

Annual Report 2000

HIGHLIGHTS OF THE YEAR

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The year 2000 marked our tenth anniversary as an organization. Over these past ten years we have touch the lives of over 1000 affected families. This amazing number is even more impressive in light of the fact that many of us were told that our child's syndrome was very rare with possibly only several dozen other affected families in the world. Our families include children as well as adults with chromosome 18 abnormalities. We have found each other across oceans and language barriers, yet are all bound by the common trail that we are blazing to help individuals with chromosome 18 abnormalities live better lives.

Our accomplishments of the past year are again derived from the talents and energy of our members. Claudia Traa chaired our annual conference that brought families together from all over the world. The story and pictures are inside. We sponsored our first annual San Antonio charity golf tournament, directed by Jim Traa. This

fundraiser was fabulously successful, even though this was its inaugural year.

In 2000, we also launched a completely revised web site. This site is our primary mechanism for reaching new families, their educators, and health care providers. The index page art (pictured below) was created by Bruce Steinke (Dylan, 18q-) and the web master who designed and maintains

the entire site is Robert Moss (Zachary, 18p-).

In addition, a seven-minute introductory videotape was completed by Dawn Scibilia (aunt of Jason Sassono, 18p-). The tape is available free to our members to use at educational and fundraising events. The video features our own members explaining who we are and what we are doing to help our affected loved ones.



The mission of **The Chromosome 18 Registry & Research Society** is to help individuals with chromosome 18 abnormalities overcome the challenges they face so they might lead happy, healthy and productive lives. We do this through service, education and research.

OUR MEMBERS TOUR THE CHROMOSOME 18 CLINICAL RESEARCH CENTER FACILITIES AT THE UNIVERSITY OF TEXAS HEALTH SCIENCE CENTER AT SAN ANTONIO



One of the highlights of the conference was a tour of the University of Texas Health Science Center at San Antonio. Families were welcomed to the Health Science Center by an important group of university officials; Dr. John P. Howe, III, President of the Health Science Center, Dr. Anthony Infante, Associate Dean for Research, Dr. Celia Kaye, Chairman of the Department of Pediatrics and Dr. Brian Herman, Chairman of the Department of Cellular and Structural Biology. After the introductions, the adults were taken on a tour of the

Health Science Center facilities and the children stayed in the auditorium and were entertained by Daisy Bee the clown.

The tour included visits to the various laboratories and the hospital. Eillen Lundeen (above and to the right), Research Coordinator for the General Clinical Research Center, shows off the facilities that are housed in Audie L. Murphy Veteran's Hospital. This is where families who participate in the clinical research studies stay



during their visit to the Chromosome 18 Clinical Research Center. The staff of the General Clinical Research Center very proudly display pictures and thank you notes on their bulletin board (left). These have all been sent from the families who have participated in the chromosome 18 studies.

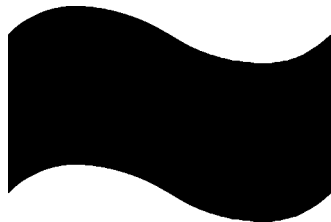


During the tour of the various laboratories, families view cells through a microscope (upper left). Dr. Robin Leach (upper right) shows how cells are grown in the incubators behind her and are kept frozen in liquid nitrogen for storage. Dr. Teresa Johnson-Pais (right) explains her plans for making mice with 18q-. Xavier Reveles (lower left) discusses how chromosomes are analyzed under the microscope. Families (lower left) are shown how to interpret molecular analysis data in the laboratory of Dr. Jannine Cody.



The Chromosome 18 Registry & Research Society

RESEARCH ON THE SYNDROMES OF CHROMOSOME 18



In 2000, The Chromosome 18 Clinical Research Center at the University of Texas Health Science Center at San Antonio continued to broaden its scope to include all the syndromes of chromosome 18. The clinical and molecular studies on 18q- began in 1993. In 1997 molecular studies of individuals with 18p- began, followed by clinical evaluations, which began in 2000. Also in 2000, the molecular studies were expanded to include anyone with any chromosome 18 abnormality. The scope will be complete only when all of the syndromes are able to be included in the clinical studies.

