



# 5 Years of CureDRPLA

## Achievements and Advances in our Mission to Treat DRPLA

CureDRPLA was incorporated as a non-profit foundation in November 2019 by Paul and Andrea Compton. We are honoring 5 years of CureDRPLA with this impact report.

**The mission of CureDRPLA is to connect families, physicians and scientific investigators to further research on Dentatorubral-Pallidoluysian Atrophy (DRPLA) and work towards a treatment for this neurodegenerative genetic condition that can affect both children and adults.**

CureDRPLA engaged with Ataxia UK, a charity for people affected by any type of ataxia, to help further its efforts and benefit from their expertise and connections in the ataxia field.

*CureDRPLA team, from left to right:  
Andrea Compton, Paul Compton,  
Dr Silvia Prades, Dr Julie Greenfield,  
and Dr Jeff Carroll*





# 5-year Impact Report

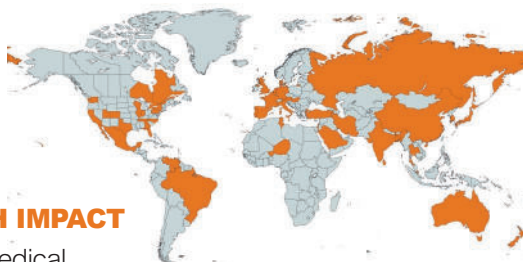
*"Our adopted son was diagnosed with Dentatorubral-Pallidoluysian Atrophy (DRPLA) in summer of 2018. This was devastating news for our family. At that time, we were told not only was there no cure available but that the development of a solution was over a decade away. We began to research the disease and attempt to identify any form of grassroots patient advocacy. Unfortunately, we found very little resource had been devoted to developing a treatment or creating a patient group of any form. **So, when we started CureDRPLA in November 2019 our objectives were simple, we wanted to build a global community of people with DRPLA and develop a robust treatment for this terrible condition.***

*While creating a global community has been more challenging in some countries than others and remains a work in progress, one thing is for sure, without data and global participation, we will not achieve our objectives. CureDRPLA has been funding scientists to advance research but this is not enough. We need the engagement of all DRPLA families to take part in research so that CureDRPLA can accomplish its objectives.*

***And, because DRPLA is so rare, we need everyone.***

*While we had our share of disappointments and frustrations, we have come a long way. However, **our mission will not be accomplished until there is a globally approved drug that is halting the progression of DRPLA.***

***Paul and Andrea Compton, Co-Founders of CureDRPLA***



## OUR GLOBAL OUTREACH IMPACT

- Reached out to over **3,000** medical professionals and organizations in **70** countries
- Our reach resulted in a **global network of clinicians** engaging with CureDRPLA from countries like United States, many in Europe, South Korea, China, Brazil, Australia, New Zealand, and more
- **People with DRPLA have been diagnosed in at least 35 countries** – this is a recollection of published information, data from our patient registry, and correspondence with CureDRPLA.

*View the map on our website: <https://bit.ly/map-DRPLAcases>*



# 5-year Impact Report

## FIRST CLINICAL TRIAL FOR DRPLA

**In February 2024, CureDRPLA reached an important milestone towards its mission – an upcoming experimental treatment for a pioneer individual with DRPLA in the United States (US) in what is known as an ‘n-of-1’.**

We are sharing with you the work of the n-Lorem Foundation, a non-profit organization that charitably provides experimental antisense oligonucleotides (ASO) to treat people with rare diseases that affect very few people in the US. This clinical trial is a collaboration between n-Lorem, Dr Jennifer Bain at Columbia University and CureDRPLA. n-Lorem has an investigational ASO specific for DRPLA and is being tested for the first time in an individual with DRPLA. In DRPLA, there is an accumulation of an abnormal protein (called ATN1) in cells, and therefore reducing the levels of this abnormal protein is thought to be a useful therapeutic approach.

CureDRPLA will provide updates on this clinical trial as it progresses. What we will learn about DRPLA from this trial and the potential treatment will help the entire community as we move forward to helping more individuals with DRPLA.

