RTS News

A newsletter for families living with Rubinstein-Taybi Syndrome around the world. Supported by the Division of Developmental and Behavioral Pediatrics, at Cincinnati Children's Hospital Medical Center, and the Jack H. Rubinstein Foundation.

Spring 2009

In This Issue . . .

Newsletter Advisory Team

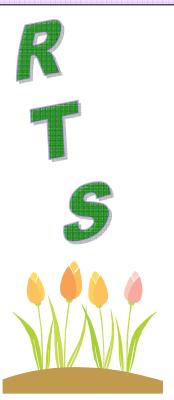
Reasons to Smile: A Dad's Perspective

Regarding the Syndrome: Genetic Testing

RTS-OKI Support Group

Resources to Share

- * RTS Brown Book
- * NORD Rare Diseases Day
- * Financial Planning for Special Needs
- * Travel Tips Planning Ahead



Newsletter Advisory Team

By Amy Clawson

Welcome Spring! This is the third issue of RTS News. So far, we have included information suggested by our Newsletter Advisory Team. I would like to share with you who they are and my appreciation of them.



The Newsletter Advisory Team consists of: Terri Burns, RTS-OKI Parent, Rebecca Riedel, RTS-OKI Parent, Jennifer Hans, RTS-OKI Parent, David Schor, MD, RTS Program Director, DDBP, and myself.

The creation of RTS News has been a true collaboration. During each planning meeting, phone call, or email, these parents on the Advisory Team share openly and honestly the focus of RTS News articles. Their perspectives are valuable to the success of this newsletter, and we in DDBP are very thankful for their time, willingness, and availability to share their insights. Most of all, because of their partnership, many families all over the world will benefit and feel connected to others living with Rubinstein-Taybi Syndrome.

Thank you to the RTS Newsletter Advisory Team! We look forward to a terrific year!

Additionally, for more information about the RTS-OKI Family Support Group, including the meeting schedule, or about the RTS Program at Cincinnati Children's Hospital, or visiting Cincinnati, please contact me at 513-636-4723, 800-344-2462 ext. 64723.

or amy.clawson@cchmc.org.



RTS Reasons To Smile: A Dad's Perspective

By Christopher Weitfle



Sophia Weitfle came into this world under the funkiest of circumstances: mv wife Maria had gone in for a routine visit with her OB/GYN, which should have been the last during her otherwise normal and uncomplicated pregnancy. I

was at home preparing a birthday dinner for the family, but instead I was called to meet her at the hospital, where she was rushed to induce labor. Our daughter was going to be born that day, three weeks early; it happened to be my birthday, and it happened to be a Friday the 13th, in August of 2004.

Now, I'm certainly *not* a superstitious person, and I can't quite subscribe to the belief that everything happens for a reason, but hours later, when I stood over her bassinet at Cincinnati Children's Hospital, alone, my throat sore and eyes wet with tears, I had a feeling of knowing that this baby came to me because somehow, somebody knew this Daddy was in place, ready to take care of her and give her a good home.

I was already that Daddy. Before she was born, I had committed myself to raising our other child, Elias. After his birth, I left my teaching job and became a full-time stay-at-home dad. It was the dream job I had always wanted, and it had been going well for him and me. We had an adventurous life and everything was sunshine and rainbows. And then the most unexpected birthday present was delivered to me, and life became a lot more

precipitous.

Those first few days with Sophia were the hardest my wife and I have ever experienced together. Before the clock had marked her first hour of life, Sophia was transported to the RCNIC unit at Cincinnati Children's Hospital, where she would remain for the next 30 days. But if there is a silver lining to that gray cloud, it is that we were fortunate to live so close to such a world-class institution, just ten minutes away in the neighborhood of Oakley. Just as fortunate, too, that we had practically landed in the lap of the man after whom her condition was named.

After genetic testing that first weekend, Sophia's diagnosis of Rubinstein-Taybi Syndrome was confirmed by the bedside appearance of Dr. Rubinstein himself, who had heard of her birth and came to visit us in the RCNIC. She was a classic case: eyes, nose, fingers, toes, but her most striking feature was her full head of thick black hair. Beauty aside, she had her share of congenital defects too, and to list them here would be tedious for the reader and painful for the author. Suffice to say, she was a complicated little girl!

