Annual Impact Report 2023

EDUCATION. EMPOWERMENT. EVIDENCE

Fondation du syndrome Loeys • Dietz
Syndrome Foundation · Canada

Table of Contents

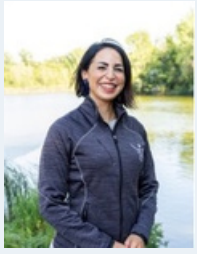
1. A Message from our Executive Director and Board Chair

This year, Loeys-Dietz Syndrome Foundation Canada dedicated its efforts to empowering our community through education, reflecting our core mission to support individuals and families affected by Loeys-Dietz syndrome. Recognizing the unique challenges faced by patients, we prioritized the development of a comprehensive family planning booklet. This resource aims to provide essential information and guidance, helping individual living with LDS make informed decisions about their futures while navigating the complexities of the syndrome.

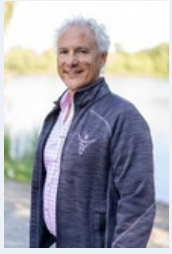
In addition to this significant milestone, we organized several educational conferences throughout the year. These events brought together leading medical experts, researchers, and patients, fostering an environment of. knowledge sharing and collaboration. By focusing on education, we have strengthened our community's understanding of Loeys-Dietz syndrome, enabling better management of the condition and improving the quality of life for those affected.

We are especially proud to announce that our main educational event, the Heritable Aortic Disorder Symposium, returned this year after a three-year hiatus due to COVID-19. The symposium was a resounding success, reuniting our community and providing a platform for cutting-edge research presentations, patient stories, and expert discussions. The return of this flagship event underscored our unwavering commitment to education and the importance of staying connected and informed, even in challenging times.

Our commitment to education has been the cornerstone of our initiatives this year, and we are proud of the positive impact these efforts have had on the lives of our patients and their families. Together, we continue to build a more informed, resilient, and supportive community.



Jida El Hajjar, Executive Director



Joseph Galli, Chair

2. Problem Statement

Loeys-Dietz Syndrome (LDS) is a rare connective disorder that has been associated with extensive systemic involvement including craniofacial, skeletal, cutaneous and vascular (arterial tortuosity, aneurysm formation and dissection) abnormalities. Approximately two-thirds of LDS patients develop an aortic root aneurysm, and nearly all have some dilation of their aorta. The most serious of these symptoms stems from the high risk of aortic root aneurysms, which, due to the friable nature of the arterial tissues, have an extremely high likelihood of bursting. When the aorta, the largest blood vessel in the body, bursts or dissects, it causes rapid and extremely deadly internal hemorrhaging within minutes.

In addition to severe vascular issues, LDS patients often suffer from a range of systemic symptoms. These can include loose joints, severe orthopedic problems such as scoliosis and kyphosis, dental and ophthalmological issues, as well as digestive problems. Moreover, the natural progression of the disease is still being studied, with new symptoms, such as increased cancer incidence, being identified through patient reports. The varied presentation of symptoms necessitates individualized care involving multiple specialists, posing a significant challenge for patients and their families who must navigate the healthcare system on their own. LDS is currently **incurable**.

The prevalence of Loeys-Dietz syndrome is **unknown**. LDS arises from mutations altering the transforming growth factor β signalling pathway. It is a genetic disorder that can be inherited from a parent (25% of cases) or can develop spontaneously due to a gene mutation (75% of cases). Consequently, patients' families and offspring are also at risk. When LDS is identified, it is crucial to test immediate relatives and offspring, as LDS is inherited in an autosomal dominant manner, meaning each child of an affected individual has a 50% chance of having the disorder. This not only complicates family planning but also affects the ability of affected parents to be caregivers, especially if their children are also impacted.

The Need for Education and Empowerment

Patients living with LDS in Canada face significant challenges, particularly in the realm of education and empowerment. A critical area where this gap is most evident is in family planning. Many individuals and families affected by LDS lack access to comprehensive, accurate information and support necessary to make informed decisions about their reproductive futures.

2. Problem Statement

The complexity of LDS, combined with its hereditary nature, makes family planning a daunting task. Patients often struggle with understanding the implications of genetic testing, the risks associated with different pregnancy options, and the possibilities of surrogacy or adoption. Without adequate resources and guidance, families are left to navigate these intricate and emotionally charged decisions on their own. This lack of education and support can lead to feelings of isolation and anxiety, as well as potentially uninformed decisions that may not align with the best interests of the family. Empowering patients with the knowledge and resources they need is crucial for their well-being and the well-being of future generations. Addressing this issue is a fundamental step towards improving the quality of life for those living with LDS in Canada.

Our 2023 Educational Initiatives

Despite advancements, there remains a gap in awareness and education about LDS among both the public and healthcare professionals. This gap has led to delayed diagnoses and inadequate support for affected families. Recognizing this critical issue, the foundation implemented two educational events and developed a family planning educational booklet in 2023. These initiatives aim to raise awareness about LDS, provide vital information to patients and their families, and ensure that healthcare providers are better equipped to identify and manage this complex disorder.

3. Mission and Vision

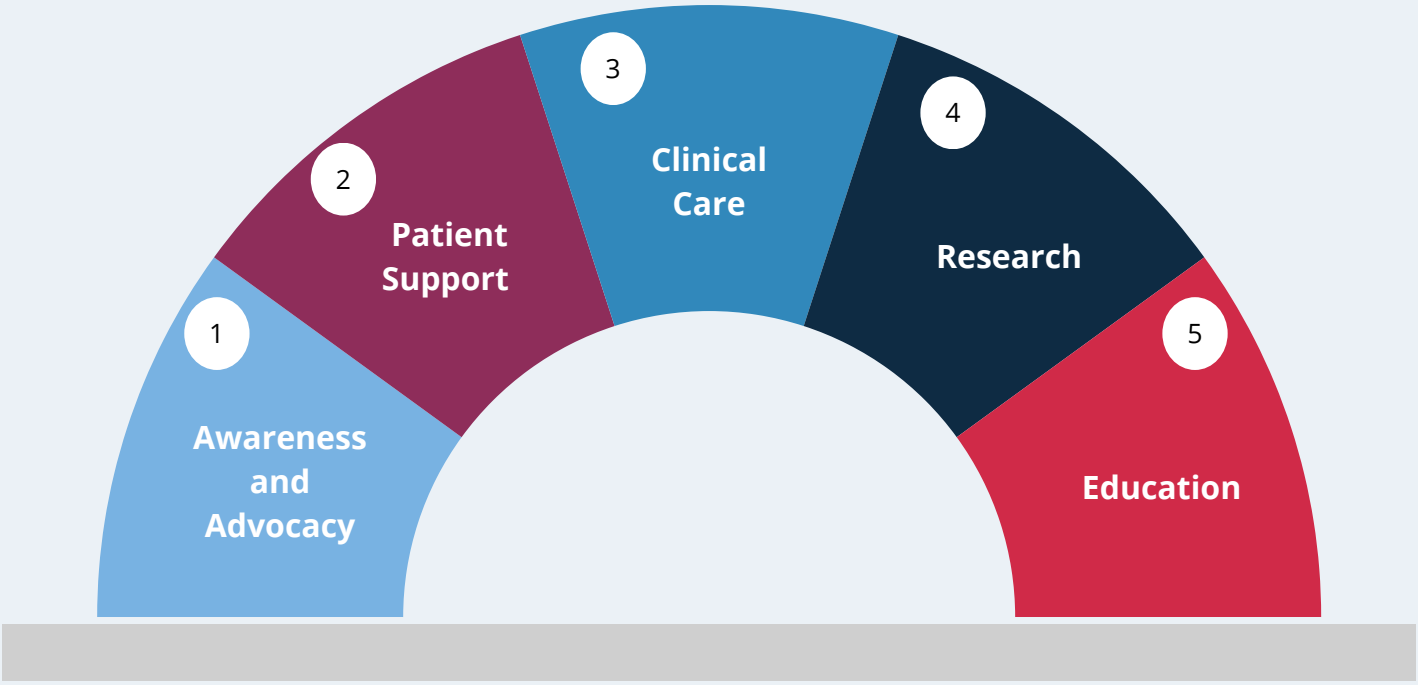
The Loeys-Dietz Syndrome Foundation Canada (LDSFC) is a national charitable organization committed to advancing research, awareness, education, and support for individuals affected by Loeys-Dietz syndrome (LDS) and related heritable aortic disorders.

