Transitions
FROM THE EDITOR
BY JEAN HOUSE

Recently, my family embarked on the Kindergarten Project. I asked friends who have children with special needs how to best handle this big step. Jonah’s preschool teachers, therapists, and hab-tech workers also offered valuable advice. We already had useful tools on hand for a smooth transition, including but not limited to toy telephones, Raffi CDs, and The Jonah Book, the 8-page (and growing) guide to Jonah that I give to every new friend who enters his life.

Here are some of the helpful hints I received:

- Even if there won’t be an answer and he isn’t familiar with the word, talk about Kindergarten throughout the summer.
- Ask the occupational therapist to work especially on school skills, such as scissoring and marking paper with crayons.
- Visit the school playground in the summer so he becomes comfortable there.
- Public school is a whole new ballgame. Read the school’s website, and jot down any questions about policies and procedures.
- Arrange a one-on-one meeting with the teacher the week before school starts to introduce yourself, point out the most important parts of the IEP, and pass along instructions.
- Sit down with the school nurse to explain medical issues and drop off medicine (in our case, multiple severe allergies were the main topic of conversation).
- Photograph the classroom, teacher, and school entrance so you can look at the pictures together, and talk to your child about the new place for a few days before going.

Attend the open house; it’s an opportunity to meet the other students and their parents, as well as spend time with the lead teacher and assistants. It also gives your child an opportunity to get familiar with the new space with parents close at hand.

Pack familiar favorite items for your child as well as supplies and toys for the classroom.

Several of the helpful tips on the list could translate to other life transitions for people with dup15q syndrome: Get workers and therapists on board. Ask those who may no longer be in your child’s life what the people in the next stage of his life will need to know. Visit unfamiliar places with your child before sending him on his own. Always pack a bag of tricks filled with favorite, calming items. Inform yourself and prepare your child.

In this issue of The Mirror, you’ll find other families’ stories about some of life’s big transitions. Also be sure to check out the latest news about strides in scientific research, the growing Dup15q International Registry, recent family gatherings, and more. As Dup15q Alliance continues to grow and thrive, you can depend on the organization to support your family through life’s transitions.

Jean lives in Wilmington, N.C., with her husband, Chris, and son Jonah, 3 (dup15q). E-mails are welcome at jean@editorhouse.com.

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.

http://www.dup15q.org
Dup15q Alliance held its 2012 Annual Scientific Meeting August 9–10. Researchers and clinicians gathered from around the country for the two-day meeting in an auditorium at Children’s Hospital Boston (CHB). While the alliance has organized scientific conferences before, they were closed, one-day meetings of perhaps a dozen professionals—normally, our professional advisory board members plus a handful of invited researchers. So the Boston meeting was the first of its size and scope, attracting more than 70 participants—including members of the press—to the CHB auditorium to hear 20 scheduled speaker presentations and four moderated panel discussions.

Participants gave presentations on a range of topics, including the effects of gene dosage at a cellular level, current and prospective animal models of the syndrome, clinical characterization and treatment of seizures and autism phenotypes, novel brain activity imaging techniques, insights from brain samples, and the unusually high levels of pain tolerance among those afflicted with dup15q syndrome.

The speakers included several researchers with interests in Angelman syndrome and Prader-Willi syndrome, related “deletion” syndromes which are due to anomalies in gene expression for the same stretch of chromosome 15 genes that are involved in dup15q syndrome (with Angelman, the corresponding genes on the maternal chromosome are not expressed, whereas for Prader-Willi it is the genes on the paternal chromosome).

Much attention was given to a gene in this region called UBE3A, which is understood to lie at the heart of Angelman and Prader-Willi and is strongly suspected to contribute to autism-like symptoms in dup15q. Stormy Chamberlain discussed a compound her lab had developed which suppresses UBE3A over-expression, while Ian King was part of a team that developed yet another compound that switches on paternal UBE3A in neurons (where it is normally silenced). Note that UBE3A is the gene that is overexpressed in the autism mouse model developed last year by Matt Anderson and also in the next-generation mouse model currently being developed by Larry Reiter and Scott Dindot with the support of a Dup15q Alliance research grant.

