

AUTUMN ■ 2005

Conference Perspectives

JANE TRUE

was the conference coordinator for *Growing Up Together*, the 3rd International Conference on Isodicentric 15 and Related Disorders

AS YOU ARE READING THIS, OUR 3RD INTERNATIONAL CONFERENCE HAS BEEN WRAPPED UP AND DECLARED AN OVERWHELMING SUCCESS.

From the opening **Children's Parade** to the **Taste of Chicago** dinner to all the conference sessions, the evaluations were very enthusiastic. Whether or not you were able to attend, much of what occurred at the conference will be of interest to you. We try to bring you a little taste of the conference with this issue of the **MIRROR**.

Many many thanks are due the conference Planning Committee and all the many individuals who gave so much of their time, money, in-kind services, and enthusiasm to make the conference such a success. We had 250 people in attendance, including

46 individuals with idic15 and related disorders. In this issue you will find perspectives by siblings, grandparents, first-time attendees, the Activities Director, a Registration Volunteer, the hotel staff, and others. There is also a summary of the Research Roundtable, which was convened by **IDEAS** on the day before the conference officially started.

From the *Medical Information Day*, to the *Education Day*, to *Family Life Day*, much valuable information was exchanged. For a summary of each session consisting of the presenters' notes, please see the Conference Binder ordering instructions in this issue.

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).



Be sure to mark your calendars now for our **4th International Conference** to be held in Boston June 28th through 30th, 2007!

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SAVE THE DATE...

JUNE 2007

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A First-Time Attendee...

KAREN SALES

first-time attendee, is the mother of William, a 2-1/2 year-old boy recently diagnosed with idic15.

MY SON WILLIAM is now 2-1/2 years old. We were learning of the Isodicentric 15 diagnosis just as William suffered a big seizure last summer. I was excited to find this group online and learn about everyone else's experiences. Since the conference was in Chicago and I live in Iowa, I decided it was a perfect opportunity for me to learn more and meet some of the people I've talked with on the listserv. I invited my mother to join me for moral support.

When I first walked in, I was surprised by the large number of people present. I was feeling pretty intimidated by it all when Donna Bennett personally welcomed us, relieving some of my anxiety. The

orientation session gave me answers to questions I'm frequently asked about idic15. It was comforting to know that my questions and my family's questions were the same questions others were asking.

The three days were FULL of valuable information ranging from scientific/medical to education (from birth through adulthood) to daily activities like potty training. It was pretty overwhelming and emotional, but wonderful to know I was with people who understood! It was a relief that I didn't need to explain and they understood the frustrations and the joys I feel. It was enlightening to hear about and see the spectrum of how our children are affected by idic15 and that no matter where your child falls on the spectrum there is a caring community here.

While all of the sessions were great, I found I needed to

concentrate on a few topics that I was currently dealing with. My brain just couldn't absorb it all! Luckily the conference binder contains everything covered during the three days, so I can use that as a resource later.

Personally, I found the presentations on *Sensory Integration* to be the most helpful. I've been trying to implement some of the things discussed and I'm seeing some improvement.

Mom and I stayed with family trying to save money, but I found I missed out on a lot of socializing with other families. Next time I will stay at the hotel, and bring William and his extended family so they can share this experience.

Attending the conference was beneficial because I learned more about my son, learned more about myself, and learned that I wasn't alone. It's an experience that I will most certainly repeat.

A Grandmother...

BRENDA "MAMMA" MACKAY

is the grandmother of Evan, one of the children who worked with Soma at the conference to demonstrate for us the Rapid Prompting Method. In Brenda's words, "I really can't put into words what I got out of the conference. It was great."

FROM THE PARADE of "our" children with idic15 at the opening ceremony to the end where pictures were shown of the children having a great time while there, the conference was a learning, crying, joyful experience.

Evan, my "special" grandson, had the privilege of spending time with



that wonderful lady, Soma, who refused to take no for an answer concerning her son and his ability to learn. It was through wet eyes that I watched and heard with my ears that Evan's brain does function and he

knows a lot of things and we need to work harder with him to get it out.