This Center will be able to bring together the critical mass of information that will be necessary to learn about and treat the chromosome 18 syndromes. This information will come through the combined efforts of many scientists from a variety of medical specialties in concert with the participation of as many affected individuals as

possible. Together, families and researchers will overcome the two major obstacles of the rarity of the syndromes and the complexity of a chromosome abnormality.

Highlights of some of the specific findings from the Chromosome 18 Clinical Research Center are:

In individuals with 18q-:

- Virtually everyone has a unique breakpoint on his or her chromosome 18.
- 14% of the people who were told they had deletions of the end of the chromosome actually had more complicated rearrangements that were interstitial deletions, partial duplications or involved other chromosomes.
- Mental ability is related to the size and location of the deletion. Those with the biggest deletions have the lowest

mental ability and those with the smallest deletions have the highest.

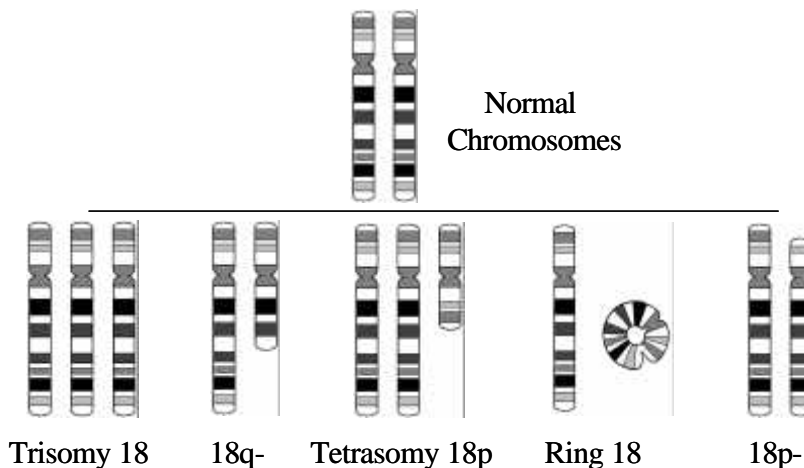
- 68% qualify for growth hormone replacement therapy.
- Individuals treated with growth hormone have performance IQ changes of between 0 and 47 points, with an average increase of 23 points.

In individuals with 18p-:

- Only 20% have mental retardation, yet 100% have speech delay.
- 72% have similar breakpoints near the centromere.
- 23% qualify for growth hormone therapy.

In the coming years, we will add information to this list about every syndrome and increase the depth of information.

The Syndromes of Chromosome 18



PROGRAMS AND SERVICES

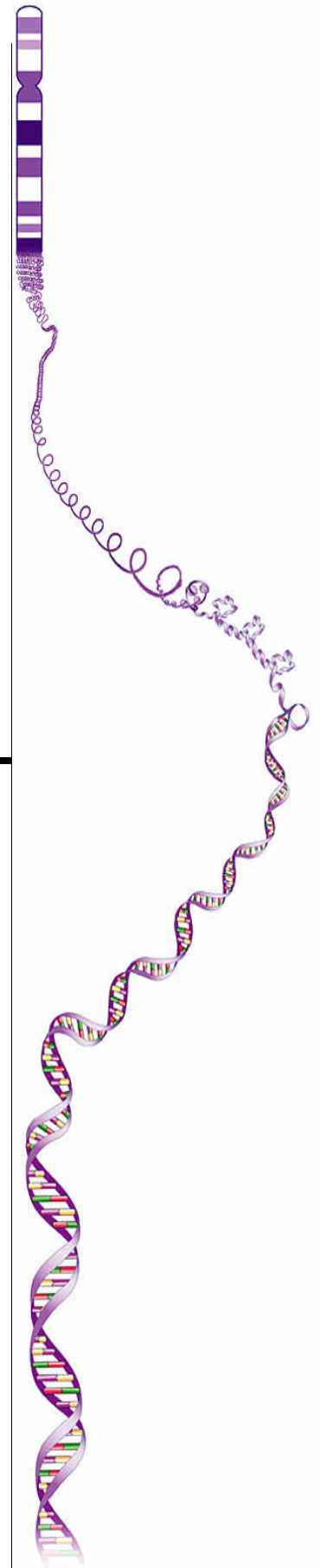
Our mission is to help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy, happy and productive lives. This mission is accomplished through *service, education* and *research*. Our service activities include an annual conference and numerous newsletters. *The Chromosome 18 Communiqué* is the most widely circulated of our newsletters. *Memos for Members* is a member's only newsletter containing information about upcoming events and ongoing projects. Our syndrome coordinators (one for each of the five major syndromes) provide a syndrome-specific

