

WINTER • 2008

FROM OUR EDITOR  
EKOW ONK EDITOR

## Taking Flight

by Doreen Gladis

*"Parents give their children roots and wings;  
Roots to know where home is, wings to fly away  
and exercise what's been taught them."*

Jonas Salk

When Jane asked me to be the guest editor for the winter edition of The Mirror she said the first thing I needed to do was to select a theme. I chose to focus on Residential Services because it was something that IDEAS had never done before and it had recently touched our lives.

In this issue, four families share their experiences regarding residential services. They write about how they searched for programs, the emotions they felt preparing for the move, the adjustment of their child/adult, the effect it had on siblings and family life. They express how they love their child and even though they are not living at home they are still a part of their daily lives. They continue to advocate for their child, remain involved in their program, share their successes and feel bad for them when things don't go as planned.

Most young people eventually move out of their parents' homes, but for persons with special needs the move is more complicated. Some may not be able to live independently and may need continued support as adults. This can be true even for children without special needs, but the reasons may be different. People with special needs may leave home earlier or later than the average person, depending on the circumstances. They may leave at a younger age when appropriate educational programs are not available in their community or due to medical conditions or even behavioral issues. They may leave at a much older age if support services, community programs or suitable housing is not available. Long waiting lists may also be a factor. But all of our families felt a tremendous sense of pride seeing their child/adult experiencing life as independently as possible. Our articles focus on families whose children received residential services at a younger age or after high school.

*My name is Doreen, my husband, Tom and I live in Georgia and our son Pavel attends a residential school in Florida.*

*Chromosome 15q duplication syndrome (dup15q) is a clinically identifiable syndrome which results from duplications of chromosome 15q11-13. These duplications most commonly occur in one of two forms. These include an extra isodicentric 15 chromosome, abbreviated idic(15), which results in an individual having 47 or more chromosomes instead of the typical 46. Individuals with an interstitial duplication 15 are born with the typical 46 chromosomes but have a segment of duplicated material within their 15th chromosome.*

This edition also includes an Update on Sudden Unexpected Death in dup 15q. We are told, "The Autism Tissue Program (ATP) researchers are working to understand how and why the brains of individuals with dup15q are experiencing a sudden and unexpected death". Dr. Jerzy Wegiel, Ph.D., updated IDEAS professional advisors in January, 2008.

Finally, welcome to all the new members who have joined the IDEAS family in the past year. We are here to share in your journey; to laugh, to cry, to exchange information, and to help you advocate for your child. "Behind the Scenes at IDEAS – Come Join Us" is an article outlining the supports that IDEAS provides for its members and news of an exciting new program that will be launched later this year.

### INSIDE THIS ISSUE

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  - Birthdays

## Leaving Home for the First Time

by Tom and Doreen Gladis



**D**riving from Georgia to Florida every month gives me time to think about where we were four years ago, today, and the future. Our 17 year old son, Pavel, has severe autism, moderate cognitive impairments and not much speech, but we have always reached for the stars. From a very young age we took Pavel everywhere, exposed him to new situations, and provided endless hours of behavioral, sensory, and speech therapy. However, as puberty set in Pavel began to change from a mostly compliant little boy to a teenager with very challenging behaviors. Not all children regress at puberty, some merely plateau, or have no problems at all. Maladaptive behaviors began occurring more frequently and with greater intensity at both school and home. Soon these issues prevented us from the familiar routines like going to church, eating out, grocery shopping, and visiting family. Pavel's world was becoming smaller. Finding respite workers became impossible and during a melt down it would often take two of us to restrain and calm him. Pavel went from being a fourth grader who could get out of the car and walk independently to his first inclusion class to a child who couldn't be contained in a classroom.

**O**ur school district is small, but affluent, and located in a major metropolitan area. The special education department was undergoing a reorganization that had them rethinking their ability to handle students with difficult behaviors. By the time Pavel reached middle school things were not working out as hoped and he could no longer attend public school in our district. We believed that Pavel would be better served in a larger system with more experienced teachers, familiar with autism, so we enlisted the assistance of advocates and attorney's to get an out of district placement. What

we got instead was an IEP recommendation that Pavel needed a residential treatment facility and a list of three schools to consider, all out of state. My heart sank. I knew by now that Pavel would never live independently, but my dream for him was to complete high school and then find a group home where he could live with peers. This was happening too fast. I wasn't ready for him to leave home, he was only 15, and I still had more to teach him. Pavel had only spent two nights away from home when he was nine to attend a summer camp. I was angry. Why were there no private schools like these in our state? I felt like we had failed him. I worried that he would think we had abandoned him and he would be scared or that he would later hate us. How would I know if he's being cared for so far away from home. How could I ever explain this to him? Everyday I thought about this and often cried.

