

SUMMER ■ 2006

## Medical Care Issues

JANE TRUE

MIRROR Editor, serves on the IDEAS Board, and lives with her husband Jim in Kansas City, MO

**I**N THIS ISSUE OF THE MIRROR we explore the role that parents play as partners with the medical community in providing medical care for our children with idic15 and related disorders. Upon getting our child's diagnosis, many of us have experienced the frustration of looking for information about the disorder and discovering that there was very little published. We have then turned to our medical providers and found that they actually relied upon us to bring information to them. And each time we bring our child to a

physician we experience again the difficulty of having an exam performed on a child with a duplication of chromosome 15q.

We as parents and family members are the acknowledged "experts" on this condition, whether we want to be or not. So what can we "experts" do to improve medical care for our children? We interviewed a pediatrician who has a child with idic15 in his practice, and got insight into how we can help the treating physician. Several parents share their strategies for getting through visits to the doctor.

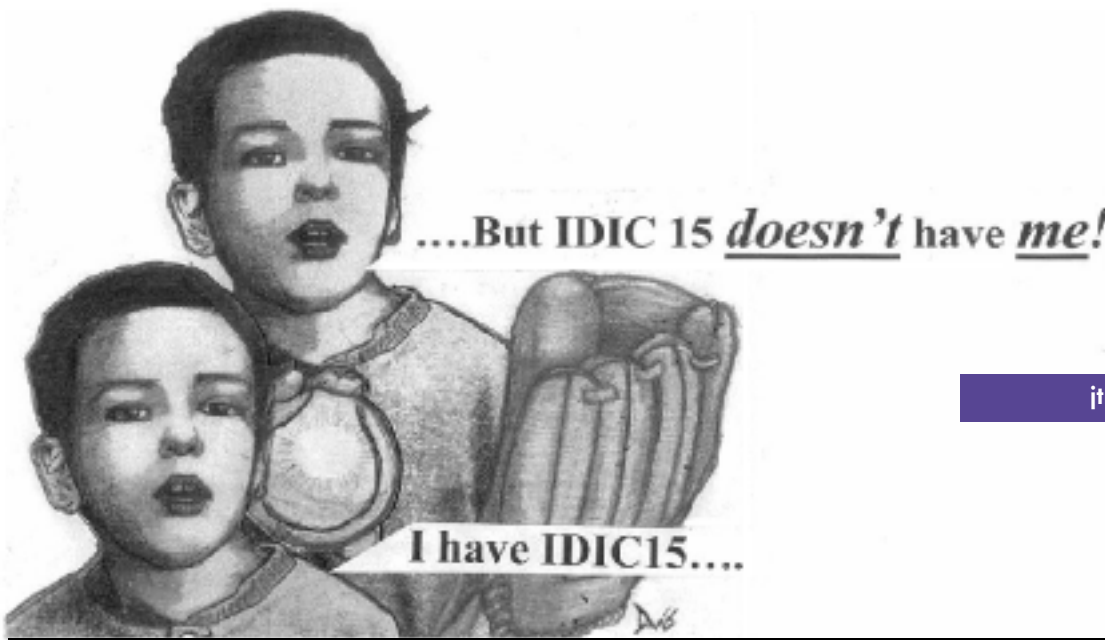
*Isodicentric 15, abbreviated idic(15), is a genetic disorder in which a child is born with extra genetic material from chromosome 15. In people with idic(15), the extra chromosome is made up of a piece of chromosome 15 which has been duplicated end-to-end like a mirror image. It is the presence of this extra genetic material that accounts for the symptoms seen in some people with idic(15). People born without an extra chromosome but who have a segment of duplicated materials within chromosome 15 (known as an interstitial duplication 15q) can have features similar to those with idic(15).*

And we explore the very key role that families play in helping to provide better information about duplications of chromosome 15q. There are several important research opportunities discussed in this issue. Supporting good research is a key way we can help increase the understanding of how idic15 affects our children, leading ultimately to better targeted treatments that may improve their lives.

Enjoy reading the family story and the reflections from a sibling. If you have ideas about a theme you would like to see covered in a future MIRROR please contact the editor at

[jtrue@kc.rr.com](mailto:jtrue@kc.rr.com)

Warmly,  
 Jane True  
 MIRROR Editor



# Working with Pediatricians: An Interview with Dr. Mark Myers



**MARK MYERS, MD, FAAP**

is a board-certified pediatrician in Manchester, New Hampshire who has been in practice for 11 years. Currently he has one patient in his practice who has idic15. He was interviewed by Jane True for this article.

