

Tuberous Sclerosis Alliance

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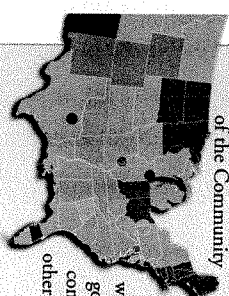
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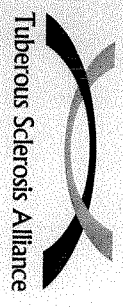
Andover, MN 55304

EMPOWER... (continued)

Community Alliances - Organized groups of volunteers offer support and increase community awareness of TSC. Raising revenue to ensure the mission and programs of the TS Alliance are implemented is an important goal of the Community Alliances Program.



Fund-raising events include walk-a-thons, golf tournaments, comedy nights and other special events.



Tuberosclerosis Alliance

801 Roeder Road, Suite 750
Silver Spring, MD 20910
(800) 225-6872 / (301) 562-9890

Fax (301) 562-9870
www.tsalliance.org
info@tsalliance.org

WE ARE...

The Tuberosclerosis Alliance, the only national nonprofit organization dedicated to finding a cure for tuberous sclerosis complex (TSC) while improving the lives of those affected. Tuberosclerosis complex is a multi-system genetic disorder that can cause tumors to grow on most organ systems. TSC knows no, racial, cultural or economic barriers.



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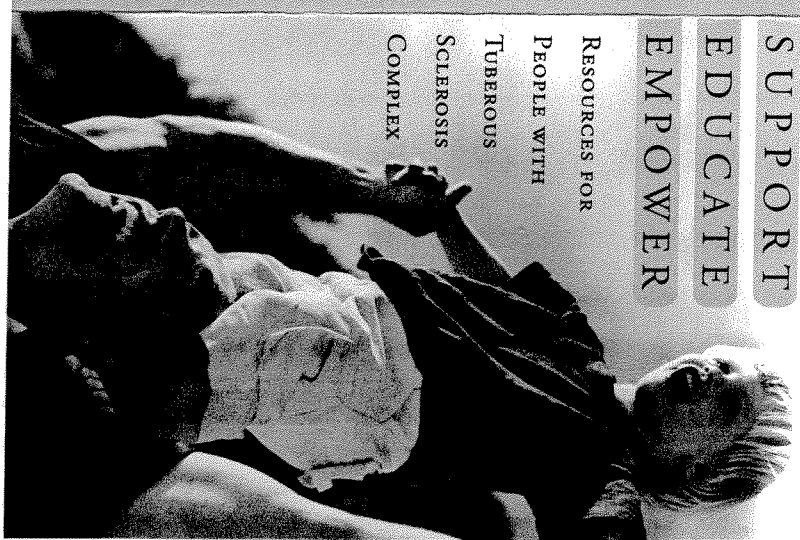
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Tuberosclerosis Alliance

SUPPORT
EDUCATE
EMPOWER

RESOURCES FOR
PEOPLE WITH
TUBEROUS
SCLEROSIS
COMPLEX



The Tuberosus Sclerosis Alliance (TS Alliance) offers a variety of resources to support, educate and empower people with tuberous sclerosis complex (TSC), their families and caregivers.

SUPPORT...

Counseling, intervention and referral services to appropriate community services are provided via telephone by a full-time, licensed, clinical social worker. Call us at (800) 225-6872 for professional assistance in this area or to connect with other families.

Connect immediately with others through online discussion groups (list serves) on the TS Alliance Web site. These groups connect users with others via e-mail who may be experiencing similar challenges.

Track doctors appointments and keep TSC related medical information in one place in the **Tuberous Sclerosis Journal**—An Organizational Diary for the Care and Treatment of TSC.

Living with Tuberous Sclerosis— A book of stories and inspirational writings by adults with TSC, and parents of children with TSC.

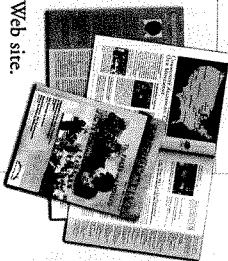


EDUCATE...

TS Alliance Web Site - Descriptions of current programs and initiatives, including updates on legislative developments and TSC research advances can be found on the Web site at: www.tsalliance.org. The Web site continues to evolve as new content is added regularly. Newsletters, fact sheets and other resources can be downloaded from the site. Individuals interested in learning more about participating in outreach and awareness activities or making a financial contribution can also do so online.

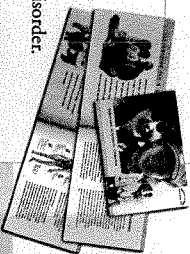


Perspective - News about the TS Alliance, research and legislative updates and profiles of families living with TSC are provided in this quarterly newsletter. The newsletter can also be accessed on the TS Alliance Web site.



TSC Resource Exchange - This electronic newsletter provides brief overviews of various disability-related topics such as community integration, transition and other topics with links to additional information.

An Introduction to TSC - Offers basic information about TSC including how it is diagnosed and the clinical features and genetics of this disorder.



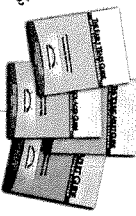
My Child Has TSC (Spanish version) - Provides an overview of TSC and answers some of the most commonly asked questions about the disorder.

Videos - Presentations from the 2001 TS Alliance National Family Conference are available on video. Please visit: www.tsalliance.org/activities/TapeOrderWeb.pdf for a complete list of titles.

Journal Articles - Articles from the *Journal of Child Neurology* and NIH Consensus Conference that address clinical diagnostic criteria and evaluations.



Life Segues Program - Detailed information about many of the non-medical challenges experienced by people with disabilities. Examples of topics covered include: early intervention services, special education, legal rights, assistive technology, housing, employment, transition, end of life issues and financial planning.



Fact Sheet Series - Fact sheets are available on a variety of TSC medical and related topics. Refer to the Resource Order Form or the TS Alliance Web site for a complete list of fact sheet titles.



EMPOWER...

TS Alliance Support Network - A national support network of trained individuals provide experience based support. Volunteers are available to talk with those seeking local resources, or experience-based information about living with TSC. Volunteers also organize educational meetings, family events and support groups at the local level.



Matching Program - Matches individuals and/or families across the country who have lived through or who are living with similar challenges and provides experience-based information and peer support. Utilizing information collected from participating volunteers, a match is made when an individual needs a specific type of information or assistance.

Government Relations Program - The goal of this important program is to increase federal research funding and awareness of TSC. For more information about how you can become involved, visit our Web site.

OVER PLEASE

Tuberous Sclerosis Alliance



at a glance

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Tuberous Sclerosis Alliance

Tuberous sclerosis complex

What is Tuberous Sclerosis Complex?

Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumor growth in vital organs such as the brain, heart, kidney, lungs and skin. It is often first diagnosed because of seizures, the appearance of white spots on the skin, or because a child is not reaching developmental milestones. Although the tumors are rarely malignant, the tumor can disrupt the proper function of an organ. The lesions on the brain may cause seizures, varying degrees of developmental delay or behavioral challenges, including attention deficit hyperactivity disorder (ADHD), autism and sometimes mental retardation. Although some individuals may have their seizures controlled through medication and/or surgery, diets or by the aid of a medical device(s), some are unfortunate and do not respond to any method of treatment.

Who Gets TSC?

There are more than 1 million people worldwide with tuberous sclerosis complex and about 50,000 cases in the United States, with two additional affected children being born each day. Because so many cases go unrecognized, undiagnosed or misdiagnosed, these numbers may be higher.

Children have a 50 percent chance of inheriting the disorder if one of their parents has the condition, but at this point, only one-third of TSC cases are known to be inherited. The other two-thirds are believed to be a result of a spontaneous mutation. The cause of these mutations is still a mystery.

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Research

The Tuberous Sclerosis Alliance (TS Alliance) has been funding research of tuberous sclerosis complex (TSC) since 1984. The Center Without Walls (CWW) research initiative started 14 years later and is now an international consortium composed of researchers from the United States, United Kingdom and The Netherlands.

Collaboration in Research

CWW was created to enable researchers to share information and resources, thus expediting research results and their translation into improved clinical care of individuals with tuberous sclerosis complex. With more than \$1 million in grant awards provided annually, CWW research has focused on the development of the diagnostic genetic test for TSC, the development of model systems to study TSC, understanding the mechanism(s) of tumor growth in individuals with TSC, and the function(s) and interaction(s) of the two TSC genes—the TSC1 gene on chromosome 9 and the TSC2 gene on chromosome 16. Understanding the function of the tuberous sclerosis genes will lead to better diagnostic tools, treatments and an improved quality of life for all individuals with TSC.

Importance of the TSC Model

Tuberous sclerosis complex offers an excellent model for the ongoing search for understanding of the mechanisms of cancer and tumor development, as well as for the development of new treatments and therapies.

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Development

The Tuberous Sclerosis Alliance (TS Alliance) depends on the financial support of individuals, corporations and foundations to provide support services and programs to those affected by tuberous sclerosis complex (TSC). There are many giving opportunities available to an individual or corporation that is interested in supporting the TS Alliance. The following giving opportunities are available:

Membership

Members provide unity, presence and a voice as the organization solicits government and private funding for the development of programs for those living with TSC. As a membership organization, the TS Alliance depends on the continued financial support of individuals who are committed to the organization's cause. Additionally, members serve in a volunteer capacity by hosting support groups, organizing special events and delivering the organization's message to heighten awareness of the disorder in their community.