But mice weren’t the only animals on show at the meeting. Larry Reiter presented insights from his work with drosophila (fruit flies), which—despite appearances—share many of the same genes as people and so hold valuable insights for parsing out the gene-dosage dynamics underlying dup15q syndrome. Likewise, Robert Nicholls discussed his research into Prader-Willi syndrome using miniature pigs, whose genome in some respects more faithfully models humans’ in the critical region of (human) chromosome 15.

It appears that these animals hold much promise for modeling dup15q syndrome as well.

In a comment on Reiter’s presentation, David Ledbetter mentioned soon-to-be published data suggesting that dup15q syndrome and interstitial duplications in particular are perhaps more common than previously thought, accounting for 1 in 500 children referred for genetic testing for undefined developmental delay, intellectual disability, or autism (leading to an estimate of 1 in 12,000 births). Ledbetter also thought it likely that the interstitial population outnumbered the isodicentric population, although this hypothesis was met with some skepticism from others—Brenda Finucane proposed a pinky bet.

In their presentation about the Dup15q Registry, Kadi Luchsinger and Lori Eisenhauer announced a new researcher portal and reviewed preliminary analysis of registry data. Many in the audience were intrigued by some of the takeaways—especially the unusually high levels of pain tolerance among those afflicted with dup15q syndrome (which prompted some researchers to speculate whether similar tolerance would be manifested by dup15q mice).

Emily Singer from Simons Foundation Autism Research Initiative (SFARI) News attended the meeting, leading to some excellent coverage of the proceedings (http://sfari.org/news-and-opinion/conference-news/2012/dup15q-scientific-meeting-2012). Moreover, as of the time of writing, Dup15q Alliance was poised to issue its own press release about the meeting. And for those who want to consume the unfiltered content, the good news is that the proceedings were videotaped, and most of that footage, along with slides from presentations, will soon be available for download and/or streaming from the alliance website, www.dup15q.org.

Dup15q Alliance would like to thank Autism Speaks and Lineagan for financial support of the 2012 meeting, and Children’s Hospital for use of their auditorium and facilities free of charge. We also are deeply grateful for the many donations from dup15q families and friends, without whose contributions none of this could have happened.
Registry Update
By Nicole Cleary

Thank you to the families who have joined the registry and entered their children’s information. You are helping to create a powerful resource for families, medical professionals, and researchers. We share updated registry statistics with the Dup15q Alliance community in each newsletter to celebrate how far we’ve come in creating the Dup15q Registry and to encourage families who have not yet done so to join us.

As the accompanying graphic illustrates, so far 276 of Dup15q Alliance’s 726 families have registered. Of those registered, 41 surveys (approximately 15 percent), are incomplete or were never begun. We would love to get complete data on all 41 of these people! Also, 79 of the records (approximately 29 percent) have incomplete contact information. Please make sure to complete your registry data and provide current contact information.

Have you registered? If not, please do today. It takes less than an hour, and the reward is an easy-to-access record of your child’s health history for you as well as for researchers. If you’ve already registered, please check in to make sure your survey and contact information are complete and up-to-date. The more completed records we have gathered, the more we can learn about the population of people with dup15q as a whole!

We are already getting some exciting coverage of our registry! The following link to an article on the Simons Foundation Research Initiative website includes information about the registry and explains the latest analysis of autism related to duplications of chromosome 15q11-13:

Summer Regional Gatherings

BY NICOLE CLEARY

Northwest Regional Gathering at Camp Prime Time


New England Regional Gathering BBQ & Swim

Fifteen families enjoyed a day of sun, fun, and relaxation at the home of Len and Joanne Poore on August 4. Families included the Barnetts, Bopps, Cohls, Davises, Gormleys, Hadayas, Marcouxes, Marstons, Poore, Rasmussens, Rivards, Santarpias, Wises, Rabbs, and Robbinses.

