INTO-HLH REGISTRY NEWSLETTER

WELCOME TO OUR SECOND ISSUE: A COMMUNITY OF HOPE

Dear Friends,

Welcome to the latest edition of the INTO-HLH Registry newsletter. This issue is a celebration of something truly powerful. Our community.

Behind every data point and research milestone are people: physicians dedicating their expertise, registry team members working tirelessly to abstract critical information, advocacy groups standing beside families, and most importantly, patients and their loved ones, who bravely share their stories. Together, they form the heart of what we call our Community of Hope.

In this issue, we've asked members of this community to share their "why" - what drives them to be part of this work, and what fuels their hope. Their words are moving, inspiring, and deeply human. We hope you see yourselves in their reflections and feel proud to be part of this growing, compassionate network.

Thank you for being here, and for being part of the story we're writing together, one of science, strength, and shared purpose.

With gratitude,
The INTO-HLH Registry Team

ENROLL HERE

GET INVOLVED

Your Story Matters — Share It!

One of the most meaningful ways you can support others affected by HLH is by sharing your story. Reach out to intohlh@cchmc.org to submit your or your family members story and photos for the INTO-HLH Registry website. Don't underestimate the power of your experience — share it today and inspire someone else on their HLH journey!

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Shining a Light on HLH

How Advocacy Groups Are Making a Difference

HISTIOCYTOSIS ASSOCIATION™
A Rare Community™

I felt for the other families and was determined to create an ongoing support network so that other families did not have to face their journey alone...We developed a hotline, and our kitchen table became a source of hope and connection, helping to answer questions and bring families together.

– Jeffrey Toughill
Founder of the Histiocytosis Association

The Histiocytosis Association was founded in 1986 by Jeff and Sally Toughill after their infant daughter Bethany was diagnosed with LCH, a rare and little-understood disease at the time. Faced with isolation, scarce information, and no clear treatment path, they launched the Association to create a support network for families like theirs. Over the years, their mission grew to include global research funding, education, and advocacy, resulting in over $7.5 million in research support and resources reaching more than 10,000 families annually. Today, the HA continues to address the urgent gaps in awareness, diagnosis, and treatment for histiocytic disorders—including HLH—by providing critical resources, fostering community, and driving scientific progress.

Histiocytosis Association of Canada

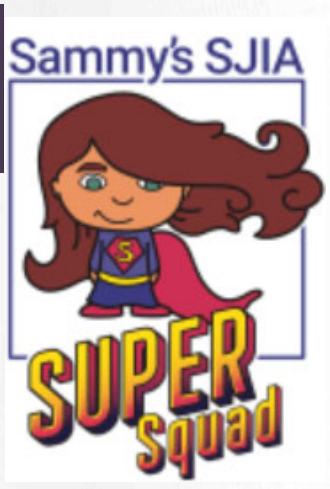
Joining the INTO-HLH Registry is important because the more patients or families that register, the higher the chances of gathering a stronger voice to help change educational and pharma protocols on research and drugs.

- Claudio DiGirolamo, President of the Histiocytosis Association of Canada

Histiocytosis Association of Canada (HAC) was reignited by a father whose daughter survived HLH thanks to a rare moment of recognition by an ICU doctor. That experience sparked a mission to support families facing this devastating disease. In partnership with HA and dedicated volunteers, HAC now provides advocacy, education, and physician referrals. With a focus on clinician collaboration and global partnerships, HAC is building awareness, expanding access to care, and urging families to join the HLH Registry to amplify the voice of this rare community and drive progress in research and treatment.

The course of an SJIA flare can be incredibly difficult to navigate and being aware of the possibility of MAS and symptoms to be on the lookout for can literally be the difference between life and death.

- Ronny Bachrach, Sammy's Mom & Founder of Sammy's SJIA Super Squad



Sammy's SJIA Super Squad began as a fundraising effort to raise awareness for systemic juvenile idiopathic arthritis (SJIA), a rare subtype of arthritis. It has since evolved into a platform to educate others about SJIA and its serious complication, macrophage activation syndrome (MAS) which is a form of secondary HLH. Run by Sammy's parents, the Squad shares up-to-date research, symptom awareness tools, and has raised over $100,000 for the Arthritis Foundation. They urge families to join the HLH registry, as data is key to driving research, improving early diagnosis, and advancing treatments for HLH/MAS.

