

# IN RARE FORM



Amanda Pitre

## My Body is Tired of Fighting But My Mind Isn't

by Jenny Picciotto

Jim Broatch, Executive Director of RSDSA, recently introduced me to Amanda Pitre, a young woman who developed Complex Regional Pain Syndrome (CRPS) when she was sixteen. After completing an inpatient program at Children's Hospital in Hartford, she discovered that her hometown of Milford, Connecticut was also home base for RSDSA. Since then Amanda has been active with RSDSA events such as the Achilles Walk in New York City, and growing community through Young Adult Weekends.

Currently living with her parents in a duplex apartment and working part time as an aide for children with special needs, Amanda has her sights set on the future. Her goal is to save enough money to purchase and modify a home where she can live independently. Monies raised through her Go Fund Me page and recycling drives are shared with RSDSA. This year she has donated over \$1,000, which is earmarked for Pediatric Pain Week 2022 at the Center for Courageous Kids Camp (CCK) in Kentucky.<sup>1</sup>

Our conversation took place over Zoom and has been lightly edited. We began our discussion with the onset of her symptoms.

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“I was sixteen when everything started”, she told me, “but was in a wheelchair for a whole year before I was correctly diagnosed. By then I couldn’t walk. Being misdiagnosed two or three times didn’t help.”

While most people with CRPS can point to a surgery or injury as an inciting event, Amanda’s symptoms came on without warning. She developed leg pain, which her mother attributed to “growing pains.” The pain persisted and progressed until she “woke up one morning and couldn’t move from the waist down.”

It wasn’t until she was referred to Dr. Neil Schechter, Director of the Pain Relief Program at the Connecticut Children’s Medical Center in Hartford, that she was diagnosed and started a program of intensive physical and occupational therapy.

Unfortunately, it is common for people with CRPS to see several doctors before being diagnosed. Lack of experience with the condition among medical professions and ruling out differential diagnoses can contribute to delays in treatment.

According to **Pediatric Complex Regional Pain Syndrome: a Review:**<sup>2</sup>

“Musculoskeletal pain is the most common reason for referral to pediatric rheumatologists. Causes of chronic musculoskeletal pain include a wide variety of inflammatory or non-inflammatory conditions, such as arthritis, hypermobility, fibromyalgia, growing pains, complex regional pain syndrome (CRPS) and more.”

Dr. Pradeep Chopra, M.D, Assistant Professor (Clinical) at Brown Medical School, and Director of the Interventional Pain Management Center of Rhode Island, emphasized the following recommendations at his presentation at the RSDSA conference in Denver Colorado, 2019:<sup>3</sup>

- Start treatment immediately, even if you suspect CRPS
- Must be evaluated by a Pain Medicine specialist or a physician who is very familiar with it, to start appropriate therapy
- Multidisciplinary approach -teamwork

When she was seventeen, under Dr. Schechter’s care, Amanda spent two months in residence at Children’s Hospital in Hartford. The pain management rehabilitation program included twice daily physical therapy and occupational therapy sessions, complemented by psychological counseling once a day. It was tough.

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## RSDSA's 2nd Annual Virtual CRPS Awareness Walk



RSDSA is very excited to announce that we will be holding our [2nd Annual Virtual CRPS Awareness Walk](#) this year on **Saturday, June 26th, 2021!**

Last year we had 33 teams with participants from 40 states and three countries. Our goal was to raise \$30,000 and we far exceeded this by raising 142% more!

Unfortunately, there is still no cure for CRPS even though the condition has been described since before the Civil War. Living with a chronic disease, and constant pain, is daunting, but to see everyone come together to raise

awareness for this rare condition is very touching. Seeing the support from so many reminds CRPS warriors to keep fighting!

The money raised helps RSDSA support the CRPS community in many ways including:

- Hosting our ongoing Facebook Live series to learn about the latest therapies and treatments.
- Hosting a virtual conference to hear information from researchers and clinicians and allowing time for CRPS Warriors to network and connect to others battling the same condition so they do not feel like they are going through this alone
- Supporting this new monthly mini newsletter that updates you on current research, news, CRPS initiatives, and special events.

