

THE MIRROR

The Newsletter of IDEAS - IsoDicentric 15 Exchange, Advocacy and Support

IDEAS Board of Directors

IDEAS (IsoDicentric 15 Exchange, Advocacy and Support) is a parent support group that was created by Donna Bennett, Mother of a son with Inverted Duplication 15 and Brenda Finucane, genetic counselor at Elwyn, Inc. Elwyn is a non profit organization in Elwyn, PA serving adults and children with a wide range of special needs. When Donna and Brenda originally formed IDEAS, there were very few families who had a diagnosis of idic(15). Now that genetic screening is becoming more common, there are many more of us who have our children's diagnosis and we have become a much stronger network thanks to the Inverted-Dup15 listserv created by Erica Jackson (Mom to Cheyenne) and facilitated by Paul Rivard (Dad to Megan).

Because of the growth of IDEAS families, we were able to hold our first national conference in June, 2001 in Philadelphia. At this conference, a group of parents decided to form a Board of Directors to provide leadership and guidance for IDEAS. The term of the first Board is from June 2001 – 2003. Board members include Nicole Iseli, Chair; Brian Gazewood, Co-Chair; Paul Rivard, Finance Officer; Heather Bruce, Secretary; Donna Bennett, IDEAS Founder and Board Member; Jodi Miller, Board Member and Patti Rubel, Board Member.

Since the conference, the board has developed a mission statement, a set of bylaws to guide our work and a committee structure to carry out most of the activities for IDEAS. The mission statement for IDEAS reads:

IDEAS is a network of parents and professionals providing information and support to families of children and adults diagnosed with IsoDicentric 15 or other abnormalities of Chromosome 15. Our mission is to serve our membership by uniting families, researchers, and professionals, and promoting awareness and understanding of IsoDicentric 15/Chromosome 15 abnormalities.

The five committees that have been launched in 2002 include: 1) Fundraising; 2) Education/Public Awareness; 3) International Membership; 4) Meeting Planning; and 5) Research. The committees are open to any parent or professional that wants to work with us. Following are brief descrip-

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Isodicentric 15 idic(15), formerly known as Inverted Duplication 15, is a disorder in which a child is born with extra genetic material from chromosome 15. Symptoms may include: poor muscle tone, developmental disability, seizures, and autism. Although the exact prevalence is not known, Isodicentric 15 is one of the most common causes of autism.

**ATTENTION
idic(15)
Families and
Friends!**

Mark Your Calendars!

The next International idic(15) Conference is currently being planned. It is scheduled for June 19-21, 2003. The conference will take place at the Philadelphia Airport Marriott in Philadelphia, Pennsylvania.

Watch for more information to come soon from the IDEAS Meeting Planning Committee!

tions and contact information for each of the committees.

Fundraising Committee: Show Me The Money!!!

We are looking for a few good people that have some fresh ideas on how to go about raising money to help support IDEAS related activities. In 2002, we will establish a mechanism for collecting funds and we will identify a few events as a focus of fundraising efforts (things like future conferences, outreach materials, distribution of research reports to families, etc). We will brainstorm different ways to become a self-sufficient entity by having the finances to support ourselves in the future. Whether you have an interest to be on the committee, or just want to share an idea or two, please let us know!

Contact Paul Rivard, committee chair, at <privard97@attbi.com>.

Education/ Public Awareness: Lots Of Families Are Raising Special Kids With idic(15)

We will develop a public awareness plan to educate families, geneticists, neurologists and other medical professionals about the existence of IDEAS and our listserve. Let's join together to find ways of preventing future diagnosed families from feeling alone and isolated with this rare genetic disorder. There are also a lot of families out there who do not yet have a diagnosis of idic(15) for their special needs child. We can act as a catalyst to get the word out about the importance of genetic testing, especially for kids who show some of the hallmark idic(15) features: hypoto-

nia, mental retardation, autism, and seizures.

Contact Ruth Kross, committee chair, at <ruthkross1@aol.com>.

International Membership: Who We Are And How We Connect

The International Membership Committee will focus on developing an international database of families raising children with idic(15). This database will be SECURE and will not be published on the web. The goal will be to store contact information and some additional information on families raising children with idic(15). The information will be used by board members and



regional representatives to assist families in networking and finding other families with similar situations. The international database is purely voluntary and families can choose whether or not they want to be listed. The international membership committee will work closely with the regional representatives to keep track of all members of IDEAS. (regional reps are invited/encouraged to join this committee).