Thanks to the remarkable generosity of our donors and the expertise of our esteemed Medical and Scientific Advisory Board and Patient Partnership Committee, the Foundation actively pursues its mission. Our focus revolves around fortifying and empowering the LDS community through the establishment of a robust support network for all those impacted. Additionally, we aim to broaden the horizons of cutting-edge LDS research, pushing the boundaries of knowledge and innovation.

3. Mission and Vision

By working altogether, we envision transforming the future of Loeys-Dietz syndrome and related heritable aortic disorders, ultimately saving and enhancing the lives of patients and their families. The collaborative efforts of the Foundation and its valued partners will lead us to a brighter and more promising tomorrow.

We aim to attain our mission through these 5 strategic pillars:



4. Team

JIDA EL HAJJAR
Executive Director

KENDALL TILTON
Finance Director

LINDSAY PARSONS
Research and Patient Support Coordinator

CHARLOTTE BAYLIS
Education and Communication Coordinator

5. Board of Directors

JOSEPH GALLI
President and Co-founder
President, Pentor

SALVATORE FRATINO
Secretary and Co-founder
Rosdev

YVES GLAUDE
Retiree

ALEXANDER JESION
Chief Financial Officer
Cummings Centre

DR. NANCY FOURNIER
Director and Chair of Risk Committee
Senior Project Manager

NATALIE COLPRON
Director and Chair of Investments Committee

RICHARD JOLY
Director and Chair of Governance Committee
Partner, Leaders International

6. Medical and Scientific Advisory Board:

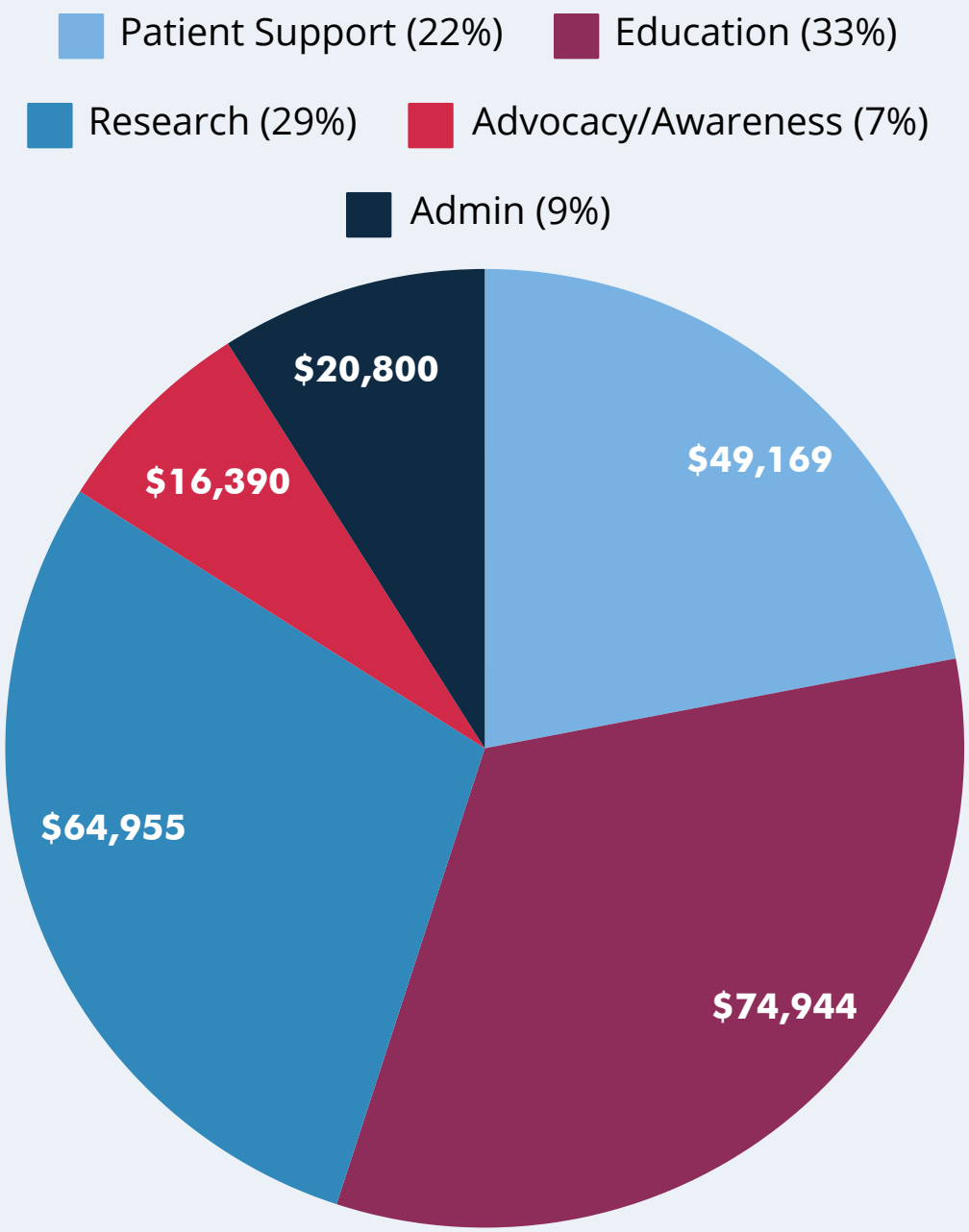
Dr. Gabrielle Horne, Chair (Cardiology).
Dr. Maral Ouzounian (Aortic Surgery)
Dr. Laura Drudi (Vascular Surgery).
Dr. Ismael El-Hamamsy (Aortic Surgery)
Dr. Dion Peppelassis (Cardiology)
Dr. Jean Ouellet (Orthopedic Surgery)
Dr. Tiscar Cavalle-Garrido (Pediatric Cardiology)
Dr. Maureen O'Connor (TGF Beta Expert/Industry)
Dr. Michelle Keir (Cardiology, Quality of Life)
Dr. Julie Richer (Genetics)

7. Programs and Activities:

The Loeys-Dietz Syndrome Foundation Canada (LDSFC) is dedicated to supporting individuals and families affected by Loeys-Dietz syndrome through advocacy, education, and research. Our mission is to improve the quality of life for those living with this rare genetic disorder. The past year has been filled with impactful events and initiatives that have brought us closer to our goals. Our programs and activities fall under our five main pillars, as demonstrated by the activities outlined below. Here is a summary of resource allocation for our key activities for 2023:

7. Programs and Activities:

Our programs and activities fall under our five main pillars, as demonstrated by the activities outlined below. Here is a summary of resource allocation for our key activities for 2023:



7. Programs and Activities:

EDUCATION

I - March 2023: Connective Tissue Disorder (CTD) Symposium in Partnership with the Canadian Spine Society and the Canadian Paediatric Spine Society

On March 1st 2023, in Quebec City, we sponsored and co-hosted a medical educational symposium in partnership with the Canadian Spine Society (CSS) and the Canadian Paediatric Spine Society. The Canadian Spine Society is a collective of spine surgeons and healthcare professionals dedicated to promoting excellence in spine patient care, research, and education.

This event focused on the spinal manifestations of Loeys-Dietz syndrome, providing a platform for medical professionals to share knowledge, research, and best practices. Attendees included orthopedic surgeons, geneticists, and other healthcare providers who are at the forefront of treating spinal issues in Loeys-Dietz syndrome patients. As part of the program, the presenters explored the spinal conditions afflicted or associated with connective tissue disorders. This knowledge focused on the following 4 learning objectives of the material presented during the symposium:

1. Gain new awareness of connective tissue disorders and how these impact the spine;
2. Be able to identify and prescribe appropriate work-up for patients with spinal conditions and connective tissue disorders;
3. Be initiated to the management of patients with connective tissue syndromes and spinal conditions;
4. Know what type of local and national resources are available for patients with connective tissue disorders.