Community Fund-Raising Events

Volunteers take action and raise money for the organization by becoming involved in national fund-raising efforts or by organizing a special event in their community. Volunteer special events generate more than \$1 million annually, which represents 40 percent of the overall revenue that the organization raises. For this reason, the TS Alliance continually recruits and provides resources to those individuals who support the TS Alliance through these events in their communities across the country.

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How is TSC Diagnosed?

Currently, diagnosis of tuberous sclerosis complex is based on clinical findings. Diagnostic testing includes a brain MRI or CT Scan, renal ultrasound, echocardiogram, eye exam and a Wood's lamp evaluation of the skin. Molecular genetic testing is available for families affected by TSC on a research basis through clinically certified laboratories in the U.S. A commercially available diagnostic genetic test is expected in 2002.

The medical community is challenged in diagnosing TSC due to the many manifestations of the disorder. Because TSC is expressed so differently in each individual, there is no specific course of treatment available for all patients with TSC.

Is There a Cure for TSC?

There is no known cure for the many symptoms of TSC, but the two genes that are known to cause the disorder (TSC1 and TSC2) have been identified. Understanding the function of these genes will lead to better diagnostic tools, treatments and an improved quality of life for all individuals with TSC.

Early intervention helps to diminish developmental delays, while surgery to remove tumors helps to preserve the function of affected organs. Improved technology is helping to pinpoint the exact portions of the brain stimulating seizures, and is creating new therapies to help control seizures.



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There are extensive resources available through the TS Alliance Center Without Walls that are in use by this specialized research community. These resources provide a strong support for the development of new treatments and therapies to improve the lives of those affected.

Awards and Grants

In addition to the fellowship, junior and senior investigator awards available through the CWW, in 2001 the TS Alliance announced the Rothberg Award for Courage in Research. This \$100,000 award was established to recognize outstanding scientific research that advances the knowledge toward finding a cure for the many aspects of TSC. Up to four Rothberg Courage Awards may be presented annually. These awards are a complement to the TS Alliance's research grant awards program. The Rothberg Award for Courage in Research will be presented to scientists identified and selected by an awards review panel or through nominations received by the TS Alliance.

Future plans for CWW include broadening the base of funding to include research on the social, behavioral and cognitive issues in TSC, and on epilepsy in TSC. The Tuberous Sclerosis Alliance is committed to stimulate further research through other organizations and institutions, such as lobbying for increased federal dollars for TSC research through the National Institutes of Health.



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Major Gifts, Corporate and Foundation Support

Major gifts from individuals, corporations and foundations provide significant support to fund initiatives as determined by the TS Alliance Board of Directors. These gifts provide a solid financial base as the organization develops new programs to serve the growing number of constituents. In addition to providing financial support, the relationships developed through major giving enables the TS Alliance to engage the corporate community in creating awareness about TSC.

Endowment Fund

The Tuberous Sclerosis Alliance Endowment Fund was created in 1994 with an initial donation of \$27,000. The fund has grown significantly since its inception and exceeded \$1 million during the 2001 fiscal year. The first distribution of the endowment fund to the Tuberous Sclerosis Alliance occurred in March 2001 with a contribution of \$25,000. The endowment fund will continue to make contributions as the fund grows through donations and investments.

Planned Giving

Planned giving enables donors to arrange charitable contributions that minimize the donor's tax obligation while maximizing the individual's personal philanthropic goals of assisting the organization in funding vital programs. There are many types of planned gift opportunities, including bequests, life income gifts, gifts of retirement plans, gifts of insurance and charitable trusts.



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Tuberous Sclerosis Alliance

The Tuberous Sclerosis Alliance (TS Alliance) is the only national voluntary health organization for the genetic disorder known as tuberous sclerosis complex (TSC). It is also the lead organization for the funding of medical research related to TSC. Such medical research has included the breakthrough discovery of two genes (TSC1 and TSC2) that are known to cause the disorder.

The TS Alliance was established in 1974 as the National Tuberous Sclerosis Association (NTSA), which it remained titled until 2000. Four mothers of children with TSC founded the organization in order to provide fellowship, generate awareness, pursue more knowledge and provide hope to those who share the common bond of facing the daily challenges of TSC.

Current Focus

Today, the TS Alliance is overseen by a board of directors, whose role is to preserve the integrity of the TS Alliance, set policy, and support and promote the organization. The staff implements policies and priorities through the delivery of programs and services.

During its nearly 30 years of existence and growth, the TS Alliance has expanded its mission to improve the quality of life for individuals and families affected by tuberous sclerosis complex through the stimulation and sponsorship of research, the development of programs, support services and resource information, and the development and implementation of public and professional education programs designed to heighten awareness of TSC.

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Resources

The Tuberous Sclerosis Alliance (TS Alliance) acts as a clearinghouse for families and physicians seeking the most up-to-date information about tuberous sclerosis complex (TSC), providing resource information on an array of care, treatment, education and other challenges that may be encountered by those caring for an individual with TSC. The organization also serves to connect individuals throughout the nation and beyond, creating a network of informed constituents and physicians.

Patient Advocacy and Education

Because TSC is often difficult to diagnose and proper management of TSC is essential for optimum health, the TS Alliance also continues to place focus on educating medical professionals through participation in medical conferences and alliances with a number of patient advocacy programs. The TS Alliance is aggressively seeking federal government support for research and increased awareness in the medical community.

Patient advocacy skills have become essential for individuals and families to obtain adequate insurance, as well as medical, educational and other services in order to maximize the quality of life of those living with tuberous sclerosis complex. The TS Alliance provides resource referrals to help parents and others obtain necessary services and support they need to improve the quality of life for TSC patients.

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Volunteer Outreach

Dedicated volunteers across the country donate their time, talent and energy to support the Volunteer Outreach Program. Volunteers play an integral part in providing enhanced services and in raising revenue to support the mission and programs of the Tuberous Sclerosis Alliance (TS Alliance). The program enables volunteers to create awareness of tuberous sclerosis complex (TSC) in their community, to provide peer connections through the support network and to participate in fund-raising events and campaigns. The TS Alliance provides the necessary training and materials so that volunteers are successful in their mission in taking action and championing for our cause.

Support Network

The Tuberous Sclerosis Alliance Support Network is an organized partnership of individuals whose lives have been affected by TSC. The national network of volunteers provides enhanced individual/family/caregiver support by being the link between person-to-person connections, offering caller's experience-based information and, if desired, coordinating or leading a support group.

Matching Program

This unique program is designed to match individuals and/or families from across the country who are experiencing the same or similar challenges. The matching program provides the necessary peer support that may not be available in their community. The TS Alliance collects information as it pertains to each family participating in the matching program so that when an individual is in need of peer support it is readily available.

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Programs and Support Services

The TS Alliance has an active government relations program through which it is striving to increase visibility of TSC in Congress and within the National Institutes of Health (NIH). The organization's goal is to engage government institutions in basic scientific and clinical research on causes and remedies for TSC.

The TS Alliance builds networks through online services, conferences and volunteer outreach programs to give the TSC-affected population a sense of community. Members and donors provide the necessary resources to meet the overall goals and objectives of the organization by their volunteer efforts and contributions.

Outreach and Advocacy

The TS Alliance outreach and advocacy program collaborates with individuals and families in their efforts to obtain entitlements (social security and medical benefits), appropriate educational opportunities (placements and vocational services), and transitional issues, which include housing and community connections.

Professional Education

The professional and medical network of the organization provides a consensus regarding diagnosis and follow-up procedures. Members of the professional advisory board, both scientists and clinicians, make time available to provide resource information, discuss treatment with other medical practitioners, and provide their services to the TS Alliance in a voluntary capacity.



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Publications

The TS Alliance produces informative brochures, "Fact Sheets" and other resources to educate doctors and other practitioners on identification, diagnosis and treatment protocols for TSC patients, as well as resources to assist parents or family members with the range of issues confronting patients with TSC. The organization's quarterly newsletter *Perspective* provides families with current news about the TS Alliance, research updates, government relations actions, reports on special events, and educates readers on various advocacy issues related to their health or their child's health and education.

Information

The Tuberous Sclerosis Alliance explores the use of technology to provide current medical information to patients and doctors. The organization's Web site serves as a primary source of information to a worldwide community of doctors and families seeking up-to-date information about TSC. The Web site also serves as a networking tool for families to unite together in support and information through various list serves.

Conferences and regional meetings link the TSC community and medical professionals by building a strong foundation of individuals who want to make a difference in the lives of those affected by the disorder.



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Community Fund Raising

Volunteers organize special events in their community that are marketed to a variety of target groups. Community special events are integral in building a foundation of donors and supporters in communities that the national office is unable to reach. Organized grassroots efforts ensure that the TS Alliance reaches a mass group of donors to obtain their contribution and educate the community about the complexities of the disorder. Special events also place a face and a family or an individual's personal story with tuberous sclerosis complex.

Outreach and Awareness

The TS Alliance depends on individuals to educate their community about tuberous sclerosis complex by distributing information to the medical community, school systems and community agencies. Volunteers are provided with information packets that are developed to specifically target a particular group of individuals or professionals. Additionally, materials are sent to train the volunteer on the information that is needed to present a face-to-face meeting.