Please contact committee chairperson Paul Rivard at <privard97@attbi.com>.

Meeting Planning: Let's Get Together!

The Meeting Planning Committee will be the place where IDEAS get-togethers are planned. Our goal is to

plan and implement some local and regional meetings, and begin planning for our next national/international conference in June 2003 in Philadelphia, PA. We will plan meeting details such as sending out invitations and locating appropriate speakers and materials. Do you love planning parties? Then this is the committee for you!

Contact Vicki Miller, committee chair, at <VickiDevon@aol.com>.

Research Committee: What We Know And What We Are Learning

The Research Committee will keep track of current research on idic(15) and related disorders (autism, seizure management, etc.). We will collaborate with researchers and other professionals who are working on research and treatment issues for people with idic(15). We will also facilitate communication between researchers and idic(15) families by distributing progress reports on current studies and keeping families informed of ongoing research opportunities in which they can participate.

Contact Jodi Miller, committee chair, at <j3smiller@aol.com> .

The IDEAS Board is really excited about the work ahead of us. Our efforts will have a direct benefit to our families and the families we share so much with. We hope you will join us in the year ahead!

-Nicole Iseli



Unique Family Day Out

September 8, 2001

I can't tell you how much we had been looking forward to this day out. The day started off very disappointing weather-wise. I thought, "Oh Dear, here comes the rain". It rained almost halfway there and then miraculously the sun came out!

We had prearranged a meeting point inside Gulliver's with other families, so at 11:30 a.m. we could all meet up for lunch at the big castle in the centre of Gulliver's. On entrance to the park I had met a few families already. By the time 11:30 came 'round there was one family, then another, then another. I was overwhelmed. Everyone seemed to be coming towards us all at once. It was great!

Twelve families attended and we spent some real quality time together with all of our children. Everyone got on so well with each other. It was like meeting up with old friends. After lunch I managed to get everybody together for a group photo. We then arranged to meet for afternoon tea back at the castle at 4:00 pm. We separated for a couple of hours to wander 'round the park having a good time on all of the rides. Every now and then as you went 'round a corner you'd bump into another family. We shared lots of rides together.

Afterwards, we met up for tea and doughnuts. The rest of the time was spent having a good chat, finding out about each other and comparing our children's similarities. As always, we realized that while our children are similar they are indeed "Unique."

The day ended at Gulliver's with a singing and dancing parade around the carousel and we all joined in. It was very hard to go

home and leave all of these special people. It isn't until you go home that you realize how isolating having a child with a rare chromosome disorder is. We can look forward to meeting up again next year!

(The family day out consisted of nine idic(15) families, one deletion 15 family, one translocation 15/21 family, one partial duplication 15 family, and one ring 15 family.)

-Marion Mitchell

ROB'S POEM

(PLEASE DO NOT JUDGE ME)

Please do not judge me
Please do not stare
I don't know I'm different
Do you think that's fair?

I am just a child
Who loves as any other
If you stare at me
You are staring at my mother

Please try to understand that
Not everything in life is the same
I cannot help who I am
No one is to blame

I want to enjoy my life
As do my Mum and Dad
So when you do not understand
It makes me very sad

Please stop and say hello
Please ask my name
I may not be a "normal" child
I may not be the same

I cannot speak in words alone
I cannot talk at all
But please come and talk to me
And stop me feeling small

Please do not judge me
Please do not stare
Please accept me for who I am
When I am standing there...

*- Marion Mitchell
Written in 2002, on behalf of
her son Rob, aged 7,
who has idic (15)*

Information on Unique

UNIQUE – The Rare Chromosome Disorder Support Group

P.O BOX 2189, CATERHAM

SURREY, CR3 5GN, ENGLAND

HELPLINE TEL NO: +44(0)1883 330766

Email: info@rarechromo.org / Website: www.rarechromo.org

- Main Contact is Beverly Searle, Development Officer.
- Unique is a registered charity (number 1024624), which provides support to families whose children have any RARE chromosome disorder.
- Membership of Unique is free but voluntary donations are always welcome. Members will receive our thrice-yearly newsletter.
- Unique hosts an annual conference.
- Unique has an international membership of 2800+ members in 50 different countries. We help to link families with similar disorders as well as linking families living near one another.
- The Unique team is made up entirely of parents/relatives whose children have rare chromosome disorders.
- Unique has adopted and works to an equal opportunities policy and is registered under the Data Protection Act (Registration Number A2760128).