7. Programs and Activities:

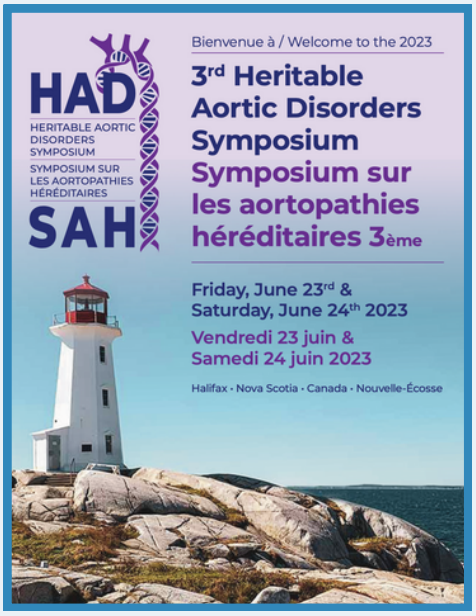
The agenda of the CTD symposium:

Time	Duration	Topic	Speakers
1300	15 minutes	Broad overview of the Genetics of Connective Tissue Disorders + case presentations	Dr. Julie Richer Assistant Professor, University of Ottawa Clinical Geneticist
1315	15 minutes	Broad overview of Cardio/Non-Musculoskeletal Presentation of Connective Tissue Disorders + case presentations	Dr. Tiscar Cavalle-Garrido Pediatric Cardiologist, Montreal Children's Hospital
1330	5 minutes	Questions-Genetics & Non-Musculoskeletal Presentations	
1335	15 minutes	Patient Perspective	Alex Galli Patient Partner
1350	40 minutes	Spinal & Orthopaedic challenges in managing these patients	Dr. Jean Ouellet Pediatric Orthopaedic Surgery AND Dr. A. Noelle Larson Pediatric Orthopaedic Surgery Mayo Clinic
1430	10 minutes	Questions-Spinal & Orthopaedic Challenges	
1440	15 minutes	National and local, medical and paramedical resources for patients with CTD	Dr. Jida El Hajjar, Executive Director Lindsay Parsons, Project Coordinator Loeys-Dietz Syndrome Foundation Canada
1455	15 minutes	BREAK	
1510	5 minutes	Questions- Resources (Dr. J. El Hajjar & L. Parsons)	
1515	60 minutes	Interactive Clinical cases discussions· • Cervical case • Deformity case • Peri op – Complications	Dr. Jean Ouellet Pediatric Orthopaedic Surgery

7. Programs and Activities:

We welcomed 110 attendees. Based on the post-event evaluation survey, 90% of responders agree or strongly agree that the CTD symposium met each of its four objectives. The event fostered valuable discussions and paved the way for future collaborations to improve patient outcomes, including two new research projects.

II - June 2023: Hereditary Aortic Disorders Symposium in Halifax, Canada




The 3rd Heritable Aortic Disorders Symposium – 2023 – Le 3ème Symposium sur les aortopathies héréditaires

FEATURING OUR KEYNOTE SPEAKER: METTANT EN VEDETTE NOTRE CONFÉRENCIER PRINCIPAL :

Dianna M. Milewicz, M.D. Ph.D., is the President George H.W. Bush Chair of Cardiovascular Medicine, Director of the Division of Medical Genetics and the John Ritter Research Program, and Vice-Chair of the Department of Internal Medicine at McGovern Medical School at the University of Texas Health Science Center at Houston (UTHealth). Her career focuses on research to identify the genetic and environmental triggers for acute aortic dissections, with an overarching goal of using this information to prevent aortic dissections.

In addition, her research has identified the altered genes triggering other vascular diseases and identified the molecular links between the defective genes and the resulting vascular diseases. She also chairs an international aortic consortium, the Montalcino Aortic Consortium, to define precision medical care to prevent acute aortic dissections based on the underlying genetic triggers.

Dr. Milewicz has received numerous honors and awards for her research, including Doris Duke Distinguished Clinical Scientist Award; American Heart Association Merit Award; and Rice University's Distinguished Alumni Award. She was inducted into the honorary societies for physician scientists, the American Society of Clinical Investigation and the Association of American Physicians, and currently serves as an AAP Council member. Dr. Milewicz has also participated in educating and increasing awareness for acute aortic dissections, including participating and chairing boards for the Canadian Genetic Aortic Disorder Association, the John Ritter Foundation, the American Heart Foundation, and the Marfan Foundation.

Dianna M. Milewicz, M.D. Ph.D., est le président George H.W. Chaire Bush de médecine cardiovasculaire, directrice de la division de génétique médicale et du programme de recherche John Ritter, et vice-présidente du département de médecine interne de la McGovern Medical School du University of Texas Health Science Center à Houston (UTHealth). Sa carrière se concentre sur la recherche pour identifier les déclencheurs génétiques et environnementaux des dissections aortiques aiguës, avec un objectif primordial d'utiliser ces informations pour prévenir les dissections aortiques.

De plus, ses recherches ont identifié les gènes altérés déclenchant d'autres maladies vasculaires et identifié les liens moléculaires entre les gènes défectueux et les maladies vasculaires qui en résultent. Elle préside également un consortium aortique international, le Montalcino Aortic Consortium, pour définir des soins médicaux de précision pour prévenir les dissections aortiques aiguës en fonction des déclencheurs génétiques sous-jacents.

La Dre Milewicz a reçu de nombreuses distinctions et récompenses pour ses recherches, notamment le prix Doris Duke Distinguished Clinical Scientist ; Prix du mérite de l'American Heart Association ; et le prix Distinguished Alumni Award de l'Université Rice. Elle a été intronisée dans les sociétés honorifiques des médecins scientifiques, l'American Society of Clinical Investigation et l'Association of American Physicians, et est actuellement membre du Conseil de l'AAP. Le Dr Milewicz a également participé à l'éducation et à la sensibilisation aux dissections aortiques aiguës, notamment en participant et en présidant les conseils d'administration de l'Association canadienne des troubles génétiques de l'aorte, de la Fondation John Ritter, de l'American Heart Foundation et de la Fondation Marfan.

Friday, June 23rd & Saturday, June 24th, 2023
Vendredi 23 juin & Samedi 24 juin 2023
Page 2

Our other major medical educational event of the year was the 3rd edition of the Hereditary Aortic Disorders (HAD) Symposium, held in Halifax on June 23rd and 24th, 2023. After a three-year hiatus due to COVID-19, this symposium brought together leading experts in the field of genetic aortic disorders. The program featured specialists from across Canada and internationally, who discussed new advances in genetics, diagnosis, and management of hereditary thoracic aortic disorders.

7. Programs and Activities:

Topics covered included the latest research findings, advancements in surgical techniques, and the development of new diagnostic and treatment protocols. The first day was dedicated to updating medical professionals, while the second day served as a community education forum for patients and families affected by HAD. We welcomed 104 attendees (50 patients and family members, and 54 healthcare professionals).

