

Sound Advice

This is an edited transcript of a telephone interview recorded in March 2010.

Dr. Paul Carbone is a general pediatrician at the University of Utah, where he provides primary care to children with a focus on those with disabilities. He also has a consultative clinic through the university that focuses on the care of children with autism spectrum disorders. His son, Ben, has autism.

Q: Dr. Carbone, when did you first notice a difference in your son, and what were the differences?

Dr. Carbone: Well, I think from a very early age, Ben seemed to show many of the red flags for autism. By that I mean during even early infancy my wife and I noticed that he was somewhat irritable and difficult to console. Later in infancy we noticed that he seemed to smile less, perhaps, when we looked at him, and later we noticed that he wasn't babbling and attaining some of those early language milestones.

After 12 months we not only noticed he was not picking up early language, but he didn't seem to make an effort or be motivated to communicate around that, by using gestures like pointing and waving that some other late talkers will usually do. We also noticed that he didn't seem to respond well to his name by looking at us when we called his name, even though we were fairly certain that he could hear other things quite well.

At about 15-18 months, we noticed as well that he didn't engage in much pretend play. For example, we noticed a lot of other 15-18-month-olds picking up their parents' cell phones and pretending to have conversations with their grandparents. Ben preferred to play with the parts of objects. So he looked at our phone, he pushed the buttons repetitively, but didn't have that pretend conversation that we were looking for. And then after that he really began to engage in a lot of repetitive behaviors that you hear of with children of autism. He developed preferences for odd objects, like my watch, instead of other traditional toys, tops, things of this sort.

I think for parents, it would be important to note that the early signs of autism can be a bit subtle because, again, we're talking about some of these social skill milestones that I just spoke of, that social smile early in infancy, turning or orienting when you call your child's name, pointing. These are sometimes more difficult to tease out than some of the motor milestones that we parents are used to looking for, like sitting and standing, things of that sort.

Children also can have delays in other areas, such as language, and some of these repetitive activities that I spoke of, but they seem to be a little less specific for autism. You might notice, for example, that your child may have this insistence on sameness, so making a transition from one activity to another can be difficult. Also, we noticed that he engaged in a lot of hand flapping when he was excited, but these can show up a little bit later. And I think I would also remind parents that there seem to be a subset of children with autism who for some reason have a more normal developmental trajectory than I spoke of with Ben and that you may notice a backsliding

or a losing of some skills that were previously obtained, and we've noticed that that can happen in maybe 20 or so percent of children where they might lose some language or social skill milestones at about 18-24 months.

Q: Is that what's known as regressive autism?

Dr. Carbone: Exactly, that's correct.

Q: How was your son diagnosed?

Dr. Carbone: Well, he was diagnosed at about 2 years of age, and that was probably 12 or so months after we really began to have concerns about autism, and that also included about three months of waiting for an appointment with a psychologist. And even though that seems like a lot of waiting, I still consider my wife and I lucky when you consider that the average age of diagnosis for children with autism spectrum disorders in the U.S. is about 4 1/2 years of age. I think it's important then to note that if parents do have concerns about the behavior or development in their child, that they really talk to their pediatrician or primary care provider.

We're trying to move away from this wait-and-see approach to a more proactive way of addressing parental concerns about behavior because we've now come to learn that although parents maybe aren't the best at diagnosing their children's problems, they do have that gut instinct that is predictive of later developmental issues. So we're trying to listen more to parents, and I think when parents do bring up these concerns with their pediatricians, hopefully, the doctor and the parent can develop a concrete plan of action.

And then if you still have concerns and you think that those aren't being adequately addressed, I would recommend that if your child's less than 3 years of age that you call the local Early Intervention Program. This is a free program in most areas and doesn't require a referral from a healthcare provider and consists of professionals with expertise in child development. They can come directly to your house, evaluate your child, and if there is a significant delay in development, they'll work with you to address those issues with therapy.

If you do have concerns and want to reach the Early Intervention Program, you can do so through the National Dissemination Center for Children with Disabilities at 800-695-0285. If your child happens to be older than 3, you can call the special education program in your local school district. If the pediatrician does have a concern when you bring these issues up, he or she might refer you on to a specialist, like a developmental pediatrician or a local team that can do more specific diagnostic evaluation.