We are hoping to make this year even more successful with your help!

We cannot wait to see what amazing things happen when the CRPS community bands together to raise awareness this June!

You can register as an individual or you can make a walk team. Registration is \$21 for adults and \$10 for children. Personalize your walk page and share it with family, friends, neighbors, and colleagues. Each U.S. and Canadian registrant will receive an RSDSA CRPS Awareness Walk medal.

You can also post your Walk page to Instagram or Facebook and create a fundraising page. Be sure to join us as we walk in our communities for RSDSA on Saturday, June 26, 2021 and learn more at [rds.org/walk](https://rds.org/walk).

## RSDSA's Advocacy Committee Wants to Hear From You

Our Advocacy Committee started the month of March by participating in the RDLA program with [Rare Across America](#). The program culminated in virtual meetings state by state with our Senators or their aides and our Congressmen to push for support for rare disease legislation.

Jim Doulgeris and Nancy Shurtleff, members of the RSDSA Advocacy Committee, were asked to lead their state delegations.

The advocacy committee needs your help, as we want to hear from the community:

1. Did you write to your Congress, Senate or House?
2. Did you have appointments with your representatives?

If yes, the Advocacy Committee would like to hear about your experience.

Please email [advocacy@rsds.org](mailto:advocacy@rsds.org), to tell what state you represented and who you met with.

Thank you,  
Jeri, Jim, Megan and Nancy

## Create an Instagram Fundraiser for RSDSA

Instagram now allows all users to launch fundraisers from their profiles!

When posting a photo or video to your Instagram profile, you'll now see an "Add Fundraiser" option under the space where you write your caption. Once you select that option, you'll be taken to another page where you can search for "RSDSA" and add our donation information to your post.

From there, users will then have the opportunity to set a goal amount for the fundraiser (just like on Facebook). Additionally, a line stating that you are raising money for RSDSA will show on your Instagram profile for 30 days. [Learn more here](#).

## RSDSA Game Night

RSDSA is hosting a game night with fellow Warriors on Tuesday, March 23rd at 7p Eastern. Keep an eye on our website, newsletters, and social media channels for details on how to join us!

# National Economic Burden of Rare Disease Study

A landmark new study by the EveryLife Foundation for Rare Diseases has released a new study on the estimated economic cost of rare disease in the United States. The *National Economic Burden of Rare Disease Study* estimates the economic cost at \$966 billion in 2019. This is the most comprehensive study of the economic cost of rare disease. Annie Kennedy, chief of policy and advocacy at EveryLife Foundation said, “Based on this data, rare diseases represent an urgent public health crisis that demands additional research, enhanced awareness, and improved access to diagnosis, care and treatment.”

The study included 379 rare diseases, although there are estimated 7,000 rare diseases total. The study was for one year, 2019. The report found that incurred direct medical costs account for \$418 billion. These costs include inpatient hospital care or outpatient care, physician visits, prescription medications, and durable medical equipment. The total also includes indirect or non-medical costs at an estimated \$548 billion. Some of the indirect costs which were not included in previous studies include lost productivity in the workplace and lost social productivity like community participation and volunteer work.

For the first time, the Rare Disease community has quantifiable economic data to show the devastating impact of rare disease on our community. We can show that appropriating money to focus on treatments and cures for CRPS is a fiscally responsible action. The data in this study just brings to light what was already known by the rare disease community, i.e. that costs of rare disease is devastating to individuals, families, caregivers, and society.

The *National Economic Burden of Rare Disease Study* was sponsored by EveryLife Foundation. EveryLife Foundation commissioned the Lewin Group, a company that provides health care and human policy research, analytics, and consulting to clients. It is part of OptumServe and UnitedHealth Group family of companies.

Funding for the study came from Alexion Pharmaceuticals, Amicus Therapeutics, Argencx US, Inc., AVROBIO, Chiesi Global Rare Diseases, Enzyvant Therapeutics, Gentech, Mallinckrodt Pharmaceuticals, PhRMA, Pfizer Inc, REGENXBIO Inc, Sanofi Genzyme, Sarepta Therapeutics, Spark Therapeutics and Travele Therapeutics, Inc.