The 3rd Heritable Aortic Disorders Symposium ~ 2023 ~ Le 3ème Symposium sur les aortopathies héréditaires

DAY 1: SCIENTIFIC SESSIONS
(Medical Professionals)

Note: Each lecture will reserve 15% of its time for interactive audience participation. ***Lectures will be given in English.***

FRIDAY, JUNE 23rd, 2023

		Speakers & Moderators
7:30 am	**Registration (Coffee/Tea)**	
8:30 am	**OPENING REMARKS** with Dr Gabrielle Horne and Dr Jeremy Wood	
8:40 am	KEYNOTE ADDRESS: **Heritable Thoracic Aortic Disease:** Molecular Mechanisms and Gene-Based Management	Dr Dianna Milewicz, University of Texas
9:10 am	**Diagnosis and Risk:** Putting it all together	Dr Gabrielle Horne, Dalhousie University
9:40 am	**Non-syndromic Aortic Disease in Children:** Who is at risk of childhood events?	Dr Shaine Morris, Baylor College of Medicine
10:10 am	**Whole exome sequencing, VUS:** What does it all mean?	Dr Hanna Faghfoury, University of Toronto
10:30 am	*Panel discussion*	Dr Nanette Alvarez & Dr Tim Bradley
10:50 am	**Networking & Nutritional Break**	
11:10 am	Medical Therapy & Surgical Thresholds for Heritable Thoracic Aortic Disorders	Dr Michelle Keir, University of Calgary
11:30 am	**Imaging in Genetic Vasculopathy:** How can we help?	Dr Bruce Precious, Dalhousie University
11:50 am	**Worrying about aortas:** Supporting patients in the Psychological Implications of Hereditary Aneurism Syndromes	Dr Sulaye Thakrar, Dalhousie University
12:10 pm	*Panel discussion*	Dr Dan Belliveau & Dr Gabrielle Horne
12:30 pm	**Lunch**	
1:30 pm	**Pregnancy:** When to worry	Dr Chris Nash, Dalhousie University
1:50 pm	Proximal Aortic Surgery in Patients with Hereditary Thoracic Aortic Disease	Dr Maral Ouzounian, University of Toronto
2:10 pm	**The Aortic Arch and Beyond Post Aortic Dissection:** Do you need to extend the repair in patients with connective tissue disorders?	Dr Michael Moon, University of Alberta
2:30 pm	*Panel discussion*	Dr David Bewick & Dr Zlatko Pozeg
2:50 pm	**Networking & Nutritional Break**	
3:10 pm	Stent grafts in Genetic Aortopathy	Dr Rob Berry, Dalhousie University
3:30 pm	Surgical Approaches to the Fragile Tissues in Heritable Aortopathies	Dr Sherene Shalhub, University of Washington
4:15 pm	Insights from Patients and Program Support	Lindsay Parsons, LDSFC & Lindsey Rusche, GADA Canada
4:35 pm	*Panel discussion*	Dr Christine Herman & Dr Jeremy Wood
5:00 pm	**Adjourn**	

The 3rd Heritable Aortic Disorders Symposium ~ 2023 ~ Le 3ème Symposium sur les aortopathies héréditaires

DAY 2: COMMUNITY EDUCATION FORUM
(Patients, Families and Friends)

Note: Each lecture will reserve 15% of its time for interactive audience participation. ***Lectures will be given in English.***

SATURDAY, JUNE 24th, 2023

		Speakers & Moderators
7:00 am	**Registration**	
8:00 am	**OPENING REMARKS** with Dr Gabrielle Horne and Dr Jeremy Wood	
8:05 am	Genetics of Aortic Disease	Dr Dianna Milewicz, University of Texas
8:35 am	Putting the Pieces Together to make a Diagnosis	Dr Gabrielle Horne, Dalhousie University
9:05 am	Medical Therapy for Aortic Disease and Lifestyle Issues	Dr Michelle Keir, University of Calgary
10:30 am	*Panel – Questions from the audience*	
10:00 am	**Break**	
10:30 am	Surgery for the Aorta	Dr Maral Ouzounian, University of Toronto
10:50 am	How Do I Stop Worrying About My Aorta?	Dr Sulaye Thakrar, Dalhousie University
11:10 am	**Workshop 1:** *(Choose one)* • Preparing for Aortic Surgery MODERATORS: Dr Michael Moon, & Dr Maral Ouzounian, & Dr Jeremy Wood • Medications, Exercise, Pregnancy and Aortic Health MODERATORS: Dr Nanette Alvarez, Dr Gabrielle Horne, Dr Michelle Keir, & Dr Dianna Milewicz • Stress, Worry and Family Dynamics in Aortic Health MODERATORS: Dr Sulaye Thakrar & trainees • Pain Management MODERATORS: Shanna Foster, Dr Monique MacFarlane Conrad, & Leslie Marcotte	
11:40 am	**Workshop 2:** *(Choose one)* • Preparing for Aortic Surgery • Medications, Exercise, Pregnancy and Aortic Health • Stress, Worry and Family Dynamics in Aortic Health • Pain Management	MODERATORS: Please refer to the Moderators listed at 11:10am in Workshop 1.
12:10 pm	**Lunch & Meeting new friends**	
1:30 pm	Insights from Patients and Program Support	Lindsay Parsons, LDSFC & Lindsey Rusche, GADA Canada
2:00 pm	**Panel:** Living well with an Aortic Gene, Tips and Stories	Lindsey Rusche, GADA Canada, Joseph Galli, LDSFC & others
2:40 pm	Music Therapy Workshop OR Networking	Mackenzie Costron, Find Your Voice Music Therapy
4:00 pm	**Conclusion**	

Feedback from the second day, gathered through post-event surveys, highlighted that: 91% of patients and their families that responded to our survey agreed that their knowledge increased on topics related to genetics, diagnosis of aortic disease, medical therapy, lifestyle issues, and coping with stress related to their condition. 86% of responders indicated that the educational workshops provided space for learning and conversation.

7. Programs and Activities:

Other positive feedback notes from participants included:

Just fantastic. Truly well done.

Gratitude for the opportunity to attend and gained knowledge.

Appreciation for the conference and the information received.

Positive feedback on the chance to interact with doctors and learn more.

Appreciation for the symposium & participation of experienced doctors.

A mention of feeling less alone after attending.

And for suggestions for future HAD symposiums, the following were shared by participants:

DAY 2 FEEDBACK SURVEY

1 DIVERSE TOPIC COVERAGE

Expand the range of topics tp include symptoms and issues beyond the aorta, such as mitral valve issues, gut health, allergies,a dn other common concerns among patients.

2 COPING AND LIVING STRATEGIES

Include sessions on coping with non-surgically treated dissections, managing life after surgery, and dealing with ongoing symptoms.

3 COMMUNICATING WITH HEALTHCARE PROVIDERS

Offer workshops on how to effectively communicate with doctors who may not be familiar with LDS.

4 PATIENT-CENTRED SESSIONS

Incorporate patient-led sessions where experienced patients can share their journeys and offer advice to newly diagnosed individuals.

5 PHYSICAL ACTIVITY AND LIFESTYLE GUIDANCE

Offer detailed advice on safe exercises, weight management, everyday life with LDS,work-life balance, mental health support, and applying for disability benefits.

6 INTERACTIVE AND ENGAGING FORMAT

Implement icebreaker activities, group discussions, and more time for patient interaction to foster connectivity and support among attendees.

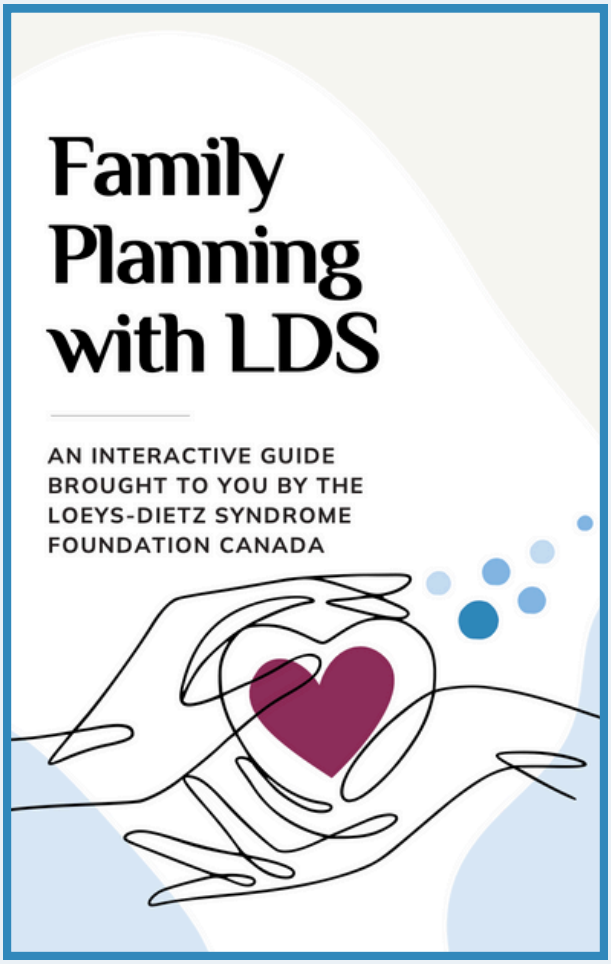
7 SPECIALIZED RESOURCES AND NAVIGATION

Provide guidance on navigating the healthcare system, accessing regular scans, and connecting wtih specialized services. Additionally, consider private consultations with specialists after their presentations.