**I**knew that without a good school program there was no way we could manage Pavel's behaviors. Then a behaviorist said something that made a lot of sense. Pavel is still school age and the school district has a legal obligation to educate him. When he's an adult, parents have no leverage. If his behavior is unacceptable no one will take him into a group home as an adult. Why not see if his behavior will improve in another setting? Maybe a residential school with structure and consistency in their



programming would improve the quality of his life when he's an adult and we are no longer there to care for him.

**I**n the spring of 2006 we took Pavel and visited two schools in neighboring states. Although we liked a school that was only two hours away, through the school district's recommendation we chose an ABA school 400 miles away with a program that would help Pavel learn new more appropriate behaviors. For several months I prepared Pavel by telling him that "he's a big boy now, and when you become a teenager you go to school and stay overnight. Mom and Dad will visit you and bring you home for vacations". I don't know if he understood any of this, but I kept telling him anyway.

**P**avel began attending his new school in June 2006. We could not visit him for six weeks as he acclimated, so I tried to stay busy. We sent cards with candy and snacks. We tried to call him and talk to him, but Pavel really doesn't really like talking on the phone. The first time we went to visit I think he was surprised to see us. I send him a calendar every month with a note and snacks. The calendar has pictures on the days when we will visit. Pavel's teacher reads the note and puts the calendar up on the bulletin board. Each day Pavel crosses out the days until we come. His teachers say he gets excited as the time gets nearer and says mom and dad a lot. We visit monthly and go to his favorite places like McDonalds and the mall.

**P**avel has done very well at his new school. We have observed him in his classroom working independently and participating with other students in group activities. We get weekly reports on his progress which continues to improve. Monthly we receive graphs that chart

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# Spreading Wings

by Diane Creeley



his progress on behavioral goals. As maladaptive behaviors are decreasing his appropriate behaviors are increasing. We have frequent meetings with staff and the educational committee, as well as the scheduled IEP's with the school district and determine his placement on an annual basis.

At school Pavel lives in a dorm, so when he comes home for holidays and vacations he especially enjoys kitchen privileges and eats non-stop like other teenagers. I tell Pavel that we are so proud of him going to a "big boy school" and he smiles. This Christmas we even flew to Pennsylvania to visit grandma and grandpa, something Pavel could not have done in the past several years. On our return trip the baggage was late and a flight was delayed, but Pavel was very patient. Pavel seems to be perfecting the art of doing nothing. At home family commented on how calm he had become and more mature. He's not always on the go like he used to be. Although Pavel is with peers daily he still hasn't made any friends, but maybe he will with time. He has become very attached to his teacher and many of the staff people. He really enjoys our visits and coming home for vacations and when it's time to return to school, he goes back without a problem. I think it is now just part of his routine. Tom said recently that what we thought a year and a half ago as the worst possible outcome may have been a good thing for all of us.

*Johanna will be 26 next month. She still lives at home and while I love having her here, my dream is for her to spread her wings, be more independent, and enjoy the typical life of a 26 year-old!*



For many years, we've been exploring various living arrangements that might make sense for Johanna. We've attended many workshops and informational sessions about housing and transitioning young adults into new living situations. We've made numerous calls to different agencies to learn about any benefits and services available to her. Johanna's Area Agency Case Manager has been very helpful in making recommendations about people for me to call, programs for us to visit, etc., so that we'd have a better sense of what to expect. I've also learned a lot through my involvement with the State and Regional Family Councils over the years. And I talk to anyone and everyone about this, wanting to learn from others' successes, as well as their failures.

Jo has been receiving 40 hours of day support (each week) through our area agency since shortly after she left the public school system and has been on a waitlist for additional residential funds for nearly two years. We were recently notified that she will soon receive these additional funds, so we've kicked our search up a notch.

Aiding us in our search has been a 'profile' that we wrote up years ago that speaks to who Johanna is, how she functions, what she likes/doesn't like, who/what is important to her, what makes her happy/sad, what activities she enjoys, her skills, her challenges, etc. We update it every year and share it with those involved in her daily life but it also serves to remind us of the optimal situations for Jo and keeps us on track as we work our way through each new maze.