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- Infantile Spasms in Children with TSC
- Intellectual & Behavior Development Problems in Preschoolers with TSC
- Learning Disability and Behavior
- Making the Most of Your Doctors Visits
- Mental Retardation in TSC
- Ophthalmic Findings in TSC
- Neuroimaging in TSC
- Processing Information in the Brain
- Psychiatric and Behavioral Issues and the Adult with TSC
- Psychopharmacology and the Child with TSC
- Pulmonary Manifestations in TSC
- Renal Manifestations of TSC
- Seizure Types
- The Brain and TSC
- The Spectrum of Attentional Problems in Individuals with TSC
- Treating Skin Disorders Associated with TSC
- TSC and Autism Spectrum Disorders
- Tuberous Sclerosis Facts for Teachers
- Tuberous Sclerosis Q & A
- Understanding the Genetics of TSC
- Warning Signs in TSC

Life Stages Program

- Adult Guide of the Life Stages Program (ages 21 and above)*
- Young Adult Guide of the Life Stages Program (ages 14-21)*
- School-Aged Guide of the Life Stages Program (ages 6-13)*

- Early Years Guide of the Life Stages Program (ages 0-5)*

Journals

- Tuberous Sclerosis Journal—An Organizational Diary for the Care and Treatment of Tuberous Sclerosis

JOURNAL ARTICLES

- "Tuberous Sclerosis Consensus Conference: Recommendations for Diagnostic Evaluation," Journal of Child Neurology, January 1999
- "Tuberous Sclerosis Complex Consensus Conference: Revised clinical diagnostic criteria," Journal of Child Neurology, September 1998
- "National Institutes of Health Consensus Conference: Tuberous Sclerosis Complex." Archives of Neurology, May 2000

BOOKS

- Living With Tuberous Sclerosis
- Tuberous Sclerosis Complex—Developmental Perspectives in Psychiatry, Third Edition, Copyright 1999. Cost: \$76.00. Major credit cards accepted.

To place your order, please call, e-mail, mail or fax your request to:

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 (301) 562-9890
 (800) 225-6872 (toll-free)
 (301) 562-9870 (fax)
www.tsalliance.org
info@tsalliance.org



Please print or type clearly. Unless otherwise indicated, there is no charge for shipments mailed to U.S. addresses. International orders require an additional charge.

NAME: _____

ADDRESS: _____

CITY: _____

STATE: _____

PHONE _____ (home) _____ (work)

E-MAIL: _____

Please provide the information below so that we may keep our database current. Any information provided will remain confidential.

NAME OF INDIVIDUAL(S) WITH TSC: _____

DATE OF BIRTH(S) (month/day/year): _____

DATE OF DIAGNOSIS (month/day/year): _____

Relationship to Individual with TSC (please check one): Self Parent Sibling Spouse
 Grandparent Guardian Other (please specify)

Are you currently on our mailing list?
 Yes No Not sure



RESOURCE ORDER FORM

Please check the item(s) you wish to receive. To receive electronic newsletters, be sure to provide your e-mail address in the space provided at the end of this form. Items marked with an * can be viewed and downloaded from the TS Alliance Web Site.

NEWSLETTERS

- Perspective* (print newsletter) Current Issue*
- Resource Exchange* (electronic newsletter)*

BROCHURES/VIDEOS

- An Introduction to TSC*
- My Child Has Tuberous Sclerosis* (Spanish)
- 2001 National Family Conference Videos*
www.tsalliance.org/activities/TapeOrderWeb.pdf provides a list of available titles and order form. Please list the title(s) you wish to order on the lines below. There is a \$16.00 charge for each video ordered.

TITLES: _____

REFERENCE MATERIALS

Fact Sheet Series

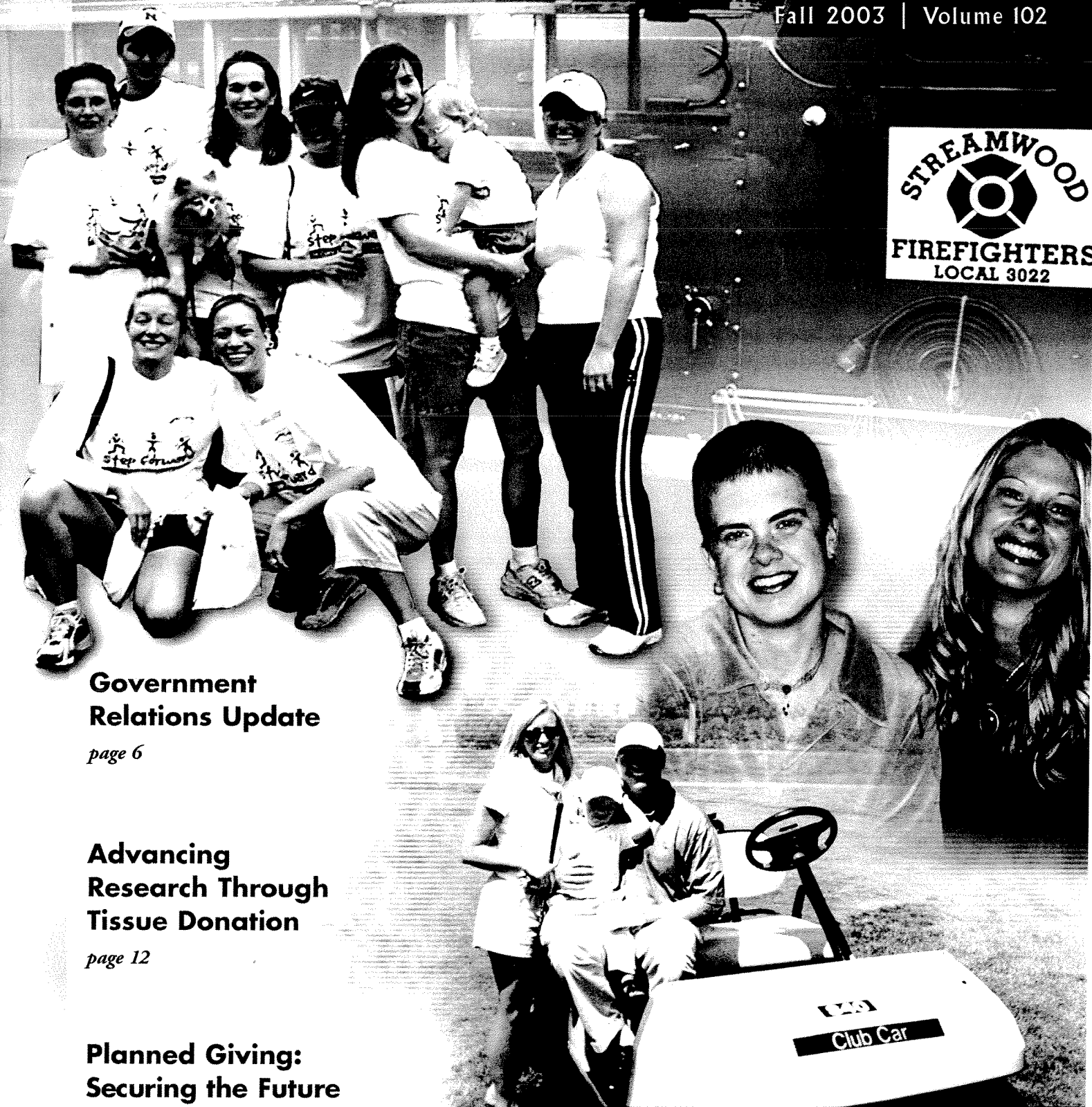
(Note: all fact sheets can be downloaded from the Web site)

- Adolescents with TSC
- Attention Deficit Hyperactivity Disorder and TSC
- Cardiac Manifestations in People with TSC
- Epilepsy Surgery for TSC Patients
- Frequently Used Seizure Medications
- Genetic Testing for TSC
- Geneticists—Why Do I Need Them?

Tuberous Sclerosis Alliance

PERSPECTIVE

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Advancing Research Through Tissue Donation

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Planned Giving: Securing the Future

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FALL 2003 • Volume 102

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Marybeth Leongini
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Director of Communications

If you have opinions, questions or articles for *Perspective*, we would like to hear from you. Please contact the managing editor to obtain a submissions form and guidelines.

Perspective is intended to provide basic information about tuberous sclerosis complex. It is not intended to, nor does it, constitute medical or other advice. The Tuberous Sclerosis Alliance (TS Alliance) does not promote or recommend any treatment, therapy, institution or health care plan. Readers are warned not to take any action without first consulting a physician. Commentary expressed herein reflects the personal opinions of the author and does not necessarily reflect the official views of the TS Alliance. Information contained in the TS Alliance database is confidential and not provided nor sold to third parties.

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CHAIRMAN'S LETTER

Scott Johnson
Chairman of the Tuberous Sclerosis Alliance

As you may recall, I closed my last letter mentioning that our daughter, Annie, (age 21) was about to have seizure surgery at UCSF. I'm happy to report that all indications are that the surgery was a HUGE success. No seizures since (versus two to three per week for 20 years); major improvement in mood; and notable increase in intellectual curiosity. We're thrilled and relieved. We're also grateful for all the information and support we received from the TS Alliance and associated professionals. While surgery may not be the right treatment for most, it has become the best option for many. Our involvement with the TS Alliance was enormously helpful in arming us with the information we needed to make an informed decision. I might also add that Annie's brain tissue (i.e. the portion removed) is now hard at work helping several TSC researchers at no fewer than six research centers throughout the United States via the TS Alliance's tissue donation program. OK, enough about the Johnsons. Here is what is happening at the TS Alliance.