closed circulation newsletter for the exchange of information and personal stories among families. Each of our ten Regional Coordinators oversee local events such as family get-togethers as well as local public and professional education. Our newly revised web site is our primary means of public outreach.

We have two main avenues for parents/guardians to share information: the Parent Network and listserves, both open to current parental/guardian members. The Parent Network is simply a contact information list. The online listserves (18q-, 18p-, and Tetrasomy 18p) are maintained by parent volunteers.

We believe there are

no incurable conditions, only those that have not yet been understood. Our goal is to make sure that the syndromes of chromosome 18 are understood. This understanding will bring treatments and therapies that will lessen the burden of these syndromes. The path to understanding the chromosome 18 syndromes is paved with research dollars. Clinical research is very costly, which is why we devote so much effort to fundraising. The price may be high, but the potential rewards are even greater. Research will bring us the answers to the many questions we have about our loved ones. It will give us insight and will guide our actions.



FINANCIAL STATEMENT

for the year ending December 31, 2000

Revenue and Support

| | |
|---------------------------|----------------|
| Membership Dues | 9,119 |
| Interest Income | 3,122 |
| Conference Income | 23,093 |
| Merchandise Sales | 7,852 |
| Fundraising | 138,433 |
| Combined Federal Campaign | 10,279 |
| <u>Contributions</u> | <u>55,315</u> |
| TOTAL | 247,213 |

Expenses

| | |
|------------------------|----------------|
| Program Services | 93,609 |
| Research Grant | 80,000 |
| Management and General | 5,480 |
| <u>Fundraising</u> | <u>8,844</u> |
| TOTAL | 187,933 |

| | |
|--------------------------------|----------------|
| Net Assets, Beginning of Year | 91,361 |
| Net Assets, End of Year | 150,643 |

Prepared by Cavazos and Coleman, P.C.
 Certified Public Accountants

2000 FAMILY CONFERENCE

Last June we held our annual family conference near the Registry's home base of San Antonio, Texas. Tapatio Springs golf and conference resort in Boerne, Texas, proved to be a wonderful forum for both education and fellowship. The conference began on Sunday, June 25, with a successful charity golf tournament and silent auction. This was followed by a combination awards ceremony and welcome reception. It was a wonderful opportunity for local supporters to get a glimpse of our families and our wonderful kids. Since The Registry is headquartered in San Antonio, it is essential the city know our organization and our mission. The timing was wonderful from a media standpoint because our conference coincided with the announcement that the entire human genome was mapped. This resulted in local news coverage and two informative newspaper articles about The Registry.

Early Monday was the official start to three days of numerous, exciting speakers. First we debuted our 7-minute video produced by Dawn Scibilia, aunt of Jason Sussano. Brad Prewitt, counsel to United States Senator Thad Cochran from Mississippi, officially started our program. Brad has been an ally to The Registry prior to our meeting with him during the Washington, D.C. conference in 1999. He spoke to us in a sincere manner about policymaking and how it applies to both our organization and as individuals with special needs children. He taught us that we had each a voice to be heard on Capitol

Hill, and encouraged us to be loud and clear. He shared with us the language, which he suggested appear in the committee reports, which spoke directly to *syndromes of chromosome 18*.

Don Meyer, Director of The Sibling Support Project, went above and beyond as a speaker. He held a "Grandparents only" workshop, where these wonderful members of our families were able to talk openly about their concerns, hopes and fears. Their discussion remained top-secret, but insiders reported that many sighs, tears, smiles, as well as stories of pride and joy were shared.