My original vision for Johanna was that she would have a home of her own and share it with one or two of her longtime developmentally disabled friends and one or two 'roommates' who would fill my role, in return for their room and board. I quickly realized that the easy part of that vision is finding a home; the hard part has been finding the right people to live in the home with her! People who'll commit to a long-term arrangement and be as responsible and enthusiastic and energetic and supportive as Jo needs (and I expect!), are few and far between, especially in our rural area. I haven't totally abandoned that vision, but I've come to realize that I must be open to modifying or delaying it a bit.

We've determined that it's important for Johanna to stay in her hometown area where she is so comfortable and happy. It will also provide me with additional eyes and ears (friends and neighbors) to make sure she is safe and well-cared for after she leaves home. We have visited several 'group homes' in our area, but for one reason or another, none of them fits the bill for Jo. I've applied for Section 8 housing assistance for Johanna (though that waitlist is about 3 years!). We're also exploring a 'home provider' setting, where a family, or couple, or individual(s) would welcome Johanna to live with them and be compensated through the residential funds she'll soon receive. This is a challenge, too, that will require some luck in making the right

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## How Clare Went To College

by Jane True

I always had the dream that Clare would go to college. During a particularly stressful IEP in middle school, we were advocating for academics and much of the rest of the team was pushing for the Laundry Track. I would like to thank the teacher or therapist who said, "Why do you want academics? Clare's never going to college!" I knew from that moment that it was a done deal. I just needed to find the appropriate place.

Clare had gone on college searches with her brothers, so she understood what it was all about. When we were visiting Oberlin College, she had a meltdown. When all was said and done, she had thought we were going to leave her in Cleveland. So we made a deal.

We would look together and she would never go anywhere unless she first saw it and agreed.

I had heard of a college experience in New Mexico, but the individuals I knew who had attended there had Downs syndrome and didn't have many medical needs. Clare needed a place where she could live in a home with other women who were her peers, move about freely, attend classes, have opportunities for social activities including her music, and have excellent medical support. Through word of mouth we found Stewart Home School in Kentucky.

When I inquired about admission requirements, the director said the only requirements were that we visit and decide that it was the right place for Clare. She promised we would know within 10 minutes. Clare, her dad, and I made the first visit. I think we knew within 5 minutes. A few months later Clare and I went back accompanied by two very close friends who went over the place with a fine-toothed comb. We all agreed that this place was perfect for Clare. So Clare began to get very excited about going to college.

We had planned for starting in the fall, but seizure management issues arose. The school had about 100 students with seizure disorders

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## The Right Fit for Joshua

by Donna Bennett

We started looking for an approved private residential school placement when Joshua was age 11 because the services in the community were not meeting his needs. We went to the ARC and searched catalogues and the internet to see what was available and found the residential schools were at least two hours away from us (either Pittsburgh or Philadelphia).

We visited our first choice school and found we needed to have approval from our school district, our special education unit, and our county Mental Health/Mental Retardation program which finances the program. We also found the school had an opening but they chose a child from California because they didn't feel they could meet Joshua's needs. We then visited Elwyn and found they had a placement available.

We had a meeting with the education system and MH/MR and told them about Joshua's needs and our concerns. We had our attorney and our advocate from ARC present at this meeting. We were told they would make a decision over the Labor Day weekend. It was the longest week we ever spent and they stated all the children in their program could benefit from this type placement. My husband said "but they are not requesting this placement that Joshua needs to progress to the best of his ability". They called and approved the private residential school placement.

There has always been a waiting list in York County for services and the ones on the top priority list are the chosen ones to get attention. The funding comes from federal, school district, and the county MH/MR programs.

We talked to Joshua about the placement but did not know how much Joshua understood the move. We were not allowed to visit the first five weeks. We feel the



relationship his sister (Elizabeth) shared with Josh changed the day we left Elwyn. Every time we visited Joshua she told him what happened at home after he left until the next visit.

It was really hard to pull away from Elwyn and leave him there. We don't think anyone can do what we can do for our special child so we naturally are afraid for them. When they can't tell you when they hurt it really hurts you as much as it hurts them. We made friends with a family who also brought their child to stay at Elwyn the same date. This child was verbal and when we

visited Josh he would tell us about Josh's hair cuts and dental appointments which really helped all of us who could not be there to hold Josh's hand or wipe his tears away when he was scared.