Illinois Dup15q Golf Outing

Five families gathered for a day of golf and fellowship on August 17. The Burkes, Rozovics, Boyds, Krosses, and Quinlans attended.

FINANCIAL REPORT

BY TOM DOYLE
FINANCIAL OFFICER

As of the time of this writing, near the end of August, Dup15q Alliance has $21,190 in checking and just over $257,000 in savings and money market accounts. We had a very successful scientific research meeting in the summer and continue to make plans for the biennial conference in Minneapolis for the summer of 2013. We once again are grateful to all who have contributed to Dup15q Alliance. As we approach the end of the eighth month of the year, we are already at 77 percent of what we budgeted for income for the year. Special thanks to so many of you who have companies that provide matching funds for your donations; that really multiplies the size of your gifts. Thanks also to the many families who have chosen to plan and host a fundraiser—everything from family runs to bike races to golf tournaments to garage sales and many, many more. You are helping us further research, develop our registry, and prepare for the next conference. Thank you!

We are also at about 85 percent of our budgeted expenses, but you will remember that we decided to fund a mouse model for research after the budget was set. That 85 percent of expenses includes the $40,000 that we set aside to kick-start some specific research. So we are in good shape on the spending side of the budget at this time.

In addition, please help us make the registry a valuable tool for Dup15q Alliance by completing the registry process if you haven’t already. This is a major expenditure for our organization and one that will reap rewards for us down the road, so we encourage you to participate. The researchers and doctors who attended the Research Roundtable in August all indicated how important it is for us to focus on getting that data collected so that it can be used more effectively for research and assisting our families.

We continue to be grateful to the many volunteers who make Dup15q Alliance work and to our families who so generously support our efforts. Thank you for all of your efforts on behalf of Dup15q Alliance.

Please feel free to contact me directly at tom.doyle@dup15q.org should you have specific questions about how Dup15q Alliance is allocating its funds or if you have suggestions on what other information you would like to see reported in this quarterly financial report.

Dup15q Alliance is looking for volunteers for our committees. Do you have a special talent to share? Are you interested in holding a fundraiser? To learn more about opportunities to volunteer, contact info@dup15q.org.
ANNUAL BOARD MEETING REPORT
BY KADI LUCHSINGER
EXECUTIVE DIRECTOR

On August 11, 2012, the Dup15q Alliance Board held our annual meeting. We were extremely pleased to receive almost 200 responses to our family satisfaction survey in advance of our meeting, and we reviewed what our members had to say. Seventy percent of families said that the Dup15q Alliance meets their family’s needs. We were happy to learn that 92 percent of families find The Mirror helpful and informative, and 87 percent of families report that research is communicated in terms they understand. We are looking at opportunities to help make the alliance an even better resource for families.

The board is considering how to best meet the online communication needs of our families. We will be redesigning the website to make some resources (such as the emergency tab) easier to find. We are also discussing how to best communicate with families, the use of e-mail, and the role of BigTent, Facebook, and other social media.

We also have some exciting new projects to announce to our members. We have formed an International Committee for the Alliance and are in the process of developing international chapters. Our initial countries include Australia, the U.K., Italy, and Spain. We look forward to working with additional countries in the future as families express interest.

We will be expanding our very successful Dup15q Clinic, where Dr. Ron Thibert has seen 40 children, to multiple sites throughout the United States and one international site. We are in the process of identifying clinics and possible clinical directors to expand this work. We are also looking at possible grants to support clinic coordinators. Clinical directors will share information about the trends they are seeing in their clinics, supporting even richer research discovery. Families will have greater access to doctors who know about dup15q and can provide the most relevant care.

We are continuing to explore ways that Dup15q Alliance can directly fund and support research. We have grown our scientific meetings into two-day meetings that include media coverage (please see the Registry Update and Scientific Meeting Update for more information). Our registry is an invaluable resource for researchers; as we approach its one year mark, participating families will begin receiving requests to update their child’s information.