For more information about The *National Economic Burden of Rare Disease Study*, like methodology, Study contributors and Study limitations, visit the study website at [EveryLife Foundation](#).

# Mast Cell Activation Syndrome and Its Role in Pain

## What are mast cells?

Mast Cells are normally found in our body. They are an important part of the initial defense system. These cells are found in most body tissues and tend to accumulate in tissues that have contact with our external environments, such as mucous membranes, the lining of airways, intestines, skin, and bladder lining, as well as being within the spine and brain. Mast cells contain many granules rich in heparin and histamine. When mast cells are activated they release compounds (also known as mediators) which cause inflammation (redness, swelling, pain). So, for example, if a person was exposed to a trigger (infection, allergy causing agent) or injury, the immune system of the body activates mast cells. These mast cells then head over the affected body part and release mediators that cause inflammation, resulting in pain, swelling and redness.

## What is MCAS?

Mast Cell Activation Syndrome (also known as MCAS) is a condition where mast cells are inappropriately activated, leading to a wide array of symptoms. In MCAS, these mast cells start misbehaving. The inappropriate activation of mast cells causes persistent inflammation that is widespread in the body. It is important to note that the number of mast cells are not increased in MCAS. There is a condition called Mastocytosis where the number of mast cells are increased. MCAS and Mastocytosis are two completely different conditions. This article is about MCAS only.

## Signs and Symptoms

MCAS can present differently in people. It is essentially a painful condition because it causes inflammation of tissue. Some of the symptoms are:

1. Rashes: unexplained, random rashes, patches of redness
2. Itching: unexplained itching in random areas of the body. They may complain of an itching sensation deep inside their body. Itching can be triggered by water (usually warm water), sun exposure.
3. Multiple chemical intolerances or sensitivities. They may be intolerant to perfumes, chemicals, foods.
4. Skin flushing after a shower
5. Temperature instability: patients report a sense of being either too cold or too hot.



6. Fatigue: the fatigue is very disabling. Patients report a feeling of 'flu-like' symptoms.
7. Sweating: they may have unprovoked sweating, especially at night
8. Gastrointestinal symptoms: may vary from loss of appetite, bloating, nausea, alternating diarrhea and constipation, belly pain.
9. Weight: they may have weight gain or weight loss or fluctuations in their weight without any change in life style.
10. Headaches
11. Acid reflux
12. Hair loss, dry hair
13. Dry eyes
14. Ringing in the ears

However, one aspect of MCAS that is far too often underestimated is the amount and severity of pain it can cause. The prostaglandins released by activated mast cells can lead to significant bone, muscle, and joint pain. Tryptase, another substance released by inappropriately activated mast cells, can further increase pain. Other mediators released during this activation process can lead to headaches, fatigue, brain fog, and muscle cramping. Pain from inflammation is a prominent symptom in patients with MCAS. Mast cells are involved in causing and contributing to inflammation in both chronic and acute pain. MCAS can cause inflammation in almost any tissue including muscle, nerves, joints, intestines.

### **Mast Cells and CRPS**

The immune system in our body is in constant communication with the brain and spinal cord (Central Nervous System). They communicate by 'text message' called pro-inflammatory cytokines (it means 'text messages' that promote inflammation). Over 70% of the brain is made of glial cells. These glial cells pack around the nerves in the brain. Mast Cells can activate the glial cells to release inflammatory chemicals that cause the neurons in the brain to become inflamed. This is the basis of some of the cases of CRPS-I. Not all cases of CRPS – I are a result of MCAS. Mast cells communicate with glial cells. Chemical mediators such as tryptase and histamine can activate glial cells. Once the Mast cells activate the glial cells, the activated glial cells then release other mediators (pro-inflammatory cytokines IL-1 $\beta$ , IL-6 and TNF ) that cause neuroinflammation of the nerves in the brain, causing a sensation of severe pain. On the flip side, the inflammation as a result of misbehaving Mast cells can be reduced by medicines that inhibit mast cells.