Q: Can you access Early Intervention Programs for your children before they have an official diagnosis of autism?

Dr. Carbone: Absolutely, you do not need to have a diagnosis, nor do you need to have a referral from a healthcare provider to call and get an evaluation through the Early Intervention Program, nor do you need a diagnosis necessarily for receiving therapy through the program.

Q: If families have concerns, where should they go for resources and information about autism?

Dr. Carbone: There's a number of useful resources that families can go to beyond simply talking to their pediatrician first. As far as Web sites go, the American Academy of Pediatrics has a nice Web site that parents can go to and get lots of useful information about how to proceed if they have concerns about autism or other developmental issues. And the other Web site that I would recommend is the "Learn the Signs. Act Early" Web site through the Centers for Disease Control. Again, a nice Web site where parents can learn about some of these more subtle social skill milestones that we've been talking about. And then it provides you resources on how to bring up those concerns with others.

Q: Do you have other kids? Are they on the spectrum?

Dr. Carbone: We only have one child. He seems to keep us busy enough for now, but you do bring up something that's very important for parents of children with autism spectrum disorders to know, and that's the risk of recurrence. We know that ASDs have a strong genetic component. Generally, we tell parents that after having one child with an autism spectrum disorder, the risk of having a second child with the same condition is anywhere from 10 to even 25 times higher than the general population. It's one of the reasons why we are making such efforts to get children diagnosed earlier because parents can then be aware of the risk of recurrence and make better decisions about family planning.

Q: Can you talk a little bit about the therapies you have done with Ben and how they've helped your son? What was the most effective therapy that you've used? Have you tried any therapies that you thought were not effective?

Dr. Carbone: We've done a number of behavioral therapies that have used a method known as applied behavioral analysis. A lot of people shorten that just to ABA. I think these have been the most effective for Ben. It's a method of therapy that teaches skills by breaking tasks down into their smallest parts, teaching every one of those parts until it's well-mastered, and then you string those parts together until they look, hopefully for all intents and purposes, like a skill that a typically developing child would do. The therapy is based on some well-established principles that really we all use to shape behavior, and that is that behaviors that are rewarded tend to increase, and those that are not tend to decrease.

There's been a number of studies showing it to be an effective method of helping young children with autism develop skills that help them function at a higher level. So specifically, if you have two similar groups of children with autism and one gets this fairly intensive behavioral therapy using this ABA method and the other group gets the standard therapy method that's offered within the community, the ABA group of children with autism does significantly better after a year or two on tests of intelligence, language, and self-help skills.

And like the children in these studies, Ben really has made great strides in his ability to interact with others. He has friends at school that he can have meaningful social interactions with, and it's helped him in just so many practical ways. His language has improved quite a bit so that he

can let others know about needs, wants, desires. He's less frustrated because he can communicate what he's thinking much better than before he started the therapy. It's also sort of opened up his world because now he can answer a question when he's asked one, and he can have better conversations with others. And we've, meaning my wife and I, have been active in the therapy as well, so it's given us a set of skills that we can use to address challenging behaviors that come up so that we can effectively teach him more appropriate coping skills, rather than inadvertently or accidentally reinforcing or rewarding the inappropriate ones.

I don't mean to say that as physicians we've always made perfect choices. We've gone down the road and done some ineffective therapies as well. My wife and I as physicians have not been immune from trying a few that haven't worked out. Like many parents, we hear of lots of therapies through the Internet, through other parents, that promise great results, and they are at times alluring. One of those that we did was the gluten-free, casein-free diet. After developing some specific goals for what we were hoping to see with the diet, we decided after awhile using it that it really wasn't helping him much reach those goals, and so we subsequently stopped the diet. And now, years later, there's been a fairly well-designed study that showed the same things that we found with Ben.

Q: How long was Ben on the diet?

Dr. Carbone: Ben was on the diet for about 9-12 months from what I can recall.

Q: And it just didn't seem to improve his symptoms?