7. Programs and Activities:

Overall, the HAD Symposium was a resounding success, fostering knowledge exchange and community support, and reinforcing our commitment to education and empowerment for those affected by hereditary aortic disorders.

III - December 2023: Launch of Bilingual Family Planning Educational Booklet



Recognizing the need for comprehensive information on family planning for those affected by Loeys-Dietz syndrome, LDSFC launched a new bilingual educational booklet in 2023. This resource covers important topics such as genetic counseling, pregnancy planning, post-partum care, and the potential risks and considerations for families. Our goal is to promote education, awareness, and empowerment for individuals and families so that they can take suitable and informed decisions.

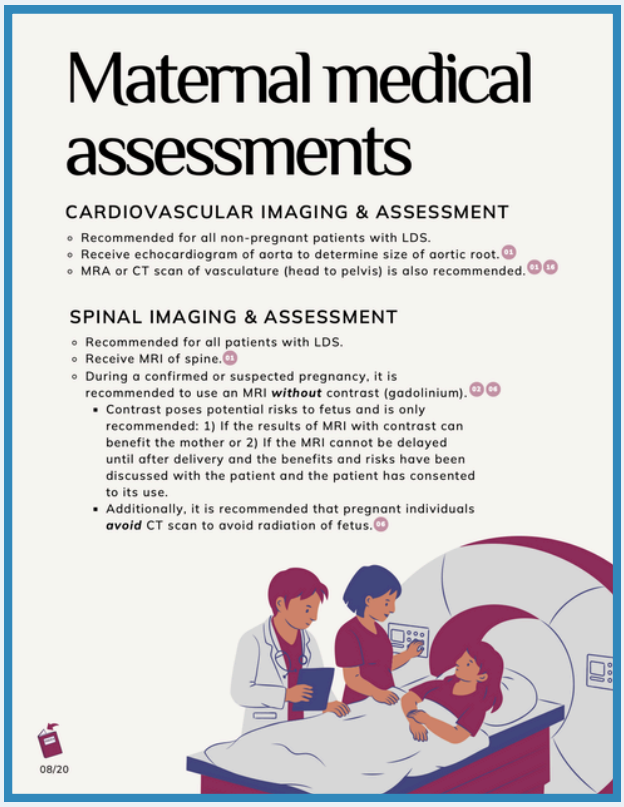
Individuals with LDS have a variety of family planning options available, including fertility preservation treatments, surrogacy, gamete and embryo donation, adoption, and natural pregnancy. We have provided a comprehensive guide to family planning that can be tailored to meet individual needs and circumstances.

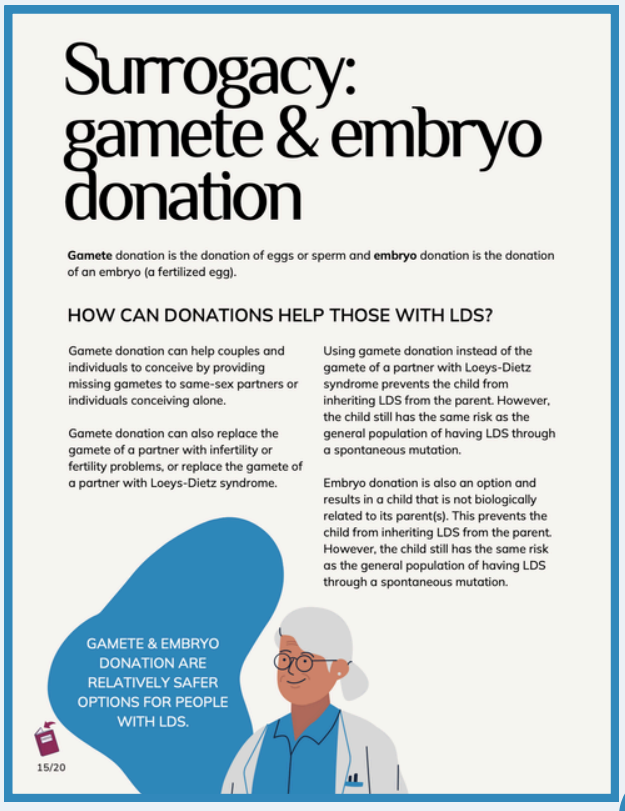
7. Programs and Activities:

We recognize that it is also crucial to manage the mental challenges that can accompany this journey. To mitigate these challenges, we have incorporated information on connecting with medical professionals, the LDS community, or equipping oneself with the right information to help combat feelings of isolation, depression, and anxiety. These connections provide a support network that is essential for emotional well-being during the family planning process.

Collaborating with a specialized, multi-disciplinary medical team is necessary to maintain a holistic wellness perspective and to attain desirable pregnancy outcomes with acknowledgement of the high maternal risk and the hereditary nature that LDS poses.

Through this guide, we have encouraged individuals to connect with their doctor at the pre-conception stage to have better chances of a healthy pregnancy and delivery. Therefore, continuous medical support and monitoring are indispensable for ensuring the best outcomes for both mother and child.
The booklet was developed in collaboration with subject experts to ensure accuracy and relevance. It has been well-received by the community and is available both in print and online through our social media platforms.





7. Programs and Activities:

Additionally, it is shared with patients during 1-on-1 support sessions and distributed in clinics through our scientific advisory board, ensuring broad accessibility. This focus to develop patient resources enables our foundation to step foot into providing accessible helpful resources that promote the health and quality of life of current and future generations with Loeys-Dietz Syndrome.

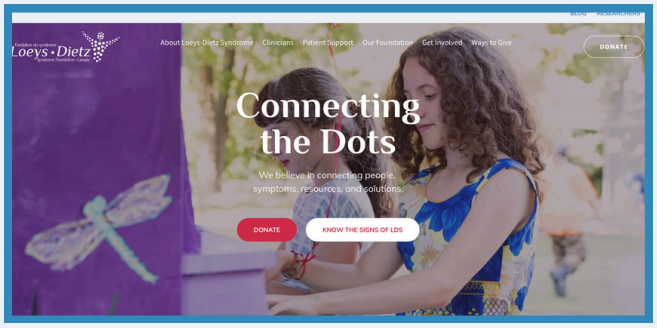
Once we launched the family planning booklet, we monitored and analyzed the engagement it was receiving through our online resources page on our website. The number of visits on the Family Planning with LDS segment during 2023 was 18 visits, with an average of 2.25 visits per user.

We also received questions from patients and families regarding certain topics developed in the booklet.

IV - 2023: Digital Communication and Learning

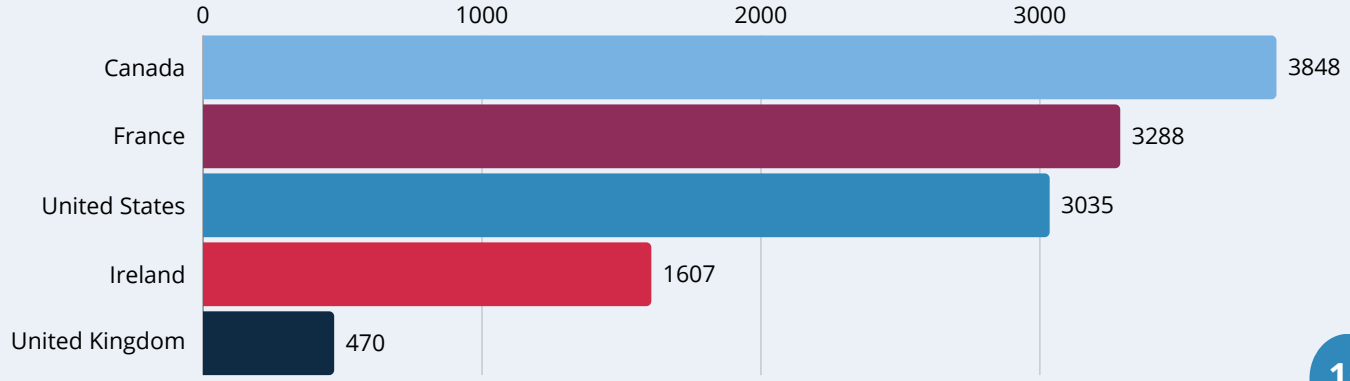
Our 2023 year focused on maintaining our virtual presence, whilst launching new educational resources. We also continued to moderate our new website and added new evidence-backed resources for the community to access.