Who would have thought we needed to first teach our children the difference between wet and dry before attempting toilet training? The price of sponges will probably rise as they have been known to be a great help in teaching the wet and dry method. We can't wait to try this experiment.

Not only did my daughter-in-law and myself learn a lot of new things at the conference, and we look forward to the next one, but Evan has been singing a new word ever since, "AMERICA, AMERICA!"

Through a Sister's Eyes...

SARAH MILER,

age 12 is sister to Jake Miller, idic15.

MY NAME IS SARAH MILLER and my brother, Jake Miller is six years old and has idic15. In June, my family and I attended the 3rd International Conference on Isodicentric 15 and Related Disorders. I would like to share my experience with you.

First of all, planning the conference was tons of fun! Having your mom on the conference planning committee requires a lot of extra work. I don't mean work like "this is so boring," but work like "this is actually really fun!" There are so many tasks to be completed in order to have a conference for almost 300 people.

We had a fundraiser at *Max and Erma's* (a local restaurant) which was a blast! We had a raffle, music, and tons of great food. Because of the fundraiser, **IDEAS** made a lot of money to go towards the conference.

The conference was very exciting and there was never a dull moment. There were tons of activities that were a lot of fun. We went swimming, played games, did crafts, and a lot more! Another fun highlight was the Taste of Chicago dinner. Everything served there was from Chicago. It was really good!

Another highlight of the conference for the kids was the sibling support group with Dr. Margot Touris. At first, when we all walked in we felt as if we were obligated to attend. We started by telling each other about ourselves. After a few minutes, Dr. Touris had us all talking about how we felt about



living with siblings with a disability. We learned all about how to express our feelings, how to deal with our siblings, and most of all, how to accept them. We all walked out with a good feeling, and felt that the kids' support group was a good experience after all! I would definitely suggest having another sibling support group at the next conference.

Being around all of these special kids with special needs was very emotional. They are all so sweet and loving. I grew very attached to each one of the kids and it was hard to say goodbye at the end of the conference. I truly wanted to take each one of them home with me. I also really enjoyed being with siblings of idic15 kids.

Going to the conference was a positive experience. I was very inspired by the entire event. I think if every kid put his or her mind to it, we could get a lot done. Kids can have fundraisers just like adults can. We can help raise awareness and funds for more conferences, speakers, equipment, and activities. I enjoyed the conference, meeting other families, making new friends, and realizing I am not alone in our idic15 experience.

The Hotel's...

PRIOR TO THE START of our 2005 conference, two sensitivity training sessions were presented to the hotel staff. These sessions were prepared by Ruth Kross and presented by Ruth and Jodi Miller. Clare True, idic15 assisted on one of the days. Ruth's presentation covered such areas as using people-first language, how to address and communicate with our children with respect and dignity, possible behaviors to be prepared for, how to physically prepare the session rooms so that our children would be most comfortable, what items we might need in extra supply (eg. pillows and blankets), how to handle a medical emergency such as a seizure, and in general how to best be of service to our group. Approximately 30 hotel employees attended each session, and at the end the questions and answers were quite extensive and sometimes emotional. These sessions were very well-received, and it was generally agreed that these sessions helped to prepare the Hyatt staff to be comfortable with our group before actually meeting us, and to be alert and responsive to our needs upon our arrival, as well as being great opportunities to increase awareness of Isodicentric 15 and related disorders. Thank you Ruth and Jodi!

On behalf of the Hyatt family, we would like to thank you for giving us the privilege and pleasure to host the IDEAS Group. The Sensitivity Training gave us an insight into Isodicentric 15 and an admiration of your strength and patience. It also gave us a new sense of purpose, understanding, and pride at getting the opportunity to work with you so closely. You have heightened and enriched our experience and have indeed made a valuable and visible imprint on our hotel. Hyatt will continue to welcome diverse clientele, and we will always strive to exceed each group's expectations.

Thank you so much for trusting us with your Group and we hope we will have the pleasure to welcome your wonderful families again soon.



An Outsider Looking In...

