Annual Report 2005

Proud of our Progress

INSIDE THIS ISSUE:

Make no little plans; they have no magic to stir men's blood.

- Daniel Hudson Burnham

2005 was an important year for the members of the Chromosome 18 Registry. This summer we saw the publication of the first of what we expect to be a long list of specific therapies that will help people with chromosome 18 abnormalities. The article entitled, "Growth Hormone Benefits Children with 18q Deletions" was published in the August issue of the *American Journal of Medical Genetics*. This work demonstrates that children with 18q deletions who are growth hormone deficient have improved growth with replacement therapy, but they also have other improvements as well. These improvements include changes of non-verbal IQ scores by as much as 47 points. This is a huge improvement that will dramatically change the life course of those who have this magnitude of benefit. There are many questions yet to be answered about why this happens and why it does not happen for all children equally. But the work is ongoing and will continue to improve the lives of people with chromosome 18 abnormalities.

This did not happen because we waited "our turn" for someone at some drug company or university to find a new market or treatment. It happened because the members of the Registry banded together, raised the money, hired the staff, participated in the research and saw that the concerns of our families were taken seriously. This is just the beginning. There are more challenges ahead, but there are many more answers as well. This year we clearly demonstrated the power we have to improve the lives of people with chromosome 18 abnormalities – if we just do it!

This Annual Report is filled with the many ways our members "just do it." They hold educational forums, fundraising events, conferences – and have a good time while they do it. This report is also filled the names for those who have helped our members in so many ways. Acknowledging their support here is but one small way we can say thank you.

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.

"You can not do great things. You can only do small things with great love."

~ Mother Teresa

Educational Events by Registry Members

One of the major goals of The Chromosome 18 Registry & Research Society is to educate the public concerning the varied syndromes of chromosome 18. Some of our very best educators are our families. During 2005, our parents made it their responsibility to share with others the knowledge they gained by being members of The Chromosome 18 Registry & Research Society. Following is a summary of just some of the educational events that took place in 2005 and who conducted them.

Jordon Alper of NY

- In November in Dix Hills, Jordon made a presentation to all 4th grade classes in her school about what a chromosome 18 deletion is, especially 18q-, and how it affects her sister.

Deb Ammann of FL:

- In July, presented to seven school personnel what 18q– is and its effects on her son, Kevin, especially in the areas of depression, autism and social issues.

Deb Bader of IA:

- In October, was part of a panel of mothers of children with special needs who presented to a class of college students entering the teaching profession. She discussed the needs of children with 18p– and their need for inclusive education and for a greater awareness for communication with school personnel to obtain services.

Marlene Brightwell of Australia:

- In October, in Sydney, there was a conference hosted for Australian families of children and adults with chromosome 18 abnormalities attended by over 100 persons. Conference consisted of speakers and workshops that gave families and professionals the latest information on chromosome 18 educational and research pursuits. Guest speakers were Drs. Jannine Cody and Daniel Hale.

Catherine Burzio of VA:

- Hosted in May in Berryville a regional meeting and activity day at Kiwanas Pavillion for 25 children and adults connected to The Registry. Shared and discussed the therapies, educational pursuits, progress and accomplishments of their children..

Kristen Earl of MI:

- In August in Portage, Kristen spoke to school personnel about chromosome 18 abnormalities, their effects on young children and their learning issues. She shared information about the work and research of The Registry.

Liz Grossman of FL:

- In January in Ridgwood, Liz opened a dance school for children with differing needs, both typically developing and those with special needs. Activities included performances, publicity and a trip to a NYCB performance.

Jennifer Kaiser of NM:

- Jennifer (18p-) was the speaker at her graduation from the ACCESS program. She described her problems associated with 18p– and how the Access prlgram helped her with the skills she needed.
- Jennifer attends a weekly adult dinner group where she, her friends and their parents meet separately to both discuss issues and enjoy being together.

Norma Lennon of NC:

- Norma spoke to a group of daycare and homecare providers at Smart Start in Kenansville on how to better provide services for children with special needs, especially those with 18p-.

Monica McDivitt of TX:

- In April, Monica set up and manned a table at a Houston Aeros game, where she provided information and literature concerning chromosome 18 abnormalities and the services provided by The Registry.
- In October in Katy, Monica held an in-service meeting for therapists and teachers concerning chromosome 18q deletion and its effects on her childSamantha.

David Martin of St. Albert, Alberta, Canada:

- David, brother of Samantha Martin (Tetrasomy 18p) received 3rd place in his school's science fair for a project on Tetrasomy 18p. His goal was to create interest in and explain chromosome 18 abnormalities, especially Tetrasomy 18p so that more people would understand and appreciate how this condition affected his sister, Samantha.