Website 2023



Page Views: 75,485
Active Users: 15,981
Average duration per session: 2m 15s
Average pages viewed per session: 4.72

Users by Country:



7. Programs and Activities:

Pages with the highest number of visits:

Page title and screen class	Views	Users	Views per user	Average engagement time
Home – Loeys-Dietz Syndrome Foundation Canada - Loeys-Dietz Syndrome Foundation Canada	10610	3032	3.50	37.37
Signes et symptômes - FSLDC	8871	4276	2.07	72.97
Signs and Symptoms - Loeys-Dietz Syndrome Foundation Canada	3748	1593	2.35	83.10
Living Well with LDS - Loeys-Dietz Syndrome Foundation Canada	1585	602	2.63	174.01
What does Loeys-Dietz Syndrome Look Like? - Loeys-Dietz Syndrome Foundation Canada	1393	624	2.23	138.60
Syndrome de Loeys-Dietz - FSLDC	1325	599	2.21	72.56
Qu'est-ce que le tissu conjonctif ? - FSLDC	1199	562	2.13	108.34
About Loeys-Dietz Syndrome - Loeys-Dietz Syndrome Foundation Canada	1190	507	2.35	61.53
La génétique - FSLDC	1163	499	2.33	78.58
Receive a Diagnosis - Loeys-Dietz Syndrome Foundation Canada	1148	414	2.77	97.27
Genetics - Loeys-Dietz Syndrome Foundation Canada	1117	473	2.36	78.02
Home - FSLDC	889	384	2.32	25.74
Clinicians - Loeys-Dietz Syndrome Foundation Canada	876	355	2.47	62.09
Find Help - Loeys-Dietz Syndrome Foundation Canada	816	301	2.71	83.48
Loeys-Dietz Syndrome and Inflammatory Bowel Disease - Loeys-Dietz Syndrome Foundation Canada	742	307	2.42	136.82

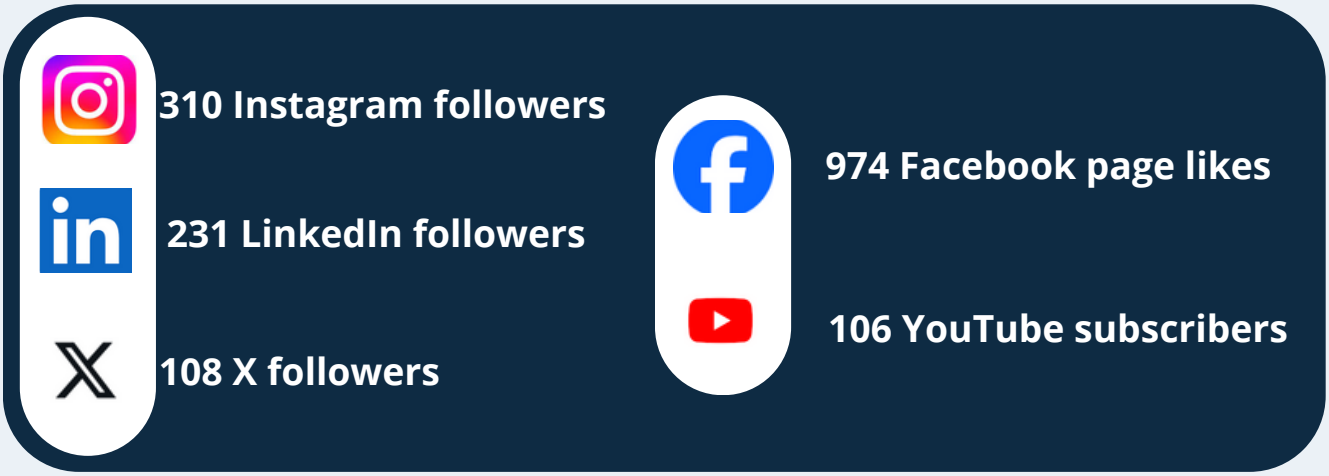
7. Programs and Activities:

Newsletter 2023

Theme	Opens (%)	Clicks (%)
Our New LDS Website is Here	28.3	2.6
Don't Miss Our 2023 LDS Events	28.8	1.9
SAVE THE DATE for the 2023 Heritable Aortic Disorders (HAD) Symposium	30.5	1.1
We're Celebrating Rare	35.9	7
Find Support for Loeys-Dietz Syndrome	35.7	6.4
Register Now for HAD	37.7	9.8
Register Now for HAD: Hotel	36.0	8.8
Allergies, Asthma, and More	25.9	0.9
Register Now for HAD: Testimonials	41.7	17.7
Register Now for HAD: Agenda	39.0	13.7
Website Repairs	28.7	0.4
Heritable Aortic Disorders (HAD) Symposium 2023	23.4	0.8
Embrace Connection in our Patient Support Group	31.3	0.6
Research Roundup: New Grants & Conference Highlights	31.6	1.7
Law 25 on Recent Privacy Provisions& Newsletter Sign Up	34.9	0.4
Reminder: Law 25 on Recent Privacy Provisions& Newsletter Sign Up	32.5	0.4
Advancements in LDSFC's Mission	74.4	33.4
Empowering Parenthood with Our New Guide:Family Planning with LDS	65.5	5.1

7. Programs and Activities:

Social Media



RESEARCH

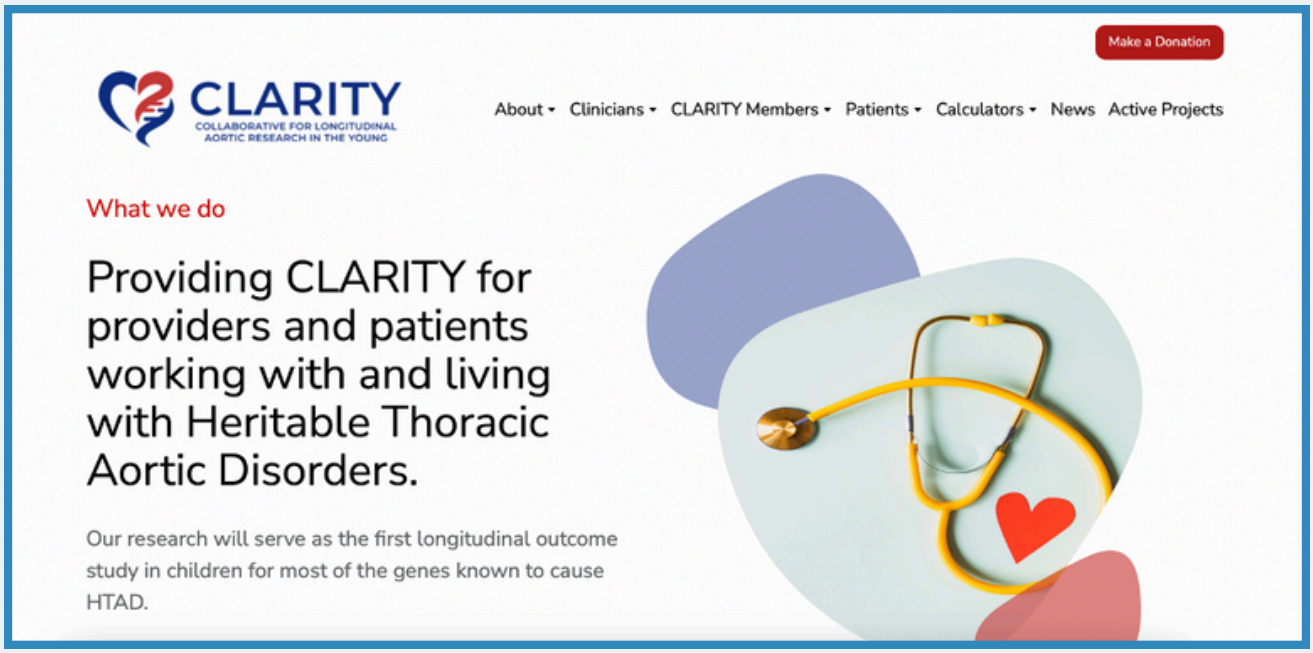
A key objective in our mission is to advance research. In 2023, we had a main program wherein this pillar was demonstrated. Namely, through the support of CLARITY.