The move helped Joshua's sister to appreciate her brother. She is very proud of his accomplishments, how he never forgets people, and how he watches out for the staff members he loves. The move was a very hard adjustment for all of us - I got a part time job because I could not stand to sit home and think about him and how he was doing each day.

We feel the best way to monitor the services in a residential setting is to visit unannounced. We visited both announced and unannounced.

We feel the reason he is in a group home in our community is because we kept the MH/MR community informed and invited them to all the parent/teacher conferences while in the residential setting. We told them there would need to be a slot available in the community upon graduation (age 21) due to his aggressive behavior.

Joshua has been in three group homes since returning to our community.

The first group home was a split-level house with four other clients. A change in managers of the home caused an incident. He ended up in the Psych ward and he had a seizure and fell down the steps and was taken to ER for stitches on a holiday. We had a meeting and requested removal to a safer house.

The second group home was a ranch with three clients and it seemed appropriate for a short time. One of the clients would scream a lot and want most of the attention. Every time the MH/MR caseworker visited the house and this client would scream, Joshua would pinch him trying to communicate - "you have got to get me out of here I cannot deal with this noise any longer". An incident resulted and we met with the group

home provider and they decided to move Joshua to another home.

The third group home was a ranch with three clients - two females. His behavior completely turned around. Not too long ago one client got very aggressive toward him and this time the group home decided to move the other client to another group home. This client was replaced by a male and the match seems to be working well.

The group home adopted a dog in his house and all of the clients have adjusted well to the dog. The dog goes to each of their bedrooms at night to make sure they are okay and then settles down in his cage for the night. ■

## CHEERS TO OUR VOLUNTEERS!

### TERRI POWERS

*Terri is the cousin of Nicholas DeLorenzo (idic 15)*



Nick, Terri, and Kelly

My name is Terri Powers and I raised over \$200 to donate to Idic15. I raised money by making friendship bracelets and the idic15 live strong bracelets. I sold these bracelets at a town harvest festival. The reason I did this is because my little cousin, Nicholas DeLorenzo has idic15 and he is the most cutest two year old ever.

At my school, Saint Francis de Sales in Barrington, NJ, I am always taught to do things for others. I know idic15 is a condition that affects only a few children in the world. That means to me that these people are special, just like my cousin Nicholas is to me. Even if these kids are different from you and me I thought everyone in the world was different.

## WINTER CLEARANCE

Water bottles, travel mugs and calendars reduced to \$5 each, while supplies last!



IDEAS news flash

# BEHIND THE SCENES AT IDEAS



COME JOIN US!

*A big welcome to all the new members of the ideas family!*

IDEAS has now 345 members. We provide family support, promote awareness, research and targeted treatments for chromosome 15q duplication syndrome. We have nine committee's soon to be ten, operating behind the scenes to improve the lives of our families. This issue will feature four of them plus our new one "Parent Talk".

## • PARENT MATCH –

This program matches new or old families with mentors from the IDEAS family. The mentors have gone through a training program and are available to talk with you. Many families find it helpful to talk to someone directly who can answer some of your questions first hand and offer you support.

You may benefit from the Parent Match if your child has been recently diagnosed with a duplication of chromosome 15q or are going through a challenging time in your child's life, such as the on onset of seizures, difficult new behaviors or problems with education or therapy. You may wish to talk to another parent of a child with your child's exact same diagnosis. You simply need to email the Parent Match coordinator Donna Bennett at [dbenn17354@juno.com](mailto:dbenn17354@juno.com) or call 1-888-64-IDEAS

## • REGIONAL GATHERINGS –

One of the ways IDEAS meets its mission of providing support to families is by promoting regional gatherings. It's an opportunity to make new friends, renew old ties, and spend some relaxed time with other families who understand the challenges and joys of raising a child with a chromosome 15q duplication.

Consider a Potluck at the home of an IDEAS family, a picnic and a day

playing at a state park, a weekend at a camp ground, a more formal meeting with professionals, or a night spent at a centrally located hotel. To HOST a regional gathering contact Paula Davis at [xpdavis@comcast.net](mailto:xpdavis@comcast.net) or call 978-392-8474. She can give you a list of other families in your area with idic15 children and ideas about putting one together.

## • FUNDRAISING–

"Thank you IDEAS families and friends, we met our 2007 goal of raising \$106,479.00! 2008 proves to be a very productive year as we begin to plan the 2009 conference and start to create our registry. In our efforts to support these projects, we need your help.