LAURA CASHMORE, twin sister KAREN & niece CHRISTEN

volunteered as Activities Directors for the Family Break Room during the conference, and helped to organize the opening parade. Both Laura and Karen are professional gymnasts, operate their own gymnastics schools in their respective communities in Missouri and North Carolina, and are committed to providing services and assistance to those in their communities who have special needs.

THIS WAS MY FIRST experience at a conference with other people who share a disability, idic15, with one of my very close friends, Clare. I left with a great feeling of community, an increased awareness of this condition and all that it involves, and with the courage to see that people are no longer alone in facing their child's disability.

My twin sister Karen, her daughter Christen, and I had the privilege of joining with people and sharing their experiences with them. I have been Clare True's long-time friend and fitness instructor. I joined in the conference experience so I could share this meaningful opportunity with Clare and her family. I am a part of her world and it was a great honor to be there as her friend.

The first thing I gained from the conference was a sense of *community*. It was a great feeling talking with people about their thoughts and ideas pertaining to idic15. As a group of people, you have created a great resource for one another. People from all walks of life came together to share their experiences in raising their child. Comparing notes with each other can

be a great source of strength when surviving the day to day struggles of raising a child with idic15.

The area of appropriate and effective *education* was the second area that I found very informative at the conference. I realize how hard it was for new parents to be there, but you now have resources of doctors, and research that was not available 15 years ago. Various methods for communication are being accessed for children who struggle with their speech and communication. The road is easier to travel when you are not alone. Hook into your listserve, chat and share your struggles with people who are survivors. There are good stories from incredible people who participated in the conference.

And finally I would like to applaud your *courage* for being there. The families there represent people who care, and want to make a change. So many people worked so hard to pull all of you together so we all can make a difference. That's why I attended. One person can make a difference and all of you represent the leadership needed to survive.

My very best to all of you.
I look forward to seeing you all again some day.



**3RD INTERNATIONAL
CONFERENCE ON
ISODICENTRIC 15
& RELATED DISORDERS**

Binders Available

Conference binders containing speaker presentation materials are available for purchase.

Order forms are available on the IDEAS website...

<http://www.idic15.org>

If you do not have internet access, you can write to Lori George at the address below. All binders shipped within the U.S. cost \$20 per binder. International orders are \$25 per binder. All payments should be made in U.S. currency.

Checks should be payable to IDEAS and sent to: L. George
18 Kings Road
Canton, MA 02021

A Volunteer's...

DANIELLE WAGNER

volunteered with the 2005 conference

LAST YEAR, MY GOOD FRIEND, Jodi Miller, asked if I would have an interest in participating in the 2005 IDEAS conference by creating a slide show presentation. While I knew this would be a very worthwhile project and accepted the task without hesitation, I had no idea at that point just how rewarding the experience would prove to be. As the months went by and the pictures started flowing in, I was overwhelmed by the beautiful children who appeared on my computer screen each evening. Their smiles and bright eyes warmed my heart. Arranging the photos gave me a sneak peek into their lives and made the anticipation of meeting them in person grow as the conference drew closer.

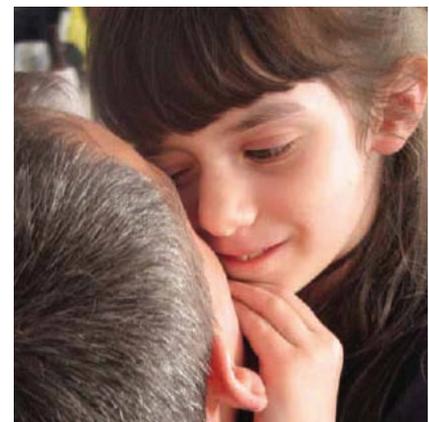
When the first day of the conference arrived, I showed up ready to take my place at the registration desk. This was absolutely the best way to meet the families who had traveled from down the street and around the world and to finally meet the boys and girls I had already

fallen in love with. The buzz and energy that filled the corridor was instantly uplifting and I was touched by the way families who had never met previously instantly connected with one another. I listened to parents telling other parents of their highs and lows, sharing helpful tips and I saw tears shed by parents of newly-diagnosed children who it seemed had the weight of the world lifted off of them by simply finding others who knew what they were going through.