When the skin in patients with CRPS was studied, it was shown to have increased Mast cell tryptase ( one of the many chemicals released by Mast Cells). Mast cells in the skin are a major source of chemicals such as cytokines, histamines, and prostaglandins that causes inflammation. They sensitize the nerve endings in the skin.

It should be noted that not all patients with MCAS present with CRPS. In patients who already have pain from an injury, if they develop MCAS, the pain may increase many fold. Unfortunately, pain is also a trigger for MCAS.

For people with MCAS, not all days are the same. There are some days when the mast cells are not as activated, which would show up as relatively low pain days. They may have some days when the mast cells are misbehaving and overly active. Those days, patients would have a lot more pain. A lot would depend on avoiding triggers, keeping the mast cells stabilized and blocking the effects of inflammatory chemicals released.

## **Diagnosis**

The diagnosis of MCAS is frequently a diagnosis of exclusion. Patients present with array of baffling multi system symptoms. As a result they wind up seeing multiple physicians, who in most cases look at the patient from the lens of their own specialty and not holistically.

Due to the unpredictability of this condition on a day-to-day basis, testing may be unreliable. Consequently, many tests that are routinely ordered may come back as being within normal limits. This does not dispute the fact that many patients have debilitating and disabling symptoms. Yet, there are a number of tests for MCAS that can be performed, including a 24 hour urine collection for N-methylhistamine and Prostaglandin D2. Another test is tryptase levels. The tryptase levels in MCAS is normal in 85% of cases, which makes the test unreliable. Blood tests for histamine, Prostaglandin D2 and Chromogranin A are very difficult. The blood has to be drawn while the patient is going through a flare. The blood has to be chilled immediately. The patient has to be off a lot of medications, including ones that are used to treat MCAS.

For patients who have undergone a gastro-intestinal biopsy (usually done because MCAS affects the GI system a lot), a specialized stain for mast cells looking at the count and shape of the mast cells may be a far more useful test. In general, the diagnosis of MCAS is based on clinical symptoms and not as much on laboratory findings.

## **MCAS and COVID-19**

MCAS may be a sign of many underlying autoimmune dysfunctions. Patients with MCAS are at increased risk when exposed to COVID-19. As mentioned earlier, mast cells release inflammatory mediators such as cytokines. In patients with severe Covid-19 there is a cytokine storm leading to a state of hyper-inflammation. There is a suspicion that patients who develop a cytokine storm may have had a pre-existing MCAS, either as a full blown



condition or as an underlying condition, making these patients more prone to the disastrous effects of COVID-19. Patients who have their MCAS well controlled may have a less severe time with COVID-19.

### **Management of MCAS**

The management of MCAS has to be individualized to each person. A discussion on managing MCAS is very complex and has to be based on the patient's underlying condition, but it can be broadly based on the following steps.

1. Anti-histamines.
2. Mast Cell Stabilizers
3. Avoiding triggers.
4. Management of any underlying autoimmune dysfunction.

by Pradeep Chopra, MD, Assistant Professor (Clinical) of Brown Medical School and Stephanie Carroll, RN of the Pain Management Center in Pawtucket, Rhode Island

## **The Miracle Violinist That Had Judges in Tears**

This [uplifting video](#) of a girl who overcame severe hardship is an inspiration for those facing difficulty.

You may notice the fluttering petals of cherry blossoms falling as she plays, joy shining through her sparkling eyes. Despite all she has experienced, she is immersed in the expression of her heart's delight.

Sakura, or cherry blossom, is the national flower of Japan. The bloom is short lived, only two weeks each Spring. It is a celebrated occasion, called hanami, when families and friends gather for cherry blossom viewings under the boughs. The delicate petals fall like snowflakes, blanketing the earth.

Sakura embodies Buddhist principles of impermanence, mindfulness, and mortality. The celebration of the beauty and fleeting quality of our lives inspires us to focus on what truly matters, to let go of petty regrets, and to be present in the moment.