Don presented to all attendees, *Meeting the Unique Concerns of Siblings of Children with Special Needs*. His presentation was insightful and moving and drew our focus to the brothers and sisters of our kids. He addressed the sibling's possible feelings of isolation, ambivalence, guilt, and pressure to achieve. In addition, he explored their feelings regarding their increasing role as caregiver, especially for older siblings, and their fear of what the future holds in store.

Finally, Don took a group of about twenty siblings ages 8 to 16 and held a sample *Sib Shop*. The only attendees permitted were siblings. No adults!! Once again, Don provided a safe environment for the kids to offer each other peer support. He accomplished this within a lively, recreational context that emphasized a "kids-eye-view". Unique, offbeat fun games were the communication tools used in this support project.

In the science portion of our conference, Janine Cody, President, gave us a valuable lesson in Genetics 101. Even for repeat attendees, this portion always sheds light on something new or clarifies something we thought we knew. We were fortunate to have two distinguished guests from The National Institutes of Health (NIH). Dr. Mary Lou Oster-Granite, Ph.D., is a Health Science Administrator at National Institute of Childhood Development. She spoke to us about the NIH, specifically: what is it, how is it organized, and how the various institutes set priorities. Dr. Monique Monsoura, Ph.D., is Policy Analyst to Voluntary Health Organizations at the National Human Genome Research Institute. She spoke to us about the very newsworthy genome project, gene sequencing, and what this might mean to our families. A highlight to our conference is always the Research Update portion of the program. This year we were fortunate to have Drs. Dan Hale, Robin Leach, Peg Semrud-Clikeman, and Celia Kaye present an update of the research findings and progress.

Related to the science portion of our conference, the families boarded busses and got a tour of the University of Texas Health Science Center, San Antonio (UTHSCSA), and Chromosome 18 research facilities. Various faculty and department heads including University President, Dr. John P. Howe, also welcomed us (see pictures on pages 2 & 3). This allowed our families, many of whom only imagined what was going on at

Conference Statistics

Attendees included:

249 individuals
73 families
5 affected adult
37 extended family
members & friends

They came from 24
states and the following
countries:

| | |
|------------|--------|
| Australia | Canada |
| Costa Rica | Mexico |
| Scotland | |

UTHSCSA, to take away a more concrete picture of the important things happening there. We then took the buses downtown for some R&R at San Antonio's beautiful Riverwalk.

Paul Burton, J.D., M.S. Ed., educated us about parent's rights under the *Individuals with Disabilities Education Act* (I.D.E.A.). This very interactive presentation was designed to acquaint parents, educators and involved parties with specific sections of I.D.E.A. that address parent involvement.

Kathleen Doria, by trade a professional fundraiser, was an extremely dynamic, motivating individual who spoke to us about how to get involved in FUND-raising in our organization. She helped us understand that we are all capable of and responsible for fund-raising.

Since all of our children seem to have speech delay in common, we invited Shawn Williams, Speech-Language Pathologist to address our group. She discussed terms we hear all the time like language, vocabulary, articulation, fluency voice and oral motor. She analyzed oral motor, feeding and speech development during the first three years of life. She also armed parents with tips to use with their children to help with speech and language development.

Rick Guidotti, President of Positive Exposure was an "out of the box" addition to our conference. Rick, a former high profile fashion photographer, shared his vision and project "The Spirit of Difference" and "Redefining Beauty" and totally enchanted us. Rick decided he was weary of society's rigid definition of beauty. He immersed himself among people affected by the genetic condition known as

albinism. In a way that never exploited these people but rather helped to build self-image and self-esteem, he photographed them in all corners of the world. His mission is to show them and the world that they are, in his words, "amazing and gorgeous". We could all relate because our children are all, by Rick's definition and our own, truly beautiful. He photographed our kids at the conference and they felt pretty special. Visit his website at www.rickguidotti.com.

This year we were able to continue the tradition of Syndrome Breakout Session with each of the various syndrome groups meeting together to discuss problems, accomplishments, hopes, and fears. This is always a favorite part of the conference because we learn so much from each other.