Fundraising remains a high priority for the board. In order for the alliance to continue maintaining the registry, expand the Dup15q Clinic, and host research meetings and annual conferences, we must continue to raise funds at an aggressive pace. Thanks to all the families who help with fundraisers and other activities that keep us on track to meet our financial goals.

The Dup15q Alliance Board is working on a strategic plan for 2013–2016, which we will complete by December 2012. The voices of our families and the needs of the organization guide this work. With 726 families in the alliance, it is a very exciting time for this organization. We look forward to building an even more responsive organization to support your family in the years to come.
As you may have read in previous issues of The Mirror, Asociación IDIC 15 was created to better serve and support families who live in Spanish-speaking countries. While the group has incorporated in Spain, its reach extends to any family whose first language is Spanish, regardless of their country of residence.

After the successful incorporation of Asociación IDIC 15 as a not-for-profit organization in February 2012, the group has begun to gather momentum. Currently, 30 families are signed up! The very first activity the group undertook was the first edition of Asociación IDIC 15 T-Shirts in April, with 2,000 shirts printed to date and now practically sold out!

And following in the tradition of Dup15q Alliance regional gatherings, the first Organizing Committee was formed in May to plan the first-ever weekend get-together. At the event, families would have a chance to meet and get to know each other, and parents and children alike would have an opportunity for fun and learning about therapies and communication tools for people with dup15q.

The meeting was named “Juntos Aprendemos” (“Together We Learn”) and was attended by 8 families for a total of 30 people.

The meeting was held August 17–19 in Cuenca, a small town about 120 miles east of Madrid in a rural setting with cabins nestled among pines in a densely shaded area with playgrounds and (thank goodness!) a beautiful swimming pool. Despite the extremely high temperature (102 degrees Fahrenheit), the swimming pool in the complex provided the refreshing experience everyone needed. There was plenty of time for both parents and children to play, and while the parents were attending the instructional sessions on augmentative communication and musical therapy, the children attended workshops and played games supported by the volunteer members of AKAFI, a not-for-profit volunteer association based in Las Rozas, Madrid, and family members who offered their help.

At the parent meetings we had the opportunity to hear from the following:

- Francisco Arjona, our secretary and IT expert/webmaster, shared his family’s experiences raising Lara and the strategies she is following using picture exchange communication system (PECS). Francisco, along with his dear wife, Francis, is deeply involved in several organizations and is unconditionally enthusiastic and tireless in pursuing the best therapies available for their daughter. We are all indebted to Francisco, our role model for following the program and sticking to it.
- Miquel Giner presented an overview of music therapy, the current approaches, and the strategies that can be followed to implement a musical program in the home setting that helps our children enhance their communication skills. Miquel Giner, an expert in music therapy, is also very involved in our association. Keep an eye on his Music15q project creation, which we will be talking about in the coming months!
- Finally, David Romero, our expert in augmentative communication devices, provided us with invaluable tools for providing additional communication channels for our children with iPod and PC support (soon to become available for Android). David is a founding member of ARASAAC, Aragonese Portal of Augmentative and Alternative Communication, a public entity funded in part by the regional government of Aragon, which promotes the use of communication tools to improve the lives of the disabled. They designed the pictograms for the Asociación IDIC15 website.

Also, the following was shared with the membership:

- The website is now live: www.idic15q.es. Please visit and share your thoughts about the page with us—in any language!
- Access to the Dup15q Alliance International Registry is supported by simultaneous English–Spanish page translation, intended to support the correct introduction of data in the registry database.

Finally we should attribute the success of the first get-together to the following people and their families, who helped make this meeting possible: Maite Coto, Elena Fernandez, Vanessa Torralbo, Francisco Arjona, and Miguel Angel Orquin.
The volunteer of the quarter is Greg Keller. Greg plans and coordinates the Northwest Dup15q Alliance family weekend at Camp Prime Time, a camp near Clear Lake in the beautiful Wenatchee National Forest of Washington State. Camp Prime Time seeks to enrich the lives of families with children who are seriously ill or disabled by providing an outdoor wilderness experience where they can enjoy themselves in a supportive atmosphere without financial burden. Congratulations, Greg, and thank you for your hard work and commitment to our families.