Martha Sakre of LA:

- In January, Martha met in Shreveport with the staff of LSU Health Center Rehab Clinic and spoke about the services of the Registry and the activities of the Chromosome 18 Clinical Research Center.
- In July in Shreveport, Martha met with and made a presentation to a financial planner and an attorney concerning the establishment of special needs trusts for children and adults with chromosome 18 abnormalities.

Stephanie Shively of IL:

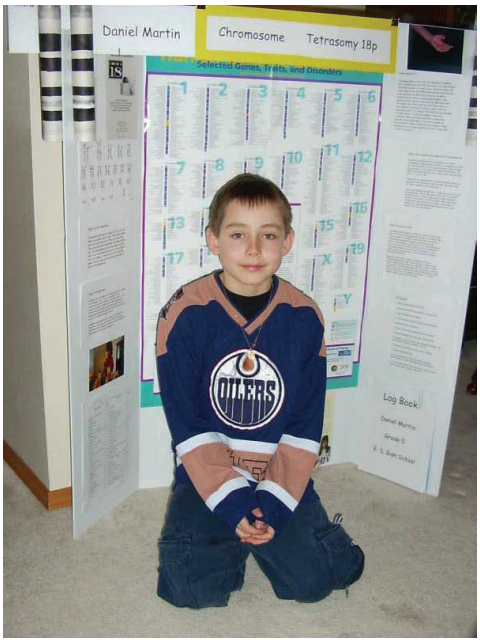
- In June at Gifford, Stephanie organized a display of Chromsome 18 materials and brochures at Gifford State Bank and spoke to interested persons concerning chromosome abnormalities, especially Tetrasomy 18p.
- June 2005 – Paxton - Organized a display of Chromsome 18 materials

and brochures at Paxton Carnegie Library and spoke to interested persons concerning chromosome abnormalities, especially Tetrasomy 18p.

- June 2005, Bayles Lake - organized a display of Chromosome 18 materials and brochures at Bayles Lake Ladies Auxillary.
- July 2005 – Paxton - Organized a display of Chromosome 18 materials and brochures at Paxton 4th of July Festival.
- July 2005 – Gibson City organized a display of Chromosome 18 materials and brochures at Bank of Gibson City.
- July 2005 – Buckley - Organized a display of Chromosome 18 materials and brochures at Buckley State Bank.
- July 2005 – Peoria – made power point presentation to about 35 members of the Adult Weslyean Class, First United Methodist Church concerning chromosome 18 abnormalities, current research and organizational goals.
- August 2005 – Loda - Organized a display of Chromosome 18 materials and brochures at Federated Bank of Loda..
- August 2005 – Cissna Park Organized a display of Chromosome 18 materials and brochures at Prairieland Quilts meeting.
- August 2005 – Cissna Park Organized a display of Chromosome 18 materials and brochures at Old Settler's Festival and spoke to interested persons concerning chromosome abnormalities, especially Tetrasomy 18p.

Kristi Street of TN:

- In August, Kristi presented to endocrinology, pediatric and pediatric orthopedic residents at the local hospital in Hampton background information about The Chromosome 18 Registry and chromosome 18 abnormalities.



Daniel Martin's 5th grade science project. Daniel is the brother of Samantha who has Tetrasomy 18p.

Member Summary

Identified Families:

18q-	520
18q– mosaic	24
18q– interstitial	20
18p-	265
Ring 18	123
Ring 18 mosaic	10
Tetrasomy 18p (includes partial)	134
Tetrasomy 18p mosaic	8
Trisomy 18	396
Trisomy 18 (mosaic)	24
Trisomy 18q (includes mosaic)	30
Trisomy 18p (includes mosaic)	12
Other	193

Our identified families live in:

United States	1369
Europe	181
Canada	91
Australasia	67
South America	7
Central America/Caribbean	4
Africa	7
Asia	10
Mexico	7
Middle East	6
Oceania	1

"We must be the change we seek to create."

~ Gandhi

Research Update

The Chromosome 18 Clinical Research Center

Department of Pediatrics
University of Texas Health Science Center at San Antonio

Jannine D. Cody, Ph.D. and Daniel E. Hale, M.D.

The important thing in science is not so much to obtain new facts as to discover new ways of thinking about them.

- Sir William Bragg

Mission

To provide families affected by chromosome 18 abnormalities with comprehensive medical and educational information with a focus on treatment options.

Goals

- To be the international medical and educational resource for the families of individuals with chromosome 18 abnormalities.
- To perform and facilitate both clinical and basic research relating to the syndromes of chromosome 18.
- To devise treatments to help these individuals overcome the effects of their chromosome abnormality.