*“Our most exciting deliverable to date has been the commencement of the n-of-1 trial utilizing an ASO created by n-Lorem. This is indeed a very exciting milestone. We must now strive to create a pathway to a global rollout.”*

**Paul Compton, Co-Founder of CureDRPLA**

*“I have been collaborating with the n-Lorem Foundation to treat individuals with ultra-rare neurological disorders using ASO treatments, including DRPLA. In our study, we administered an experimental ASO treatment to an 18-year-old male to improve the ataxia and prevent further disease progression.*

*He has received four doses via lumbar puncture, which injects the experimental medication into the spinal fluid to reach the brain. The injections have been well tolerated without any safety concerns. While there does not appear to be any significant effect on seizures, there has been improvement in motor skills, speech, and overall ability to perform activities of daily living. The individual will now receive the medication every three months and be monitored closely for side effects or clinical changes.”*

**Jennifer Bain, child neurologist at Columbia University and New York-Presbyterian Hospital**



# 5-year Impact Report

## OUR RESEARCH IMPACT



**\$5 million+**

**funded by CureDRPLA**  
in research projects and activities  
aimed at advancing the  
organization's mission

**50+**

**scientists** have worked on  
**DRPLA projects**



**15**

**grants awarded to  
advance research**

Research groups who received  
the largest amounts of funding:

**Dr Timothy Yu,**  
**Dr Vikram Khurana,**  
**Prof Paola Giunti,**  
**Prof Henry Houlden,**  
**Dr Yael Shiloh-Malawsky,**  
**Dr Claire Miller**  
and **Dr Jeff Carroll**

**51**

**engagements**, including:  
**17 academic groups** doing  
**research on DRPLA,**  
**13 companies** working on  
**DRPLA research projects,**  
**21 pharmaceutical and biotech**  
**companies** pitching DRPLA as a  
**potential treatment area** and  
explore feasibility of **developing**  
**therapies for DRPLA**

**5**

**scientific articles** published by  
**CureDRPLA staff**  
and other scientists



**2**

**humanized ATN1 mouse**  
**models** developed, one line with  
**112 CAG repeats** which  
expresses severe symptoms and  
another one with **70 CAG repeats**  
which is less affected



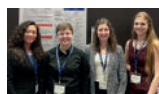
**Cell lines developed** from at least **17 people with DRPLA**



**Launched DRPLA Centers of Excellence accreditation** to  
recognize those physicians **with expertise in DRPLA** and  
help families find medical teams near them that understand  
the **various needs of those diagnosed with DRPLA**



Junko Shiozawa  
talking to  
healthcare  
professionals  
at the National  
Center of  
Neurology and  
Psychiatry in  
Tokyo, Japan



Scientists from  
Dr Jeff Carroll's  
lab presenting  
a poster at the  
International  
Congress for  
Ataxia Research  
in 2022 in Dallas,  
United States.  
*From left to right:*  
*Aliza Ben-Varon,*  
*Velvet Smith,*  
*Emma Jerome,*  
*Sage Berry.*

# 5-year Impact Report

## PARTICIPATING IN RESEARCH STUDIES

It is vitally important for people with DRPLA to take part in research studies as it can provide invaluable information, helping scientists understand DRPLA better and develop effective therapies. We wish to drive progress, improve future care, and offer hope to the entire DRPLA community. CureDRPLA shares all opportunities to participate in research on its website, newsletter and social media channels.

This study collects health information over three years, requiring **annual appointments at the clinics where the neurologist will do some clinical assessments** (e.g. ataxia scales, brain MRI, blood collection, etc.). The study is recruiting people with and without DRPLA to allow for comparison between these groups.

This study is led by Prof Paola Giunti and Prof Henry Houlden from University College London and Dr Yael Shiloh-Malawsky from the University of North Carolina at Chapel Hill. It is expanding through collaborations with other neurologists to include participants from Austria, France, Spain, Italy, South Korea, Turkey, and more.

**So far, 45 people with DRPLA and 25 people without DRPLA have been enrolled in this study.** We extend our heartfelt thanks to all the families and doctors participating in this study. **Recruitment is still open** and it will end by September 2026.

