being aware that there are some vulnerabilities to some other conditions, and not saying, "Whatever it is, it's just the Down syndrome."

The whole point of the guidelines, in a sense, is to overcome the notion that "if anything is going wrong, it's Down syndrome" rather than a treatable and definable condition. That's unacceptable.

What effort is being made to inform health care professionals about the need for specific and proactive treatment for people with Down syndrome throughout their lives?

The health supervision guidelines from the American Academy of Pediatrics are published in Pediatrics. Members of our group present this information at pediatric grand rounds locally and nationally whenever we get invited. A colleague, Dr. David Smith, has written an article for Family Physician about adult health care. There are presentations at continuing medical education conferences.

There are big conferences about health care that are often held by parent groups. I caution parent groups that primary-care physicians are not going to take a day to learn about all the problems of kids with Down syndrome. It's just not that salient for them.

When I participated in the National Down Syndrome Society's conference in San Diego a couple of years ago, I arranged with a colleague at a local hospital to give pediatric grand rounds, which is the primary teaching conference of the week, at the Children's Hospital Medical Center. It was an hour-long conference attended by perhaps 130 people. They got the capsulized information. I did a similar program at St. Barnabas Hospital in Livingston, New Jersey, in November of 2003. There are regional conferences as well, and many of the members of the Down Syndrome Medical Interest Group are involved in this education.

But I will tell you that the most effective thing we do is educate parents to be the educator for the physician.

We know that people who are born with Down syndrome today can live longer, fuller and more active lives than those born as recently as 20 years ago. Would you say that is due, at least in part, to a more informed and interested healthcare community? Is it also a result of parents and caregivers taking an active role in assuring the best possible medical, social and educational opportunities?

It certainly is a combination of factors. Clearly, early identification of serious medical problems-cardiac disease being number one-has made a huge difference. When you look at the actuarial studies of lifespan in individuals with Down syndrome that start back in the '50s you'll see that there are basically two groups - the individuals with cardiac problems, and those without. Individuals with cardiac problems that were not identified or not satisfactorily treated might die in the third decade, in their twenties, as opposed to the other group that would live through their sixties and beyond.

Today, the recommendation is that everyone be evaluated for congenital heart disease, however that's available in the community. Certainly we recommend evaluation by a pediatric cardiologist, or, if possible, an echocardiogram done on a screening basis.

The notion that withholding care for children with Down syndrome is an option has, I think, passed. Certainly the idea that a family can choose not to operate on an operable lesion, such as a duodenal atresia, is no longer an option.

The situation is also a result of the closing of institutions, of raising children in communities, and of children participating with typically developing kids in a whole variety of activities - in athletics, Cub Scouts, going out to restaurants, being educated alongside of them. It also reflects an increasing awareness of the value of people with differences, and the fact that instead of with shame, parents are approaching Down syndrome with a sense of acceptance and perhaps even pride.

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