As I write this, we are three months into our new fiscal year and we're preparing for our fall board of directors meeting. Together, the board and staff have established the following as our key objectives for this fiscal year.

Raise \$3 million. While our mission statement "Finding a cure for tuberous sclerosis while improving the lives of those affected" does not mention raising money, the truth is that funding is the greatest obstacle to the accomplishment of our mission. \$3 million represents a 13% increase from last year, a larger increase than most non-profits are targeting in this still difficult economy.

So, what can you do? Consider: Bumping your own contribution up 20% or more. Organize a fund raising event – or volunteer to assist in one. Participate in this year's Stamp Out campaign. Put us in your Will. As they say, don't give until it hurts, give until it feels good – and encourage your friends to do the same.

Help us secure federal funding of TSC research. As an NTSA/TS Alliance veteran, I'm happy to report that TSC research is no longer sponsored strictly by "family and friends" (although these contributions remain critical). Recently, we've made very substantial progress with the National Institutes of Health and the Congressionally Directed Medical Research Program within the Department of Defense. Last year, these agencies spent \$8.4 million on research specific to TSC (compared to the \$1.3 million that the TS Alliance funds directly). Next year, that number will grow to \$10 million or more. Virtually none of this would have occurred without the efforts of the TS Alliance. As important, it will be the TS Alliance that will guide what research projects these funds will be invested in. Big opportunity! Big responsibility! We're up to both.

Absolutely essential to our efforts with the federal government is creating a visible presence with our representatives on Capitol Hill. To that end, we will be establishing and institutionalizing an early spring Congressional lobbying campaign by TS Alliance volunteers, both on Capitol Hill and the district/home offices of our Representatives and Senators. We did a pilot run of this last spring, utilizing our Community Alliance Chairs. It was very effective. As a personal participant, calling on my own Congresswoman (with whom I'm not typically aligned politically), I found the experience to be more interesting, more rewarding and a lot less intimidating than I imagined. You will too. You'll be hearing more about this in the next issue of *Perspective*. We all

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SPECIAL EVENTS



TOUR OF CHAMPIONS

The Maria L. Anderson TS Alliance Charity Golf Classic – *Centreville, Virginia*

The fourth annual memorial golf event was held on August 22 at the Fairfax National Golf Club in Centreville, Virginia. This tournament was held in memory of Maria Anderson who died after a life-long battle with tuberous sclerosis complex (TSC). Congratulations to Bud Anderson and all of his friends and family who raised over \$22,000 for TS Alliance research!

TS Charity Scramble – *Cincinnati, Ohio*

The 12th annual TS Charity Golf Scramble took place on July 29th at the Shaker Run Country Club in Cincinnati, Ohio. Golf chairman, Doug Loftus reports that this year's

event raised an incredible \$20,000 for the TS Alliance. Doug and his wife Linda started the event out of Doug's passion for golf and the desire to raise money for the TS Alliance in honor of their daughter, Alyssa. Doug and Linda began the tournament with a handful of friends, family and co-workers and now have a loyal group of golfers that include other TS families. Since 1991 this annual golf scramble has brought in over \$300,000 for TSC research.

Ocean Properties Ltd. Golf Tournament – *New Castle, New Hampshire*

Tom Gwinn is the director of purchasing with Ocean Properties Ltd. and each year they sponsor a golf tournament. This year's event was held on August 24-25 at the newly remodeled historic hotel Wentworth by the Sea. The tournament raised \$25,000 and was held in honor of Tom's son Trey who has TSC.

Golf for Tuberous Sclerosis – *Mahtowa, Minnesota*

Thank you to Candy Beaulieu who organized the inaugural Golf for Tuberous Sclerosis Tournament in Northern Minnesota held on August 24. The event, which took place at Twenty-Nine Pines Golf Course, included 17 foursomes and raised \$3,400 to support the TS Alliance.

Caddy Shack Golf Tournament – *Monroe, Connecticut*

The 16th annual Caddy Shack tournament took place on Aug. 19 in Monroe, CT. A special thank you to volunteer Fran Hillier for her faithful dedication and longstanding support of the TS Alliance with this special event.

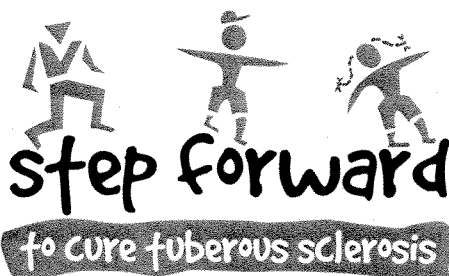
Foster, Gundy and Associates Golf Tournament to benefit the TS Alliance – *Grand Rapids, Michigan*

Volunteers Eric and Kimberly Hyde organized the inaugural Tuberous Sclerosis Alliance Charity Golf Outing sponsored by Foster, Gundy and Associates. The event was held at the Thorneapple Golf Course in Grand Rapids, Michigan on July 18. One hundred golfers participated and raised over \$7,000 in honor of the Hyde's son Foster.

Courtney Leigh Small Steps Tournament – *Parsippany, New Jersey*

Frank Gallagher, past chair of the TS Alliance Board of Directors, hosted his 9th annual tournament on August 7, 2003 at the Knoll West Country Club in Parsippany, New Jersey. In attendance were 140 golfers who came out to help raise funds for the Courtney Leigh Small Steps Fund Research Award.





step forward
to cure tuberous sclerosis

Rocky Mountain Region Hosts First Walk-a-thon

The TS Alliance of the Rocky Mountain Region held their first walk/5K run, Step Forward to Cure TS, on Sunday, August 24. The walk



was chaired by Bill Youmans and raised more than \$32,000. The 5K race was officially started by Denver Nuggets head coach, Jeff Bzdelik. In addition, Cindy and Jim Lynch were



featured on local network news and Denise Lovato appeared on the front page of the *Denver Post* with her daughter Kylee.

The TS Alliance of Greater Chicago



The TS Alliance of Greater Chicago held the second Step Forward to Cure TS "Walk to Liv" on Saturday, September 13. The walk, chaired by Jenny Smiley, raised more than \$31,000 with over 225 participants. Many thanks to the entire Smiley (Jenny, Sandy, Jay, Lisa and Livy) family and the incredible walk committee for all of their work on another successful walk and to the Streamwood Fire Department for their support.



GIFT-WRAP CAMPAIGN

It's time again for the Annual Gift-wrap Campaign. This campaign continues to raise money for the organization by engaging TS Alliance volunteers and members to sell and/or buy gift-wrap.

It is the perfect opportunity to stock up on gift-wrap for birthday presents or to get ahead of the game and prepare for winter holiday gift giving. Whatever your needs may be, the Annual Gift-Wrap Campaign, organized by Gift-Wrap Campaign chair Milly Augustine, is here again to help you box and wrap those special gifts with beautiful, high-quality gift-wrap paper and products.

Sally Foster® provides seller kits which enable the participant to show potential buyers the vast gift-wrap collection. Buyers place their order with the seller and the TS Alliance retains 50 percent of the proceeds.

Your continued participation in the successful gift-wrap campaign also helps support the Tuberous Sclerosis Alliance's mission to help those who live with TSC. If you are interested in supporting the event and would like to receive a seller's kit or purchase gift-wrap, please contact Milly Augustine directly at (626) 286-4322 (PST) or leave a message on her voice mail (800) 225-6872 ext. 26 or send an e-mail to: m4-ts@pacbell.

TS ALLIANCE RECEIVES AWARD FOR ADVOCACY EFFORTS

The Child Neurology Foundation selected the Tuberous Sclerosis Alliance as recipient of the second annual Child Neurology Foundation Advocacy Award of Merit. Recipient organizations are nominated by members of the Child Neurology Society and selected by the board of directors of the Child Neurology Foundation. The award is presented in recognition of a patient support organization associated with pediatric neurology. The 2003 award was presented in Miami on October 2 at the 32nd Annual Meeting of the Child Neurology Society. The TS Alliance was selected and recognized for its outstanding achievements on behalf of patients and families, with a special emphasis placed upon the TS Alliance's efforts to create increased public awareness of this specific neurological condition. TS Alliance President and CEO Mike Coburn received the award on behalf of the organization. The TS Alliance, in conjunction with the Child Neurology Society and the National Institutes of Health also presented a special day-long symposium on tuberous sclerosis complex as part of the Child Neurology Society's annual meeting. The Child Neurology Society is the pre-eminent non-profit professional association of pediatric neurologists in the United States, Canada and worldwide devoted to fostering the discipline of child neurology and promoting the optimal care and welfare of children with neurological and neurodevelopmental disorders.



PRESIDENT'S MESSAGE

Mike Coburn
President and CEO of the Tuberous Sclerosis Alliance

In each issue of *Perspective* we take time to introduce our readers to members of the scientific community through our "Scientific Spotlight" (see pgs. 8-9). There is, however, a growing number of medical researchers, physician/scientists and other skilled scientific professionals working on a broad array of scientific discovery related to tuberous sclerosis complex (TSC) that deserve our ongoing gratitude. While some individuals come to TSC research by way of mentorship in laboratories of seasoned TSC researchers, others become increasingly interested in the science of the TSC genes by virtue of their work in other areas that lead them to wonder as we do, what role these genes play in disease, why mutations in these genes cause such variable expression and how protein products of the TSC genes relate to tumor development and growth. TSC research has now become a beacon of interest to a much broader group of scientific professionals than in the past.