How did you first learn about Camp Prime Time?
When our son Andrew was 2, we were receiving early intervention services and were asked if we would like to attend Camp Prime Time. We were in desperate need of some family outing time, so we agreed to go. When we arrived, it was awesome. Then after talking to the Prime Time staff and learning more about their services and mission, I discussed it with my wife, Carol, and we decided to spearhead a dup15q family camp. This summer marked our fourth dup15q family camp at Camp Prime Time.

What are the greatest benefits of bringing Dup15q Alliance families to Camp Prime Time?
I feel the greatest benefit is undoubtedly the experience of having our kids among other families in a relaxing situation where they can be fully accepted as who they are without a second look or comment. The parents can share their experiences, benefits, lessons, and growth with other parents who have different gifts. We can use those natural and learned skills to better our own lives and families.

How much time does coordinating Camp Prime Time take, and what is involved?
A great deal of time e-mailing, contacting by phone, explaining, and coordinating. The process is a little hairy at first. Families need to know dates of camp as soon as possible due to school, vacations, and general planning; however, the Camp Prime Time Board does not choose the year’s camp session until the first week of March. By that time, the number of families that will attend needs to be available, but the families won’t know until they get dates whether they can attend, so we start with a guessing game. When the date is assigned, the fun begins: contacting each family and getting them all the information, from maps to supplies needed. The greatest challenge is finding an available date that as many families as possible can attend without a family being left out due to a schedule conflict.

What is your greatest joy in coordinating Camp Prime Time?
Seeing all the families relax, share stories, and enjoy good company and good old-fashioned family time. And most of all, experiencing the growth of each child and knowing that the other parents understand the pure joy in the small steps forward, like walking, talking, potty training (hurray!) and the ever-so-slight advancements that are often overlooked with typically developing children.

What is the benefit of the Camp Prime Time weekend for your family?
Our family, like all others, has its own challenges, and in the hustle and bustle of daily life we don’t often get the chance to spend time together, so this is that opportunity to avoid the mindless idiot boxes (video games and television) and enjoy family time. Also, this is the time that the siblings can make friends, vent, and discuss the challenges and successes unique to them, as well as watch the siblings of other children with dup15q interact appropriately and lovingly with their sisters and brothers. This camp is a full team effort. I may be at the center of the coordination, but I would be completely useless without my wife, Carol, and all her hard work taking care of three children and keeping us all focused and as organized as possible. Other families—the Lowells, Clearys, Gazewoods, and many others—jump in and spread the news, make contacts, and give feedback and especially encouragement. Without the families, I would be coordinating nothing. And a special, deep, heartfelt appreciation to the Camp Prime Time staff and volunteers, who not only kindly tolerate my endless calls and questions but also those of many other coordinators from other groups. The patience of angels they have. And the camp staff and volunteers who cook; clean; and operate the camp, boat rides, horseback riding, campfires, and sing-alongs—to all of them the gratitude is deep.
Beyond High School

BY MELANIE O’NEILL

My son Brenden celebrated his 21st birthday in May. This meant he would graduate from school and attend his senior prom and other senior class activities. Unfortunately, though, he would no longer be able to attend Club Aspire, an after-school program he attended for 15 years where he would work on academics and social skills, celebrate holidays and birthdays by making crafts and goodies, practice using the computer, and play in the gym. The gym was Brenden’s favorite place. He had his favorite spot where he would look out the window and watch the cars go by. There was even a train that would go by once a week.

Brenden has required 24-hour supervision. He has exhausting seizures that have caused profound delays. Despite these challenges, he wakes up every morning with a smile on his face and is very loving. Although he is nonverbal, he can communicate and knows exactly what he wants. He loves to play with his trucks and cars, hum nursery rhymes, watch his Thomas the Train DVDs, and look at books and magazines. He can run faster than anybody I know.