Approach

1. Understand the unique characteristics (phenotype)
   Employ the most sophisticated medical technologies and assessment tools to define as precisely as possible the medical and educational characteristics. (e.g., instead of global IQ assessment, evaluate more detailed subsets of features such as executive function, spatial reasoning and temporal processing need to be assessed). This alone may lead to treatment options. For example, knowing that the short stature is caused by growth hormone deficiency leads to an immediate treatment option.
2. Characterize the genetic differences (genotype)
   Define exactly which genes are present in something other than 2 copies in each individual with a chromosome 18 abnormality. This will utilize the latest information on the genome and will employ cutting-edge molecular genetic and molecular cytogenetic techniques.
3. Correlate characteristics with the genetics
   By identifying which genes are associated with which components of the phenotype, a better understanding of the underlying biology will be gained. This can then be used to inform the search for the most effect treatments. This process is facilitated by a secure custom-designed database for the storage and retrieval of the study participant's medical records as well as study-generated phenotype and genotype information.

Significance

- This Center will be the prototype for the development of other centers across the country (and indeed the world) focused on other chromosome abnormalities.
- Since chromosomal abnormalities are responsible for half of the cases of mental retardation, these Centers will be key in ameliorating the personal, social and economic impact of these conditions.

The Chromosome 18 Clinical Research Center and the families of the Chromosome 18 Registry have a unique and productive partnership. The Center exists to find answers for the affected families. The families provide the vision, direction and a substantial part of the funding so that the Center can do the work of finding answers. Sometimes we all get so focused on the questions for which we do not have answers that we forget how far we have come and the things we have learned. So it is important to take a moment and reflect on how far we have come and what we have learned.

When we began this work, it was not known that children with 18q- and 18p- could have growth hormone deficiency. We knew they were all short,

but we were the first to report the cause. The cause is treatable.

We have also shown that those children with 18q- with growth hormone deficiency have an average nonverbal IQ increase of 18 points after they start growth hormone therapy. Some children have no increase and some children have twice that increase. We are trying to understand why this is.

We have shown that the amount and location of the missing part of chromosome 18 is different for each child with 18q-, so it is no wonder that they are each unique in their strengths and weaknesses. Interestingly, the majority of people with 18p- have the same breakpoint so they should be more alike as a group making it easier to make group generalizations.

We have shown that many of the people with 18q- develop hypothyroidism, therefore they should all have their thyroid levels checked every year.

We found that only 20% of people with 18p- and 68% of those with 18q- have mental retardation. This is not even close to the 100% that was reported in the literature before we began, or the expectation that we were given at the time of diagnosis.

We found that almost all the adults with 18q- have depression or bipolar disorder and respond well to treatment once appropriately diagnosed.

We have shown that hearing impairment is a much more prevalent problem than was in the literature. 70% of children with 18q- and 55% of children with 18p- have some level of hearing impartment. It is therefore important that parents have their child's hearing assessed.

We have shown that 100% of the people with a deletion of a certain region of 18q have dysmyelination of the brain. We have not yet determined the consequence of this finding in terms of the functional significance.

We are just beginning to evaluate individuals with tetrasomy 18p with the expectation that we will gain new insight that will lead to treatment plans.

We continue to have more questions than we have time, money and staff to answer, but we are making steady progress and are making a positive difference in people's lives. However, we have a long way to go and many more questions to address. This is reflected in our mission and goals as spelled out below.

The mission of the Chromosome 18 Clinical Research Center is to:

To provide families affected by chromosome 18 abnormalities with comprehensive medical and educational information with a focus on treatment options.

The goals are:

To be the international medical and educational resource for the families of individuals with chromosome 18 abnormalities.

To perform and facilitate both clinical and basic research relating to the syndromes of chromosome 18.

To devise treatments to help these individuals overcome the effects of their chromosome abnormality.

The approach is:

Understand the physical and functional characteristics to inform treatment decisions

We employ the most sophisticated medical technologies and assessment tools to define as precisely as possible the components of their characteristics. This alone may lead to treatment options. For example, knowing that the short stature is caused by growth hormone deficiency lead to an immediate treatment option.

Characterize the precise genetic abnormality at the molecular level

We define exactly which genes are present in a number other than the normal 2 copies in each individual with a chromosome 18 abnormality. This will utilize the latest information on the genome and will employ cutting-edge molecular genetic and molecular cytogenetic techniques. The implications of this are additional treatment insights resulting from an understanding of the basic biology of the involved genes.

Correlate the two sets of findings listed above

By identifying which genes are associated with which components of each individual's characteristics a better understanding of the underlying biology will be gained. This can then be used to inform the search for the most effect treatments. This process is facilitated by a secure custom-designed database for the storage and retrieval of the study participant's medical records as well as study-generated information from the activities listed above.