As soon as Sophia was home, my life as Dad became much more complicated too: I was now taking care of a medically-fragile infant as well as an 18-month old toddler. The fantasies I'd had of being the "Super Dad" were tempered by harsh reality; I had to shed that cartoonish ideal and instead step up to be the more heroic everyday Dad. This little girl had come to me, for whatever reason, and I had to do everything possible to keep her safe, help her thrive, and allow her to grow up in the most normal way possible. (I later shed *that* ideal too; there was never going to be anything considered "normal" unless one used the word in a sentence like: "Oh, you're going out of your mind? *That's* normal.")

Fast-forward four years —four long and trying years— and Sophia *still* has that full head of thick

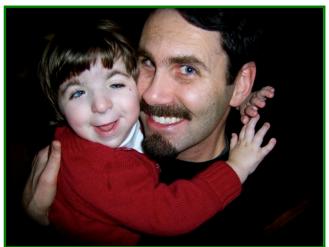


hair. But where we have been, and how we have arrived at this place, has been a series of enormous challenges and small victories. Some of her insurmountable difficulties have passed like water under the bridge, and it seems like such a far-away time when we remember... how we had to snake 100 feet of vinyl tubing from oxygen tanks on the first floor up the stairwell to her crib on the second floor... how we got so frustrated with filling bags with formula and programming a pump for nighttime enteral feedings... how she endured countless MRIs, CAT-scans, and ultrasounds... how it took her almost an hour to drink an ounce of formula without aspirating... And I remember when I thought she would never roll over... I remember when I thought she would never taste solid food... and I cried when I thought of her never walking, talking, or twirling on the dance floor with me... But I remember when I could clutch her little breathing body within my forearm, and soothe her to sleep, and put my fears to bed for one more night...

And now, how she has grown! She's feeding herself bottles of milk, and swooping up the curve on the growth chart. She is on her feet and clumsily cruising along, reaching out for any handhold she can find. She's developing a playful sense of humor, and reacting with gusto to peek-a-boo and tickles on her tummy. She is infatuated with her big brother, and generous in doling out bear hugs. She bangs the keys on her toy xylophone with as much enthusiasm as she bangs the kitchen cabinets. She is doing everything she wants to do, and everything she's able to.

We just wish she were able to do... more.

As I write and re-write this, I'm continually thinking of my relationship with Sophia and my role as a stay-at home dad, and how I can relate my tale from a "Dad's perspective." But I keep coming back to the same realization: that this relationship is not unique, despite the circumstances. Fathers and mothers want the same things for their children. We all want peace, love, safety, and good health for our children. We want to give them security, happy homes, and memorable



experiences. We want to give them every benefit so they can grow up to be good-natured, well-rounded, and independent people. Fathers and mothers the world over would likely agree to this. But for we parents who have children afflicted with RTS, sometimes it's hard to convince ourselves that what we're doing for our children is the same that any other parent would do. Because it's not.

It is different, much different, for parents of special-needs children. We have burdens that nobody seems to understand (or appreciate), and though it is true that there is *always* somebody who's got it much worse than you, you feel like you've got it the worst. (At least I do, from time to time — especially on sick days, when it is raining...) But there is a silver lining around that gray cloud, too, and it is the comfort of knowing that we are not alone.

Maria, Elias, Sophia, and I: we will always be a family, and I hope our foursome lasts forever. For whatever reasons, our extended families are small in number and scattered near and far, living their own lives. Our neighbors are sympathetic and yet unknowing of our challenges. Church gives us serenity and a temporary strength; and school is a playful respite wrapped up in endless bureaucracy. Who can understand what we are going through? Who can relate to the difficulties we face? Who can accept why we are restricted in our social or recreational pursuits? Sometimes, it seems for us that the only person to lean on for support is the one staring back from the mirror. But, it is going to



sound very trite when I say (speaking for myself) that the one place I have found the greatest solace and easiest camaraderie is within the walls of Cincinnati Children's Hospital Medical Center.

Here I have found physicians, nurses, specialists, and therapists who have all improved the course Sophia's life has taken. All have welcomed Sophia and me, and helped us understand the complicated panoply of conditions affecting those with RTS. It may seem overly gratuitous of me to say this, but throughout the past four-and-a-half years, when I felt like I had no one who understood my daughter, with nowhere to go for support, I knew there was always a "home away from home" ten minutes away at CCHMC.