Recently, I have been very anxious about how Brenden would react to his last two weeks of school and his transition from school and Club Aspire into the day-habilitation world. He attended his senior prom in June. The school asked whether he could be accompanied by his community habilitation worker, as parents were not allowed once the party started. That night, I helped him with his dressing and realized I couldn’t knot a tie very well. I called my son Kevin for direction, and he suggested I go on YouTube. So there I was, knotting a dress tie, thanks to the gentleman on the computer screen. Brenden looked so handsome, his light blue tie matched his blue eyes. He looked like a Wall Street broker.

When we arrived at the prom, we were met by Brenden’s worker, who commented on how handsome he looked. The prom’s theme was Depths of Forever, and the gym looked like a wedding reception hall with great food and music. After waiting in the portrait line for five minutes, Brenden wanted no part of it, so outside we went. We walked through the parking lot and started singing songs. After 20 minutes of singing, I actually had to hide between the cars so his worker could quickly walk him back inside. Three hours later, they were home and I was told he had a great time. One girl pulled him onto the dance floor and he danced. Whenever his worker felt that Brenden was starting to get a little overwhelmed by the loud music, they would step outside for a few minutes. Brenden came home with his prom picture in hand and a smile on his face, and he went straight to bed.

A few weeks later at a family function, I saw pictures of my nephew’s junior prom. He and 200 other students were waiting for limo buses to arrive. For Brenden’s prom it was me, him, and my Dodge Caravan. I couldn’t help but feel a little sad, and on my way home I allowed myself to cry. I realize we all have our own journey, but I couldn’t help think about the “what if’s.”

Brenden’s graduation was a week after prom, scheduled for a Thursday at 9 a.m. I was worried that he would have a seizure early that morning and be too tired to be at his graduation on time. Fortunately, that was not the case and he proudly walked into the auditorium with his 1:1 aide and classmates. He was seated on stage for two hours in the back row. His aide gave him a snack when needed, and he held his favorite musical toy. He received a speech award, which he proudly accepted. Brenden was the last person to receive a diploma. He walked up to the podium with his aide behind him and was greeted with loud applause from family, friends, and members of the after-school program. Pictures were taken and a party followed in his classroom. Staff members gave him a Thomas the Tank Engine puzzle, a big bottle of bubbles, and strands of graduation beads, which Brenden loves to squeeze and hold onto.

Brenden’s graduation day was also the last day of his after-school program. They invited my other son, Kevin, who used to work there, and me to join them in a party for Brenden, where cupcakes were served and more pictures were taken. There was one particular person who worked with Brenden who had a beautiful voice and would often sing to him. She wore a button of Brenden’s graduation picture on her shirt and cried throughout the whole day. I had heard from other people that she kindly offered to adopt him. In his yearbook she wrote “You will always have
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a special place in my heart and I will never forget you.” We exchanged numbers, as I wanted Brenden to be able to see her again. I wondered if he understood that this was his last day. As we left, I cried because Brenden wouldn’t be in this program anymore, although a possible Saturday program for adults 21 and older was in the works.

Throughout the past year, my main concern has been transitioning Brenden to a day-hab that would meet his needs. I started this process one year before graduation, as I was told there was a long waiting list for most programs. After visiting many programs I noticed that most of the customers were of geriatric age and wondered where the young adults were. Were they all high-functioning and working? Another issue of concern was safety. I was told that they couldn’t lock doors to the outside because it was in violation of fire code laws. I finally found a program where they met the needs of all people with different abilities. They specifically had a program that dealt with consumers who are on the autism spectrum. The building was huge, and I liked that the rooms were large so clients could roam around. Attached to the main room was a smaller room where Brenden could receive 1:1 teaching in a quiet environment. He is easily distracted and has a short attention span, so he requires this type of learning environment. To make this transition work, the behavior specialist from the day-hab observed Brenden in school, spoke to the teacher, and saw some of the programs they were working on. I also suggested she visit him during his favorite class, speech, to see how much better he works in a quiet environment.