The last day before our farewell Cowboy Bar-B-Que, we tried something new. We held two panel discussions. We had a panel of doctors present and then answer questions. Since our children are affected in different ways, we decided several specialists would be the

most effective way to address at least some of our medical concerns. The panel consisted of Dr. John V. Mumma, pediatric ophthalmologist, Dr. Kenneth Bloom, pediatric cardiologist, Dr. Ted Knicker, allergist, (*discussed his theory on how certain food affects behavior*), and one of our members Dr. Susan Sheinkop, pediatrician, Mom to Ellie, Ring 18 mosaic (*discussed medical advocacy*).

Our other panel was of our own very talented members, who have so much to offer. Panel members included, John McGing, *Dad to Sean, tetrasomy 18p*, Catherine Burzio, *Mom to Pauline, 18p-*, Juana Carter, *Mom to Geoffrey, 18p-*, and Torrie Bryant, *18q-*. This was an extraordinary segment to our conference. Members shared their struggles, accomplishments, advice, talents and heartwarming stories. Tears and smiles were abundant during these presentations. It just goes to show we don't have to look very far to gain insight and wisdom. Our own members have tremendous information to share.



A panel of our members shared experiences and information with the conference attendees. Catherine Burzio (l—r), John McGing, Juana Carter and Torrie Bryant.

MEMBER SUMMARY

Identified Families:

| | |
|---|-----|
| 18q- | 360 |
| 18q- mosaic | 19 |
| 18q- interstitial | 12 |
| 18p- | 186 |
| 18p- interstitial | 1 |
| Ring 18 | 89 |
| Ring 18 mosaic | 5 |
| Tetrasomy 18p (includes partial) | 81 |
| Tetrasomy 18p mosaic | 4 |
| Trisomy 18 | 319 |
| Trisomy 18 (includes partial & mosaic) | 30 |
| Trisomy 18q (includes partial & mosaic) | 24 |
| Trisomy 18p (includes partial & mosaic) | 11 |
| Other | 148 |

Our identified families live in:

| | |
|---------------|------|
| United States | 1014 |
| Canada | 78 |
| Mexico | 3 |
| Australia | 36 |
| Europe | 133 |
| South America | 19 |
| Africa | 6 |

ONE FAMILY'S STORY

It's hard to believe that over a year has passed since Talia came into this world. Like the hurricane that threatened the very foundation of St. Mary's hospital the day after her birth, Talia's start in life was a rocky one. And while the winds have calmed and seas settled, these past twelve months have left all us a bit weathered. It's been one battle after the next with the end far from over, but we're charging uphill and that's what really matters. Fortunately the Chromosome 18 Registry & Research Society has been there for us every step of the way, furnishing the kind of guidance and support that keeps us trudging along and coming back for more. Thanks to them, we've made enormous progress that still leaves us amazed. As for our little girl, she's a true wonder. Never could a family feel more empowered. Never could a family feel more blessed. Never could a family feel more thankful to have such a daughter as Talia. In Hebrew

Talia means "Heaven's Dew." There's no doubt -- our little dewdrop was heaven-sent!

An easy pregnancy, an easy labor, and an easy delivery ended with an emergency suctioning, a chest X-ray, a low birth-weight and all sorts of questions. Even the honorary umbilical cord-cutting was usurped by the obstetrician while daddy stood by helplessly waiting.

As Hurricane Irene's wrath left the aged hallways flooded, monitors flickering and back-up generators burning, we spent the next few days going back and forth to the Neonatal Intensive Care Unit. All those hours I had spent pouring over pregnancy and parenting magazines were to no avail. None of them had breached the subject of brown scrubbing soap, 48-hour neonatologist duty, miscommunication and chromosome tests. Rather they were all peaches and cream with a bit of postpartum depression on top. Lucky for us,

the well-wishes of family and friends helped keep our spirits alive despite the heartache we felt beneath. Forever, we will be grateful!