Here, we have received all the support we could ever want, and if the services of this world-class facility were not enough, there is the icing-on-the-cake: the RTS-OKI Support Group. *It* has been the most beneficial of therapies, and immeasurably useful in helping us appreciate the variances of Sophia's condition. Among like-minded people facing similar challenges, I am able to hold my head up and confidently accept the role of parent of a child with RTS —and I get a collective shoulder to lean on. Here we all can share, discuss, even celebrate the challenges we face having sons and daughters affected by this rare syndrome. Amongst friends, the peculiarities of RTS fade away, and Sophia is Sophia, and I am her Dad.

...And where I once shied away from drawing attention to her "otherness", I now accept it as an irrefutable part of her being: yes, she has RTS, and yes, she is a special-needs child, but foremost she's still a *child*. And she's *my* child, my blue-eyed, birthday-present, Daddy's-little-girl with the oh-sotight squeezes and hugs, the hugs that seem to say:

"Hold on to me and love me and never let me go, because I came to you, and you were here for me."

Sophia Weitfle her brother, Elias mom, Maria, and dad, Christopher live in Cincinnati, OH.

RTS Reasons to Smile... RTS Reasons to Smile... RTS Reasons to Smile.....RTS Reasons to Smile



RTS Regarding The Syndrome: Genetic Testing for RTS

By Elizabeth Schorry, MD



Dr. Elizabeth K. Schorry is Associate Professor of Pediatrics in the Division of Human Genetics at Cincinnati Children's Hospital Medical Center. Dr. Schorry provides medical management, genetic assessment and genetic counseling for children with a wide range of genetic disorders and congenital anomalies. The article mentioned below is: Schorry EK, Keddache M, Lanphear N, Rubinstein JH, Srodulski S, Fletcher D, Blough-Pfau RI, Grabowski GA. **Genotype-phenotype correlations in Rubinstein-Taybi syndrome.** *Amer J Med Genet*

Advances in research have helped us to better understand the genetic cause of Rubinstein-Taybi Syndrome. We know that there are at least two different genes, called CREBBP and EP300, which

can cause RTS. There is likely at least one additional gene which has not yet been identified. Mutations, or changes in the chemical structure of the gene, cause the gene to malfunction and to subsequently cause the features of the syndrome. In the large majority of cases this is a new mutation in one of these genes in the egg or sperm cell which forms the child with RTS, with neither parent having the gene change. Current research shows that about 50 - 60% of people with RTS have mutations in the CREBBP gene, and a much small number (about 3%) have mutations in EP300. In the remaining cases, no gene change has yet been identified.

We recently published our data on genetic changes in patients with RTS in the American Journal of Medical Genetics (Volume 146A, 2008). We studied mutations in the CREBBP gene in 93 patients who contributed blood samples at the International RTS Family Conferences held in Cincinnati in 1998 and 2003. We found mutations in CREBBP in 52 of these patients (56%). We compared features of RTS in patients who had CREBBP mutations versus those who did not and found that overall the two groups were very similar. Some of the differences found were that patients with CREBBP mutations were more likely to have seizures, and patients without CREBBP mutations had lower height and weight compared to those in the CREBBP group. Further details are available in the article cited below for those who wish to read it.

Is genetic testing now available for all patients with RTS?

Testing for the CREBBP gene is now available at several different laboratories including the University of Chicago and Gene Dx in Maryland. Testing will also be offered at Cincinnati Children's Hospital in the near future. Sequencing of a complete gene such as CREBBP is very time-consuming and costly, and costs currently range from \$3,000 to \$5,000 per patient. In most cases insurance will cover the testing unless your policy has a clause excluding all genetic testing or you have a very large deductible. It is always a good idea to check with your insurance company before proceeding with the testing.

Reasons to consider genetic testing

Not every patient with RTS needs to have genetic testing. For those in whom the diagnosis is certain and has been confirmed by a physician experienced with RTS the testing may not be necessary. However, testing can be very helpful for those whose diagnosis is not completely certain or if there is concern about a parent or other family member also having features of RTS. It is important to remember, though, that a major limitation of the test is that it only finds CREBBP in about 50% of patients with a known diagnosis of RTS. Therefore, a negative result (no mutation found) does NOT mean that the patient does not have RTS. It is



also important to know that sometimes minor changes (variants) can be found in the CREBBP gene which are not the cause of RTS. Sometimes blood samples from both parents as well as the affected person may be requested in order to find out if a gene change is a true mutation or instead a normal variant than can run in a family.