1. Would you like to serve on the fundraising committee?
2. Do you have an idea for a fundraiser that you would like to share?
3. Would you be willing to hold a fundraiser for IDEAS in your community?
4. Would you be interested in joining forces with other IDEAS families in your community to organize a regional fundraiser?

***"I can't thank you enough for all that have contributed not just money, but their time. In order for us to help our children, we need all of the help we can get". Please email me at [kadi@twcny.rr.com](mailto:kadi@twcny.rr.com) or call 1-888-64-IDEAS.***

***And don't forget May 15th*** is our second annual Phantom Tea! This was a very successful fundraiser for us last year with 15 participants we raised \$4,500. Start getting your list together now!

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• **IDEAS WEBSITE** – [www.idic15.org](http://www.idic15.org)

This expansive, well organized site contains topics such as a description of idic15, genetics, and a brochure you can print. It includes family issues and resources, information for doctors, teachers and therapists. The IDEAS section includes our mission statement, archived newsletters, conferences, European resources and ways to support IDEAS including the IDEAS store. The research section is full of published articles and

research currently being conducted. Finally there is a message board, where you can ask questions and get answers from those who have been there.

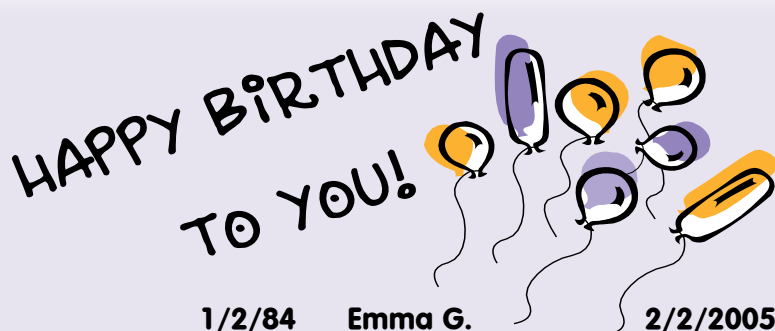
**You can see there's a little something for everyone.** I was surprised to find so much new information had been added since my last visit, so check frequently you never know what you'll find!

Contact Len Poore at [jmpoore@comcast.net](mailto:jmpoore@comcast.net) or call 1-888-64-IDEAS for questions

about registering or using the site.

IDEAS also operates a message board at:  
[inverted-dup15@yahoogroups.com](mailto:inverted-dup15@yahoogroups.com).

*If you are interested in participating in a program, volunteering on a committee, or have further questions about a program, please feel free to contact our coordinators.*



Joseph B.	1/2/84	Emma G.	2/2/2005	Sara H.	2/26/2006
Khalid E.	1/3/95	Jacob P.T. F.	2/3/98	Kathryn G.	2/28/93
Naomi G.	1/5/71	Riley McCabe	2/4/07	Raphael B.	2/28/2006
Shane R.	1/7/2000	Ryan T.	2/7/2002	Payton F.	2/29/2000
Frederique D. 	1/8/92	Nicholas S.	2/8/2004	Allison G.	3/1/97
Ethan L.	1/8/2001	Nathan H.	2/8/85	Dylan M.	3/1/94
Christopher O.	1/9/92	Graham R.	2/9/2000	Fabio C.	3/1/93
Sarah G.	1/9/90	Mathias W.	2/9/2000	Kameron R.	3/2/2000
Mason B.	1/12/96	Emma D.	2/9/99	Jacob C.	3/3/2003
Angela B.	1/13/94	Abby E.	2/9/90	Lis R.	3/6/98
Pavel G.	1/17/91	Joshua B.	2/9/76	Samuel R.	3/8/83
Genevieve M.	1/17/91	Robin R.	2/13/85	Shawn K.	3/9/93
Nathan L.	1/18/2000	Nicholas D.	2/14/2005	Laura W.	3/12/2004
Sarah B.	1/24/94	Thomas Q.	2/15/2000	Benjamin R.	3/13/98
Nicholas L.	1/25/89	Noah H.	2/16/99	Kathryn B.	3/14/2000
Alexa V.	1/25/87	Johnathan P.	2/16/99	Christopher S.	3/14/96
Kian S.	1/26/94	Tristan W.	2/17/98	Patience V.	3/15/99
Matthew B.	1/26/89	Johanna C.	2/17/82	Jacob L.	3/16/2000
Kate B.	1/27/98	Ben I.	2/20/85	Michael H.	3/17/80
Ryan M.	1/27/86	Connor L.	2/22/94	Jacob G.	3/19/99
Avery J.	1/31/2004	Aidan L.	2/23/2004	Christina M.	3/20/85
Elleni P.	2/1/2002	Stuart H.	2/25/99	Joshua R.	3/21/96
				Joseph G.	3/21/88
				Jason K.	3/23/90
				Jon B.	3/26/2007
				Katherine C.	3/29/97
				Cody K.	3/31/99