Each individual who comes directly into TSC research or becomes interested in TSC from a different direction brings incredible human resource to our common vision of finding treatments and a cure. Creativity, curiosity, discipline, ingenuity, intellect, innovation and resourcefulness are but a few words to describe the skills and attributes of the scientific professionals working daily to find missing pieces of a puzzle that one day will improve the lives of those living with TSC.

Years of schooling and hands-on training combined often with mental and sometimes physical challenge become part of the investment the research community has made in furthering the science of TSC. While many of the dedicated research professionals work strictly in laboratories, there are those too that split their time between the lab and the clinical setting. Physician/scientists are talented individuals who strive to bring results from laboratory bench work to the often referred "bedside". We can illustrate this clearly through the current clinical study of the drug Rapamycin. Research scientists find that through laboratory experience, Rapamycin may shrink renal and lung tumors. Physicians who also work closely with science, are now using this scientific evidence in the clinical trial with patients. It is this combination of basic science and clinical science that brings hope for new and effective drugs to treat aspects of TSC.

I know that each and every family that has encountered TSC joins me in appreciation of the growing scientific community that is dedicated to TSC research. We look forward to working closely with scientists and physicians in all fields related to TSC as we push for increased scientific and medical knowledge. We are indeed grateful to those clinicians who seek innovation through science to improve treatments for TSC and especially thank them for always keeping their colleagues in the lab mindful of the human aspect of science and medicine.

GOVERNMENT RELATIONS UPDATE

NIH PUBLISHES FIRST COMPREHENSIVE RESEARCH PLAN FOR TSC

The TS Alliance Government Relations Committee is pleased to report that continued progress is visible! During the past year and a half, the TS Alliance has been working with friends in Congress, most notably Rep. Sue Kelly of New York, Rep. Jerry Lewis of California and Sen. Arlen Specter of Pennsylvania to encourage the National Institutes of Health to develop and implement a research plan for tuberous sclerosis complex (TSC). On August 7, 2003, NIH Director Elias Zerhouni, M.D., transmitted to Rep. Kelly the "Research Plan for Tuberous Sclerosis". The plan is a direct result of last years House

Concurrent Resolution sponsored by Congresswoman Kelly and also results from the September 2002 research conference cosponsored by the TS Alliance and the National Institute of Neurological Disorders and Stroke.

Acting director of the National Institute for Neurological Disorders and Stroke (NINDS), Audrey Penn, M.D., in her letter to TS Alliance president Mike Coburn stated "the plan offers a long-range, forward thinking vision for the tuberous sclerosis research community. NIH and the broader TSC research community will use these goals (outlined in the plan) as a guide for developing focused research projects, programs, and infrastructure, and as a yardstick for measuring progress."

The TS Alliance will continue to work with the leadership and other

officials within NIH to implement all aspect of the research plan. The TS Alliance will also utilize the NIH plan as part of an overall blue print or master plan for research in order to engage more scientists, physicians and other researchers and institutions in acceleration of scientific and medical knowledge of TSC. We expect, as a result of our success in getting report language included in the NIH appropriations bill for 2004 that NIH will report back to congress on progress of the plan and we will continue to cultivate our relationships with individual Members of Congress to ensure that the NIH research plan is indeed a work in progress.

A copy of the NIH research plan can be viewed by visiting the TS Alliance Web site, www.tsalliance.org

LATE BREAKING NEWS!

On September 30, 2003 President Bush signed the Department of Defense Appropriations bill for 2004 which includes \$ 3 million in new funding for the Tuberous Sclerosis Complex Research Program managed by the Congressionally Directed Medical Research Program (CDMRP).

This new research funding for the CDMRP brings total new research dollars through the Defense Department research program to \$6 million over the past three years. The efforts of the TS Alliance Government Relations Committee, members of our Community Alliances and volunteers from throughout the country were instrumental in gaining increased support for the program.

The Department of Defense Tuberous Sclerosis Complex Research Program was established in 2001 (for the FY 2002 fiscal year) by Joint Appropriations Conference Committee Report No. 107-350, which provided the initial \$1M for TSC research. The program is managed by the United States Army Medical Research and Materiel Command (USAMRMC) and is a peer reviewed research grants program.

The TS Alliance government advocacy efforts were quite successful this year—a published NIH research plan, report language in both the House and Senate NIH appropriations bills, questions about TSC from lawmakers to NIH officials at appropriations hearings and this \$3 million in new funding.



TS ALLIANCE CALL FOR NOMINATIONS TO THE BOARD OF DIRECTORS

The Tuberous Sclerosis Alliance (TS Alliance) is the only national voluntary health agency committed to finding a cure for tuberous sclerosis complex (TSC) while improving the lives of those affected. Governance of the TS Alliance is provided by a volunteer board of directors comprised of up to 31 individuals who individually provide expertise and guidance and who collectively establish goals, objectives and monitor progress of the organization. The board works in partnership with staff to accomplish annual plans and has financial and legal responsibility for the health of the organization.

Ideal candidates will possess leadership skills, provide outreach to resources, help raise funds for the organization, have a passion and commitment to help those affected

by TSC and provide sound analysis and judgment to organizational issues. Board members are expected to attend face-to-face board meetings twice each year, participate in several board teleconferences and serve actively on at least one committee. Members of the board of directors may serve two consecutive three-year terms or may be elected for a minimum of one year. In keeping with best practices guiding non-profit organizations, approximately one-third of individual board members' terms expire each year, providing for a rotation of terms.

Nominations or applications for board service will be considered by the nominating committee upon receipt of a letter of interest outlining the nominee's interest and qualifications to serve on the board. Because there are limited board positions available,

nominations will be reviewed seeking knowledge and experience that will add dimension to the board as a whole. The Nominating Committee is especially interested in identifying candidates with an expertise in the following fields:

**Finance /Accounting
Public Relations
Science /Medicine**

The nominating committee may request additional information during the review process. Nominations are presented to the annual meeting in March for election.

Nominations may be received no later than December 1, 2003 and sent to the attention of: Mike Coburn, President, Tuberous Sclerosis Alliance, 801 Roeder Road, Suite 750, Silver Spring, MD 20910.

CALL FOR NOMINATIONS

MANUEL R. GOMEZ RECOGNITION AWARD

The Tuberous Sclerosis Alliance (TS Alliance) invites nominations for the fifth annual Manuel R. Gomez Award. Any individual or group may submit a nomination to the TS Alliance no later than November 15, 2003. Mail all nominations to Holly Knorr at the TS Alliance at 801 Roeder Road, Suite 750, Silver Spring, MD 20910 or send by e-mail to holly.knorr@tsalliance.org.

Award recipients will be selected on the basis of two principal criteria:

- *Achievement for either a single significant breakthrough, or for going above and beyond the call of duty in caring for individuals with tuberous sclerosis complex (TSC) during their career.*
- *Achievement of national or international scope.*

Information about additional selection criteria and what materials need to be included with your nominations can be found on the TS Alliance Web site – www.tsalliance.org.

A panel of experts will be convened later in the year to review all nominations and to select the recipient of the award.

This award is made possible by a generous donation from Mr. and Mrs. Harold Aronson in honor of their son Peter Aronson, M.D. Peter is currently a member of the TS Alliance professional advisory board and has also served as a member of the TS Alliance board of directors in the past. The organization will present the award in December of 2003.

SCIENTIFIC SPOTLIGHT

An Interview with Christine Wade, project coordinator, Brain and Tissue Bank



WHAT ARE YOUR MAIN RESPONSIBILITIES AT THE TISSUE BANK?

My main responsibility is creating awareness of the Brain and Tissue Bank through support groups and working with families in order to make sure the tissue donation process is simple and successful. Other responsibilities include establishing positive relationships with pathologists and researchers inside and outside of the United States.

WHAT LED YOU TO THIS POSITION (EDUCATIONAL BACKGROUND, INTERESTS, ETC...)?

I have always been a strong advocate for research particularly in pediatrics. I feel that we need to help the future and this would start with our children and our children's children. I am still pursuing my education; concentrating in developmental psychology. I have a very unique family background. With

my grandfather and father licensed as funeral directors and having served in the military medical corp., my current position seems a natural fit. My father has been the director of the Maryland State Anatomy Board for the past 30 years. I spent many summers assisting families and promoting the body donor program. As a result, I grew up realizing the benefits of advancing medical studies.

APPROXIMATELY HOW MANY DISORDERS ARE REPRESENTED BY TISSUE AT THE BANK?

Currently, we represent over 183 different developmental disorders. We work closely with 30 various support groups including the National Organization for Rare Disorders (NORD), the Genetic Alliance and the Genome Institute at NIH.

WHAT TYPES OF TISSUE DOES THE BANK COLLECT?

Since we work primarily with genetic and developmental disorders the majority of the collection is brain tissue, although tissues collected can vary depending on the type of disorder and its' effects on the individual. Also, we take into account what type of tissue is needed for the researchers.

WHERE IS THE TISSUE STORED?

The tissue is stored in two different environments on site. Frozen tissue is kept in freezers; we currently have seventeen freezers here at our facility with tremendous amounts of tissue. We have more freezers on the way

in 2004 in order to accommodate all of the donated tissue. Fixed tissue is kept in two different rooms at room temperature in formalin on shelves fully stocked, so you can only imagine what researchers are provided!