I wanted Brenden to have some familiarity as he grew comfortable with the new program, so I planned his transition to day-hab carefully. Throughout the years I have sent positive reinforcements, including books, musical toys, and beads to school. On the last day of school, I picked up these toys, updated what was needed, and packed them in a container ready for the day-hab program. I also had the teacher make copies of some of his programs to be provided to the new staff. For the first week of day-hab we visited the program for only two hours each day so Brenden could adapt to his new environment and staff could get to know him. At times, I left the room and let them work with Brenden while I observed through the window. His program includes academics in the morning and time out in the community in the afternoon.

I was told that the day-hab requested an aide specifically for Brenden, which our caseworker and others told me was unheard of. But this day-hab is very cautious and saw how Brenden can bolt right out of the room. For safety reasons, the aide was approved; I couldn’t have been happier. Transportation was another concern, as most day-habs do not provide aides on the bus and usually transport many consumers at one time. I shared my concern, and the agency provided a van for Brenden with a staff member from his group who does the driving. Brenden is the only consumer on the van. They did include an aide on the van but later found it wasn’t necessary.

I was unemployed during the time Brenden was transitioning from school to the day-hab, so I had an opportunity to speak with different staff members daily when they picked him up and dropped him off. One of the staff is a musician who plays six instruments. I shared with him Brenden’s love of music and asked if he could teach him to play an instrument. He is now attempting to teach Brenden the organ, since he enjoys his toy piano. I shared with another staff member how Brenden stands at the end of the slide at the park and loves to watch when the kids come down. On that same day, the group went on a picnic to a local park, where Brenden stood right in front of the slide. With no kids around, this 20-something-year-old staff member went up and down the slide while Brenden clapped and jumped up and down.

I’ve also requested that the day-hab staff write me a daily note about Brenden, as I am returning to work and will no longer have the luxury of speaking with them. The staff has shared with me that they feel Brenden is very smart. I can tell from what they’re sharing with me that they have a good understanding of him and will push him beyond expectations. This new environment has exceeded my expectations, and I feel that Brenden has adapted very well. The staff members and I made these changes work. I’m also very proud of Brenden, and I know that angels are watching over him. I’m gearing up for his next transition, which will be to a group home, but for now we will deal with one transition at a time.

Melanie and her two sons, Brenden, 21 (dup15q), and Kevin, 23, live in Monroe, N.Y.
A Lifetime of Transitions

By Coco M. D’Ascola

While biking with my son, Santiago, around our neighborhood at the end of the summer, we rode past last year’s preschool. He immediately insisted that we turn around, wiggling to get out as he reached toward the gates. In a few weeks, he would be starting at a new school in a specialized pre-K program, and I was hoping this visit wouldn’t confuse him.

I opened the gate and let him run straight past me to his beloved playground. But as I sat on the swing, a sudden feeling came over me. Tears started streaming down my face as I watched him run around so comfortably and happily.

Memories of his first school year flooded my mind. It was a mainstream school that had accepted him and worked with us to meet his needs as best they could. He attended with a private aide and progressed in leaps and bounds. We were heartbroken to leave, but aware that it was time to move on. So after much preparation and collaboration with Team Santiago, we got everything we requested in his first IEP.

We felt optimistic about the upcoming changes, having heard great things about his new teacher and program. But it was not until that one moment on the playground that it hit me: change would be our new norm. I started thinking about transitions and the emotional impact they have on our lives.

Life is all about change, and I know that every parent deals with it. But, as a parent of a child with dup15q, we don’t have an “expected” path to follow, a natural comfort in knowing there is a certain flow to things. We have to chart our own courses as we go. Navigating life requires flexibility, patience, preparation, and faith. We have to be ready to adapt and make difficult decisions.

My husband and I work hard to provide everything our son needs, and Santiago works even harder at growing and learning every day, pushing though all the changes and making the journey worthwhile. But sometimes, we need to stop to let the impact of the work, progress, and success sink in.

That afternoon at his former school, I realized how important it is to not only acknowledge all the effort we have put in, but to be realistic about the work it will take to ride through countless transitions ahead. There is great peace in accepting this. We are forever dedicated to a life of change with Santiago and hopeful that we will always find the strength and grace we will need along the way.