The continued success of the Chromosome 18 Clinical Research Center is possible because of the dedication of the families as an active part of this partnership. Thank you all for helping to make and keep this partnership strong.

See our website at :
www.pediatrics.uthscsa.edu/chromosome18/

Fundraising Activities

We are on the web!
www.chromosome18.org

Registry-wide Fundraisers

The Registry has, at this point in time, four separate fundraisers each year that are designed to include the entire membership. The rational behind this is that, in order to press forward with research to better the lives of our children, we must have a system in place that will consistently raise the funds to aid in that research. These events are meant to encompass all member families and extended families. Ideally, each and every member family should participate in one or more of these events which are varied enough that members can choose the event that best fits their style. These four events are:

- Phantom Tea
- Birdies for Charity
- San Antonio Golf Tournament & Silent Auction
- Run for Research

San Antonio Golf Tournament

The Fifth Annual San Antonio Golf Tournament took place at Tapatio Springs Golf Resort on June 18, 2005. It was billed as "Come Play 18 for Chromosome 18 on June 18th." This was another great success, with the event clearing **$26,433.69**. Our wonderful corporate sponsors and faithful individual players were back again, with the only goal being to benefit our children.

2004-2005 Phantom Tea

The annual Phantom Tea campaign always crosses over two calendar years, starting in October and ending the following summer to make it convenient for our families to have their Tea at almost any time of the year. The tenth annual event that ended in 2005 was very similar to those of the last few years. Our families and their relative and friends donated **$136,043.33**. There were 689 donors who gave in the names of 58 of our children and adults. This total represents 13.9% of the families who were sent invitations.

Birdies for Charity

The **Birdies for Charity** is an event held in conjunction with the PGA Tour Pro Tournament in San Antonio that raises substantial dollars for charities in the San Antonio area. Preparations begin in July and end with the tournament in October. 2005 was the sixth year that the Registry participated, and it brought in **$8,522.41** in pledges, straight donations and contest winnings. It should be pointed out that Golf San Antonio who runs this event does not get the final total and donor list to us until February of the following year, so most of the dollars and donors are actually recorded in the following year's financial annual report.

Run for Research

Three amazing Registry families took part in the 2005 **Run for Research,** the same faithful families who have been doing it for years. On October 1st, the Shively family from Buckley, Illinois, hosted their fourth Run and they (including Aunt Karen Lanning) and their many friends walked, ran, biked, etc. to bring in **$25,419.00** in honor of Sara who has Tetrasomy 18p. If that was not enough, they set up a golf scramble on September 24th, presumably to get ready for the Run a couple weeks later!

In the community of Katy, Texas, the McDivitt family hosted their third **Run for Research** on March 5th. John and Monica McDivitt, along with their supportive family and friends, walked and ran their way to **$8,603.14** in honor of their daughter, Samantha, who has 18q-.

Pamela Del Angel, who lives in Houston, organized her 2nd **Run** in the Valley where most of her family lives. The date was March 26th and the place was Weslaco, Texas. Pamela's efforts and that of her many family members and friends resulted in raising **$5,389.00**.

All three of these families spare no comfort, time or effort to bring about such amazing success which they do, of course, to benefit the research that will help chromosome 18 children.

Individual Fundraisers

In addition to our four Registry-wide fundraising events, individual families are invited to take on the responsibility of creating their own fundraiser. The Chromosome 18 Registry & Research Society has been blessed to have some families who are not only energetic, but very creative. They enlisted the help of their families, friends and communities to raise awareness of chromosome 18 syndromes abnormalities and raise money to fund the research that is changing and improving the lives of their children.

During 2004, there were thirteen individual fundraising events hosted by the family and friends of our affected children and adults. We wish to acknowledge those of you who went out of your comfort zone and helped raise research funds your children.

Doug and Julie Masterson (Jake, 18q-) from Pacific Palisades, California, were on the golf course for the 6th time to host what has become a much-anticipated golf tournament/silent auction attended by friends and colleagues. The event took place on October 11th. Their hard work always results in a very successful event that continues to be a very important spoke in the fundraising wheel each year. We are so grateful, Doug and Julie, and we thank you for your loyal and important participation.

Stephen and Ellen Botello from West Barnstable, Massachusetts, (twins Cameron and Joshua, 18q-) continue their amazing success by hosting their fifth golf tournament. We thank

you sincerely for all your efforts which seem to always be carried off without a hitch. Special thanks to Jean D'Olimpio whose skills and hard work help make this event so successful. Thank you, Stephen, Ellen and Jean.

Mike Reilly from Columbia, Missouri, is a friend of **Dan and Katie Schilly** (Remy, 18q-), and he hosted his fifth golf tournament to benefit The Registry. Here is an example where not just family, but friends of our children, host fundraisers to help in the research efforts. Thank you, Mike Reilly.