Two weeks later, Seth and I - with Talia in hand sporting an adorable Hannah Andersson Swedish special, don't-cha-know - sat facing our pediatrician whose sullen face was enough to make the knots in my stomach do a few extra loops. He had contacted us the day before with the results of the chromosome test, explaining that while the test out-ruled Down syndrome, it revealed another chromosomal abnormality. But, since he did not like to discuss such matters over the phone, he insisted on meeting us the next day at 6:30 p.m. -- after his office had closed. Click. So there we were twenty-four hours later, seething with anger from having to wait, our insides twisted, thoughts numb, throats dry.

After a moment of

awkward silence, he began, "How long have you been married?" Whew! Finally a chance to speak, let alone breathe. "A year, but jeez it feels like more," we joked. "We've known each other forever and, let me tell you, it sure does feel like it!" Any hope for comic relief at that point in time was quickly extinguished as the doctor shook his head and continued forth with his dispiriting preamble. "Well, you may think your marriage is strong, but I want you to know that these type of things break up marriages. Divorce is common. 80 percent common. Your lives will be forever changed. Happiness will be hard to come by. And you know those friends you thought were friends? Well, they won't be there for you anymore. They'll run away and hide. There are always a few that do. It will be lonely struggle - a difficult one. You'll never be the same again. It will be a strain on you. It will be a strain on your entire family. "

Fifteen minutes later, he finally got to the core of it. "Your daughter has a rare chromosomal abnormality called 18q- syndrome. It's very rare, very severe. Actually, I'd never heard of it. We're talking 1 in 500,000 or something like that. In fact, most kids with 18q- are typically miscarried. It's not good. It's possible that your daughter will never grow - that she will remain in a frog-like position and vegetative state for the rest of her life. She might live two years. She might live 80. But chances are that she will be bed-ridden for the rest of her life, never responding to you, never doing anything, her IQ non-existent"

Take that one and swallow, then fast-forward 12+ months. Can hear Talia's squeals and laughter as she babbles at the TV set in the adjacent room. Craning my neck around the corner away from my computer terminal, I look to see what all the fuss it about. Wouldn't you know, it's Tinky-Winky, that flamboyant purple Tellytubby again. This time he's chucked the purse and grabbed a fluffy white tutu instead. As he pirouettes around Tellytubby land, Talia flings her arms about and bangs her blue-casted feet against the chest next to her. She oohs. She ahs. She babas and das. I swear she's hootchy-cootchy-ing it. She's loving it. I'm loving it.

A year ago, I never would have thought that Talia would be in a play group with Alexander, Max, Fraser, Annika and Lindsay. Six months ago, I never thought that she'd go swinging and sliding at our neighborhood park with across-the-street friend Cayley --or that she'd explore our now infamous Palm Beach County with Boca pals Spencer, Sam and Alex. But she does all that and more. And it's just such a joy to see her interact with her buddies as they do all the things that wee ones like to do. They certainly are the cutest toddlers around and we're fortunate to have met them. Recently, it's been one birthday party after another. Next to Talia's social calendar, mine is non-existent!

Being a ballet dancer, you can't help but notice a person's feet, especially when it's your own daughter's. I detected something was wrong from day one in the NICU. As the weeks progressed, my suspi-

cions only heightened. Never mind her prominent heel or her rocker-bottom feet, let's talk flexibility. I couldn't point Talia's feet. I struggled to position her feet in a neutral position. Instead, it remained almost rock-hard in a flexed position. Thanks to the Chromosome 18 Registry & Research Society, we were able to confirm our suspicions and get Talia's feet and ankles squared away before it became too late.

Finally after convincing the doctor to send her in for some X-rays, we got the results. "Nothing wrong, other than flat feet," we were told. "All babies have flat feet. Adults have them too. It's really no big deal."

Well, I didn't train all those hours for nothing. Call it years of bunions, blisters and calluses. Call it years of comparing one arch after the next. Call it years of trying to get that arch higher and higher and higher. Call it what you will. The fact of the matter was simple: My daughter couldn't point her feet. But where do you turn?

Fed-up, I sat down at the computer and wrote out a description of Talia's feet to post on the Chromosome 18 Registry & Research Society's parent on-line support group. I had no clue what the response would be. Within minutes - and I'm talking minutes -- I was bombarded with emails from other parents. Vertical Talus, Vertical Talus, Vertical Talus. It was unbelievable how fast those responses came flying in. It was unbelievable how common Talia's foot abnormality was to some of the parents on this internet site. One West

All those hours I had spent pouring over pregnancy and parenting magazines were to no avail.