What if I donated blood to the research study in 1998 or 2003?

If your child or family member with RTS donated blood to the Cincinnati Children's Hospital research study in 1998 or 2003 and you would like to know if a mutation was found, you can contact Dr. Elizabeth Schorry (not to be confused with Dr. Schor!!) at 513-636-4760. If we identified a mutation, we will be glad to share that with you. However, we always recommend that research testing be followed-up by testing at a clinical lab.

Additional information from Dr. David Schor, RTS Program Director, DDBP, Cincinnati Children's Hospital Medical Center:

Dr. Schorry is lead author of a medical paper published late last year describing changes in a specific gene found in some children who attended RTS conferences in Cincinnati. In the accompanying article, she addresses the current understanding regarding the genetics of RTS and various questions surrounding testing. She points out testing is helpful in some circumstances but also has limitations. Please contact her for further information regarding your child's testing.

If you would like a copy of Dr. Schorry's RTS article (Genotype-phenotype correlations in Rubinstein-Taybi syndrome. *Amer J Med Genet* 2008;146A:2512-9), please contact me.

If you are considering testing, I recommend going through a regional genetics center in order to have current recommendations about the testing and where to send the blood specimen.

RTS Regarding the Syndrome... RTS Regarding the Syndrome...RTS Regarding the Syndrome



RTS-OKI

2009 Family Support Group Meeting Schedule



Saturday, March 14 Saturday, June 13

Saturday, September 12

Saturday, December – CCHMC's Special Santa Day – tbd

11:30am Social, Pizza, Check-In Noon – 2:00pm

at Division of Developmental & Behavioral Pediatrics Cincinnati Children's Hospital Medical Center 3333 Burnet Ave., Location E. 4th Floor, Room 353

This group is for any child who has Rubinstein-Taybi Syndrome (RTS), and their family members and caregivers who live in Ohio, Kentucky, Indiana, or other nearby states. Would you like to meet other families and talk with those who understand life with RTS? You and your entire family are welcome to attend!

Childcare is available with pre-registration.

To register, please contact Kari Edwards at 513-803-0052 or 800-344-2462 or email kari.edwards@cchmc.org.

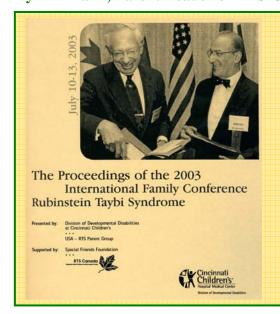
These events are sponsored by RTS-OKI with support from the Division of Developmental and Behavioral Pediatrics, at Cincinnati Children's Hospital Medical Center.

For more information about Rubinstein-Taybi Syndrome or RTS-OKI please contact Amy Clawson at 513-636-4723 or amy.clawson@cchmc.org, or Bill Mann at 513-708-5611 or bmann@theppsgroup.com.



RTS Resources to Share

What is the RTS Brown Book? By Bill Mann, Parent Leader of RTS-OKI



The "Brown Book" is the common name for the 2003 international RTS conference proceedings. It is a book of all the medical articles and studies done on RTS for the conference. Most of the information is condensed in the RTS medical guidelines document that is available as a pdf and has been circulated on the RTS listsery.

The Brown Book (cleverly named for the color of its cover) is not available in digital form. However, families can request a free copy of the Brown Book through Amy Clawson at Cincinnati Children's Hospital via email: amy.clawson@cchmc.org

Global Rare Disease Day!

The National Organization for Rare Disorders (NORD) organized an observance of rare diseases on February 28, 2009 in the United States. The ultimate goal is to have global rare disease day for the last day of February each year! Families, parent organizations, medical societies, and advocates were asked to partner with NORD in raising awareness for rare diseases, research, treatments and cures. Many states, including Kentucky and Indiana, wrote letters asking their governors to issue a state proclamation for Rare Disease Day. For more

information about NORD and the U.S. Rare Disease Day on the NORD website, please visit:

www.rarediseases.org/rare disease day/rare disease day info or the Global Rare Disease Day website: www.rarediseaseday.org.

Mark your calendars for February 2010!