DESCRIBE YOUR DAY YESTERDAY.

Wow...yesterday was a definite coffee day for many others and me! To start, I received an urgent page directly from Vicky Whittemore around 3:45a.m. concerning a donor who was to pass within hours. This case was extremely critical with regards to timing of tissue recovery for the research protocol. Needless to say, I woke many people up in the middle of their REM cycle; most were very understanding. I then scooted to the office around 7a.m. and spent another two hours on this case. My day finally ended around 5:30p.m. Unfortunately, this case did not happen to work out. No one was really to blame; but had I received all the pre-registered consent forms the outcome would have been different!

DO YOU OFTEN WORK OUTSIDE OF BUSINESS HOURS? DESCRIBE THE LATEST AFTER HOURS TISSUE RECOVERY.

That would be a definite "yes"! I am usually on-call 24 hours 7 days a week – everyday! There have been many after hour and holiday recoveries, but there is one that comes to mind vividly. It was Saturday 11:30p.m. and I found myself driving down the interstate to meet a pathologist on the roadside at a designated exit. I gathered the specimen (tissue) and drove

off to the Brain and Tissue Bank at the University. I arrived there around 1:00a.m. and procured the tissue. I will never forget that night!

WHAT DO YOU LIKE MOST ABOUT THIS POSITION?

I enjoy working with families and assisting in the research to help the future. This to me is personally satisfying. Every situation that is brought to the Brain and Tissue Bank is uniquely different. This helps me to stay on top of things and bring closure and/or peace of mind to most families.

WHY IS IT SO IMPORTANT TO PRE-REGISTER WITH THE BANK? HOW DOES THIS MAKE TISSUE RECOVERY EASIER?

In pre-registering, all of the consents have been completed and are kept on file for future donations. Pre-registering is ideal in the sense that there would only be one phone call or communication made to the Brain and Tissue Bank initiating the entire process. In every case, whether it is a surgery or a death, we are not able to set-up recovery without these forms. If we do not have these forms completed and on file, we have to find an alternative way (usually faxing) to send them to the donor and/or family and in most cases fax machines can be hard to come by at this critical time. Consequently, most pathologist and/or physicians will not begin any type of tissue recovery without these forms completed. Verbal consent is out of the question in most circumstances. Consent forms are a fundamental key in obtaining tissue donation due to the fact that they are acknowledged by most pathologist and/or physicians in the United States.

HOW DO RESEARCHERS NORMALLY OBTAIN TISSUE? WHAT'S THE MOST INTERESTING REQUEST YOU'VE HAD FROM A RESEARCHER?

Researchers that would like to obtain tissue would contact the Brain and Tissue Bank directly. They would then complete a Tissue Transfer Agreement (TTA), an informal summary of their particular type of research and a request form listing their parameters (what type of tissue, age, etc).

There have been some very unique and unusual requests in the past for tissue. In speaking with our pathologist, Robert Vigorito, there was one instance where a researcher was researching for a toxicology study. He requested fingernails in order to complete his project.

HOW HAS THE TS ALLIANCE HELPED YOU IN DOING YOUR JOB IN RECOVERING TSC TISSUE?

The TS Alliance has made my job easier in recovering TSC tissue. First and most importantly, the lines of communication are completely open. As soon as there is word of potential donor tissue for research, they immediately contact me or have the family contact me directly. Also, they have made every effort in promoting pre-registration with their members to simplify the process. As discussed before, pre-registration is extremely critical to successful tissue recovery. Not only does the TS Alliance assist in communication with families, but also with the researcher(s) that they know will require and utilize the tissue for TSC. Without the assistance of the TS Alliance, the Brain and Tissue Bank would have greater difficulty obtaining this precious tissue that fulfills the needs of medical researchers.

REGISTER TO BE A TISSUE DONOR TODAY!

The National Institute of Child Health and Human Development (NICHD) has funded development of the NICHD Brain and Tissue Bank for Developmental Disorders.

We encourage all individuals with TSC to be proactive and register with the tissue bank. This will allow for efficient collection of tissue at the time of a scheduled surgery or in the unfortunate event of death. Registration is easy. Just request a donation form from the TS Alliance by sending an e-mail to info@tsalliance (subject: tissue donation) or call (800) 226-6872. Thank you for becoming a registered TSC Tissue Donor today!



ASK DR. BONNIE



Bonnie Gould Rothberg M.D. is the medical director of the Rothberg Institute for Childhood Diseases and a member of the TS Alliance board of directors. The Rothberg Institute for Childhood Diseases is a non-profit organization dedicated to finding a cure for children suffering from tuberous sclerosis complex (TSC).

Dear Dr. Bonnie,
My child with TSC has intractable epilepsy and his neurologist suggested we have him evaluated for seizure surgery. What does the evaluation involve? How do I decide if this is something that is right for my child - brain surgery seems so drastic!

Thank you,
Ken

Dear Ken,

Thanks for your post requesting information on seizure neurosurgery for TSC and the process of a surgical evaluation. As the parent of a child with TSC whose seizures have become increasingly difficult to control over the past six months and who is in the midst of her own surgical evaluation, I recognize not only the magnitude of the decision to perform brain surgery on our children but also the insecurities and fears that parents and other loved ones undergo during the process.

The goal behind epilepsy surgery is to remove the piece of brain tissue that is at the source of the seizures. The expectation is that with the source gone, the rest of the brain will have no further stimulation to have a seizure and will resume its normal functions, uninterrupted. Because neurosurgeons are precise by nature, they strive to remove the smallest piece of brain tissue possible that can account for all of the seizure activity - the rest of the brain will be spared. Post-surgery pathology studies have also shown that, in the large majority of cases, the brain tissue removed was diseased or abnormal and unlikely to have contributed to any key brain function in the individual. Nonetheless, this does not diminish the point that brain surgery for seizure control is highly invasive and side effects, while not very common, can be extremely devastating. For this reason, surgery is only considered when all non-invasive methods of seizure control have failed.

In cases of TSC, brain surgery is currently considered only for those patients with 'intractable epilepsy'.

Two conditions must be met to have 'intractable epilepsy'. First, the seizures are typically not responsive to any of the available anti-epileptic medications or other non/minimally invasive therapies such as the vagus nerve stimulator or the ketogenic diet. Second, the seizures cause a negative impact on the patient's development or quality of life (e.g., the patient is falling behind on their developmental curve, is losing skills he/she had previously mastered or is unable to fully interact with their typical environment due to extreme post-ictal sleepiness). Since parents and caregivers are most likely to notice new or increased developmental difficulties, they should not hesitate to share these observations with their neurologist.

Once a TSC patient is identified as medically appropriate for surgery (i.e., he/she has intractable epilepsy), the next step is to identify whether a patient is surgically appropriate for the operation. Surgical appropriateness requires meeting two criteria: 1) all sources of epilepsy (a.k.a., the epileptogenic focus or foci) can be identified and their boundaries defined with confidence and 2) removal of these foci will not compromise major brain functions, especially vision, hearing and motor capabilities. The process of determining surgical appropriateness is called the "pre-surgical evaluation". Because this process is considered 'elective', in many geographic regions the scheduling of the suggested diagnostic tests can take weeks if not months. It is therefore recommended to start the process as soon as is medically appropriate and not wait until the patient has had a substantial change

LISTING OF TS CLINICS

in their quality of life such that the work-up and surgery become medically urgent.

The first step of the pre-surgical evaluation is to determine where in the brain the seizure focus resides. TSC research has suggested that, in the majority of cases, the source of the seizure corresponds to the location of a tuber and that the disorganized nerve cells inside that tuber cause the seizures. However, this is not always the case and TSC doctors have identified and successfully operated on patients where seizures are coming from brain regions outside of tubers or even in patients who do not have tubers. In order to complete this part of the evaluation, doctors need to get information on where the seizures are coming from and what parts of the actual physical brain this corresponds to. This is accomplished largely by completing two types of testing: 1) the electroencephalogram (EEG) that identifies the electrical source of the seizure and 2) a series of imaging studies that look at the actual structure of the brain, some of its basic functions (e.g., blood flow, glucose metabolism) and how some of these functions change during a seizure.

I hope this is helpful information. My future columns will continue on this topic as I will discuss topics including EEGs, PET scans and other issues related to determining if brain surgery is the appropriate measure to take.

I wish you lots of luck,

Thanks,
Dr. Bonnie

BOSTON, MASSACHUSETTS
Massachusetts General Hospital
To schedule an appointment contact
Christina Anagnos at (617) 726-6540

CHICAGO, ILLINOIS
University of Chicago
To schedule an appointment contact
Sharon at (773) 834-8064

CINCINNATI, OHIO
Children's Hospital Medical Center
To schedule an appointment contact
Cindy Tudor at (513) 636-4222

DALLAS, TEXAS
Texas Scottish Rite Hospital for Children
For appointments contact either
Lori Batchelor at (214) 559-7824 or
Catherine Thompson at (214) 559-7818

DENVER, COLORADO
The Children's Hospital Neurocutaneous Clinic
This is a genetics/diagnostic management clinic only. For more information,
contact Jean Jirikowic at (303) 861-6974

HARTFORD, CONNECTICUT
Connecticut Children's Medical Center
To schedule and appointment contact
Carman Ayala at (860) 545-9586

LOMA LINDA, CALIFORNIA
Loma Linda Health Care Tuberosus Sclerosis Clinic
To schedule and appointment, contact
Teresa Serna at (909) 558-2383.