*If you are interested in participating, please visit our website:*  
**<https://bit.ly/NHBS-article>**

**DRPLA**  
**Natural History & Biomarkers Study**

**51** participants from **11** countries  
Japan, United States, United Kingdom, Brazil,  
South Korea, Canada, Italy, New Zealand,  
Netherlands, Saudi Arabia, Spain

**17** people with **adult-onset DRPLA**,

**26** people with **juvenile-onset DRPLA**,

**5** asymptomatic, and **3** unknown

 **CURE  
DRPLA**  
**Global  
Patient Registry**

Join the registry:  
**<https://bit.ly/DRPLA-registry>**



# 5-year Impact Report

## OUR EVENTS & MEETINGS IMPACT



3

**CureDRPLA Research  
Conferences hosted,**  
**83** attendees in total

4

**research update talks**  
for people with **DRPLA**  
and their **caregivers**

**450+**

attendees for co-hosted **Externally-led  
Patient-Focused Drug Development  
meeting** with the **National Ataxia  
Foundation**

**130+**

meetings with  
**Ataxia UK**  
and **CureDRPLA**



Attended **13 research conferences** relevant for our mission



**Joined key organizations** in movement disorders, neurology  
and epilepsy to stay current and expand our network



Organized the **largest in-person gathering of DRPLA  
families in Cardiff, United Kingdom**



Participants  
at the 2023  
CureDRPLA  
Research  
Conference  
in Boston,  
United States

*"A great symbol of our broad-based progress is that this year at the 2024 International Congress for Ataxia Research in London there will be one scientific talk on DRPLA and 7 posters highlighting the outstanding body of global research."*

**Andrea Compton, President and Co-Founder of CureDRPLA**

What families said about the meet up in Cardiff:

*"Thank you for organising this event. I don't think any of us stopped talking. I've never met any other family with DRPLA in the 24 years I've known about this condition. Let's hope we can do this again."*

**Emma Davis**

*"What really hit home for me was the families sharing their experiences and support difficulties of caring for a loved one who is suffering. This brought all the memories back of my own family's journey many years ago. We were alone and 18 years on, these people feel the same. I hope they will find some solace in being in touch with each other and the knowledge of the wonderful work you are all doing."*

**Katherine Roberts**



CureDRPLA  
booth at  
the 2023  
International  
Congress of  
Parkinson's  
Disease  
and Movement  
Disorders in  
Copenhagen,  
Denmark

# 5-year Impact Report

## VOLUNTEERS ORGANIZE BOOTH AT A NEUROLOGY CONFERENCE IN TOKYO

*"I am grateful to have been able to attend the AOCN 2024 conference and to have been given this really valuable opportunity. I was able to meet and talk with fellow mothers, and I was reminded that we all feel the same way. Many doctors came from far away and told us more about the treatment drugs, which gave us more hope than ever before."*

**Masako Shingai (新開昌子)**

*"It was a very, very good experience. Above all, it was a great pleasure to meet other patient families for the first time. I had met them on videocalls, but I had never been able to talk to them face to face. I have a little bit of hope for the future. I am grateful for this opportunity. Thank you very much. I pray that clinical trials will be successful. And I hope that drugs will reach us soon. Please keep up the good work."*

**Etsumi Hosogai (細貝悦美)**

*"It was reassuring to know that I was not alone and that there were people who understood me. I also learnt that there are many people with DRPLA in various parts of Japan, and I hope that more people will join the DRPLA Family Association."*

**Yasuko Kamata (鎌田康子)**





# 5-year Impact Report

## OUR SOCIAL MEDIA IMPACT

**www**

**6,750**

visitors in the **CureDRPLA website** in 2023, a **443%** increase from 2020

Top countries in website visits: United States, United Kingdom, Japan, Australia, Korea, Portugal, Canada, Italy and Brazil



**1,597**

views on **CureDRPLA YouTube channel**

**1,000,485**

views and **2.28K** subscribers on **Cure Disease Kuri Channel** run by our Advisory Board Member, **Junko Shiozawa**



**135**

**social media posts** and reached **533** people on **Facebook** in the first half of 2024

**162**

members on the **closed DRPLA Facebook group** for people with DRPLA and their families with at least **54%** active members