Financial Planning for Those with Special Needs

Provided by the Special Needs Resource Directory at Cincinnati Children's Hospital http://www.cincinnatichildrens.org/svc/alpha/c/special-needs/resources/estate.htm

Estate Planning

Parents and other family members often want to put money aside to provide lifetime care for their child with special health care needs. As parents age, it may be more difficult for them to provide care. In addition, the individual with a disability may want to be more independent. Having assets in the name of the child with special health care needs, regardless of their age, often disqualifies them from important government programs including Medicaid and Supplemental Security Income (SSI) that would provide many needed services. While it is natural to want to leave everything to your child with disabilities, having more than \$2000 in the child's name can eliminate many government benefits.

Luckily there are special estate planning techniques that can be used to protect your child from losing government assistance.. As part of estate planning, families should look into establishing special needs trusts and / or leaving funds to a trusted family member or friend to be used only for the care of the child with special needs. If done properly, these could still provide the necessary funds to help the individual without disqualifying them from government programs. Funds may also be used earlier in the child's life for such needs as home or vehicle modifications without affecting eligibility for state programs. They can pay for things not covered by government benefit programs, including entertainment, trips and vacations, computer equipment, education, recreation and other items

that can add to the individual's quality of life. Any approach should be carefully discussed with a knowledgeable attorney to see what would work best in your situation.

Letter of Intent

It is often recommended that parents take the time to decide what kind of future life they would like for their child and then describe it in a letter of intent to be included in their estate planning documents. The letter can address their hopes and dreams and reflect what they want for their child in all of the major life areas including living arrangements, child's likes and dislikes, recreation, social and community interests. While not legally binding, the letter can help the future caregiver gain an understanding of the family's vision. The letter of intent can include:

- Medical history of the child including doctors, allergies, medications and any other health related information
- Housing preferences
- Daily needs, life skills and personal care needs
- All About Me Form can help share information about the child's likes, dislikes, activities, friends and special interests
- Hopes and dreams that the family has for the child's future

Share the letter with family members and trusted friends to help build support around your vision. Review the letter on a regular basis and keep it updated to reflect your child's needs and your changing goals.



Special Needs Trusts

When set up properly, a special needs trust can provide the funds to supplement, not replace, government funded benefits. It is critical that you discuss your options and set up a special needs trust with a qualified attorney. The special needs trust provides the ability to pay for items and services beyond the basic necessities of medical care, room and board, employment, basic personal needs, care and supervision funded by government benefits programs. Funds from the special needs trust can be used to provide enrichment, such as recreation, trips, electronic equipment, gifts, medical / dental expenses not covered by Medicaid, and other personal expenses. For example, if your child's computer or television broke, money to purchase replacements could come from the special needs trust. If your child enjoys going on vacations, this too could be paid for by the trust.

- Exceptional Parent Magazine offers online publications about financial planning and legal considerations for the child with special needs.
- Federal Citizen Information Center publishes Planning for Your Special Needs Child, an online pamphlet produced by the MetLife Consumer Education Center.

While government agencies recognize special needs trusts, they have developed complicated rules and regulations. Special expertise is required in estate planning because laws affecting individuals with disabilities can change frequently.

This is why it is critical that you consult an attorney experienced in estate planning for families who have a child with special needs, not just one who does general estate planning. One wrong word or phrase can make the difference between an inheritance that really benefits the person with a disability and one that causes the person to lose a wide range of needed government services and assistance.

Don't be afraid to interview special needs attorneys who have a thorough understanding of your state's disability laws. Ask questions before deciding who would be a good match for your individual situation:

- Is special needs the main area of the practice?
- Does the attorney have a proven record of designing and implementing special needs trusts?
- How many special needs trusts has the attorney written?
- Is the attorney thoroughly knowledgeable about current government benefit programs including SSI and Medicaid?
- Is the attorney familiar with recent developments and changes in disabilities laws and regulations?
- Does the attorney have a strong understanding of the language used by special needs professionals?

Consider asking other parents for references or check with local advocacy groups, such as Easter Seals or Arc, before making a selection.