**// Dr. Myers, thank you for agreeing to discuss with me today how parents might help facilitate the diagnosis and treatment of their child with idic15."**

**● JT | How does a patient with a rare genetic disorder generally get identified?**

**■ Dr. M |** Two ways. Possibly a family has already had genetic testing so they have a diagnosis. In this case it is helpful if the family can inform me of available resources. But more often my first clue is the child missing developmental milestones, together with my instinct. In the case of my patient with idic15 I recall something about her eyes set off an alarm. That together with developmental concerns caused me to recommend referral to a neurologist. That particular neurologist didn't feel there was a problem. I disagreed. The family was then sent to early intervention. Usually a front-line person would recognize that something's not adding up. Then they would be referred for further testing.

Some pediatricians might rely on physical presentation but for me the biggest clues are developmental in nature. Things like poor muscle tone and poor feeding are often clues, but the key question is "When is this abnormal?". Throughout my practice I place great emphasis on

listening to parents but for diagnosis with first-time parents it's tougher. They have nothing to compare to and often aren't aware of what normal development is.

**● JT | How is having a child with idic15 in your practice different from having a typical child? How is it the same?**

**■ Dr. M |** Obviously both children have the same illnesses and common conditions, such as ear infections. What is different is the exam and administering some treatments to a highly fearful and defensive child. For a child who is highly fearful of exams and wants to leave immediately, the fear often doesn't get better over time. Also having to coordinate with multiple specialists is different.

**● JT | What can parents do to help the visit go more smoothly?**

**■ Dr. M |** For a first time visit I require copies of all medical records ahead of time. Parents should get all records from specialists to me in advance so I have time to review them.

If a parent knows that certain times of day are better for their child they should make that known at scheduling time. Certain appointment times allow me not to be rushed: the last appointment before lunch and the last appointment of the day. Also the first appointment of the day and the

first one after lunch I know I will be running on time. I note electronically once I know that a certain patient requires more time.

I am not a fan of medicating a child to prepare him/her for a medical exam. I have always been able to get the exam done without that, though it isn't always pleasant. My patient with idic15 seems to do better when her siblings are present. She still has anxiety but not as much—her whole demeanor is different. It may also help to bring the child when it's the sibling's turn for an exam and the challenged child has nothing done.

**● JT | How can parents help coordinate medical care for their child when there are multiple specialists involved in the ongoing care?**

**■ Dr. M |** Obviously by making sure all specialist's records and recommendations make it back to the primary care provider. Coordination depends, to an extent, on your area. Large tertiary care centers have a coordinator to provide this service. If you don't have this in your area you must rely more on your primary care provider. I see these patients more often—maybe twice a year instead of just once. I also refer to a developmental pediatrician on occasion just to get his assessment, to evaluate appropriateness of therapies, etc. But the parents and child are always my #1 source of information.

● **JT** | *When a child ages out of a pediatric practice, how can parents get a good match for their child's special health care needs in an adult practice?*

■ **Dr. M** | Word of mouth referral from other families in similar situations is helpful, or from medical people who know and understand your child's issues. Also there is a new, in the last 5 or 10 years, Medicine-Pediatrics Specialist. This physician has completed a 5 year residency, half in internal medicine and half in pediatrics. This training is more extensive than a 3 year Family Practice Residency. If you have such a physician in your area and he/she is a good match for your child then you could avoid aging out of the practice.

● **JT** | *How can our group help pediatricians identify and treat a child with idic15 in their practice?*

■ **Dr. M** | It would be helpful to have a link on your website for providers. Include such things as what we should expect to see, photographs, common experiences of parents, what things most of us have in common most of the time, what are the resources in an area. This is important because with rare conditions I have to rely more on the parents, so compiling those experiences would be helpful. Also if I get a new diagnosis it would be helpful to have another physician to contact who has a similar patient.

# Getting Through Medical Appointments

*ELLEN DOXTATOR and her husband Peter are parents to Cheyenne (idic15, age 13) and Samuel (11). They live in Ontario, Canada.*

**G**etting through medical appointments has been a source of stress for our family. This has been ongoing since birth so we have lots of first-hand experience. Not only is it extremely stressful to deal with the stares and the judgmental opinions of the public but also sometimes the contentious attitudes of the nurses and doctors. And often the inappropriate behaviors of the child are blamed on the parents and we are seen as inept. At these times I have learned when to step in and speak up for Cheyenne and when silence and a good stare are my most effective weapon!!!

For us, having a child with idic15 has meant hundreds of appointments at the hospital. Cheyenne is seen for many different reasons: genetics, immunology, feeding clinic, orthopedic surgeon, neurologist, hearing and vision clinic, metabolics, blood work and many more. As the frequency of behaviors has increased over the years, so has the severity of the behaviors. When Cheyenne was younger she used to be non-compliant. Now the anxiety starts as soon she knows about the appointment and will lead to yelling, screaming, crying and physical aggression either during or after an appointment. In fact, for us, we have found that it is better not to tell Cheyenne about an appointment until it is time to get ready the same day. If she knows before then, she does not sleep the night before and obsessively

worries about the appointment.