Family Portraits

Anna's Story

Anna was born November 3, 1999 after a normal pregnancy. She came very quickly into the world by natural childbirth. She was kept in the Neonatal Intensive Care Unit for "non-bacterial pneumonia" for a week. We had no idea that there were any other problems. We had always noticed that she seemed cross-eyed, but passed it off as just part of being a newborn. By her 4-month check-up, I pushed a little harder to have her eyes checked. We were referred to a pediatric optometrist who prescribed glasses for farsightedness. This was to cure all of her delays in fine motor skills, though it didn't. At this point, we had just started occupational therapy. She still wasn't holding her head up. We saw a neurologist in May. Luckily, he ordered all the right tests. An upper GI revealed a mal-rotation of the intestines-supposedly unrelated to idic(15). The neurologist called us at 9pm on a Monday evening in June of 2000 with the blood test results and told us that Anna had Prader-Willi Syndrome. We were devastated. Our parents rushed over to console us. The following day, our entire family got together to help take our minds off of the pain.

The following Monday, we took Anna to Riley Children's Hospital for a swallow evaluation. Since birth, she had choked while nursing at almost every feeding. The test showed that she was aspirating. She was admitted immediately and an n-g tube was placed for nourishment. Two days later, she had surgery to place a g-tube and correct the mal-rotation. She spent a



Anna

week in the hospital. The day following the swallow study, Ron and I were taken to a conference room filled with various specialists. We were told that Anna did not have Prader-Willi, but Inverted-Dup 15. We were given the IDEAS website to find information, but told we'd only find the worst-case scenarios. We had no choice, though, if we wanted to find any information on

idic(15). We've never regretted for a moment going to the website. We found our way from there to the email list where we gained invaluable information and friendships.

Feeding issues were our main problem with Anna. She struggled to keep weight on since she was 4 months old. This was surprising after her healthy birth size of 8 lbs., 7 oz and 22 ½ inches long! The g-tube had periodically been her only source of nutrition. An OT day was added to her already full week of therapy to concentrate on oral mechanics. We used oral stimulation such as an ice-cold spoon, lemon/pickle juice, vibrating toothbrushes, and a nuk brush to "wake up" her mouth before and after a meal. At 16 months of age, she weighed only 18 pounds. We added Carnation Instant Breakfast to her milk for calories. By two weeks later, she had gained 4 pounds! She stopped using the g-tube last May and has continued to gain weight. It was finally removed at almost 23 months of age.

Anna has 5 therapies a week including occupational, speech, physical, developmental, and oral motor therapy for an hour each. We have seen the progress from each therapy. She has tactile defenses that are much more controlled since adding brushing daily. She has begun walking independently and has limited verbal skills. She has approximately 70+ words; some of

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ANNA *continued from previous page*

which she uses spontaneously and appropriately. Last August she was labeled “mildly autistic”. This wasn’t much of a surprise, though no easier to hear. She has 50% delays. The gap is beginning to widen. However, we remain encouraged by every new sign of cognitive awareness.

Anna has very recently begun “playing” with her older sister, Holly, who is without a doubt her best therapy. It’s so exciting to see Anna watching and laughing at Holly. We take great delight in watching them roll around on the floor giggling.

We have a fifteen month-old daughter, Ellie. We know that the day will come when she surpasses Anna in many ways. I’m sure no amount of planning can cushion the pain that it will cause. I know we’ll get through it, though...just as we have along this already long road we’ve journeyed with Anna. Just one smile, one little belly laugh, makes every discouraging word the doctors say fade into the distance.