MIAMI, FLORIDA
Miami Children's Hospital
To schedule an appointment contact
either Angeles Sanzano or Laura Estrada at (305) 662-8338

NEW YORK CITY, NEW YORK
New York University - Mount Sinai Comprehensive Epilepsy Center
To schedule an appointment contact
Tracy Bosley at (212) 263-8331

PHILADELPHIA, PENNSYLVANIA
University of Pennsylvania Medical Center and Children's Hospital of Philadelphia
E-mail questions about the clinic to Dr. Crino at crinop@mail.med.upenn.edu.
When sending an e-mail to Dr. Crino, please cc: Margo Aragon aragon@mail.med.upenn.edu. To schedule an appointment contact Margo at (215) 349-5312

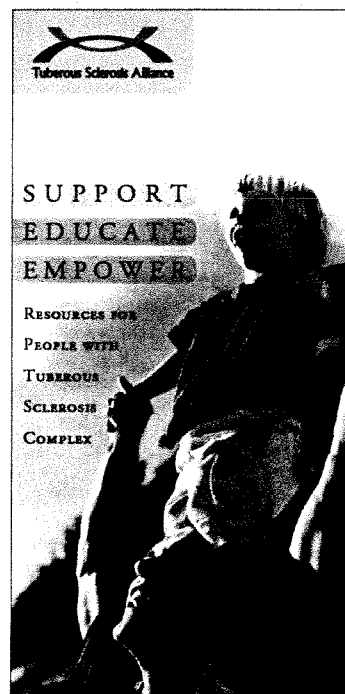
PITTSBURGH, PENNSYLVANIA
Children's Hospital of Pittsburgh
To schedule an appointment contact
Denise, Donell or Debbie at
(412) 692-5520

ST. LOUIS, MISSOURI
St. Louis Children's Hospital
Division of Pediatric Neurology
For appointments call Terri DeVault at
(314) 454-6120

WASHINGTON, DC
For information on the clinic or to
schedule an appointment contact Nancy
J. Elling at (571) 226-8343

COMING SOON...
The TS Clinic at Oakland Children's Hospital (CA) is expected to open in the first quarter of 2004. The clinic opening is made possible in part by a Children's Hospital Board grant and \$50,000 in grass roots funding. Contact Shannon Leal at (510) 483-9958 or shannonleal@attbi.com for more information. Additional details will be provided in the next issue of *Perspective*.

THE TS ALLIANCE RESOURCE BROCHURE IS NOW AVAILABLE.



This new publication lists the various programs and services offered through the TS Alliance. Available for download at www.tsalliance.org or by calling 800-225-6872.

ADVANCING TSC RESEARCH THROUGH TISSUE DONATION

In so many ways, Annie Johnson is like most young women. She loves social dancing, ice skating and being on her computer. But in one way, she is very special...she helped to make history in tuberous sclerosis complex (TSC) research! Experiments done in the laboratory of Scott Baraban, Ph.D. at the University of California, San Francisco (UCSF) using tissue removed at the time of Annie's seizure surgery are the first of their kind, ever! Other experiments utilizing tissue from her surgery are also underway in several other laboratories across the country – a testament to the importance and incredible value of donating tissue for research, as well as the increasing number of researchers who are studying TSC. All of the individuals with TSC will benefit from this research to understand the role of the TSC genes in epilepsy.

Annie was diagnosed with TSC when she was 17 months old and has had seizures all of her life. Her seizures had never been completely controlled with medication and they were getting worse, so about a year ago Annie's parents went to UCSF not expecting to consider surgery as a possible treatment for Annie's seizures. After a full year of careful testing, including EEGs, several different MRIs and implanted electrodes to monitor her seizures, Annie had surgery in August 2003 to remove a portion of her right temporal lobe – the area that doctors were able to pinpoint as the focus of her seizures. She is seizure-free two months after the surgery and has had no memory loss or other complications following the surgery. Her hair is growing back and she is already back to dancing once a week!

So, how did the tissue donation happen? The Johnson's knew that the researchers might be interested in obtaining some tissue from Annie's surgery, so they contacted the TS Alliance to find out what needed to be done and to make the arrangements. They were put in touch with Christine Wade, the project coordinator at the Brain and Tissue Bank for Developmental Disorders at the University of Maryland in Baltimore (see article page 8). The staff at the TS Alliance also knew

that Dr. Baraban was interested in obtaining tissue for his experiments so they discussed this with the Johnsons who agreed that some of the tissue would be sent to the Brain and Tissue Bank and some would be given to Dr. Baraban. The critical issue was that Dr. Baraban needed to obtain the tissue within ten minutes of it being removed or they would not be able to do their experiments.

Dr. Baraban was able to obtain tissue directly from the operating room, take it back to his laboratory where within a couple of hours he and his postdoctoral fellow were able to place electrodes into individual cells and obtain recordings from them of their electrical activity and characteristics. They identified that these cells appeared to have abnormal characteristics compared to normal cells. In order to be sure of these findings and to confirm their results, Dr. Baraban



Pictured: Annie and Megan Johnson

will need to repeat these studies in tissue obtained from other individuals with TSC and to compare it to samples from the temporal lobe from people who do not have TSC. As you can well imagine, these studies are not easy and tissue does not become available every day!

A special thanks to Annie for making history and for generously making a tissue donation that will benefit research on TSC in many laboratories. We received a tremendous response to the mailing asking individuals to register to become tissue donors receiving more than 200 responses from our members. There have been 15 tissue donations made since January 1, 2003, and more are being coordinated by the TS Alliance and the Brain and Tissue Bank.

If you are interested in signing up to be a tissue donor and/or have a surgery already scheduled and are interested in donating tissue, please contact the TS Alliance.

MANDY'S SPECIAL FARM

Jan Mintz

Our 25 year-old daughter, Dani, is severely affected by tuberous sclerosis complex (TSC). She is autistic, epileptic and although she is very smart, functions at the level of a three-year-old. I always knew that someday we would have to find a place for her to live but with her many issues and needs, who would look after her like I do?

In 2000, I happened upon an ad in a newsletter from the Autism Society of America. It was for a place called Mandy's Special Farm (MSF), a residential facility for young women with autism located in Albuquerque, New Mexico. My husband, David, and I flew from Los Angeles to meet with Ruthie and David Robbins, Mandy's parents and the farm's owners. They too had struggled with finding a living arrangement for Mandy that they felt met her needs. Through hard work, fund raising and donated services, they purchased four acres of land in their hometown of Albuquerque and built a beautiful home that could accommodate four to six people. What impressed us more than the

farm were the Robbins and their incredible vision. They want MSF to succeed because they know about the scarcity of good facilities and want to help families that struggle with this difficult decision.

Unfortunately, MSF was more than we could afford. Because it was out of state, we never thought to ask our social service agency to pay. In January, we met with them and mentioned MSF as our dream placement. As we gave details about the farm and why it would work so well for Dani, our worker said it was possible California could pay and that's exactly what happened. California's Department of Developmental Services contracted with MSF as a service provider and in April Dani moved in.

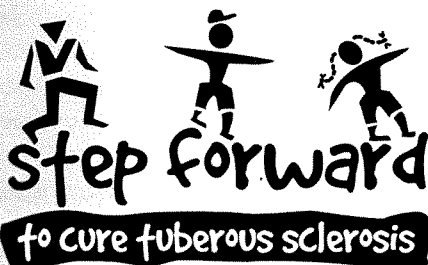
It's been a big adjustment, but I know Dani is happy. The enthusiasm in her voice when we call, the places she goes (the movies, bowling, weekly dances) and frequent talks with the staff all assure me that she is in good hands. We visit her monthly and are so impressed with how her behavior

has improved. She is benefiting from the slower pace, the wide-open spaces and being in her own house where people attend to her needs.

I share my story because MSF has an opening for another disabled young woman and my hope is to be able to help another family in the way that Ruthie and David have helped us. Information is on their Web site at www.mandysfarm.org. I would be glad to answer questions or correspond with parents at my e-mail address: spawn_boyz@msn.com.



Dani and friend



*We need you to step forward
and organize a walk-a-thon in
your community!*

The TS Alliance can walk you through the simple steps of organizing a walk. Organizers are provided with training materials, training calls, promotional materials and staff support. Join dozens of communities across the country this spring and raise money and awareness by taking a STEP FORWARD FOR A CURE.

Contact: Becky Bull or Nicole Engdahl (800) 225-6872

becky.bull@tsalliance.org or Nicole.Engdahl@tsalliance.org

ABCs: LESSONS FOR LIFE

It is estimated that in the United States 15% of the general population has some type of learning disability. Learning disability (LD) is the term used to describe a handicap that interferes with one's ability to store, process or produce information. Such disabilities affect both children and adults. Learning disabilities create a gap between a person's true capacity and his day to day production and performance. The Dictionary of Developmental Disabilities Terminology define LD as a "disorder in one or more of the basic psychological processes involved in understanding or using language, spoken or written; the disorder may manifest itself in an imperfect ability to listen, think, speak, read, write or spell." The definition of LD by the LD Association of America includes the effect of the disability on self-esteem, education, vocation and daily living activities.