During the conference I was also able to listen in on many of the speakers and learned more in those couple of days than I had ever learned in biology. I found it staggering how much has been

learned in the last decade about idic(15) and related disorders through the dedication of many professionals and the tenacity of the parents pressing to know more. The parents I met were such amazing advocates for their children...a strong lesson I took from the conference.

Now that the conference has concluded, I find that I miss working with the pictures of these terrific kids. I also miss being of service to an amazing group of people. I hope that I will be able to contribute to a similar project at the next conference so that I can see how the kids have grown and hear great stories of the progress they've made.



IDEAS Research Roundtable Explores Links Between idic(15) and Seizures

NICOLE CLEARY, MSS,
is the Executive Director of IDEAS

AS PART OF OUR MISSION, IDEAS convenes research roundtable meetings to stimulate research into duplications of chromosome 15q. These meetings bring scientists together to talk about their interests in isodicentric and interstitial duplications of chromosome 15, and to promote research collaborations. On June 22, 2005 IDEAS convened its third research roundtable meeting. This meeting represented the first time parents and scientists came together to explore the link between duplications of chromosome 15q and seizures.

The roundtable meeting had three objectives:

- 1) To review current research on idic(15) syndrome, with special emphasis on what is known about the relationship of this syndrome to seizures.
- 2) To identify priorities for future research to further characterize seizures in idic(15) syndrome.
- 3) To determine how the meeting attendees might collaborate on research priorities identified at this meeting.

Our meeting started with five parents sharing their children's experience with seizures. The scientists in attendance heard about seizures that started in infancy, in childhood and in adolescence. Families described how children

started with one seizure type and other seizure types emerged as the child aged. Some seizures were easily controlled with the first medication, other seizures were controlled for awhile and then became more complex, and a few families described seizures that have never been controlled with medication. It was helpful for everyone to begin this meeting with the realization that over half of all individuals with idic(15) will experience seizures, but seizures are not uniform in their onset, their presentation or in their response to treatment.

IDEAS Professional Advisors Brenda Finucane and Dr. Carolyn Schanen provided an overview of the current understanding of the relationship of idic(15) to seizures, drawing from the IDEAS seizure survey and Carolyn's NIH funded study on the Molecular Investigations of Duplications of Chromosome 15 in Autism.

The meeting then turned to an examination of the role of Gamma Amino Butyric Acid (GABA). GABA is a major inhibitory neurotransmitter in the brain. There are three GABA_A receptor subunit genes that are commonly duplicated in idic(15). It is hypothesized that the abnormalities in these GABA_A receptor genes may play a significant role in the seizures experienced by people with idic(15).

Dr. Martin J. Gallagher from Vanderbilt University Medical Center in Nashville, Tennessee provided a very technical presentation regarding the many consequences of abnormal GABA_A receptor expression. He showed that even minor changes in the receptor structures can lead to human epilepsy syndromes. The methods that he uses to analyze the function of GABA_A receptors in the lab could be applicable to characterizing the effects of the extra copies of the GABA_A receptor genes in individuals with chromosome 15 duplications in isolated cells.

This presentation was followed by Dr. Diane Chugani from Wayne State University in Detroit Michigan. Dr. Chugani is currently engaged in a study to measure brain GABA_A receptor binding in children with chromosome 15q11-13 mutations through the use of positron emission tomography (PET). Understanding of GABA_A receptor binding abnormalities in subjects with chromosome 15q11-13 duplications may lead to clues regarding the diverse presentation of seizures among individuals with this syndrome as well as to the autism and cognitive disabilities in these children.

Dr. Art Beaudet from Baylor College of Medicine in Houston, Texas spoke about the role of the Prader-Willi/Angelman region in determining the clinical features in individuals with duplications of chromosome 15q. Dr. Beaudet's research on genetic imprinting

explores parent-specific gene activation (ie. genes coming from the maternal vs. paternal chromosomes). He suggested there is a fair amount of overlap in the phenotypes of individuals with Angelman syndrome and idic(15). Angelman syndrome is caused by an interstitial deletion/mutation in the maternal copy of chromosome 15q11-13 and the effects of idic(15) are more severe when caused by a maternal duplication of chromosome 15q11-13.