**43** members on the **closed DRPLA Facebook group in Japanese** for patients and families

**91**



subscribers to **CureDRPLA newsletter** launched in August 2023

**New issue every 6-8 weeks.**  
**More than 70% open our newsletters**

Junko Shiozawa has been advocating for her loved ones affected by DRPLA for years:

*"It has been 16 years since my husband and daughter were diagnosed with DRPLA. I wanted to help them, so I was active on social media and contacted many doctors and researchers. But there was no hope at all. From time to time, I received inquiries from patients' families, but we could only encourage each other.*

*In the meantime, their illness progressed, and my husband passed away seven years ago. My daughter Kuri is bedridden with a gastric tube and a tracheotomy. I couldn't give up.*

*One day I received an e-mail from Andrea Compton. She told me that her son has DRPLA, and she was forming an organization to find a treatment. I had no idea that in five years we would become such a successful organization. There is now hope for the patients' families. To everyone involved with CureDRPLA, thank you so much!"*

**Junko Shiozawa, Advisory Board Member to CureDRPLA**

# Quotes and testimonials

*"My first encounter with a person with DRPLA was about 15 years ago when I moved to North Carolina. When caring for patients with this devastating condition, our ultimate goal is to be able to offer a cure.*

*It is remarkable to see what CureDRPLA has accomplished in just five years. I am fortunate to be part of the international collaboration with Ataxia UK that developed the natural history and biomarker study and set up a study site at UNC.*

*Through enrolling in the study, I have met some of the bravest and sweetest patients and their incredible families. I am honoured to be the first DRPLA Center of Excellence designated site.*

*As a pediatric neurologist, I have the privilege to team up with families and witness how parents cross rivers and move mountains to bring comfort, hope, and the possibility of a cure to their children. CureDRPLA is one of those extraordinary and most inspiring examples."*

**Dr Yael Shiloh-Malawsky, neurologist at the University of North Carolina, Chapel Hill, United States**

*"Ataxia UK's partnership with CureDRPLA has been remarkable.*

*The scope and the quality of the research programme during these 5 years is yielding great results and is changing the prospects for people with DRPLA.*

*The collaboration between n-Lox and CureDRPLA has facilitated the start of n-of-1 clinical trials in the United States with an experimental treatment. Such quick progress is unheard of in the ataxia community and sharing expertise between Ataxia UK and CureDRPLA has proven to be a great strategy.*

*I look forward to continuing to advise on the DRPLA research programme and transferring our learnings to other ultra-rare ataxias."*

**Dr Julie Greenfield, Head of Research at Ataxia UK**

*As the disease progresses day by day my daughter is no longer able to perform the daily activities that she used to be able to perform and requires full assistance. Not only my daughter is suffering, but I am also struggling physically and mentally. It is particularly difficult for me to follow her instructions and to see her behaving strangely due to the decline in her cognitive function.*

*Thank you for working to find a cure for DRPLA and supporting the activities of the Japanese family association.*

**Yasuko Kamata (鎌田康子)**





## Quotes and testimonials

*"Having worked with CureDRPLA since almost the inception, I've seen up close the remarkable global impact of this relatively small organization.*

*Their spending has been very targeted, and very smart - they've encouraged some of the best researchers and clinicians around the world to be doing focused work on this ultra-rare condition*

*In just a couple of years, they've made it so that any researcher wanting to work on DRPLA will have all the tools they need - from iPSC lines, to novel mouse models, to human clinical samples collected in a very precise and careful way.*

*None of us will be happy until DRPLA isn't a problem for anyone, but this organization has changed the outlook for people diagnosed with DRPLA today, compared with when they started.*

*It's a remarkable achievement that when someone is diagnosed with DRPLA and first searches out resources, they find a lot of exciting research, as opposed to the landscape 5 years ago."*

**Dr Jeff Carroll, Scientific Advisor to CureDRPLA**

*"CureDRPLA holds a special place in my heart.*

*My brother is battling DRPLA, and we lost our beloved mother to this devastating condition.*

*The journey has been incredibly challenging, filled with moments of despair and helplessness.*

*However, knowing that CureDRPLA is out there working on behalf of this rare illness has been a beacon of hope for our family.*