Palm Beach pediatric orthopedic surgeon, one New York Hospital for Special Surgery pediatric orthopedic surgeon and one Boston Children's Hospital pediatric orthopedic surgeon later (where we remained), we had our diagnosis confirmed three times over. Talia's condition: Vertical Talus (Tali for two), a rare foot abnormality in the world at large. But, as we surmised from all those emails we had in hand, a commonality amongst the kids with 18q-

Fortunately we found out about the Chromosome 18 Registry & Research Society from day two of Talia's diagnosis - and it's been our lifesaver ever since!

W i t h o u t knowing about my past pirouetting life, Talia's orthopedic surgeon innocently told me point-blank, "Talia will never be a ballerina, let alone a professional one at that." At first it stung, but only for a second. I realized that his comment was intended to make me feel good. Most parents probably wouldn't care less whether or not their child ever danced in a Nutcracker, wore a leotard or executed a plie. So, why should I? After all, he did say that she would be able to play and have fun like all the other kids her age. In the scheme of things, that's what really counts. I walked away quite happy.

Thanks to the Chromosome 18 Registry & Research Society, we were alerted to check Talia's hearing on a regular basis. Apparently, hearing loss in individuals with 18q- is very high - and so are very narrow ear canals. We were advised to take heed of Talia's growth hormone levels - and fast. We were not to wait until she had stopped growing. It's been nightly injections ever since - and

will be from here on forward. I never knew how someone like me (who goes faint at the sight of a needle) would be able to administer the treatment to Talia, let alone by myself. But I do. Thanks to the support of parents affiliated with the Chromosome 18 Registry & Research Society and other friends who have faced similar circumstances, these nightly injections - administered with a short insulin needle - have become a breeze for both of us (Seth does weekends) and seems to get better with each passing day. It's remarkable. She doesn't even flinch. Now brushing her teeth is a whole other matter.

Fortunately we found out about the Chromosome 18 Registry & Research Society from day two of Talia's diagnosis - and it's been our lifesaver ever since! Aside from the specialists mentioned above, the Registry recommended that Talia see others. We have, for example, learned that Talia's vision is fine. We have, as another example, learned that Talia's heart and kidneys are healthy and strong. We've even seen a neurologist at Boston's Massachusetts General Hospital. He seemed quite impressed that Talia was doing so well. Like we said, she doesn't fit the textbook definition - and especially the kind of textbook that has yellowed and crinkled with age.

As much as the Chromosome 18 Registry & Research Society has done for Talia and for us, it's also done a great deal for Talia's doctors who are eager to learn what research has shown and what parents have to say. As for our pediatri-

cian, he's become our Florida medical mainstay, always eager to listen, always eager to learn, always eager to assist in any way that he can. I now understand why he broke the news in the way that he did. He was simply preparing us for the worst, leaving the sugar-coating behind. Things have only gotten better since - and each time he sees Talia, his grin gets wider and wider and wider.

Finally, the Chromosome 18 Registry & Research Society has introduced us to parents like us, as you might already conclude. Each year, the Registry holds a conference. This year it was in San Antonio. Words cannot express what it felt like to meet the people we had met online -- to run into a college friend whose daughter has 18p- -- to watch Talia interact with others so very much like her -- to visit the research facility -- to attend workshops - to see the Alamo behind a locked gate -- to put things in perspective (things really could be much worse) - and to let our hair down and simply laugh. Words cannot express what it felt like to meet Torrie, an aspiring journalist from Massachusetts and Kathryn, a genetics student at the University of Glasgow. Both young women have what Talia has, namely 18q-.

We have no idea what the future holds for Talia. But there is one thing we do know for certain. No matter what path she'll take, we know that there will always be lots and lots of people cheering loudly behind her.

Liz Grossman

The following people are responsible for the success of our organization. Without their generosity, none of our services or accomplishments would be possible. The membership of the Chromosome 18 Registry extends its sincerest thanks to the following donors.

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