The **McDivitt** family from Houston (Samantha, 18q-) not only did their usual Run for Research, John and Monica also did some advertising of the Registry at a Houston Aeros game and also raised some funds.

Cheryl Frayne is the aunt of Talia Grossman (18q-). Cheryl and her family live in Canada. Cheryl, her family and friends hosted a second "Fall Get-Together" and it brought out dozens of interested family and friends. Thank you so much for your help, Cheryl and family.

The **Patterson Family** (Liam, 18p-) again showed movies and served the usual movie fare food after educating their friends who attended about chromosome 18 abnormalities. Thanks, Molly, Mike, Liam and Delany, for continuing your unique fundraiser!

Bruce Steinke (Dylan has 18q-) keeps a candy machine full and makes it hard for his coworkers to stay trim. The vending machine in his office at *The New Republic* brings a steady income for the Registry. Thank you, Bruce and Dina.

Jill and Rodney Lee (Owen, Tetrasomy 18p) hosted *three events* in their town of Boonville in north central New York: a bake sale, a poker walk and a golf tournament. This small community has so much heart and expends so much effort to help the Lee family raise money for the Registry and for Owen. Thanks so very much for all you do.

Eric and Allison Alper (Sydney, 18q-) of Dix Hills, New York, with the help of many family and friends, hosted their third Comedy Night/Raffle at a local hotel. They again raised tens of thousands of dollars for research. Of course this kind of event does not happen without a great deal of work, but Allison and gang are not afraid of work! The event was another truly astounding success story. Thank you to the entire Alper family for continuing to host this event that brings so many laughs to so many people and so many research dollars to our affected kids.

Jarvis and Renee Watson (Jalon, 18q-) live in a tiny east Texas town called DePort. The family and friends of the Watsons gathered at their church for a night of music. They opened their hearts and wallets to benefit the research the will help Jalon and many other chromosome 18 children. As new members, we thank you for jumping right in to help!

Ivan and Xenia Cabrera hosted a birthday party for their son, Nicholas**,** (Partial T18/Y translocation) in Miami Lakes, FL. Guests enjoyed the party and gave generously to help the work of the Registry. Thank you to the Cabrera family. There are no limits to the ideas that our families come up with!

Greg and Sue Dorsey (Carley Rose, 18p-) of Evergreen, CO, hosted a wine-tasting party to raise funds that specially went to offset the cost of the 2005 conference held in Winter Park, CO. There seems to be no question as to why this was such a popular event! Thank you so much for helping in yet another way.

Brett and Nichola Chappell (Luke, 18p-/15p translocation) held another of several golf tournaments in the area of Camden, Australia. These funds were used to help pay the costs of the 2005 Australian conference which was attended by both Drs. Cody and Hale. Thank you for all your wonderful giving and hard work over the years, Brett and Nichola.

The total of these individual fundraiser was in excess of $118,000!

Other Areas of Giving

In addition to fundraising events, there are numerous other ways that people share with the Registry Family. They include the following:
Combined Federal Campaign
Memorials
Matching Gifts
Donation with dues payment
Donors Choice
Research pledges
Charitable endowments
Charitable awards
Special events (birthdays, anniversaries and special occasions)
Workplace Giving
In-kind donations

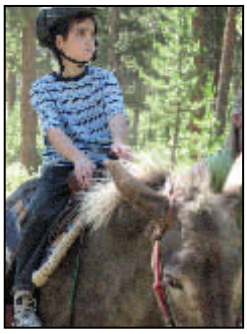
Memorials

During 2005, contributions were made in memory of the following persons:
Sarah Blanton
Ryan Bowden
Roy E. Cash, Sr.
Betty Eslick
Roland Fowler
Newton Goldberg
Shirley Goodbaum
Trinity Heard
Albert Hersh
Calum Lawrie
Cary Latham
Bert Odem
Mildred Patrick
Nancy Sarzynski
Hanna Shaner
Lloyd Wilson

Pledges

Several families or family foundations have made pledges to provide financial help for research efforts that are given over a period of years. These pledges continued to be honored during 2005. We are very grateful for their generosity.

Our sincere thanks go out to the generous and thoughtful people who gave in one of these avenues. All donors are acknowledged in the Annual Report except the Combined Federal Campaign. These donors cannot be named unless specific permission is given by them to the Registry office.



11th Annual Family Conference
YMCA of the Rockies
Winter Park, CO
June 27– 30, 2005

The glorious Colorado sunshine and breath-taking views set the pace for the 2005 Chromosome 18 Conference. The 2005 conference was about leaving the hustle and bustle of city life. Attendees got to focus their attention on relaxation, slow afternoons, and Rocky Mountain adventure at Snow Mountain Ranch/YMCA. We started things out in Colorado style with a big bonfire, smores and hot chocolate the first night.