## "Gonna Be Ok"

by Jesse Leo



Jesse, Jake age (1 1/2), Maria, Aidan age 4 (dup15q)

It was simultaneously the worst day of our adult lives and the best day my wife and I had in quite some time. The Doc and I, we locked gazes for about five seconds right after he delivered the news – the news that after a second analysis of Aidan’s blood test results they’d “found something.” If a gaze could speak, Doc’s would’ve said something like: “I’m here for you, but this is uncharted water for me, too. Best of luck.” He handed us some printouts from the IDEAS website and told us that in his 30 or so years of pediatrics he had never come across a single case of idic(15) before.

But driving home there was an amazing sense of relief. The wondering was finally over. The speech, the low muscle tone, the sleep issues, the cognitive delays... finally an explanation, a name. And we knew we were taking the first steps of a life-long marathon, but for that one afternoon there was serenity.

But an afternoon is a short time. And as everyone who reads this knows all too well – there are good days and there are bad days. The bad ones are all about the stimming. The tantrums. The floppiness. The absolute refusal to sleep. The fixations. The aggression.

But one good day erases a year’s worth of bad ones – when Aidan is on his game there’s no sweeter kid on the planet. He’ll look us square in the eyes and ask (in so many words) if we’ll play action figures with him. Or read to him. Or play video games with him. Or get him juice. Or make hand shadows on the wall. He’ll even allow Jake (2-year old brother) to share a toy or two with nary a bite attempt.

With work occupying an unhealthy amount of my time, my wife has done a Yeoman’s job in overseeing the bulk of his daily development. Pitch in an amazingly supportive family and a hard-working (if understaffed) elementary schoolcrew, and our little guy’s progression has been remarkable.

Aidan recently picked up an expression that he must’ve heard us use to soothe his cranky brother – “Gonna be ok.” At times he’ll use it when appropriate, but more often just for the heck of it. A cynic (and probably a therapist) would say he’s simply regurgitating an oft-used phrase around the house. But we prefer to think he’s sending a message, a message that we sometimes desperately need to hear.

Gluten and casein free diets, uncooperative insurance companies, mountains of research materials, pricey therapeutic equipment, Yahoo newsgroups, holistic medicines, potty training, cardiac tests, Assistance Dogs, Aqua Therapy, Equine Therapy, IPPs, OT, PT... We’re climbing Mt. Everest blindfolded. But it’s like our boy says – gonna be ok.



## Understanding SUD in dup15q January, 2008 Update

*In Memory of  
Cameron Poissant  
2000-2007*



Oh my little sunshine heart  
How it hurts to say good-bye  
Oh my little sunshine heart  
I hear you say "no cry"  
But my little sunshine heart  
I cannot stop the tears  
Oh my little sunshine heart  
I will always stop to hear  
The sound of your laughter  
Your giggles filled with joy  
Oh my little sunshine heart  
I know I must find a way  
To feel the happiness that you  
brought before I feel the pain  
I miss you little sunshine heart  
and I love you so  
Oh my little sunshine heart  
I cannot bear to let you go.

By: Kelly Poissant

IDEAS has tracked the sudden and unexpected deaths of six seemingly healthy young people with chromosome 15q duplication syndrome over a 21 month period. In this same period, another three medically fragile children with duplications of chromosome 15q11-13 died, and a child died from a severe choking incident. Several of these families donated their child's brain to the Autism Tissue Program(ATP) at the time of death with the hope that their donation might accelerate research and help other families affected by chromosome 15q duplications.

The ATP researchers are working to understand how and why the brains of individuals with dup15q are different, and why some young people with dup15q are experiencing a sudden and unexpected death. They have developed a new protocol specifically for the study of dup15q cases. Dr. Jerzy Wegiel, Ph.D. has performed neuropathological studies of the first two brain donations. He and ATP staff presented their observations to IDEAS Professional Advisors in January, 2008. The purpose of this update is to provide a brief synopsis of Dr. Wegiel's presentation.