*Having an organisation dedicated to DRPLA has provided a sense of community and support that we desperately needed.*

*The resources, research updates, and the compassionate network of individuals who truly understand our struggles have made a significant difference.*

*CureDRPLA has not only offered us practical support but also emotional solace, knowing we are not alone in this fight.*

*Looking towards the future, I see a community that is stronger and more united than ever.*

*For families like ours, CureDRPLA is a promise of a brighter future for those who suffer now and are diagnosed in the future."*

**Beth Lancaster**

# Quotes and testimonials

*"CureDRPLA has made such a tremendous impact on my family.*

*My four youngest children all have DRPLA. When I first found out about their diagnosis, I was told in no uncertain terms that there was no cure and no treatments and to take them home and "watch" them.*

*Those are hard words for a mom to hear. Over the past 7 years since they were diagnosed, much to my amazement, CureDRPLA came along and has been actively working to find cures for this terrible disorder.*

*My kids are able to participate in a study at the University of North Carolina to help them identify changes as the disorder progresses.*

*I cannot thank CureDRPLA enough for all of their hard work.*

*My kiddos outlook on life is looking brighter because of all of their efforts!"*

**Kerry Mackey**

*"Five years since my second son  
was diagnosed with DRPLA,  
The list of things he can't do grows longer,  
Day by day, he breaks a little more...  
But I desperately shift my thoughts,  
Focusing on what he can still do, while he can still do it...  
As the morning light filters through,  
I open the door to his room,  
Imagining the boy who once ran to me,  
"Good morning, Mom!" he'd say with a smile.  
Now he can't walk, and words elude him,  
Yet one more worry weighs on my heart—  
My eldest son, untouched by the illness... for now.  
He chases his dreams, fights his way in the world,  
But sometimes, his heart trembles with fear.  
The dread that he, too, might one day fall ill,  
Or the pain of losing the brother he so dearly loves,  
Leads him to despair.  
How can I hold his fragile heart?  
I can only hope for a day when I can say,  
Even if you fall ill, there is a cure—  
And believe it with all my soul."*

**Yuko Kanmate (かんまで優子)**





# What's next & how to get involved

## OUTLOOK FOR THE NEXT 5 YEARS

As we look back, we are amazed that we pushed ahead and have accomplished what we have. We were very fortunate to team up with Ataxia UK as that connected us with deep experience in natural history studies and patient registries, which were critical to advance research.

We embarked on this journey with a deep commitment to our mission. While we take pride in our accomplishments thus far, we remain aware of the challenges that lie ahead.

## OBJECTIVES FOR THE NEXT 5 YEARS

- 1. Expand and strengthen our supportive community:** continue building and growing a strong network of people with DRPLA, families, physicians and researchers at a global scale.
- 2. Ensure more DRPLA research projects get funded:** identify and pursue funding opportunities to support ongoing and new research projects, including the screening of two new potential treatments.
- 3. Promote research participation:** encourage and support participation in DRPLA research studies.
- 4. Scale research contributions towards treatments:** explore partnerships with organizations that possess greater resources to develop treatments and conduct clinical trials.
- 5. Establish subject-matter expertise:** strive to maintain our position as the leading experts in DRPLA, ensuring that all relevant stakeholders in the industry are aware of our work and contributions.

## STAYING IN TOUCH & INFORMED ABOUT RELEVANT NEWS

- 1. Subscribe to our newsletter:**  
<https://curedrpla.org/en/newsletter-archive>
- 2. Follow our Facebook page:** [www.facebook.com/cureDRPLA.org](https://www.facebook.com/cureDRPLA.org)
- 3. People with DRPLA, their families and friends can join the closed DRPLA Facebook group for peer support:**  
[www.facebook.com/groups/144167217964](https://www.facebook.com/groups/144167217964)
- 4. Do not forget to visit our website:** <https://curedrpla.org/en>

## CONTACT US

Please be in touch with us personally to share your story or any relevant information. Together, we may be able to develop a better understanding and potential treatments for DRPLA. Email: [info@cureDRPLA.org](mailto:info@cureDRPLA.org)

Newsletter  
subscription