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“I wouldn’t wish that program on my worst enemy,” she recalls. “I made two friends, but if it wasn’t for the arts and crafts, movies, and games, I don’t think I would have made it. I celebrated my eighteenth birthday in the hospital. I had a five percent chance of walking out of the hospital and knock on wood, I walked out on crutches.”

Although the experience was trying, she credits the program with enabling her to complete her walk across the stage at high school graduation without assistance. Unhappily, she experienced setbacks.

“Luckily I have a really good pediatric physical therapist I can still see. Without her, I probably wouldn’t be walking. Originally,” she explains, “it was just in my legs. But then in a college class I had a seizure and fell back onto a linoleum floor. Now I have it in my entire back, both shoulders, neck, and both legs.

“I tried to get on SSDI (Social Security Disability Insurance) for eight years, and had two lawyers, but was turned down. I can’t work. I’m losing the ability to walk and function. They don’t know about the condition; it’s not like Cerebral Palsy or Epilepsy. In 2014 I couldn’t walk, hold my head up or cut my own food. I couldn’t do anything by myself. You probably have to be like that for social security to say yes.”

Despite the challenges, Amanda graduated from a community college with a certificate in early childhood education. She works part time with children at a special needs school.

“It can be exhausting,” she says, “but I love it there. It is like everything to me. Still, I worry. What if I can’t work there anymore? I’d be devastated.”

Living with her parents in a duplex isn’t ideal, she explains, but that isn’t stopping her from planning for her future.

“(The duplex) isn’t good for people with physical disabilities. We can’t make it handicap accessible because it’s only half a house. So I have a Go Fund Me page and we collect bottles and cans for me to be able to buy a house and convert it. Half the money goes to RSDSA and half goes toward the house, to make it handicap accessible.”

I asked what kind of modifications she will need to be able to live independently. Plans include single story living, with an accessible bathroom and shower, adequate door width and a ramp for wheelchair access. She tells me how much support she has received from her community as word has gone out through Go Fund Me, her recyclables redemption drive, and the local newspaper.

“People have sent donations in the mail. I’m amazed at people’s outpouring of support. We have a chair lift now, so I can go up the stairs. It was donated by a lady from my church who had it for her mother and paid to get it reinstalled in our house. Milford is a very giving town, and people from other towns will contact us to collect their bottles and cans.”

COVID-19 led the Young Adults Weekend participants to create a team for RSDSA’s first Virtual Walk. I asked Amanda to tell me about being a member of the team, the importance of community and the challenges of isolation for people living with CRPS.

“The Young Adult weekends are amazing,” she said. “I wish they were longer than a weekend. They are priceless because you meet an actual human being sitting next to you who understands. We do art therapy, and sit around the table eating together. It feels like family. I’ve also met people through the Central Park RSDSA fundraiser walk, and I met my best friend from Ireland through a support group online. Since COVID, a bunch of us meet in virtual Zoom meet ups and I have a list in my phone of all the friends I’ve made with CRPS.”

I asked Amanda how she copes, how she remains so hopeful, and if she has any tips to share.

“I always told myself I would never give up - as hard as the condition is physically and mentally. Dr. Schechter was amazed at my mind set during those two months in hospital. I think life is precious and I’m determined not to let the disease beat me. It is hard, and easier said than done, but you can’t just give up. Just know you are not alone. It is a mind set. I can’t let this disease win. It might put me in wheel chair one day but that doesn’t mean that it’s won.

“Some days the pain is so bad I cry myself to sleep and think ‘How will I get up in the morning?’ I wonder how I just keep going. A friend with CRPS asked me if I feel like ever giving up. I told her that my body is tired of fighting but my mind isn’t. As long as I can beat the disease in my mind I don’t think it has won because I am still alive, no matter what it does to me physically.”