The final scientific presentation of the day came from Dr. Timothy DeLorey at the Molecular Research Institute, in Mountain View, California. Dr. DeLorey has been involved in research with mouse models of Angelman syndrome. He provided information about the connection between chromosome 15 abnormalities, seizures and one of the GABA_A receptor subunits. He noted that some of the issues he has heard parents of Angelman patients discuss in regards to difficulty with seizure treatment he also heard from several parents of children with idic(15) at the roundtable. He speculated that the similarities in epilepsy treatment difficulties in both Angelman and idic(15) syndromes could have the same root cause, but more research is necessary to investigate this possibility.

In terms of future research priorities, roundtable participants agreed that Dr. Chugani's study provides a critical opportunity to learn more about what is happening

with respect to how GABA_A receptors are functioning in the brains of individuals with chromosome 15 duplications. Knowing whether the duplicated genes on 15q11-13 is resulting in over expression or under expression of the encoded proteins in the brain is will strongly impact the direction of future research in regards to both epilepsy and other symptoms of idic(15) syndrome.

In addition to studies of individuals living with idic(15) and int dup(15), roundtable participants encouraged **IDEAS** to educate families about the tremendous value of donating the brains of family members with chromosome 15 duplications to one of the national brain bank repositories after death. These are NIH funded brain banks that provide tissue for research into basic disease mechanisms, which have provided powerful information as to the processes that are disrupted in patients with numerous genetic disorders. Research on post mortem brains is essential to increasing our understanding of how idic(15) effects neural structures and processes.

Finally, participants debated the value of a seizure medication survey to identify seizure medications that have been effective as well as those that were ineffective or made things worse. This would be a valuable resource to both parents and researchers. However, a seizure medication survey would require the involvement of a neurologist or epilepsy researcher because the results of such a survey must be

evaluated by individuals trained in interpreting this sort of data, who can also publish it in a well circulated medical journal.

As a result of this meeting, some exciting collaborative projects are in the works that may provide much greater information about the connection between idic(15) and seizures. Dr. Gallagher has interested his colleague Beth Malow, MD, MS, Associate Professor of Neurology at Vanderbilt (Nashville, TN) in the challenges of idic(15). Dr. Malow currently leads a study looking at the causes of insomnia in children with autism spectrum disorders. She also studies the overlap of sleep and epilepsy, including the effects of treating sleep disorders on epilepsy, the relationship of epileptic seizures and interictal epileptiform discharges to sleep, and the effects of anti-epileptic drugs and vagus nerve stimulation on sleep. We will have more to report on Dr. Malow's research in the next issue of the **MIRROR**, and on the **IDEAS** website and listserv.

The 2005 research roundtable was an exciting moment when parents and researchers came together to address the unanswered questions regarding the relationship of idic(15) to seizures and autism. **IDEAS** will continue working toward the day when these relationships are well characterized, and a discussion of targeted and effective treatment options becomes a reality.

Families Increase Research Opportunities for Chromosome 15q Duplications

IN THE WINTER, 2005 MIRROR we introduced an important new



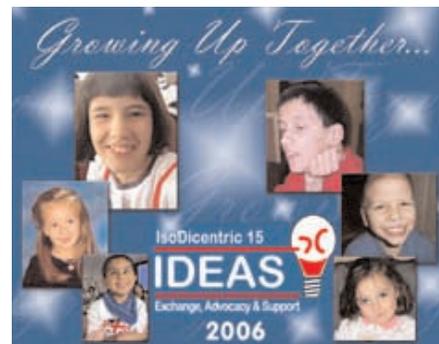
opportunity for families to help increase scientific interest in studying chromosome 15q duplications. Cell lines from patients and their families are a critical resource for researchers, but it is often a major challenge to find families with specific chromosome abnormalities, like *idic(15)* and *int dup(15)*. Researchers often turn to tissue and cell "banks" for access to samples from patients with a specific diagnosis. These samples are often in the form of a cell line, which are derived from a blood sample that has been specifically treated to allow the cells to be grown in culture for extended periods of time.