- Heather Bruce

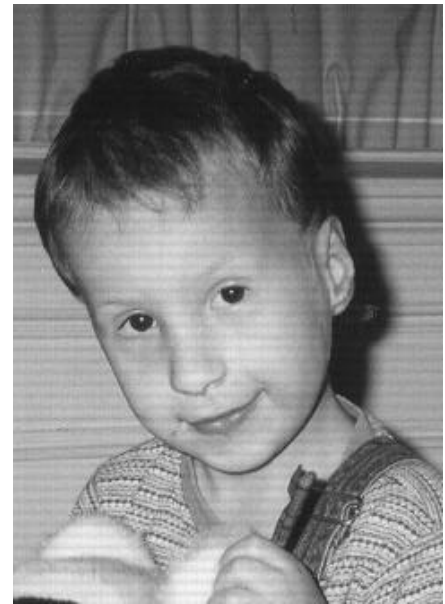
Joshua’s Story

Over the last eight years, people have asked me how my husband Robert and I were able to deal with the news of our Joshua’s genetic difference *idic(15)* and his initial challenges. I respond that he was our first and our only experience with a newborn and that experience was visiting him in NICU for three weeks. He under-

went open heart surgery for aortic stenosis. We had never known anything else. The therapists and specialists evaluated him: his inability to nurse (poor muscle tone), his tightly curled body (hypotonia), his conjugated uvula (a mild version of cleft) and the multitude of tests and x-rays that ensued - it was a new world we had entered. This introduction to parenting made us realize that there are many things in life we just have no control over, and we have to let go and let God take over.

Joshua is seven now. He functions at about a two-three year level, is not quite potty-trained and not really verbal. He can say about six words and sign about ten more. He has a five year-old brother, Jacob, and a one year-old brother, Jonah, who has become his best buddy. In having more children, it has been so amazing to realize how fast most children learn and imitate. For Joshua, every milestone has been achieved through therapies and patience, in his own time. After three years of EIP and two years of public school TMH program, I was fortunate to be referred to a program called NACD (National Academy of Child Development), or nacd.org. I feel like this organization has been such a blessing to us. Joshua is being home schooled for the second year. NACD has helped to empower and enable me, as the parent, to have some influence in my son’s progress - to know I am doing the best that I can do with an awesome program designed just for Joshua and his needs. The program requires sacrifice and commitment, but we are seeing progress and achieving new milestones.

As the parent of a child with *idic(15)*, there are so many different challenges to confront. For us it has



Joshua

been scoliosis, hearing impairment, digestion problems, congenital heart disease, physical and mental delays and processing issues. But early on, the heart anomaly demanded the most focus. When Joshua was one, we flew to Ann Arbor for an emergency open heart surgery to replace his aortic valve. Upon arriving and getting our first echo, we were told he did not need surgery. Instead we got a catheterization, which looked even better. All of his pressure gradients had dropped miraculously. And that is how the doctors described it. We were later told by our cardiologist that Joshua has a very abnormal heart, but obviously there is a guardian angel that is keeping his heart pumping better than they can.

And that is how I have always thought of my Joshua. He is very social, very expressive, and has a heart that just won’t quit. My days have been filled with hugs and kisses and unconditional love and acceptance. As he matures and strives to communicate, we are facing new behavioral issues and challenges with his increase in inde-

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JOSHUA *continued from previous page*

pendence. But his love is always so pure and unconditional - a constant reminder of the agape love we all would love to maintain in our lives.

Yes, most parents out there could never understand what we, as parents and caretakers of a special needs child, go through on a daily and hourly basis. It is a uniquely different life. And all I can, I stand on Romans 8:28. We live in an imperfect world. But all things work together for good for those who love God and are called according to his purpose. I look at all of the lives that have been touched by people meeting Joshua and hearing his story. He loves people in a way they do not forget. Of course, as he gets older, we are trying to teach him the appropriate handshake for strangers instead of hugs and kisses, but he's still a lover.

I've noticed a difference between siblings of those who have relationships with special needs children and those who do not. They seem to have a compassion for others that is mature beyond their age. Their mean-spiritedness is not as prevalent and they grow up having a little more understanding of others' challenges. I encourage all parents to help their children to know and spend time with a child with special needs. They need to hear these children's stories, not just pass them in the hallways. I know my other sons will be better boys and men having had Joshua for their brother.

With all of the challenges that we have faced, we know it has made us better people, better parents, and even better spouses for it. That still doesn't make it easy, but Joshua makes it all worth it.