RTS Resources to Share.... RTS Resources to Share.... RTS Resources to Share.... RTS Resources



Vacationing and Travel Tips – Plan Ahead

Provided by the Special Needs Resource Directory at Cincinnati Children's Hospital http://www.cincinnatichildrens.org/svc/alpha/c/special-needs/resources/transportation.htm

With spring break and summer right around the corner, it is important that families of children with special health care needs make preparations well before traveling to make sure that their child receives the proper care should acute problems arise. Plan ahead and keep these tips in mind to help make travel easier:

- Disability Placards can be used in other states and in rental vehicles.
- Travel with a detailed and current medical summary, plan of care and contact information on all providers, include an Emergency Medical Services form and an updated Patient/Family Care Notebook
- Check with your physician for the necessary precautions, immunizations and / or medications for your destination.
- When traveling by plane, keep your medications in your carry-on bag for easy access during the flight and to protect against loss if your luggage doesn't arrive. This will also prevent exposure to extreme temperatures in the baggage compartment, which can reduce the effectiveness of some medications.
- People with special needs have the right to board the airplane before other travelers.
- If you check durable medical equipment such as a wheelchair with baggage, make sure it is marked with your name, address and cell phone and check in thoroughly for damage before leaving the airport.
- Be prepared to provide airport security with a copy of your prescription if you use medication that requires a syringe.

- Carry your pharmacy's phone number and list of your prescriptions in case you lose your medication.
- Keep your medications out of direct sunlight if you are traveling in a hot, humid climate.
 Excessive heat can reduce the effectiveness of some medications. Avoid storing medications in the glove compartment of your car.
- Take along more medication than the number of days you plan to be away so that you will be prepared for unexpected delays.
- Call your insurance providers to clarify what coverage they have when traveling out of your network area and be specific in relating your itinerary, especially if you are traveling out of state or out of the country. Ask for written documentation and record the insurance representative's name, department and extension.
- Obtain a list of the nearest appropriate hospitals, physicians and durable medical equipment (DME) contacts for all cities / regions you are visiting. Bring an adequate supply of medications, special nutritional items and equipment before leaving.
- Thoroughly investigate all aspects of your trip to make sure that your child's disability can be accommodated, including barrier-free hotels, ADA compliance, accessible bathrooms, etc.
- Many children with special health care needs have difficulty with travel and need frequent stops as well as an ample supply of snacks, games, toys and other distractions.
- Many beaches now offer accessible wheelchairs as part of their rental choices.
 These "all-terrain" wheelchairs can be pushed in the sand and can often be delivered to your hotel room.

RTS Resources to Share.... RTS Resources to Share.... RTS Resources to Share.... RTS Resources



David Schor, MD, MPH RTS Program Director

Division of Developmental and Behavioral Pediatrics Cincinnati Children's Hospital Medical Center MLC 4002, 3333 Burnet Ave.

Cincinnati, OH 45229

800-344-2462 ext. 0541or 513-803-0052

Email: david.schor@cchmc.org

Amy Clawson, Family Support Coordinator

Division of Developmental & Behavioral Pediatrics Cincinnati Children's Hospital Medical Center MLC 9500, 3333 Burnet Ave.

Cincinnati, OH 45229

Toll Free: 800-344-2462 ext. 4723, 513-636-4723

Email: amy.clawson@cchmc.org

RTS-OKI Support Group Bill Mann, Parent Leader

513-708-5611 or Email: bmann@theppsgroup.com

RTS Email ListServ

Janet Estes, ListServ Manager

250-753-7795 or Email: janetestes@telus.net

RTS Parent Group USA Lorrie Baxter, Coordinator

P.O. Box 146

Smith Center, KS 66967

Toll Free: 888-447-2989 or Email:

lbaxter@ruraltel.net

Rubinstein-Taybi Syndrome Website:

www.rubinstein-taybi.org

Special Friends Foundation Website:

www.specialfriends.org

Michelle Farrell, Director of Family Services

P.O. Box 313

Windham, NH 03087

Toll Free: 866-316-9029 or Email:

mfarrell@specialfriends.org

Dr. Cathy A. Stevens

Dept of Pediatrics

T. C. Thompson Children's Hospital

910 Blackford St.

Chattanooga, TN 37403

To receive a copy of the blue booklet "Rubinstein-Taybi Syndrome A Book for Families", please

contact Dr. Stevens.

Save the Date!!

Annual North East Family Reunion

When: March 21st, 2009 Where: Hilton Garden Inn

> 5015 Campbell Boulevard Baltimore, MD 21236

For more details contact:

Brenda Bon Levine lev306@comcast.net 443-829-4507

To apply for a Disability Parking Placard, go to: www.dmv.org



for the Department of Motor Vehicles in your state.

RTS Resources to Share.... RTS Resources to Share.... RTS Resources