Activities abound at Snow Mountain Ranch. There was hiking, fishing, mini-golf, rock climbing, roller skating, swimming, zip lines, horses, white water rafting, and so much more! A favorite activity for the children was the craft center. This center was amazing and the kids had several opportunities to make different items. Therapeutic Horseback riding was a popular choice for many of the Chromosome 18 children and the smiles were invaluable. Families enjoyed hayrides one night and then on another we had a unique Wine & Cheese social. The wine social was a mini-fundraiser and featured a special Colorado winery. It was an evening for kids as well, with balloons and desserts. We ended the evening with a special slide show of pictures taken throughout the week by Rick Guidotti.

This year there was a twist on the silent auction. Instead of having several single items to bid on we made regional baskets made up of several great items from specific areas. We really need to thank all of the Chromosome 18 Regional Coordinators for making this a success. They were asked to designate some sort of theme to their regional area and than to contact families and ask that they go out and either ask for a donation or purchase something for their area's basket. Families throughout the United States and even Holland and Australia participated in this event. The Regional coordinators were responsible for getting all of the items from

their area to Colorado. This was a huge undertaking. The baskets were beautiful and raised $1,250.00 for The Registry. Thank you, Regional coordinators, and to all of the families who participated in this event. There were many that helped make this happen that were not even at the conference.

The speakers provided us with a lot of great information. We talked about: inclusion, osteopathy and nutritional intervention options, neurodevelopment, learning environments for our kids and testing and, as always, we had a Chromosome 18 research update. We also had a mini workshop on building a joyful life with special circumstances, our kids! One particular session that was a favorite for many was "Nourishing the Special Needs Family". Our guest speakers were Dr. Brad Beck and his wife, Lisa. They shared their story and spoke to our hearts with the memory of their special daughter, Maddie.

2005 Conference Stats

Attendees included:
211 individuals
66 families
89 children, 36 of them affected
13 affected adults

They came from 22 states and the countries of Canada, England and Finland.

The 2005 Chromosome 18 Conference came to a close with a Barbeque and Square Dance. The barbeque had good food, a park for the kids to play in and many friends saying their goodbyes until next time. The square dance ended the evening and many discovered there was a country/western side to them that was just waiting to come out!

I am exhausted all over again just going through all that we did in those four short days. This conference could not have happened without the help of many. While Robin Coursen and Angie Moss (2005 conference coordinators) started running with dates, details, and planning, there were those who simply took the batons from the coordinators and did not finish until the end. Thank you so much to the whole Colorado planning team. This team was invaluable and worked so hard. I would like to extend a special thank you to Bob Coursen, Hannah Coursen, Jeff & Chris DeVoss, Greg & Sue Dorsey, Robert Moss and Susan Wright. The 2005 conference would simply not have been possible without these great people.

Photographs by Rick Guidotti

positiveexposure.org

One Family's Story

By Caryn Chung

This is one family's story, our family's story. And it isn't actually the whole story. Let me begin by introducing us. There's James and myself, Jacob (8), Jonah (5) and Eli (2). The reason for this story is our middle son, Jonah.

Jonah was born in March of 2001. He, by all accounts, was a healthy, normal baby. Very quickly things didn't seem to add up. With having a two and a half year-old brother in the house, one would think the baby would startle. Because I was an experienced nursing mother, I shouldn't have struggled with feedings. Then we had the diapers with some blood, followed by strange eye movements. By three months old, we knew Jonah had allergies and a hearing issue. Vision was questionable. Regional and county services started to kick in. A pediatric neurologist gave us a blanket cerebral palsy diagnosis after the high resolution chromosome test erroneously came back as normal.

Weeks stretched into months with no answers. Milestones went unmet. Then there was a bout with pneumonia at nine months. The doctors at the hospital seemed to detect something our pediatrician didn't, and they suggested a trip to the geneticist.

Chromosome 18 deletion. There. We had identified the anomaly. And, because the symptoms had surfaced so slowly, we were never hit in the face with the diagnosis. It stung, nonetheless. The reality of it still stings.

We found the Registry through an internet search. We found a family. You've heard the saying, "You can pick your friends, but you can't pick your family." Same goes true for having a child with a chromosome abnormality. We didn't pick it. But we were "born into" the Chromosome 18 family, a family which we will forever be a part of.

This family, similar to ours, gave direction and support. They didn't allow for helplessness or hopelessness. Oh, if all families were like that. We got to meet our new family, just a few months after the diagnosis, in Niagara Falls. Joining this group, we were encouraged and therefore realized we, in turn, had the opportunity to be an encouragement.