We have made great strides in the past 5-10 years in our understanding of the symptoms arising from chromosome 15q duplications (dup15q). But up to now we have not had the ability to look at actual changes in the brain that contribute to the neurological and behavioral traits that are commonly found in individuals with dup15q. Most studies done to image the brain, such as MRI or CT scans, have had relatively normal findings. Studies of brain activity, like EEG, often reveal changes consistent with seizures or a risk for seizures. However, EEGs cannot tell us the basis for this abnormal brain activity.

The detailed studies of brain anatomy from the first two cases submitted to the ATP revealed several new findings that have important implications in our understanding of how chromosome 15q duplications may affect brain development and function. Importantly, while changes in brain architecture were found in both cases, variability between the two cases suggests that the duplications may differentially affect brain development. We don't know if the findings in these cases will be replicated in other cases. Until we know that, we don't know if what the neuropathologist found was unique to these two individuals or more indicative of a syndrome issue. Thus, just as we have learned from characterization of the clinical symptoms, it will be crucial to examine additional cases to understand the spectrum of changes that can occur. Then we can begin to move from speculation to observation. Better understanding the neuroanatomical underpinnings of the disorder will provide insight into potential treatment strategies down the road.

Sadly, the study of these two brains has not provided us a specific cause of death for either case. IDEAS professional advisors are still actively investigating possible causes for sudden unexpected death in dup15q. The physician advisory released in July, 2007 has been updated and is available on the IDEAS website at <http://www.idic15.org>.

Dr. Wegiel is examining another two brains, and the ATP will make another report to IDEAS Professional Advisors in March, 2008. With four completed neuropathological exams, we are hoping for a statement of preliminary findings later this year. Findings from individual cases will not be shared so as to protect the confidentiality of the children who have passed away.

## Reflections from a Sibling

This part of the REFLECTIONS

shares perspectives from a sibling

### A Journey into Chase's World

By: Jenna Gosa



How can someone say that a child with autism, Chromosome 15 Duplication Syndrome, in Chase's case, is any different from a child who does not have autism? Despite the fact that the name is a dead give away, I still see a normal boy who is the center of everyone's attention, who lives in his own world, and who loves beyond extraordinary limits. Aside from the more hectic days of climbing, searching, and chasing, he is just the same as other children his age. He has his favorite toys such as his trucks and his read along red book that is attached to him at the hip just like other boys. The way he uses his interests make him unique and distinctive from other children; it makes him who he is. Spinning the wheels and holding the book's speaker up close to his ear are just a part of his daily routine but may not be for other children. Other people may see these actions as out of the ordinary and unusual behavior; I see them as one of the normal activities of the day.

Even though he cannot say my name, I know he knows that I am his sister. There are days when my mom works that I have to pick him up from daycare. As soon as I walk outside to the playground and he sees me, I can just see his face light up as he hurries over to me. He knows that if I am there then that means that he is going home. We walk over to the car and he heads straight to the door and uses all of his strength to try and open the door. He immediately scoots over to the middle seat so he can see out the windshield and raises his hands signaling me to buckle the seatbelt. I always have to remember to put on window lock because the feeling of wind blowing on his face beats anything. When we get home, I get him out of the car and he immediately runs to the tires thinking that just maybe he can get them to spin. I pull him away and he grabs my hand and leads me inside. Where do you think he goes? He goes straight to the trucks and the red book of course. Oh, how could I forget? We have to make a trip to the refrigerator first as he climbs up on the bottom shelf and reaches for his juice. After his thirst is quenched, he then runs off to begin his infamous playing and the chase begins.



IDEAS news flash

If anyone wishes to receive *The Mirror* online through our web site, please submit your request to: [info@idic15.org](mailto:info@idic15.org)

*Understanding SUD ...continued from page 9*

In the words of Dr. Wegiel, "The opportunity to study these brains is the opening of a new era in brain research. It will help to understand the function of chromosome 15 and the role of its genes in all individuals—both those with healthy brains and those with dup15q. Even healthy people should be very grateful to those donating tissue." Autopsies and brain tissue are absolutely critical for this ongoing research. IDEAS families are strongly encouraged to consider requesting autopsy and/or making a brain tissue donation at the time of their child's death, whether it is sudden and unexpected or at the end of a more normal life span. You can learn more about tissue donation at <http://www.brainbank.org/>.

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match, but in the short term, we're hoping it'll be more doable than my original plan. Time will tell.

In the meantime, we continue to hone her daily living skills so she'll be at her best when the time comes. We continue to arrange some overnight (or longer) trips so she is more and more comfortable being away from home – for short spells anyway. I talk to her about this upcoming change and how many new opportunities will open up for her. I know she doesn't fully comprehend it all yet, but eventually.