Traveling to appointments is also exhausting. Cheyenne talks non-stop and often fixates on what will happen when she gets there. She has had so many procedures done that she is fearful of anything now and fights them all, even the simple ones. Her behaviors include: screaming, crying, kicking, physical aggression, refusal to cooperate and trying to escape the appointment.

**Here are some techniques we use to help us cope:**

- **Information:** Ensure new doctors or clinics have information on idic15 & autism and an understanding of your child's needs. Also sometimes reminding staff that your child's fears are very real to him/her.
- **Timing:** It is helpful to request the first or an early appointment in the day to decrease the waiting time and anxiety.
- **Relaxation:** We use a small laminated relaxation book to travel with which has simple relaxation exercises with visual cues which we do as we travel to the appointments.
- **Reassure:** Reassuring your child as to what kind of appointment you will have that day and focus on the positives. "Today we will see Dr. Munson—remember you had fun there last time?" Also, be honest. If there is no blood work that day, say so. This reduces the anxiety.

## Getting Through Medical Appointments

- **Reward:** For us, cooperation at appointments is rewarded. It can be as simple as a donut and a juice or sometimes she can pick a small toy or book. Cheyenne also gets to pick out something for her brother so that he never feels left out by all the attention she receives and this makes Cheyenne feel special too!
  - **Social Stories:** We also use social stories to discuss and reinforce appropriate behaviors
  - **Sense of Humor:** Having a sense of humor can really help even when things are at their worst. Like when it took 7 of us to get an IV back into Cheyenne after she pulled it out after surgery and she was hysterically crying, screaming and fighting. But once it was in, Cheyenne stopped and calmly said, "Now, that wasn't so bad, was it?" Even some of the nurses had to chuckle.
- Lastly, I always try to remind myself that as bad as it gets, as long as these appointments continue to help our daughter, then we will continue to go. And at the end of many appointments, Cheyenne has looked at me and said, "I couldn't do it without you Mommy." And I always smile and say "And I couldn't do it without you either Cheyenne," and then I laugh to myself because without Cheyenne I wouldn't be there. And so in spite of all our difficulties, Cheyenne also puts everything back in perspective, behaviors and all.

# Reflections

## What do you do to help Dr.'s appointments go more smoothly?

This column is designed to share perspectives from recent ListServe discussions regarding doctor's appointments. To subscribe to the listserve, send an email to [Inverted-Dup15-subscribe@yahoogroups.com](mailto:Inverted-Dup15-subscribe@yahoogroups.com).

### Jill Kertzman

| Cody's (idic15) & Jaycee's mom  
"I keep a 'Word' document going on my computer. When I read emails or think of something I need to discuss with Cody's Neurologist, ENT, or another important Doctor, I copy and paste or type it out. Then, I print 2 copies for the appointment, one for me and one for the Doctor. I write notes/answers on my copy next to my questions. It has been very helpful, especially using the listserve email information, since the group usually knows more than the Doctors about trends and medications with our kids."

### Doreen Gladis

| Pavel's (int dup15) mom  
"These are some things I found helpful."

1. Schedule the appointment first or last appointment of a day.
2. When Pavel was younger I always brought food or a stim toy to keep him busy during the wait.
3. We sit far away from groups of people.
4. I asked Dr. to check his ears, heart, and throat while he was sitting on a chair.
5. For outpatient procedures I advised them of the anticipated problems prior to the procedure. We were given a private room to wait in, instead of being in an area separated only by curtains. I have found most staff very accommo-dating at Children's hospitals and willing to find ways to make the procedure less stressful

6. Now that he is older going to Doctor appointments is more of a challenge. Tom has to come to all Doctor appointments to physically get him in. I have tried getting the help of staff when I'm alone, but most are hesitant to try to physically move him.
7. We have yet to find a medication that is helpful"

### Mari O'Connor

| Christopher's (idic15) mom  
"The doctor's waiting room seems to be the worse part of the visit. I ensure that Christopher has several toys that are favorites and may help distract him from the new situation. I also have little bags of snacks, drinks and favorite foods. I focus all of my attention on just him (to comfort him, *WHEN* needed—not *IF*). I try to always have my husband with me so that I can deal with Christopher and he can deal with the receptionist. I have a notebook, armed with questions, comments on development, seizure activity or anything that needs to be discussed with the doctor. Once we get in the doctor's office, my husband focuses on Christopher so that I can talk with the doctor. I make notes of everything that he tells us, suggests, recommends, etc. Christopher normally sits on my husband's lap, toy in hand, snack in mouth, and is fearful about when he will be touched by the strange man. We get in and out quickly. The notebook is the key to making it successful and useful."