Dark brown eyes that stare at you
And penetrate your mind
A smile that says "I have a secret"
That's never vocalized

He learns to say a few words to meet
his daily needs
But the language in his hugs and kisses
needs no therapies

I savor each day of our journey
Tho' victories are a slow pace
He takes my face between his hands
For a moment-I forget the race

A finite life of struggles
Yet a hope that never ends
I try so hard to teach him sounds
I fail to hear the lesson that he sends

My sweet and fragile little boy
Tell me, my God, what have I done?
To be so blessed-I cannot express
How your love speaks in my silent son

-Sophia Dentiste

*Wife to Robert, Mom to Joshua,
Jacob and Jonah*

Chicago Family Meeting

April 19 - 21, 2002
Schaumburg, Illinois

The family meeting in Chicago was very successful. Fifteen families, primarily from the Midwest, but also from the East coast gathered to visit with each other and speak with professionals, including researchers and doctors.

Families that arrived on Friday evening met for dinner. It was fun to see all of the parents and their children. Everyone had a great time and it was wonderful to have so many of the idic(15) families together in one place.

Saturday morning folks gathered for breakfast and met in a conference room at the Embassy Suites Hotel. The children played well together and the older siblings were great helpers so the parents were able to chat. At mid-day we broke for a pizza lunch and birthday party for Cody LaCombe (9), Marlana Higgins (3), and Jake Miller (3). The lunch party was at a nearby restaurant fully equipped with goodies for the children...balloons, cake and treat bags, and horns (thanks to the LaCombes) were the highlight!

After lunch we met back at the conference room to speak with doctors and researchers attending the meeting. Professionals included: Dr. David Ledbetter, Chair, Department of Human Genetics; Patti Mills, Genetics Counselor; Dr. Darrel Waggoner, Geneticist; and Laurie Weiss, graduate student; all from University of Chicago; and Dr. Bonnie Klein-Tasman from University of Wisconsin (Milwaukee).

Dr. David Ledbetter, Chair, Department of Human Genetics spoke with families about his hopes of opening a multi-disciplinary clinic for children with idic(15). Dr. Ledbetter (famous for his discovery that Prader Willi, another chromosome 15 disorder, is caused by the lack of paternal genes from a segment of chromosome 15), has dedicated many years of study to chromosome 15 research. He and his staff of genetics counselors and researchers have been studying idic(15) on a laboratory basis and now hope to study it on a clinical basis as well. By providing families with a multi-disciplinary team of doctors to provide routine medical care for our children, they will be

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CHICAGO *continued from previous page*
 able to gather more information about trends seen in idic(15). Additionally, they would like to follow trends through anecdotal infor-

everybody met for breakfast and good-byes. It was a wonderful weeked...one that needs to be repeated again...very soon!

-Jodi Miller



Pictured l. to r. are: Dr. David Ledbetter, Laurie Weiss, Dr. Darrel Waggoner, Patti Mills, Dr. Bonnie Klein.

mation such as that provided by parents in the IDEAS database.

Many local businesses and individuals donated products and goods that were raffled off to families attending. The children delighted in helping call raffle numbers and each family won at least one prize and many won more. Prizes included Bulls tickets, Packers tickets, Blockbuster certificates, a Radio Flyer Wagon, Target gift certificates, and more! All of the families were provided with a huge bag of donated products full of pop, chips, cookies, candy, Motrin, Tylenol, diapers, wipes and more. We have Ruth Kross, Mom to Marlana, to thank for all of the wonderful donations - as she solicited these by herself!

At the conclusion of the day, families met at the hotel bar for drinks and snacks and many continued on to the hotel restaurant for dinner. Some kids and parents went swimming and others sat around the hotel chatting. Sunday morning

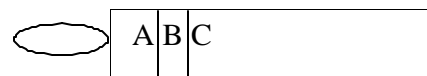
Nemours - UCLA idic(15) Project Update

Our research group officially began the idic(15) project in early 1999, when we received our NIH funding. Since then, we have been contacted by over seventy families who have children (or other relatives) with some form of duplication of chromosome 15q. We have been working on two basic questions:

1. What are the symptoms and developmental profiles of people who have chromosome 15q duplications?

2. Are there specific molecular features of the chromosome duplication that alter the risk for the different symptoms?

To simplify the molecular and cytogenetic studies, we divide the potentially duplicated region into three regions (A-C) as shown in the figure.



What have we learned?