TSC AND LD

Students with learning disabilities often find learning a difficult and painful process. Learning is a process of acquiring and retaining knowledge so it may be applied in life situations. Many children and adults with tuberous sclerosis complex (TSC) have learning disabilities. It's estimated that approximately 60% of individuals affected by TSC have some learning impairment ranging from mildly to severely affected. Often these students who have been diagnosed with TSC and a learning disability feel overwhelmed, disorganized and frustrated.

Homework and learning in general becomes a nightmare for children and adults affected by TSC and LD

when there are memory problems, difficulties in following directions, trouble with visual or auditory perception of information and an inability to perform simple paper-and-pencil tasks. Aside from the already identified challenges many of these same individuals have some form of a language disorder. More than one million students served in public special education programs have a communication disorder diagnosis. This number doesn't account for the children who have speech and language delay secondary to another disorder such as, but not limited to, TSC, Autism Spectrum Disorder or mental retardation.

ABC'S FOR LEARNING

Notwithstanding the difficulties that students with learning disabilities often experience with learning, they have the same needs as their peers who are not disabled, to acquire, skills and strategies, both academic and non-academic, that are necessary to function independently or semi-independently on a daily basis in our society. There are many practical interventions that are effective in the classroom and that will assist in preparing the student for life as an adult.

Because of the nature of learning disabilities, students need to become strategic learners; Teachers can be helpful in this regard. They can teach students specific strategies and demonstrate when and how the strategies are used. Teaching strategies can be simple or complex depending on the student's skills and strengths.

The majority of teaching methods recommend that a multi-model

approach be used for optimum success. For students with learning delays and TSC the multi-model method is frequently used to maximize their learning experience.

The basic classroom environment for students with more severe learning difficulties should do the following:

Seat the student in areas free from distraction

Eliminate all unnecessary materials from the student's desk to reduce distraction

Use a checklist to help the student get organized

Understand that break time is a necessity

Reduce visual distractions in the classroom

THE MASTER PLAN FOR TODAY & TOMORROW

All teachers know students who constantly fidget, who don't finish homework assignments, who never know what page the class is on or just don't seem to understand what is expected of them. Individuals with learning disorders are often told they are lazy or they just didn't try hard enough. The burden is on the teachers to ensure that the classroom environment doesn't perpetuate learning failure. This is not to say that all the responsibility falls on the shoulders of the educational system. Parents and caregivers play a critical role in how their children will be taught. The Individual Educational Program (IEP) should identify how each skill/goal will

be taught and reached. It's the responsibility of the parents to have a master plan for their child with disabilities.

The master plan helps parents and educators stay focused, anticipate future problems and to be prepared for the future. The master plan includes goals for the child in academic and non-academic areas including hobbies, sports, play and friendships. It's vital that the master plan is clear, focused, concise, flexible and attainable. A master plan should have the following basic elements: a vision and mission statement, goals, strategies and timelines to reach the goals. The elements of a master plan can change as the child acquires new strengths and skills or is unsuccessful in mastering a goal. The key words in a master plan are flexibility and attainability.

SOCIAL RELATIONSHIPS

Social relationships are critical to a student's quality of life and are a major determinant of whether the student will be a valued participant in his/her community. When schools deliberately assist students with and without disabilities to have social relationships, they engage in a powerful strategy for overcoming the negative effects of a disability. The most promising practices are education in the least restrictive environment and participation in the community. The most significant barriers for preventing social relationships are segregation and categorical grouping of students.

If a child has the opportunity for developing positive social relationships while in school, it is believed that she or he will have close relationships with peers, professionals and community support staff and positive interactions with family members as an adult. The

secondary gains will include, but are not limited to, cooperation with peers and teachers, getting and keeping a job and successfully negotiating participation in the community activities that are a part of daily life.

EDUCATIONAL MODEL

Social context has become an overriding framework for the attainment of both social and academic competence for children. Few if any skills are performed without reference to their social context. Children's mastery of critical life skills is integrally connected to their relationships and interactions with others.

Inclusion in life at the school and in the community throughout the developmental period also relates to positive outcomes for children with and without disabilities. School inclusions that effectively promote positive interpersonal outcomes include the following:

Quality inclusive schooling in which the child with a disability receives an appropriate education focusing on his/her IEP goals

Peer support within the general education structure

Circles of friends and collaborative problem solving in which children themselves are empowered and supported

Integrated therapy approaches that entail goals and provide therapy services in the context of naturally occurring routines

Cooperative and collaborative goals and activities that reflect children's multiple intelligences, their diverse learning styles, strengths and needs

STUDENT INVOLVEMENT IN PLANNING & DECISION-MAKING

Some of the most visible efforts to promote self-determination are those that involve students in the planning and decision-making process. For students with less severe disabilities there are models that employ self-knowledge, decision-making skills, educational goal setting, effective communication and leadership skills. For students with more severe disabilities there are models in which students, family members, professionals and others work through a process to identify goals, resources and obstacles to achieve desired outcomes. Based on this information, the student, supported by the group, develops an action plan across all areas of life.

CONCLUSION

Being a parent is a lot of responsibility; being a parent of an individual with a disability is even more responsibility. You can learn from other parents, contact community advocacy organizations for information and support, elicit independent educational consultants when possible, know your child's strengths and needs, join support groups, work as a partner with the educational system and sign onto the parent list serve at www.tsalliance.org. You don't have to feel alone on the journey; there are many who have started on the journey before you and many will follow after you. Reach out for guidance and support and offer your experiences to others. The TS Alliance is here today and will be a support system for you in years yet to come. We are part of your family now.

GRANDPARENT'S VOICE

THE CARTERS' STORY

Rebecca Cutbirth
Texas Scottish Rite Hospital for Children

"What Nicole Carter has given to this family! That little smiling face has taught us so much," Margaret Carter, one of Nicole's grandmothers, says proudly. Bitsy Carter, Nicole's other grandmother agrees. "She has brought our families together. As much as we wish that she didn't have tuberous sclerosis complex (TSC), the love that has grown in our families because of it has been such a blessing."

Nicole Carter's grandparents, Margaret and Syd Carter and Bitsy and Harold Carter, marvel at Nicole's energy level. "She is a go-er," says Bitsy. "She loves to go places and do things." Nicole has her own closet at Bitsy and Harold's house filled with books and toys, and she heads straight for it when she walks in the door. "We call it 'Nicole's Room.' She loves to pull everything out and play on the floor," Bitsy says. Margaret and Syd often take Nicole to their lake house in East Texas. "She just adores going to that lake. Swimming, riding on the boat and jet skis, jumping on the trampoline—she loves all movement." The Carters believe that sharing time like this with Nicole is the best way to show her how much they love her.

The Carters have learned to celebrate these little miracles. "We have come such a long way," says Bitsy. When Nicole's pediatrician first told the family that he thought she had TSC, they had never heard of the disorder. Margaret recalls that she called five doctors across the country that night, trying to find out more information. "From that first moment of hearing Nicole's diagnosis, we

have felt so blessed to have so many resources. But what about a family that has no one to call?" The Carter family realized that there were families everywhere facing this same situation with nowhere to turn. That was the family's motivation for getting the word out. "We just couldn't let this happen to another family."

The Carters knew that they wanted to do something, but did not know exactly where to begin. Margaret clearly remembers the day her son, Dodge, called and said, "Mom, don't get too settled in your rocking chair! I have something I need you to help me with." The family's first step was contacting the Tuberous Sclerosis Alliance for guidance in coordinating an event. "We were on the phone with the TS Alliance constantly," Margaret says. "They gave us leads for sponsors in Dallas and so much guidance." Bitsy adds, "The TS Alliance gave us the mental and emotional support and encouragement that we needed. They really made the extra effort." Working tirelessly and enlisting the support of friends and family members, the Carters created Treasure Street in 1996 as a silent auction. They sold friends' and relatives' donated "treasures" to raise money and awareness for TSC and Texas Scottish Rite Hospital for Children. "We dreamed of making \$20,000 that first year," remembers Bitsy. The event received an overwhelming response and raised more than \$150,000.

Over the past eight years, Treasure Street has raised more than \$2.6 million for the Tuberous Sclerosis Clinic at Texas Scottish Rite Hospital for Children and helped provide research dollars for TS Alliance funded researcher, Dr. Hope Northrup, who was instrumental in developing the standardized testing for TSC, adopted by Athena Diagnostics, which is now



Pictured are (standing from left to right) Dee Carter, mother; Bitsy Carter, grandmother; (seated from left to right) Nicole Carter, age 10; and Margaret Carter, grandmother.

commercially-available to identify genes in affected patients. Despite the event's growth over the past years, Bitsy says that its spirit and focus have remained the same. "Treasure Street still has that same loving atmosphere that it had the first year. Everybody is a friend of somebody's and you can feel that love."

The Carters agree that they have received immeasurably more from Treasure Street than they have given. "Having a grandchild with TSC is a double-hurt," Margaret says. "We see our children hurting over their child hurting." The Carters have discovered that in working to raise awareness of the disorder and generating support for other families, they have found a source of personal satisfaction and joy.

The Carters have embraced their role of supporting their family in coping with TSC and their love has made such a difference in Nicole's life. "We encourage other grandparents to get involved," advises Bitsy. "Be there for your family. You are not only helping your grandchild, but also your child."

The Carters' involvement in the TSC community has touched the lives of so many people and provided a source of enrichment in their own lives. "We couldn't have been luckier," Bitsy says confidently. "We haven't had to do this alone."