This part of the REFLECTIONS  
column shares perspectives  
from a sibling.

## Growing Up Together: My Big Sister

**Tyler True,**

| 21-year-old brother to 25-year-old Clare (idic15) and 27 year-old Toby

When I was little, my mom told me about a comment one of her friends had made. “Your boys are so good,” the friend said. “They’re never getting in trouble. How do you do it?” I looked at my mom and laughed. “Tell her to have a daughter with a disability. We don’t have time to get in trouble.”

At 25, my sister Clare is one of the older kids with idic15. I don’t know of too many younger siblings from the group, perhaps because the idic15 kids—the cases we know of—are so young, perhaps because idic15 parents are—well—so busy. But you don’t need to know too many such younger siblings to know that we’re a little different, even than our older non-affected siblings.

Sibling rivalry in the absence of idic15 makes us stronger, but what happens when the “stronger” sibling has a mental disability and can’t fight the same way? Clare always knew she was a little different, going to a different school, frustrating mom just a little more than the rest of us, taking all that medicine. But our family always worked because nothing was ever completely out of place; Clare went to school like my brother and me even if she went to a different school, she ate dinner with the family even if she had to take all that medicine afterwards.

So while Clare maybe sensed she wasn’t all that she could be, there was no reason, from her point of view, to suspect a problem. Not until, that is, my brother left for college and I started sitting in the front seat all the time, even though Clare was older. Not until I got my driver’s license, but Clare still couldn’t drive. Not until I

turned 21, but alcohol still didn’t mix with Tegritol for poor Clare—who does, by the way, enjoy her wine if she gets a chance. And don’t think Clare doesn’t notice this inversion: unlike my older brother, parents, cousins, family friends, just about everyone else except certain teachers and bus drivers, I have never been able to help calm her down, exert any control, or have any kind of special ability to help my sister. In fact, I have only three options for getting her to do anything:

**1. Compromise heavily:**

“Okay, you can have a cookie, but only four or five, Dad needs some, too.”

**2. Be with someone else:**

me: “Clare, don’t do that.”  
Clare: “AHHHHhhhhhhhhhh”  
Cousin: “Clare, stop it.”  
Clare: [silence]

**3. Invoke the fear of Mom:**

“If you touch that, Mom will be maaaaaadd.”

So the younger siblings, experience a strange twist on an already bizarre situation. But for us, in a way unlike anyone except the people with the disability, this is reality. There is no life without idic15, no reality of before to compare. As we get older, along with our siblings, hopefully we will be able to figure out what our slightly skewed perspective has to offer.



### The Mirror

*We are looking for siblings who would like to share their thoughts in a future issue of **The Mirror**. If you are interested please contact the editor at [jtrue@kc.rr.com](mailto:jtrue@kc.rr.com)*

# Research—Our Hope for Targeted Treatments

NICOLE CLEARY, is the Executive Director of IDEAS. She lives in Portland, OR with her husband Tim and daughters Corrina (idic15), Sierra and Jasmine.



**ONE OF THE FRUSTRATIONS FACED BY PARENTS** raising a child with a chromosome 15q duplication is the lack of standard medical treatment recommendations for the symptoms caused by isodentric and interstitial duplications of chromosome 15. While we all would like to know how to treat our child's symptoms, research and medical professionals are still working on fully characterizing the effects of chromosome 15q duplications and trying to understand how the specific genes involved in these duplications cause the developmental problems experienced by our kids. Once these associations are better understood, then scientists can begin to translate the knowledge gained from this basic research into clinical applications.

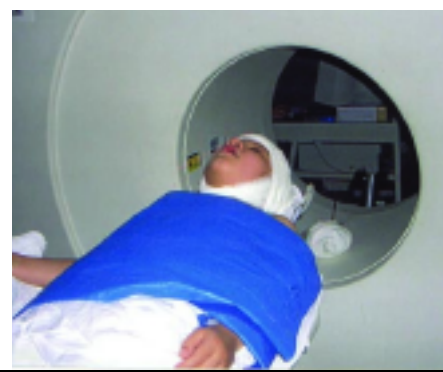
Families play a critical role in increasing the understanding of chromosome 15q duplications through their participation in research studies. **IDEAS** provides families with information about current research studies on our website, [www.idic15.org](http://www.idic15.org). Studies listed on the **IDEAS** website have been reviewed by the **IDEAS** board to ensure the proposed research meets the following criteria: 1. The research directly addresses an important aspect of chromosome 15q duplications; 2. It has been approved by an Institutional Review Board (IRB); 3. There is an opportunity for families to provide informed consent; and 4.

The research complies with all regulations governing privacy, security, and electronic transaction standards for health care information. **IDEAS** collaborates with studies that meet these criteria by making parents aware of the opportunity to participate in them. Thankfully, many parents have chosen to participate, thereby providing researchers with critical information, samples, and access to affected children.