IDEAS is collaborating with Carolyn Schanen and the Human Genetic Cell Repository at the Coriell Institute for Medical Research in Camden, New Jersey to establish a large collection of *idic(15)* and *int dup(15)* cell lines. This collection will help spur research into chromosome 15q duplications by making it much more feasible for researchers to access cell lines. The Coriell repository currently has 8 lines from Carolyn Schanen's lab. At the 2005 conference, another 21 consent forms were signed from families who are already enrolled in Carolyn's study, and 4 families who have not yet participated in Carolyn's study. A total of 33 cell lines will be available for research as soon as Carolyn's lab can make the transfers and new families can provide their blood donation. We are off to a great start!

The collaboration between **IDEAS**, Carolyn's lab and Coriell is continuing. Families who are already enrolled in Carolyn's study and would like to authorize Carolyn's lab to share their cell lines are encouraged to contact Tina Sellers, Genetic Counselor at the Coriell Cell Repositories who is coordinating the donations of these lines. Families who would like to provide cell lines to the repository but are not enrolled in Carolyn's study are also encouraged to contact Tina to learn how they can donate their samples.

Tina Sellers, MS, CGC
Genetic Counselor, Coriell Cell Repositories
Coriell Institute for Medical Research
403 Haddon Avenue
Camden, NJ 08103
856.966.5062 Phone
856.757.9737 Fax
tsellers@coriell.org

*Working together, we can build an *idic(15)* and *int dup(15)* cell repository that will bring even more research attention to this rare disorder.*



IDEAS 2006 Calendar Available Now!

Contains over 70 pictures of beautiful children and young adults affected by isodicentric and interstitial duplications of chromosome 15.

What a great way to raise awareness and celebrate the beauty of our children! The calendar contains information pages that are easy to photocopy and share with families and professionals who work with your children.

Order forms are available on the **IDEAS** website

<http://www.idic15.org>

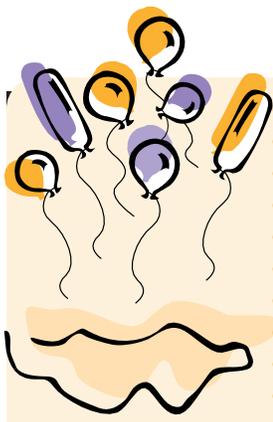
No internet access?

Write to Lori George at the address below.

Only \$15 each, order yours today!
Make checks payable to **IDEAS**
and send to:

L. George
18 Kings Road
Canton, MA 02021

Calendars will be shipped
November 18th & December 9th



Happy
Birthday
to these
special
people:

• Brenda M	09/02/89	• Joshua C	10/02/94	• Chad L	11/27/75
• Cheyenne D	09/08/92	• Riley R	10/04/03	• Lisa P	11/30/90
• Bente S	09/09/97	• Sem K	10/05/99	• Ruth O	12/02/89
• Elana K	09/09/88	• Joshua K	10/10/01	• Joseph T	12/03/96
• Dante R	09/10/98	• George L	10/17/81	• Jared L	12/04/02
• Thomas N	09/10/93	• Nathan L	10/18/01	• Naomi H	12/07/97
• Major R	09/10/98	• Makenna J	10/22/01	• Abigail S	12/09/97
• James S	09/11/93	• Renate D	10/22/92	• Matthew K	12/12/96
• Waawiiya W	09/11/93	• Jarod W	10/30/89	• Ellie S	12/12/96
• Julia W	09/12/96	• Shay P	10/31/92	• Chase W	12/13/00
• Raquel L	09/14/00	• Anna B	11/03/99	• Cody C	12/14/95
• Rachel M	09/14/94	• Richard M	11/04/86	• Corey C	12/14/95
• Breauna E	09/15/95	• Corrina C	11/05/96	• Sven D	12/16/98
• Mason L	09/15/99	• Carrie J	11/07/83	• Olivia T	12/16/94
• Jeremy L	09/17/88	• Rohan N	11/08/02	• Nicholas B	12/18/87
• Conor M	09/19/96	• Hayley P	11/08/99	• Caroline W	12/22/00
• Brandon J	09/24/97	• Will G	11/11/98	• Austin B	12/22/95
• Sarah L	09/23/95	• Taylor P	11/12/02	• Megan R	12/22/95
• Evan M	09/28/94	• William C	11/14/94	• Justin O	12/23/97
• Chase G	09/30/99	• Stephen L	11/15/93	• Chelsi C	12/23/93
• Cara V	09/30/92	• Lindsay K	11/15/90	• Chandler R	12/27/96
• Logan K	10/01/01	• Twan V	11/19/02	• Ada E	12/29/99
• Sarah D	10/02/96	• Chandler G	11/20/98	• Teddy S	12/29/90
• Adam R	10/02/91	• Melanie P	11/22/86	• Kevin C	12/30/95
		• Andrew S	11/25/01		