I'm saddened when I think about how some families view "less than perfect" prenatal tests as death sentences. Yes, some days are almost unbearable, a struggle just to stay sane. But though a struggle, we should give credit where credit is due. Helen Keller impacted me when she said, "Only through experience of trials and suffering can the soul be strengthened, ambition inspired, and success achieved." To me, the annual conference best displays the strengthened souls, inspired ambitions and successes. The emotions felt as a person sits and listens to the panel of affected young adults telling of their struggles and the successes. Wow! The pain is so deep, but the tears of joy are so much sweeter than what I believe a "normal" family encounters.

Such displays of courage and success help me to continually strive to uncover Jonah's great abilities. Please don't misunderstand me, Jonah is severely affected. He's autistic and non-verbal living without much hearing. Some missing, some mangled, he isn't working with a full set of tools. But neither was Helen Keller. Let me tell you just one thing that I stand amazed at. Jonah can play a computer game equipped with a touch screen, holding the left speaker up to his ear while he successfully, 100% of the time, chooses which instrument is being used to play a few notes. I didn't even know what a bassoon was.

Our family's story can't be fully told because it's not finished. When it is all said and done, I am most certain that our souls will be strengthened, our ambition inspired, and through our own measurement, success achieved. And maybe, just maybe, we will have encouraged another family that life can be lived to the fullest by a non-traditional set of values. Jonah, born with a chromosome 18 anomaly, can be that family's inspiration. And for our family, to impact another for good is worth the credit of a life well-lived.

The McDivitt's Fun Run for Research in Katy, TX, honors Samantha (18q-) March 5, 2005

The Shively Family's Annual Bike Ride/Run near Buckley, Illinois, honors Sara (Tetrasomy 18p) October 1, 2005

Left: Tucker Lee, brother of Owen Lee (Tetrasomy 18p) at the Bake Sale Benefit in Booneville, NY. Beside Tucker is his friend, Kyrn. Right: Owen, Rodney, Tucker and Jill Lee.

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-, Ring 18 and Tetrasomy 18p) are maintained by parent volunteers. A relative listserve has also been added, and it is moderated by Julia Wentz, grandmother of Bryan Yocum who has 18q-.

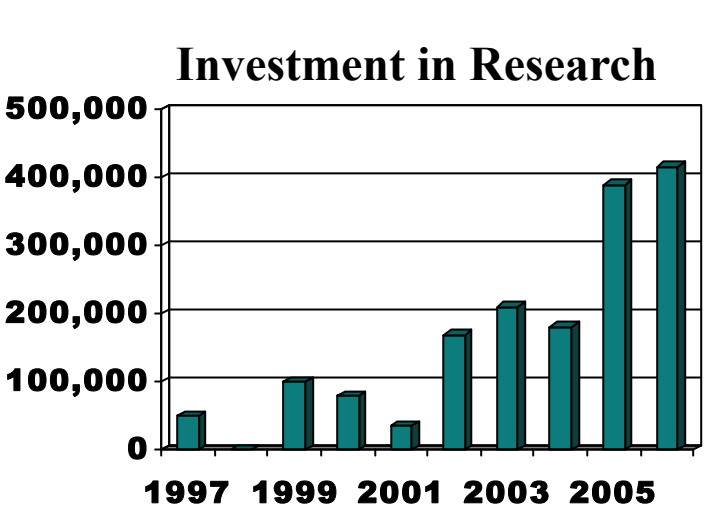
We believe there are no incurable conditions, only those that have not yet been understood. Our goal is to make sure that the syndromes of chromosome 18 are understood. This understanding will bring treatments and therapies that will lessen the burden of these syndromes. The path to understanding the 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.

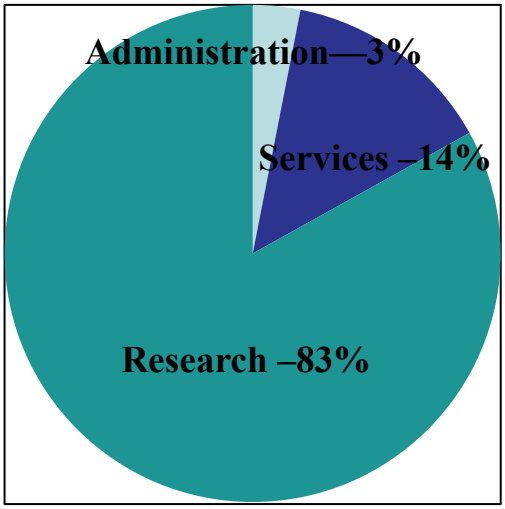
Financial Statement

for the year ending December 31, 2005

Revenue and Support	
Membership Dues	10,084
Interest Income	(31,297)
Conference Income	21,271
Special Events	123,785
Merchandise Sales	4,321
Fundraising	216,108
Combined Federal Campaign	23,731
Contributions	38,298
TOTAL	$406,301
Expenses	
Program Services	486,321
Management and General	10,142
Fundraising	2,620
TOTAL	$499,083
Net Assets, End of Year	**$ 572,727**