I've been frequently reminded that despite my best efforts to nail it, the perfect solution may not be the first one ... or the second one ... or even the third one. But that doesn't mean it won't ever happen. I know it will, some day!

I am so ambivalent about the prospect of Johanna leaving the nest – but just as it's been for her brothers and sister, I know it's the right thing for Jo – and so I'm excited about the possibilities for her!

And finally, my best advice is this: our children are never too young to begin to think about and plan towards their adult living situation.

How Clare went college...continued from page 4

so they felt ready to take Clare on, but we all agreed we wanted to wait until we had them under better control. So she waited until January to start.

It was fun shopping and collecting all the items Clare would need for her room and closet. Of course one entire suitcase was filled with her beanie babies. When the day arrived we were all somewhat apprehensive, but a huge ice storm chased us all the way to Kentucky. So for two days we were so worried about road conditions we forgot about college anxiety. Once we arrived everyone was so warm and friendly that Clare never looked back.

Clare spent 3 years at her college, and I am so thankful she had the experience. For the first time in her life, she was free to move around a beautiful campus without waiting for me. She was among 400 potential friends, and it was a thrill for me to meet her latest "best friends" every time we visited. Until she went to college she had never been able to carry on much of a phone conversation. But after a few months, when I called she would tell me jokes and describe places she had been.

There were some bumps in the road. Staff changes were the biggest challenge. During one housemother turnover her medicine was missed for several days and she ended up in the ICU. Another time a new dietitian was not informed of her dietary restrictions—that was just a 911 and an ER trip. I think for the entire three years, whenever the phone rang my stomach leapt into my throat until I was sure it was not her school calling. But it was so worth all the risk to visit Clare on campus and see the pride on her face as she introduced us to her new friends and teachers. Looking back, one of the best things about Clare going to college was that her parents and brothers learned that she could thrive away from us, and that we could survive without being joined to her at the hip. It's all about finding the right match. ■



## MONTHLY CONFERENCE CALLS FOR PARENTS

The IDEAS Board will begin offering monthly conference calls that would provide an opportunity for parents to talk and share experiences with other parents, regardless of where they live. It would be similar to the Yahoo list serve and message board except it would be on a specific topic and be more personal. This would also provide parents with an opportunity to meet other parents who share similar issues and concerns.

The monthly conference calls would be limited to a maximum of 10 people per call on a first come, first serve basis. All participants must RSVP at least 5 days in advance of each date as indicated below. Long distance phone charges will apply.

Please see our schedule of Parent Talk conference calls for March, April and May below. Based on your response, the IDEAS Board will decide whether to continue offering the conference calls on a monthly basis.

### PARENT TALK SCHEDULE:

DATE	TIME	TOPIC
Tuesday, March 11	5:30 PM Pacific time, 8:30 PM Eastern time	Potty Training
Friday, April 11	6:00 PM Pacific time, 9:00 PM Eastern time	New Families
Monday, May 19	6:30 PM Pacific time, 9:30 PM Eastern time	Teen/Adult Issues

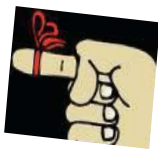
Once registered, you will be contacted regarding the format of the call and call in instructions. To save time at the beginning of each call, we will ask each person to provide us with some background information including your name, location, name of child, etc. which will be shared in advance with those registered to participate.

To participate in a Parent Talk Conference Call, please email or call Joanne Poore at least 5 days in advance of each call at [jpoore181@yahoo.com](mailto:jpoore181@yahoo.com) or 978-851-5182. Joanne will provide you with a short form to provide background information that will be shared with each participant.

If you have any questions, please contact Joanne Poore ([jpoore181@yahoo.com](mailto:jpoore181@yahoo.com)) or Kadi Luchsinger ([kadi@twcny.rr.com](mailto:kadi@twcny.rr.com))

*IDEAS is a non profit organization that provides family support and promotes awareness, research and targeted treatments for chromosome 15q duplication syndrome. IDEAS offers help and hope for chromosome 15q duplications.*

Upcoming Events:



- March 11, 2008 – Parent Talk conference call
- April 11, 2008 – Parent Talk conference call
- May 15, 2008 – Second Annual Phantom Tea
- May 19, 2008 – Parent Talk conference call
- June 25-27, 2009 – IDEAS Conference

## IDEAS

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