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

or to IDEAS
PO Box 4616
Manchester, NH 03108.

Jane True, Editor

Lin Neighbors, Design & Layout

Printing costs for this newsletter provided by Elwyn, Inc.

Announcing: The Sunshine Committee A New Opportunity to be Involved with IDEAS & Our Wonderful Children

RUTH KROSS

is mom to Marlena, *idic15* and Chair of the IDEAS Sunshine Committee

THE SUNSHINE COMMITTEE is a new committee charged with putting a smile on our kids' faces on occasions special to them. That could be a birthday, a holiday of special significance, or just a day chosen by a family to make a child feel special. An example is a recent card shower organized for a young woman on St. Patrick's day which was enormously successful and enthusiastically carried out by over 30 families, many from our IDEAS group. We would also like to celebrate in our kids' accomplishments and make them feel special by having members of the Sunshine Committee acknowledge their achievement.

The Committee will be directed by Ruth Kross, mother of Marlena. Before finalizing the role of this committee, we are looking for any input, thoughts, ideas, or suggestions that anyone may have regarding the possible role of this committee. We are also looking for volunteers to help on the committee. Anyone with ideas or anyone interested in volunteering can contact Ruth Kross at

lenasmom@sbcglobal.net

or by cell 708.691.0378. Don't miss out on this great chance to get to know our children better and to be a part of putting smiles on their beautiful faces!

Cheyenne

ELLEN DOXTATOR

is mom to Cheyenne, 13 (idic15) and Samuel 11

AS PARENTS OF SPECIAL NEEDS CHILDREN, we experience many moments of sorrow from the first moment we realize that there is something wrong, to realizing the degree of the disability, to acknowledging that our children will always be different and that there is no cure, to knowing that our lives will be forever impacted as well as our child's. We can easily get caught up in the negatives, other people's perception of us and feeling lost as we deal with the medical professionals who may know little of the Isodicentric 15 disorder, but sometimes are quick at making judgments about our child's potential.

Few people have helped me to acknowledge and celebrate my child's special needs. While I see my daughter Cheyenne as special and amazing, this is a rarity in the medical profession, the school system, in the community, or in our everyday lives. They focus on how Cheyenne looks, the fact that she is in a wheelchair, or how she is behaving. What they miss is the amazing sense of humour she has, the deep insight she has, and her ability to always use the right words at the right time.

There have been two people who have made me celebrate Cheyenne. The first was Cheyenne herself. Doctors told us she would never walk or talk or see, but from the story that follows, you will see that Cheyenne has helped us to believe in little miracles everyday.

Yet another year has passed and as always it has been busy. Cheyenne continues to have lots of medical appointments and based on average calculations, we have completed close to 880 appointments over the past 12 years. It is still quite stressful trying to coordinate the medical appointments while working full-time. But sometimes Cheyenne reminds me that it is all worthwhile. Usually when we are at appointments at the hospital and if Cheyenne is fairly cooperative with the doctors, she is rewarded with a treat.