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





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We Are All Family

By Elvie Echols

Please know that it does not matter which child is having the problem at the time, one is just as serious as the other. We are all in this together. Sometimes I feel like NEMO, and we all are swimming in this big ocean, but we have different little fish, and we are all trying to figure out what is the best area to stay in. Smile

We are all family. I do not feel one other human being in the whole world knows what I go through, emotionally, socially, educationally, with love but the familes on here, we encourage each other, we cry together, and perhaps each of us have different things we go through, we are also somewhat the same as we all have Chromosome 18, in one form are the other. We that have younger kids, look up and admire the ones that have gone before us. The little ones that are behind us, we can also support, because we to now, have gone through some other doors. Some of us are more medically fragile, while other have behavior issues, but yet we are all family. If we met someone with 18 then would be an instant bond because we know.

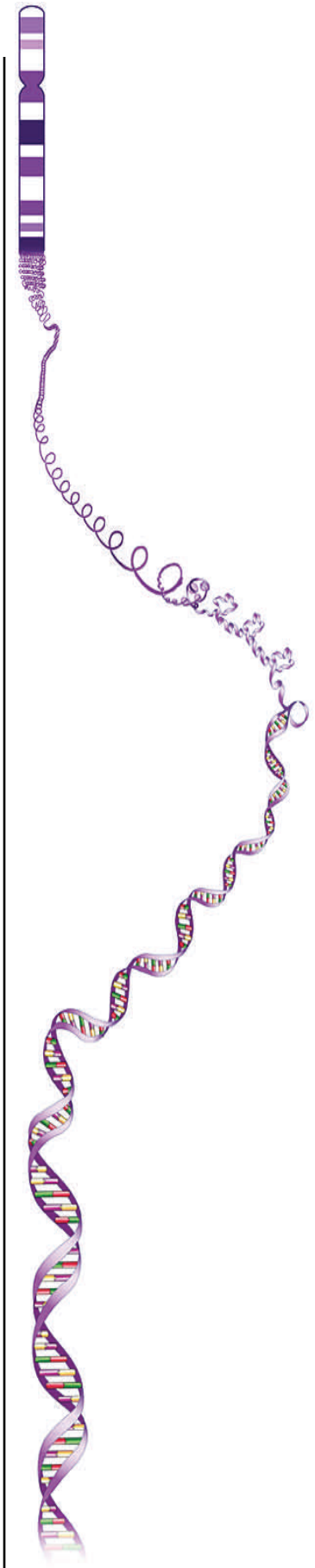
When I am at Children's Hospital, there are other heart families, that know us very well. If I am in the Cafeteria they just instantly walk up to me and say hello. Often they have seen me in there alone, and without asking were EJ was, just immediately walked up to me and hugged me, and they say we are in surgery also. With just that we know.

I said all of this to say, we all know and learn from each other. My God I am so blessed, to be able to have family all over the USA, and in other countries also.

I always dreamed of having friends, that I could go and say hello to all over, and look what God did for me, he surely answered my prayers.

With Love and God's Blessing,

Elvie and EJ



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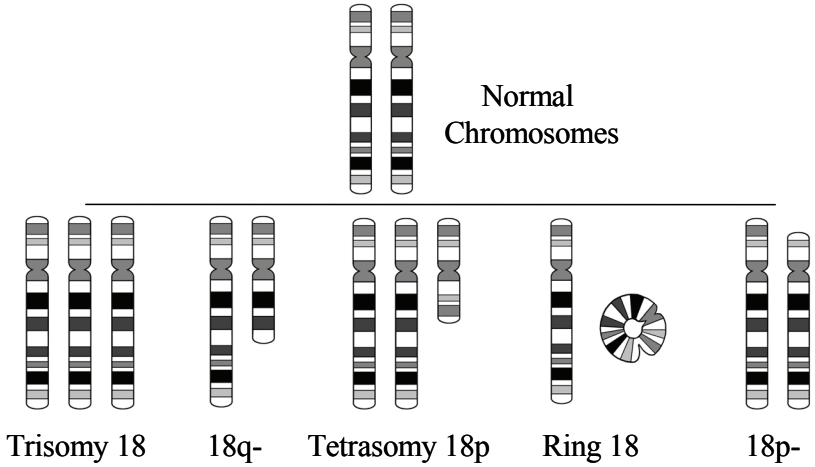
The Syndromes of Chromosome 18



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