Dr. Carbone: You know, I think we identified really some target symptoms that we were hoping to improve. Specifically with Ben, he engages in a lot of repetitive activities, bless his heart, so he engages in a lot hand flapping, a lot of pacing, a lot of walking in circles. We were hoping to help him specifically with that because we had read some preliminary studies that showed that it might help with that, but when we tracked these specific behaviors, we noticed that it just didn't seem to extinguish them any quicker than when he wasn't on the diet.

And that was okay. We tried it. We felt like we did it in a responsible way. We worked with other folks to help track his behaviors, and it just didn't seem to do what we were hoping it did, so we checked that box and moved on.

Q: How do you talk to parents who want to try either that diet or other therapies that they've read about online?

Dr. Carbone: Well, as a pediatrician I try to stay true to the science. There is a growing body of research out there looking at effective treatments for children with autism, so if there is a study that I can apply to an individual patient, I share that with the family so that together we can make the most informed decision. And that means that I definitely emphasize the behavioral interventions that we talked about earlier.

That said, you know, every family and every child is different. We have a saying in our clinic that you've met one child with autism, you've met one child with autism. So I think it's

important that we have an ongoing, trusting relationship with the family so that we can tailor treatments in a way that makes sense within the context of this particular family and this particular child.

In my own practice, I very frequently see parents who have questions about new treatments that come up and that they hear about. And my own experience that I shared with you earlier has shown me the value of being open to having these discussions in a nonjudgmental way. When parents bring up complementary alternative therapies with me, I work with them to review the evidence behind any potential risks and benefits of a treatment, and then, again, we try to make a decision together about how to proceed. And if we don't agree, which sometimes happens, I try my best to be flexible and support families while at the same time being an advocate for the child.

So, to give you an example, we might decide that in looking at a particular therapy that it might not in 2010 have evidence to show that it's effective. It's important, though, to note that that doesn't necessarily mean that it won't work. So in this case if it doesn't pose risk, we might do the same thing that my wife and I did with Ben. We call this an "N of 1" trial in which we decide on exactly what we hope the therapy will help with. We identify those things. We figure out how to evaluate if we're going forward towards those goals or away from them, and then we set up a timeline to make a decision on the effectiveness of the treatment.

And what I found is in general, families are very interested in these alternative therapies. They'd like to discuss them with their child's doctor, but they're not interested in being lectured to. So I think there's room to have respectful conversation so that parents can make the most informed decisions about treatments that they possibly can.

Q: Dr. Carbone, how did you set goals for therapies for your son? If autism cannot be "cured," how should families approach therapies and measure their success?

Dr. Carbone: Well, thankfully the therapists and the treatment team that we work with help us with making reasonable goals and setting timelines for achieving them. We as parents are intricately involved with the team, in that it's our job to let the treatment team know how autism affects Ben and our family in a very practical, rubber-meets-the-road sense. And by listening to us, then they help us develop goals that are truly meaningful to him as well as our family.

And you did mention that these days autism isn't considered curable. It's considered a life-long condition. That doesn't mean that children with autism and adults with autism for that matter don't make steady gains, and that's what we as parents really focus on. We focus on forward progress. And at the same time, my wife and I really focus on what we have rather than what we don't have, meaning that we love Ben just the way he is. His therapy really helps him reach his potential, whatever that potential is, so that he can participate in community life to the greatest extent that he wishes to.

Q: Do you have a good partnership with Ben's pediatrician?

Dr. Carbone: We do. She is always willing to advocate for Ben. She's made an effort to be knowledgeable about childhood disabilities. She's essentially devoted her whole career into helping children with disability, and we're very lucky to have her.

I do have some advice for families about how they can work with their pediatrician, and in one word, I think it's a partnership. You as a parent should feel like that your expertise, your opinion about your child is respected and recognized. And I think when you find a provider like that, like our pediatrician, working with them is relatively easy.