Molecular Cytogenetic Findings

While most reports indicated that idic(15) chromosomes were very similar structurally, we have found that at least half of the large marker chromosomes in our sample have some sort of structural difference. They can be formed from one or both copies of the parental chromosome 15; they can carry one or two (or sometimes more) copies of the A, B or C regions; they can come from either parent; and they may carry only a small part of a region.

Developmental Features

There is remarkable variability in how the kids are doing. From what we can tell with the group we have characterized so far, there is a high risk of autism or autism-like symptoms for people carrying duplication of 15q. While this has been reported to be only associated with duplications that come from the mother, this is not holding perfectly true. We have subjects whose duplications are paternally derived who are autistic and some with maternally derived duplications that are not. We are looking hard at the charac-

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teristics of these duplications to see if there is something different about them that alters the risk. We also see that language is often delayed (as you know) but importantly language and speech generally improve with age, thus we strongly encourage

seem to be a dosage effect for the B region, with better performance associated with fewer copies. (We have kids enrolled in the study with one - four extra copies of this region. Usually, there are two extra copies of the region in most of the idic(15) chromosomes). The A and C regions appear to have limited

Other news

The project is moving but continuing from both coasts

Part of our research group moved to the Nemours Research Institute in Wilmington, Delaware in May 2002. Naghmeh Dorrani, MS will remain at UCLA and continue as

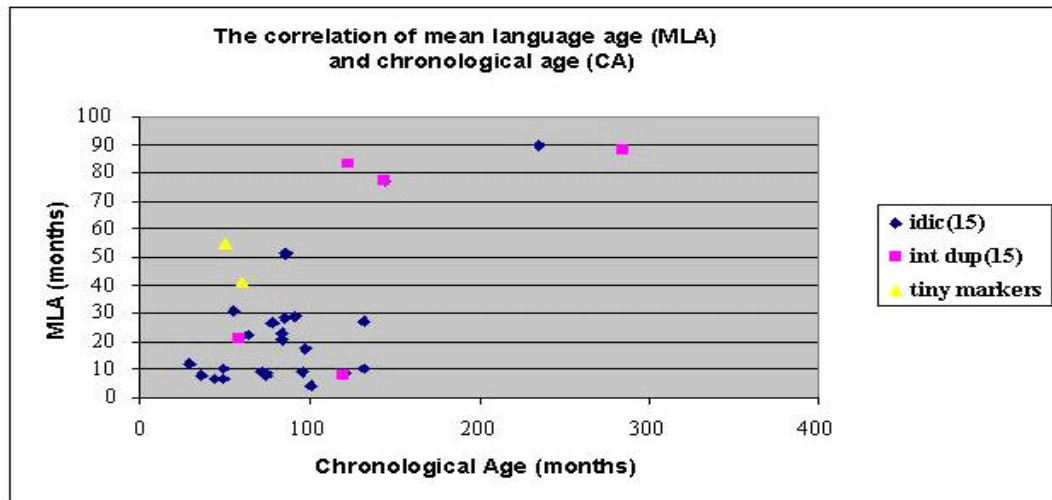


Figure 2.

ongoing speech/language interventions even into adulthood. See Figure 2.

The average mental age of the first twenty kids we characterized (who carried idic(15) and interstitial duplications) was approximately twenty-five months (at an average chronological age of 111 months) but again there is a wide variability in performance (range four - 128 months) and improvement over time.

Can we correlate the marker type with the symptoms?

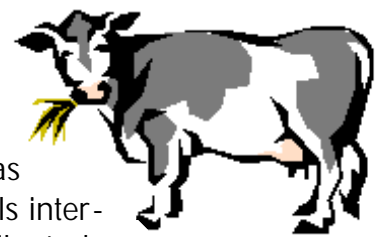
Not yet. We see some trends but don't have enough subjects completely characterized to say for sure, especially since the duplications are more complex than we thought going into the study. There does

effects on symptoms. We hope to sort out whether the complexities that we see in the chromosome studies are underlying the differences in how the kids do. To do this, we need to assess more kids with similar types of duplications of each kind.

project coordinator. The "phenotyping team" will still be based at UCLA but the molecular and cytogenetics part of the group is East-Coast bound. While we expect a little downtime while we set up the new lab, we hope to get back up to speed quickly.

-Carolyn Schanen

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 Jodi Miller, c/o IDEAS, 300 N. Pine St., Mt. Prospect, IL 60056. Or email your newsletter items to j3smiller@aol.com