I - Collaborative for Longitudinal Aortic Research In The Young - CLARITY

The **C**ollaborative for **L**ongitudinal **A**ortic **R**esearch **I**n **T**he **Y**oung is a multicenter collaborative initiative, led by Dr. Shaine Morris, aimed at improving outcomes for patients with aortic disease. By collecting the largest cohort of patients with longitudinal aortic imaging and clinical outcomes to date, CLARITY combines this data with other published information and collaborating registries to create advanced microsimulation models. These models predict dissection and mortality over the long term, tailored by gene and patient-specific characteristics, enabling a precision-based medicine approach. CLARITY supports clinicians with risk prediction and optimal care provision, aids informed decision-making regarding interventions and medications, and establishes structure for future longitudinal studies. Additionally, CLARITY's findings will help identify research targets for randomized controlled trials within the hereditary thoracic aortic disease (HTAD) population.

Throughout 2023, we supported and financed the development of the CLARITY brand, website, educational content, and communication plan. The support was both in-kind (total of 30 hours of Lindsay Parsons and 10 hours of Jida El Hajjar) and monetary (3,500$ for content creation and web development).

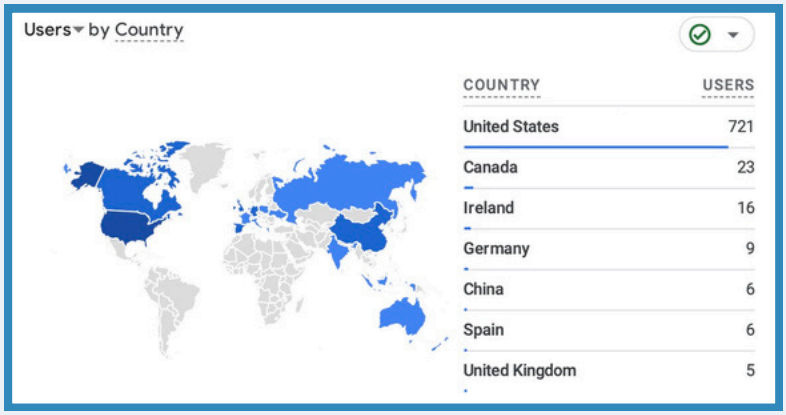
7. Programs and Activities:

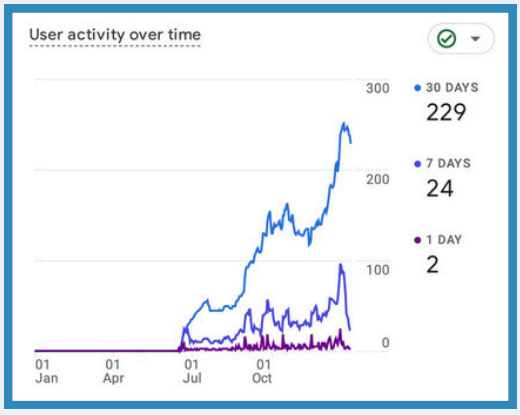


https://clarityregistry.com

Overall, 799 users clicked on the CLARITY website. The top 5 visitors were from the following countries: United States (721), Canada (23), Ireland (16), Germany (9), and China (6). Below are summary analytics exhibiting the demographic details of users by country (left), user activity (right), and the top pages accessed through the CLARITY website (bottom).

As we move forward, and as members of the CLARITY committee, we remain committed to keep supporting Dr. Morris on expanding and promoting the content of the CLARITY website.





7. Programs and Activities:

	Page path and screen class	↓ Views	Users	Views per user	Average engagement time	Event count All events
		1,457 100% of total	799 100% of total	1.82 Avg 0%	28s Avg 0%	4,339 100% of total
1	/	864	696	1.24	12s	2,784
2	/about_unused/members-and-committees/	103	61	1.69	48s	254
3	/for-patients/patient-enrollment/	72	40	1.80	1m 11s	187
4	/about/	60	32	1.88	36s	141
5	/calculators/risk-calculators/	57	33	1.73	41s	164
6	/for-patients/factsheets/	55	38	1.45	1m 05s	155
7	/for-clinicians/about-clarity/	43	30	1.43	41s	108
8	/calculators/risk-calculators/vti-calculator/	39	31	1.26	12s	122
9	/patient-enrolment/	35	22	1.59	4s	102
10	/about-2/	13	11	1.18	4s	30

II - Loeys-Dietz Syndrome Emergence Acceleration Program (LEAP) Papers

Launched in 2022, the LEAP Impact Award aims to identify, fund, and support investigators to encourage them to take a leap into Loeys-Dietz syndrome research. The LEAP Impact Award has the power to promote LDS research that also sheds light on other connective tissue and heritable aortic disorders, and to support LDS researchers as they progress to receiving larger grants and creating greater impacts.

In 2023, we are proud to have received the impact of two LEAP funded projects. The awardees have published their research papers in renowned scientific journals:

- The first article “TGFβ signaling pathways in human health and disease” is authored by the LEAP awardee Dr. Pei-Yu Chen et al. from Yale University. The article explored the cellular and biochemical mechanisms related to LDS and related disorders. The article can be found here: https://www.frontiersin.org/journals/molecular-biosciences/articles/10.3389/fmolb.2023.1113061/full. It currently has an attention score of 2, which is according to the journal considered an above-average attention score, compared to outputs of the same age and source (63rd percentile).

- The second paper “Mitral annular disjunction on cardiac MRI: Prevalence and association with disease severity in Loeys-Dietz syndrome” authored by LEAP awardee Dr. Gauri R. Karur et al. demonstrated that mitral annual disjunction (MAD) is prevalent in LDS and is related with progressive aortic dilatation and aortic events at younger age. What is very interesting is that MAD may be eventually a marker of disease severity necessitating close monitoring. Access to the full article here: https://www.sciencedirect.com/science/article/abs/pii/S0167527323011555

7. Programs and Activities:

Each of these LEAP awardees received a seed funding grant of $25,000 USD, leadership development coaching, and access to our global network of resources.

PATIENT SUPPORT

I - Patient Helpline

We are pleased to continue with our 2022 advances in expanding and fulfilling our patient support pillar of our mission. In 2023, our patient support helpline 1-888-LDS-FCAN has made great success and has allowed us to maintain a reference center of 522 resources for patients and their families.

Below is an excerpt of different specialists and healthcare providers that are available as a resource for those with Loeys-Dietz Syndrome.

Service Provider			Personal Information			
Type	Country / Province	City	Specialty	Age Treated	Need a Referral?	Language Spoken
Medical	BC	Vancouver	Electrophysiology; Cardiology	Pediatric	Y	Eng
Medical	MB	Winnipeg	Cardiology	Pediatric	Y	Eng; Greek
Medical	MB	Winnipeg	Orthopedics	Pediatric	Y	Eng
Medical	MB	Winnipeg	Aortopathy	Both	Y	Eng
Medical	MB	Winnipeg	Cardiology	Pediatric	Y	Eng
Medical	NS	Halifax	Medical Genetics; Genomics	Both	Y*	Eng; Fr
Medical	NS	Halifax	Medical Genetics; Genomics	Both	Y*	Eng; Fr interpreter
Medical	NS	Halifax	Medical Genetics; Genomics	Both	Y*	Eng; Fr interpreter

7. Programs and Activities:

The resource directory below highlights the services and resources that patients have accessed in the past year.