You can learn more about Amanda in her hometown paper, the Milford Mirror<sup>4</sup>, and at her Go Fund Me page.<sup>5</sup>

About CRPS:

According to **Pediatric complex regional pain syndrome: a review:**<sup>6</sup>

“Complex regional pain syndrome (CRPS) is a chronic, intensified localized pain condition that can affect children and adolescents as well as adults, but is more common among adolescent girls. Symptoms include limb pain; allodynia; hyperalgesia; swelling and/or changes in skin color of the affected limb; dry, mottled skin; hyperhidrosis and trophic changes of the nails and hair. The exact mechanism of CRPS is unknown.”

The National Organization for Rare Disorders (NORD) estimates that 200,000 people in the United States develop CRPS every year.<sup>7</sup>

- <sup>1</sup> Help RSDSA send Children with Pain to Summer Camp. RSDSA. January 17, 2019. <https://rds.org/help-rds-sa-send-children-pain-summer-camp/>. Accessed 2-27-21
- <sup>2</sup> Weissmann, Rotem, and Yosef Uziel. “Pediatric complex regional pain syndrome: a review.” Pediatric rheumatology online journal vol. 14,1 29. 29 Apr. 2016, doi:10.1186/s12969-016-0090-8
- <sup>3</sup> Slide 42, April 2017 Presentation Complex Regional Pain Syndrome (CRPS) Diagnosis and Management Nashville, 2017 <https://rds.org/wp-content/uploads/2017/04/Pradee-Chopra-CRPS.pdf> Accessed 2-27-21
- <sup>4</sup> Milford Mirror, January 24, 2021, Her Pain Is Like A Blowtorch, <https://www.milfordmirror.com/news/article/Her-pain-is-like-a-blowtorch-Milford-woman-15894099.php> Accessed 2-28-21
- <sup>5</sup> Buying House and Making it Accessible., organized by Amanda Pitre. Gofundme.com. <https://www.gofundme.com/f/29j2mkuj5c> . Accessed 2-28-21
- <sup>6</sup> Weissmann, Rotem, and Yosef Uziel. “Pediatric complex regional pain syndrome: a review.” Pediatric rheumatology online journal vol. 14,1 29. 29 Apr. 2016, doi:10.1186/s12969-016-0090-8
- <sup>7</sup> Complex Regional Pain Syndrome. NORD (National Organization for Rare Disorders). August 08, 2019. <https://rarediseases.org/rare-diseases/reflex-sympathetic-dystrophy-syndrome/>. Accessed 2-27-21

*Jenny Picciotto is a writer and Complex Regional Pain Syndrome patient who enjoys reading and playing the piano. She was a yoga instructor and massage therapist before CRPS changed her trajectory. She currently lives in Hawaii, where she facilitates the Oahu CRPS Support Group.*

## Join our next Facebook Live with Dr. Steven Richeimer

Our next Facebook Live will take place on Wednesday, March 24th at 7p Eastern with Dr. Steven Richeimer. The Facebook Live will focus on ketamine.

Dr. Richeimer is the Chief of the Division of Pain Medicine, a Professor of Anesthesiology & Psychiatry and the Director of the Online Master's Degree in Pain Medicine at the Keck School of Medicine at University of South Carolina.

Don't forget: If you miss one of our Facebook Live sessions, you can always watch the replay on the [RSDSA YouTube Channel](#) as we do record each one.

## Help RSDSA Find An ASL Interpreter

RSDSA would like to have an ASL interpreter for upcoming Facebook Lives in order to better serve the community.

Please send us an email at [info@rsds.org](mailto:info@rsds.org) if you have a recommendation!

## Donate to RSDSA

Have you thought about your legacy? Are you looking for a long-term way to make a meaningful difference in the CRPS community?

Please consider making a planned gift to RSDSA today. Planned giving options include:

- Gifts of stocks and bonds
- Including RSDSA as a beneficiary in your life insurance policy
- Including RSDSA as a beneficiary in your will.
- Contributing via an IRA

Tax benefits apply to each of these options. Please contact your attorney, a financial attorney, or a financial advisor for more info.

## Please send us feedback!

Please send any suggestions or upcoming events of interest to our community to [info@rsds.org](mailto:info@rsds.org) and please consider a donation to [rsds.org/donate](https://rsds.org/donate).