Sometimes participation in a research study is as simple as filling out a survey form. For other studies, blood samples are required from affected children, their parents, and occasionally siblings. In one of the most comprehensive studies on idic(15) to date, *Molecular Investigations of Duplications of Chromosome 15 in Autism*, Dr. Carolyn Schanen and her staff are collecting blood samples and conducting clinical assessments on a subgroup of children with chromosome 15q duplications. As a result of this research a lot has been learned about the structural features of the long arm of chromosome 15 that causes it to rearrange and make duplication chromosomes. Although the initial reports of idic(15) chromosomes suggested that the extra chromosome was symmetric and a mirror image, Dr. Schanen has found that the positions of the breakpoints involved vary among kids with seemingly identical duplications

(based on the clinical studies). How this affects outcome is still not known at this point.

Children with duplications of chromosome 15q can experience cognitive disabilities, autism and seizures. In the region of chromosome 15 most commonly duplicated in idic(15) and related disorders, there are 3 GABA receptor genes. Impaired GABA<sub>A</sub> receptor function has been proposed to play an important role in several neurodevelopmental and genetic disorders, and may be a significant contributor to the developmental problems in chromosome 15q duplications. Dr. Diane Chugani is currently conducting a study to investigate this. Her study, *Brain GABA<sub>A</sub> receptor abnormalities in children with chromosome 15q11-13 mutations*, seeks to understand how changes in the genes of people with abnormalities of chromosome 15q11-13 may cause changes in GABA<sub>A</sub> receptors in their brains. This study is currently enrolling children who have already been characterized in Dr. Schanen's study. It involves two procedures: a Positron Emission Tomography (PET) scan with an EEG, and an MRI scan. Ron and Heather



## Research—Our Hope for Targeted Treatments

Bruce recently enrolled their daughter Anna in this study. Heather states, “The whole process was much easier than I thought it would be. The few moments of discomfort that Anna had to go through (IV sticks) were well worth contributing to such an important research study. Anna was only the 3rd child to participate, so they definitely need more kids! **IDEAS** helped cover our travel expenses and they can help other families who are interested in participating.” I invite families interested in learning more about this or other research studies to contact me for further information: Nicole Cleary (503.253.2872) or [nicleary503@comcast.net](mailto:nicleary503@comcast.net).

The future is bright with possibilities for continued research. **IDEAS** is negotiating with the Duke Center for Human Genetics for development of a registry that will allow us to systematically collect data on the effects of chromosome 15q duplications. New investigators contact **IDEAS** regarding their interest in studying our kids, and as they secure funding for their projects new collaborations are sure to come. Research studies with our children and families today will provide the answers to basic research questions that one day will translate into the clinical applications we all want for our children.

# The Gift of Hope

In this article, **IDEAS** introduces our families to the Autism Tissue Program. The Autism Tissue Program is a campaign to understand and treat a whole spectrum of pervasive developmental disorders, such as those caused by chromosome 15q duplications.

Many of us have signed the back of our driver’s license to give permission to donate our organs after we die. We have been made aware of the importance of doing this in order to help another person, and have taken action in advance so that something good can come from our death. We generally don’t think about this kind of opportunity in regard to our children affected by a chromosome 15q duplication. Now there is a program that provides families an opportunity to donate the brain and tissue of a loved one who has just died so that researchers can access a critical resource for understanding chromosome 15q duplications.

**IDEAS** Professional Advisor Dr. Edwin Cook Jr. states, “Although many scientists have become aware of the importance of understanding what causes the symptoms of *idic(15)*, a major roadblock to further understanding is the absence of brain tissue. Scientists are able to be creative and work with cells from blood and brain imaging, but access to studying the impact of *idic(15)* on the brains of affected individuals is vital.”

The Autism Tissue Program has a worldwide network of researchers seeking to understand how and why the brains of affected individuals are different. The brain is the locus of the behaviors associated with chromosome 15q duplications (repetitive and restricted activities, lack of communication and social skills) and the seizures experienced by over half of affected individuals. Brain tissue is vital because it is the only way to see individual brain cells and read the DNA and genetic messages to get a better understanding of chromosome 15q duplications.

According to Dr. Carolyn Schanen, **IDEAS** Professional Advisor, “Brain tissue will be tremendously valuable for understanding the ways that duplication of chromosome 15 affects brain development and function, which will hopefully help us provide better treatment options for patients with *dup(15)* syndrome. It is difficult to think about the potential death of a child. However, thinking about it in advance

allows the family to discuss the issues surrounding a brain tissue donation without the incredible emotional stress that occurs when a child has died. Advance planning for a tissue donation is critical.”