This day we lined up so she could get a donut and a drink. The line-up behind us was long and as we were waiting Cheyenne noticed a doctor beside us. In typical Cheyenne fashion, she put out her hand and said to him, "Hi, I'm Cheyenne." The doctor shook her hand and said, "Hi, I'm Doug." Cheyenne, not missing a



CELEBRATING THE SPRIT OF DIFFERENCE

beat pointed to me and said, "And this is **my mother. She's the expert.**" The doctor said "oh really" and everyone in the line had big smiles on their faces. I was kind of taken aback as I didn't even know that Cheyenne knew the word "expert" or what it meant. So after we sat down at the table, I quietly said to Cheyenne, "Do you know what expert means Cheyenne?" She looked at me ever so seriously and said, "It means **Superhero, Mommy.**" And after that, all the difficult appointments didn't seem to matter. And later I thought to myself that Cheyenne reminded me that often parents forget how well we do know our children.

So maybe I can keep going for another 880 appointments and maybe the doctor will remember Cheyenne's comments next time he sees a family. Whenever I think of Cheyenne's comments, I can't help but smile... Cheyenne reminded me to celebrate the amazing progress she has made

and the wonderful person she has become.

The second person who has helped me to celebrate my daughter I met recently. At the 3rd International Idic15 conference we had the opportunity to meet and experience the amazingly talented and awe-inspiring Rick Guidotti. His previous work as a famed professional fashion photographer has led him now along a unique path: to photograph and promote positive self images of individuals with genetic differences. Through his non-profit organization Positive Exposure, he celebrates the “spirit of difference” through positive images. You could feel his energy as he photographed the kids coming in for the initial parade. Cheyenne immediately took a liking to him, even though she is fearful of having her picture taken as she is afraid of the flash. He snapped a picture of her just as she was blowing the pinwheel that each child was given as part of the parade. Even as we passed Rick to take our seats, he said “Amazing” to Cheyenne and I thought to myself, how many people take the time to acknowledge Cheyenne as the amazing young lady she is.

Rick presented his work on Albinism and his presentation clearly demonstrated how he has positively impacted individuals’ lives worldwide.

It was a wonderful way to start off the conference on a positive note as we were able to see the world through a different viewpoint, through Rick’s eyes and lens. As Rick said, so often individuals with medical conditions such as IDIC 15 are photographed against a stark hospital wall looking frightened. The doctors are more concerned with capturing the child’s physical features and not their spirit of individuality. Rick not only encompasses that spirit but is able to capture it for the rest of the world to see that spirit of individuality too.

Later, when Rick photographed Cheyenne while reading a book, he commented on the fact that she is very photogenic. This word has never before been used to describe Cheyenne after hundreds of pictures of her eyes closed and her face screwed up. Often family members did not want pictures of Cheyenne – that is how bad they were. I had resigned myself to the fact that bad pictures were the norm for her and that I just had to accept this. When Rick approached us again at dinner, I told him of Cheyenne’s fear of pictures and he said “Oh, we are not taking Cheyenne’s picture, we are taking a picture of Captain.” This was Cheyenne’s stuffed whale that she had brought with her. Rick then said “Smile, Captain” with such

energy and joy that he captured Cheyenne with eyes open, relaxed and feeling special. Subsequent pictures were taken with Cheyenne’s other stuffed animal friends and her Canadian football. Although I had previously tried this technique at home, I was not successful in achieving such wonderful pictures. Family members are thrilled to have such lovely pictures of Cheyenne. Me, I get tearful just thinking about them.

Yes, Rick reminded me to appreciate the wonderful aspects of our children with Isodiscentric 15 and seeing the joy in Cheyenne’s face whenever he was around was awe-inspiring. My only complaint is that now Cheyenne is telling me that she would like a digital camera to take to camp because they take better quality pictures!!! Wonder who she got that from? Rick, you made my girl (and Captain) smile and captured her spirit and for that I am eternally grateful. Keep your visions of hope alive and continue celebrating the spirit of difference and individuality. Together with Cheyenne, I hope that medical professionals will be able to see beyond the physical features and diagnosis. And remember, you are “the expert” and a Superhero to families like ours. Believe me, Cheyenne knew it from the moment she met you.



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