Coco, Fabio, and Santiago, 3½ (dup15q), live in Florida.
Reflections from a Grandmother

By Vivian Rojas

My Story of Kaylor

Kaylor, my 4-year-old granddaughter, has dup15q syndrome. We live 6½ hours away from her family, but we visit often. From the moment we arrive until we leave, I enjoy all of my time with her.

She is a very loving little girl who always gives me kisses. She sits on my lap, looks at me with a smile, and says “Nana.” I tell her, “Yes, baby, I’m Nana.” She runs her soft little hands over my face and through my hair, and she gives me kisses and big hugs! Kaylor also loves to sing and is always singing songs that she has learned at preschool. She can even say the Pledge of Allegiance. Kaylor really enjoys the outdoors. She loves to walk, jump, and skip. She loves to feel the warm sidewalk and rocks.

Kaylor is the second child of three. She has a younger brother, Kole, and an older sister, Kasaria, who is 14. Kaylor is blessed with wonderful parents who are always there for her. They have become very involved in Dup15q Alliance to seek help and also share their ideas with other families. I thank God for the wonderful home that her parents have provided for her and that each day she continues to learn more on her own.

Vivian, mother of Franny and grandmother to Kasaria, Kaylor, and Kole Mackling, lives in Scottsbluff, Neb., where she is a bilingual paraeducator.

Reflections from a Sibling

By Lorenzo Jannon

My Brother Luca

My brother Luca is 8 years old and he has an extra small piece of chromosome 15. I like him because he is always in a cheerful and lively mood.

He plays most of the time, and when I go near him he always listens to what I’m telling him and tries to hug me. What I do not like about him is when he scratches me and pulls my hair or when he puts his hands into my mouth, but the worst thing he does is when he slips his hands into his throat until he vomits.

I feel a close bond of love and affection for Luca. I play with him, and we cuddle each other and sleep in the same room. I’m worried about Luca’s future; I wonder who will take care of him when I’m not there. Will he ever find true love?

I remember once when Luca put a hazelnut into his mouth. I heard him choking and I began to cry until dad came and took it out. Thanks to Luca, I had the chance to go to the United States to meet other families with children like him. I had always wished to see the T. Rex at the American Museum of Natural History in New York, so my biggest dream came true.

Lorenzo, 11, lives in Italy with his family. On the occasion that his brother choked, he was only 6 years old but quickly got his parents’ help, saving Luca. He and his father enjoyed attending the 2011 international conference in Philadelphia and visiting New York and Washington, D.C., thanks to the hospitality of Dup15q Alliance members.

http://www.dup15q.org
Duplicity Alliance is a nonprofit organization that provides family support and promotes awareness, research and targeted treatments for chromosome 15q duplication syndrome. Duplicity Alliance offers help and hope for chromosome 15q duplication.

Great news! We now have a calendar on our website so you can keep track of all of the upcoming events. If you have an event to put on the calendar, please let us know! Visit www.dup15q.org/calendar.html.

2013 CONFERENCE RAFFLE TIME! By Katie Supina

With less than a year before the 7th International Conference in Minneapolis, it is time to start thinking about what you can donate to the raffle and silent auction. In Philadelphia we raised over $10,000 at the conference raffle—an amazing feat that we hope to surpass at the conference in July 2013. The raffle and silent auction will be held during the reception dinner on Thursday, July 25. We would love to include a basket for each home state and country represented. Think of what you can donate, work together, and be creative! Donations might include CDs, art supplies, jewelry, foods from your home state, sports memorabilia, toys, DVDs, quilts, shirts, gift certificates, coffee, or whatever you think people might try their luck at winning!

Watch for updates on BigTent to see the list of great raffle items coming in. Either send the baskets to the Supina family in Minnesota or bring them with you to the conference. If you plan on bringing the items directly to the conference, send me a quick e-mail so we know what to expect.

Contact me if you have any other raffle and silent auction questions!
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