Organ donation is a highly personal decision that has the power to accelerate research and transform the lives of countless families affected by chromosome 15q duplications. It reflects a family’s choice to benefit others, and honors the complex and challenging life of the affected child. You can make a tremendous difference in research into chromosome 15q duplications by participating in this program.

Registering to donate brain tissue does not mean that anyone anticipates your child will die an early death. It does mean, however, that you are prepared to act if such an unexpected and tragic event were to occur. A pathologist performs the procedure and coordinates with the funeral director. Your child is treated with the utmost respect and dignity. The procedures used to obtain brain tissue will not affect any funeral arrangements, including viewing, that you wish to make. The Autism Tissue Program assumes all costs related to obtaining tissue.

Brain tissue recovery is coordinated nationally by the Autism Tissue Program and the NIMH/NINDS Harvard Brain Tissue Resource Center. In the event of a death, contact the 24-hour hotline number: 1-877-333-0999 for immediate assistance. You will be put in touch directly with brain bank representatives who will then contact a local pathologist to assist with tissue recovery at your area hospital. But you will have to call. You can pre-register on the Autism Tissue Program website [www.memoriesofhope.org](http://www.memoriesofhope.org). Even if you don’t pre-register, donation can still be arranged with this phone call. The Autism Tissue Program coordinator can answer questions you might have about donation. All inquiries are treated with absolute confidentiality.

One Simple Step

Call toll-free 1-877-333-0999

You Can Make a Difference



# Carrie

## Carrie's Story

**DARLENE JOHNSON**

*is mom to Jared age 26 and Carrie age 22 (idic 15). Together with her husband Carl, the family lives in Kunkletown, PA*

### **T** HIS IS MY STORY ABOUT OUR DAUGHTER, CARRIE.

She is the apple of her daddy's eye, her grandparents' princess, Jared's 'dweeb' of a sister, and my shopping and going to the movies buddy.

Carrie was born on November 7, 1983, at 8:30 AM in Allentown, Pennsylvania. Her birth weight was 6 pounds 4 and one half ounces and the apgar was 10. Everything seemed perfectly normal. It was my sister-in-law who noticed that Carrie wasn't rolling over or trying to sit. When we went for one of Carrie's scheduled checkups I questioned the pediatrician. The doctor started to ask more questions and decided we should have frequent visits and that maybe

Carrie was developing at a slower pace. By the time Carrie was eight months old the doctor decided that she needed to see a specialist. We saw a neurologist who told us that Carrie had Tuberous Sclerosis.

We did have genetic testing done and even had a geneticist from St. Joseph's Hospital in Philadelphia take a look at Carrie. This doctor confirmed the neurologist's diagnosis. The genetic testing that was done 21 years ago wasn't as precise as it is now. Well, needless to say we needed early intervention for Carrie and all the help we could find to help Carrie develop and hopefully catch up to kids her own age. That was what we hoped would happen.

Carrie didn't sit up till she was six months old, crawled at twelve months, and walked at about 26 months. I don't think I could remember half of the things Jared did as well as I could remember Carrie's milestones. Carrie had to work so much harder. Carrie was my teacher, she taught me to fight and not give up, and she taught me to appreciate the little things in life.

Carrie is the most persistent person I have ever met. Carrie charms most people she meets and those she doesn't like she ignores (there are very few she ignores).

When it came time for pre-school, Carrie's early intervention program was two days a week and she attended a school district program three days a week. I sent a written request for Carrie to be tested for preschool. It took six months and many phone calls until Carrie was tested (I found out later that was illegal—she needed to be tested within thirty working days of the request).

When it was time for Carrie to go to school, her school decided that Carrie had to be seen by "The Kennedy Center for Handicapped Children" in Baltimore, Maryland. Carrie was only three years old and when we arrived there it was three days of exhausting tests and questions. By the time we left there, Carrie was running a fever and we were told Carrie's neurologist was lying to us and she would never be more than she was. The doctors made her life look

# Carrie

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bleak and I was devastated. After thinking things over, I decided no one was going to tell me Carrie wouldn't learn to talk or play. She did both. She may not be able to carry on a conversation like other kids, but she understands and she can play for hours on her own. Carrie loves movies and books and puzzles, and her Leap Pads. She colors in books and asks us to color with her, and likes us to read to her. Carrie has a good life. I sometimes think she is better off than most of us. Carrie only needs her favorite toys around her, a warm bed to sleep in, and people around her that love her—and McDonald's!

When Carrie was 18, she had a horrible year. She started her period when she was twelve years old and she couldn't deal with that, so we started her on depo provera. I really think that is what caused a lot of problems. Her behavior became erratic. She tried to break the doors down, screamed and cried, didn't sleep or eat, and I thought she would have to be put in a mental hospital. We tried to get

help and finally got to see a psychiatrist. We tried all kinds of meds; most did not work. Carrie's school wanted us to keep Carrie at home until her behavior was better.