Total Number of Resources	***522***
Medical	169
Paramedical	56
Legal	12
Financial	38
Psychological	120
Genetic Testing	16
Educational & Informational	56
Clinical Trials	4
Global Organizations	51

Throughout this past year we assisted patients and families seeking various areas of support.

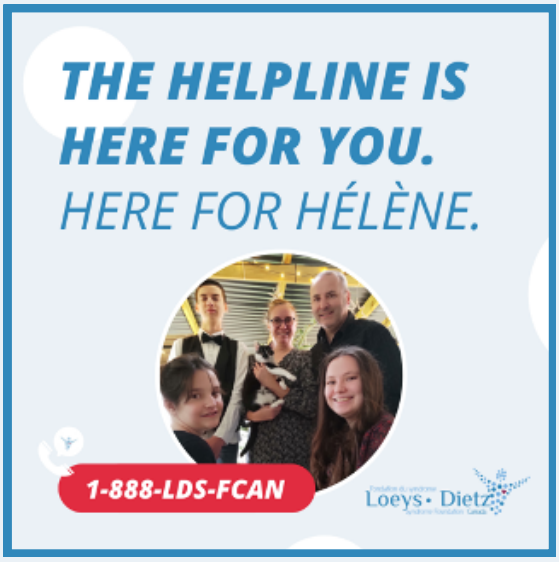
# of requests that we received and fulfilled from patients in 2023: 68

7. Programs and Activities:

The reasons of communications varied across different areas including:

seeking financial assistance: 1
insurance questions: 1
looking for reliable educational materials: 17
looking for a doctor: 23
seeking access to research: 4
seeking medical advice: 8
seeking community support: 12
looking for volunteer opportunity: 2

Our inquiries extended beyond local boundaries and gained international attention from over 6 countries, demonstrating the wide-reaching impact of our efforts.



II - August 2023: Monthly Peer-to-Peer Support Groups

In August 2023, we launched monthly virtual peer-to-peer support groups for patients and their families, for a total of 5 virtual support groups in 2023. The average attendance rate was 3 patients/caregivers per meeting.

Specific themes and discussion guides were planned for each group meeting. Each session is facilitated by trained volunteers who understand the unique needs of the Loeys-Dietz syndrome community.

7. Programs and Activities:

These groups are a cornerstone of our community outreach efforts, providing a safe and supportive environment for individuals to share their experiences, challenges, and successes.

The support groups have been instrumental in reducing feelings of isolation and providing practical advice, empowerment, and emotional support to participants. Through building a community, individuals with LDS can connect with others with similar challenges and find validation, hope, and acceptance within those support groups.

8. Collaborations and Partnerships:

I - Fundraising and Awareness Event with PPI

PPI's mission is to elevate independent Advisors to help them grow their business. PPI help Advisors assist their clients in achieving their most important life milestones with financial protection and security - through life insurance, living benefits, and investments - for education, home and family life, building a business, and retirement.

In November 2023, PPI hosted its annual networking event, raising substantial funds for the Loeys-Dietz Syndrome Foundation Canada (LDSFC) by donating the registration fees of participants. Our chair, Mr. Joseph Galli, was a guest speaker, emphasizing the importance of raising awareness about Loeys-Dietz syndrome (LDS).

The PPI event raised a total of $7,964.99 CAD, which will support ongoing research efforts and provide essential resources for affected families.

This collaboration with PPI not only provided crucial financial support but also significantly increased public awareness of Loeys-Dietz syndrome and underscored the importance of early diagnosis and management.

8. Collaborations and Partnerships

II - Advocacy with the Canadian Organization of Rare Disorders (CORD)

March 27, 2023: CORD planned a lobbying day at Parliament Hill in Ottawa, to raise awareness on the urgent unmet needs of Canadians living with a rare disease.

LDSFC, represented by Jida El Hajjar, participated in the meetings and consultations with 1) Health Canada Rare Disease Director Mr. Daniel McDonald and his team; 2) Health Canada Chief Medical Advisor Dr. Supriya Sharma; and 3) Member of Parliament Mr. Francesco Sorbara (Photo below).

By partnering with entities such as the Canadian Organization of Rare Disorders. we aim to advance the cause of rare diseases such as LDS and related disorders.

8. Collaborations and Partnerships

III - Sponsorship of the First European Loeys-Dietz Syndrome Patient and Research Networking Conference organized by Prof. Bart Loeys & Prof. Harry Dietz

LDSFC sponsored an amount of $5,000 CAD for this first edition of the LDS networking conference in Antwerp, Belgium that was held on June 24th and 25th 2023. Presentations on the patient perspective, basics and genetics of LDS, cardiovascular imaging and treatment, and more including breakout sessions were planned for patients and families.

https://vascern.eu/events/loeys-dietz-syndrome-patient-and-research-networking-conference/

9. Volunteer and Community Engagement

We are deeply grateful to our incredible community, whose dedication and passion inspire us every day. Specifically, we want to extend heartfelt thanks to our amazing 13 volunteers, whose unwavering commitment has significantly enhanced our resource directory, programs, blog articles, research inventory, website, and fundraising efforts.

The collective achievements we celebrate today are founded on the dedication and hard work of individuals like you, and we are truly thankful.

10. Transparency and Accountability

LDSF Canada is committed to maintaining the highest standards of governance and compliance. Transparency is at the core of our operations and financial management, ensuring that our organization is accountable to our stakeholders.

We recognize the crucial role of our external auditors, who work diligently to ensure that our accounting and financial reporting practices meet the highest standards. Their meticulous expertise enables us to maintain best practices, and we appreciate their support in our ongoing pursuit of excellence.

11. Future Directions: Advancing Care and Research with the CAN-ACT Registry

Looking ahead, the Loeys-Dietz Syndrome Foundation Canada is committed to expanding its reach and impact through several strategic initiatives. Key among these is the development of our CANadian Aortopathy and Connective Tissue Disorders Registry (CAN-ACT) (https://ccpcrn.ca/portfolio/canact-registry/), a comprehensive data collection effort focused to gain knowledge of these diseases in the pediatric Canadian population, in order to best identify and treat these patients, and guide advocacy at institutional and governmental levels to improve access to health care resources. CAN-ACT aims to determine:

- How many children in Canada are being treated for heritable thoracic aortic disorders (HTAD)?
- What are their genetic diagnoses and clinical features?
- How and where are they being treated?
- What is the impact of these diseases on their quality of life and physical activity?
- How can we better advocate for these children to improve their health care and quality of life?

In addition to the CAN-ACT registry, we are dedicated to enhancing our patient support services, increasing awareness and education efforts, and fostering cutting-edge research collaborations. Our goals include:

1. **Enhancing Patient Support Services:** We aim to provide tailored resources and guidance for LDS patients and families to support their mental health struggles.
2. Incr**easing Awareness and Education:** We plan to broaden our outreach efforts to educate both the public and healthcare professionals about LDS, promoting early diagnosis and better management of the condition.
3. **Strengthening Advocacy Efforts:** We are committed to advocating for the highest standard of care for LDS patients, working to ensure they receive the best possible support and medical attention.
4. **Collaborating Nationally and Internationally:** We will continue to work closely with both national and international partners, leveraging collective expertise to push the boundaries of knowledge and treatment for LDS.

By engaging with our community and leveraging the expertise of our Medical and Scientific Advisory Board, we are dedicated to transforming the future for individuals affected by Loeys-Dietz syndrome. Through these initiatives, LDSFC will continue to be at the forefront of improving lives and advancing the understanding of this complex disorder.

12. Acknowledgments and Gratitude

On behalf of the Canadian Loeys-Dietz Syndrome community, we are deeply grateful for your unwavering support in championing the importance of our rare disease.
For more information on LDS, to sign up for our newsletter, or make a donation:
please visit: https://loeysdietzcanada.org
or contact us at: info@loeysdietzcanada.org
Toll-free: 1-888-LDS-FCAN