I'd also advise that parents not hesitate to ask for their pediatrician's advice, especially when it comes to those issues that really affect day-to-day life. For us, for example, Ben's sleep was an issue for a long time, and we've gotten a lot of help from our pediatrician and others in this area. Pediatricians can be very helpful in these areas, sleep problems, gastrointestinal problems, even how to manage some difficult behavior. They may not know how to address every single problem, but you might find a real partner and an ally in your pediatrician in that they can help you find specialists and other resources in the community that can help as well.

There's a big push underway right now to make pediatric practices more receptive to the ongoing needs of children with special healthcare needs. Many practices, for example, are making accommodations like longer visits, helping families with coordinating care with the other practitioners that you use in the community. This whole concept is known as the medical home. I think for children with autism, they'll really find some benefit in some of these changes within pediatric practices.

Q: How has Ben's autism diagnosis affected your family and his relationships with other family members? Is it important to have a support network?

Dr. Carbone: Ben's autism affects virtually every aspect of our lives, but not necessarily in a negative way. Much of it is positive. I tell people that parenting a child of autism isn't better or worse, it's just different. For me as a father, those differences did mean that I had to make some adjustments that I can honestly say at first were difficult to accept, but as more time goes by, I see them as good things.

To give you an example, Ben doesn't necessarily enjoy some of the traditional father-son activities that I might've envisioned we'd do together, like playing soccer or playing baseball, but he's developed interests in other things, like running and swimming and rock climbing. We just ran our first 5K the other day, and we really, really enjoy these activities that we do together.

So he's taught me how to be a better father and a better person than I really ever thought I could be. He's made me more tolerant of the differences in people. He's helped me to become more patient. I'm definitely not as self-conscious if he's having a hard time in public because I've come to realize that much of this is because of his disability.

So my, my wife and I are very lucky to have a lot of supports, and I would recommend that families reach out for support. We have a wonderful extended family. We regularly rely on them for help. We also have some amazing friends here who are very accepting of Ben and have

become really strong advocates for him. We also have this treatment team that not only treats Ben, but really, truly cares for him deeply.

And we live in a community that offers a variety of programs for children with disabilities, and we take advantage of every single one of them that we can, not only because they're good for Ben, but because they're good for us. Wall-climbing programs for children with disabilities is one example in our community. These programs connect us with other families of children with disability, and we learn about their experiences, and we get to simply be with people who get it when it comes to the issues that we face.

And my only other advice for families would be to not be afraid to ask for help. You're not weak if you do, and likely you'll find that the people that are helping you are really getting just as much back from the experience as you are from them.

Q: How is Ben doing now?

Dr. Carbone: Oh, thanks for asking. He's doing great. He is 7 years old. I think the most important thing is that he's just a really happy guy. He's in a regular education classroom with an aid that helps him stay on task and work on his social skills. He has meaningful friendships at school, and we have his classmates over for play dates all the time. He's made great progress in the area of communication.

I don't want to paint it in completely a rosy form. He still struggles with some anxiety, especially when the unexpected occurs. He can definitely become over-fixated on certain activities and interests, but again, he's making progress all the time.

Q: What advice would you give families who are just facing a-a brand new diagnosis, and what advice do you wish someone had given you?

Dr. Carbone: That's a good question. I'm not sure any of the advice that I could talk about wasn't given to me. I think, in a sense I just had to go through the journey of autism a little bit to realize that it was really good advice and that I should take it. But some of that, early on, is that it's important to be as parents on the same page.

It's really helpful if both parents come to appointments so that you're not getting mixed messages and not moving forward in a supportive way. The diagnosis, no doubt, is a very serious one. Again, we've said that autism is a life-long, condition, and so it's important to initially take some time to grieve and support each other through the process. I think staying together as a family is probably the most important thing to do early on.

And then when you start treatment, I guess realize that it's a marathon and not a sprint. It's a long-term endeavor in most cases without easy, quick fixes. So my advice would be to think about it, start a treatment, measure your success over months and even years and not days or weeks. And take time for yourself. I see a lot of families who get so wrapped up into the world of autism and treatments that they forget about themselves and the people around them, so take time for yourself. My wife and I both have hobbies. We've kept them. Try to make sure that you

nurture your relationship with your spouse, with other children, and with other friends. I think those were all good things that we learned from others.