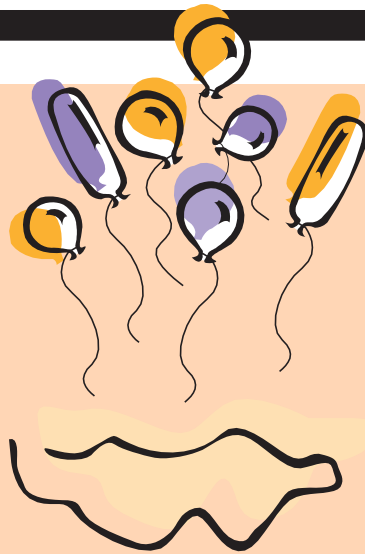
During this time Carrie's neurologist decided to do genetic testing. In about two weeks we discovered Carrie had isodicentric 15, and received a lot of information about this wonderful group and the Elwyn Institute. We were told that Carrie was never a true Tuberous Sclerosis patient. She didn't have enough markers for the syndrome. I remember telling the doctors it didn't matter, she needed an education. Carrie was in a life skills program in a private school paid for by our school district. It was the best there was at that time. I can't tell you how devastated I was at the new diagnosis. I cried and my sister-in-law got in touch with Paul Rivard (we didn't have the internet at the time). Paul called and we talked and I also called the Elwyn Institute. No one could understand how Carrie could be misdiagnosed for so long. I still feel guilty sometimes that I didn't do enough sooner.

So, this brings me to Carrie now. When she was 19 years old she had her uterus removed (no more depo provera and no more periods she couldn't handle). It was the best

decision for Carrie. She began a program at Devereux Pocono Center, where she started attending one day a week and four days at her approved private school. Then the days were gradually increased to three days a week at Devereux and two days at her approved private school. We wanted to slowly have Carrie introduced to her new life and place to live. Devereux is a wonderful choice, because there is a golf tee program for Carrie to go to during the day, they also take care of recycling the other group homes and some private homes in the area.

Carrie goes out to eat, goes to parks, picnics, dancing, shopping (her favorite) and many other activities. Carrie is taken care of by people who love her and want her to have a good life. We bring her home every other weekend and on holidays. They are great to work with and we are happy. There are still times we cry when we take her back and we miss her, but we know she will be taken care of when we are older and when we are no longer here. Carrie moved into her house on July 5, 2005. Almost a year! A year of no more fighting for what she needs and being able to sleep at night because we know she will be okay, and we will too.

Happy  
Birthday  
to these  
special  
people:



- |              |          |               |          |                |          |
|--------------|----------|---------------|----------|----------------|----------|
| • James C    | 06/01/03 | • Samuel V    | 07/09/95 | • Charlotte J  | 08/28/03 |
| • Alyssa L   | 06/02/93 | • Michael B   | 07/09/84 | • Rachel H     | 08/29/03 |
| • Kathleen M | 06/04/93 | • Clare G     | 07/09/82 | • Ashley Q     | 08/29/91 |
| • Jack R     | 06/07/02 | • Lauren C    | 07/11/00 | • Shannon N    | 08/30/94 |
| • Angel S    | 06/09/93 | • Andrew H    | 07/11/89 | • Kacey T      | 08/30/93 |
| • Chris L    | 06/11/90 | • Gabrielle P | 07/12/01 | • Brenda M     | 09/02/89 |
| • Anthony S  | 06/14/00 | • Erica A     | 07/12/86 | • Cheyenne D   | 09/08/92 |
| • Heather W  | 06/16/92 | • Jessica M   | 07/18/98 | • Bente S      | 09/09/97 |
| • Andrew B   | 06/18/99 | • Nicolas D   | 07/18/95 | • Elana K      | 09/09/88 |
| • Richard H  | 06/18/96 | • Kate S      | 07/20/96 | • Major R      | 09/10/98 |
| • James M    | 06/18/96 | • Ashtyn M    | 07/23/04 | • Dante R      | 09/10/98 |
| • Yuri P     | 06/20/75 | • Zoe N       | 07/25/02 | • Thomas N     | 09/10/93 |
| • Cheyenne J | 06/21/96 | • Clare T     | 07/25/80 | • James S      | 09/11/93 |
| • Krystyn B  | 06/22/98 | • Colin O     | 07/30/98 | • Waawiyi W    | 09/11/93 |
| • Trevor B   | 06/23/94 | • Gavin S     | 07/31/98 | • Julia W      | 09/12/96 |
| • Grace L    | 06/24/03 | • Emily M     | 07/31/80 | • Raquel L     | 09/14/00 |
| • Jonah C    | 06/24/99 | • Rachel K    | 08/04/86 | • Rachel M     | 09/14/94 |
| • Klara H    | 06/24/92 | • Devon J     | 08/07/93 | • Mason L      | 09/15/99 |
| • Douglas M  | 06/26/03 | • Jesse M     | 08/12/94 | • Breaua E     | 09/15/95 |
| • Sarah H    | 06/26/93 | • Eli C       | 08/14/92 | • Cameron P    | 09/17/00 |
| • Holly T    | 06/27/94 | • Austin V    | 08/15/96 | • Jeremy L     | 09/17/88 |
| • Mikaela O  | 06/28/00 | • Spencer G   | 08/15/94 | • Conor M      | 09/19/96 |
| • Taylor L   | 06/29/99 | • Patrick S   | 08/16/98 | • Brandon J    | 09/24/97 |
| • Alex S     | 06/29/96 | • Carly P     | 08/16/93 | • Sarah L      | 09/23/95 |
| • Erika M    | 07/01/01 | • Gavin H     | 08/17/04 | • Cheyanne F   | 09/27/93 |
| • Kyle B     | 07/01/84 | • Alicia M    | 08/19/97 | • Evan         | 09/28/94 |
| • Travis R   | 07/02/98 | • Sarah D     | 08/23/98 | • S. "Chase" G | 09/30/99 |
| • Alexis M   | 07/05/94 | • Colt C      | 08/24/93 | • Cara V       | 09/30/92 |

## IDEAS

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### GOT NEWS?

The MIRROR welcomes the stories, insights, and ideas of all parents and professionals interested in *idic(15)*. We'd also like to hear your suggestions for future articles.

Send correspondence to Jane True, MIRROR Editor at:

[JTRUE@kc.rr.com](mailto:JTRUE@kc.rr.com)

or to  
IDEAS  
6699 SE Scott Drive  
Portland, OR 97215

Jane True, Editor

Lin Neighbors, Design & Layout

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# BELIEVE IN OUR KIDS

eBay  
auction  
to support  
IDEAS

**IDEAS** is holding an eBay auction from August 6th thru August 27th, and everyone is invited to participate! We are hoping to raise over \$5,000 to support development of the International Chromosome 15q Duplication Syndrome Registry and jump start our fundraising for the 2007 Conference, **Sharing the Journey**. Both the registry and the conference will directly benefit families raising children with duplications of chromosome 15.

Auction items will be listed on the **IDEAS** eBay home page, which is found on their non-profit site (**Mission Fish**). Tell everyone you know about the auction and come find some good deals! All proceeds from the sale directly benefit **IDEAS**.

**BELIEVE IN OUR KIDS**

eBay Auction  
August 6 —August 27



go to... [http://missionfish.org/NPMMF/nhomepage.jsp?NP\\_ID=10419](http://missionfish.org/NPMMF/nhomepage.jsp?NP_ID=10419)

Paper or  
Plastic?

## Reading the MIRROR on-line?

Mailing fewer paper copies will help keep the **MIRROR** costs down. If you and your family read the electronic version of the **MIRROR** on the **IDEAS** website and do not wish to receive a paper copy, please let us know.

Email us a "We read on-line" message to: [info@idic15.org](mailto:info@idic15.org)



# Upcoming Regional Gatherings

## ● Northeast

July 22  
Cindy & Andrew Johnson's home  
Central New Jersey  
Contact:  
Cindy Johnson 609-723-7731  
cynthiaj100@verizon.net

## ● California

Date TBA  
Golden Gate Park, San Francisco  
Contact:  
Evelyn O'Dell 925-827-9054  
evelynodell@astound.net

## ● Northwest

August 25-27  
Camp Prime Time  
Clear Lake, WA  
Contact:  
Greg Keller 360-270-3167  
gregkeller1887@msn.com

## ● Southeast

September 2  
Dewey Buck Destin Park  
Destin, FL  
Contact:  
Paige Gosa 678-462-4856  
pgosa@bellsouth.net

## ● Midwest

September 3  
White Pines Forest State Park  
Contact:  
Ruth Kross 708-691-0378  
lenasmom@sbcglobal.net

## ● New England

September 9  
Paula & Ben Davis' home  
Westford, MA,  
Contact:  
Paula Davis 978-392-8474  
xpdavis@comcast.net

## ● Germany

September 9  
Bersenbrück, Northern Germany  
Contact:  
Christian Reiter  
lissyppa@yahoo.com

## ● SAVE THE DATE!

June 28-30, 2007  
Seaport Hotel in Boston, MA

*The 4th International  
IDEAS Conference  
Sharing the Journey*



is a non profit organization that provides information, education and support to families affected by isodicentric and interstitial duplications of chromosome 15. **IDEAS** promotes research, awareness and understanding of isodicentric 15 and related disorders.