Advocacy Group Spotlight

LIAM'S LIGHTHOUSE FOUNDATION

What inspired the creation of your advocacy group?

My son, Liam, was the inspiration. He went 10 months without a diagnosis as to why he was ill for so long. No specialist could diagnose him. If he had been diagnosed sooner, he might still be here with us today. The information/education about HLH just wasn't out there for us to find when Liam was finally diagnosed. I don't want other families to go through the difficult journey we went through, trying to find a diagnosis. The treatment of HLH is so harsh, resulting in higher mortality rates. Funding research for safer, more effective treatment methods leading to a cure is a priority for us. With more HLH research being done over the years, survival rates are on the rise, and physicians are becoming more aware of how to identify HLH and treat it sooner. We want to help change the outcome for those HLH patients/families that follow us.

What resources or services does your group provide?

We do much of our outreach through social media channels and email communication. We provide resources to inform families on finding the right care/hospital for them and where to get their questions answered, or if a second opinion might be needed. There is much to be said about having gone through the experience yourself. Being able to share that experience is priceless when helping families navigate an HLH diagnosis. We are in the process of re-establishing a family support fund for newly diagnosed HLH patients at Cincinnati Children's Hospital. We also hold a Meet & Greet annually as part of our 5K To Fight Histio fundraiser. This provides healing for so many of these families, whether they are survivors, still fighting, or angels.

What rewarding moments or achievements stand out since the creation of LLF?

I am proud to say we have had several rewarding moments and achievements over the past 15 years. Because we chose to share Liam's story, I have been informed that several patients have been properly diagnosed with HLH. We went back and educated the physicians that misdiagnosed Liam time after time. Liam has been the motivation for over 2,000 people joining the bone marrow registry. Because these people signed up to be a match for Liam, several have been called to donate to help save someone else's life. Of course, being able to fund just under one million dollars of research sits at the top! Each year we bring affected families together to provide support and share their stories, so they know they are not alone in this fight. Hearing what LLF has done for them in each of their journeys is the most rewarding accomplishment of all.

The Why Behind My Work:
A Journey Inspired by a Patient and Her Son

written by Adi Zoref-Lorenz

My journey into HLH research began with one unforgettable patient—a woman with chronic lymphocytic leukemia who came to the hospital very ill. She had persistent fevers, low blood counts, and signs of organ dysfunction. We suspected HLH, a rare but life-threatening immune reaction, but at the time, there were no clear guidelines for treating HLH in adults with cancer.

In the absence of established protocols, we turned to Professor Michael Jordan's "How I Treat HLH" paper in Blood, which focused mainly on the inherited (familial) form of the disease seen in children. We followed his approach, and although she initially responded to treatment, she sadly passed away a month and a half later.

What has stayed with me even more than the clinical course was her son's presence. He was an unwavering advocate—thoughtful, engaged, and determined to understand what was happening. After her passing, he continued to seek answers—not out of blame, but from a place of love and deep commitment. His questions mirrored my own and left a lasting impression on me.

That case became a turning point in my career. I knew I needed to understand HLH more deeply. That's what led me to travel to Cincinnati to train and conduct research with Dr. Michael Jordan—one of the world's leading HLH experts and now the scientific lead of the INTO-HLH Registry. Under his mentorship, I completed a PhD focused on HLH in the context of cancer. Today, I lead a laboratory and clinical service dedicated to this challenging condition.

The patient's son has remained in close contact with me throughout this journey. His support and encouragement have been a powerful source of motivation—and, in many ways, the fuel behind my dedication to this mission.

That's why I'm so committed to the INTO-HLH Registry. Together—as physicians, researchers, patients, and families—we are building a community dedicated to transforming how HLH is recognized, treated, and ultimately prevented, so that more patients have a chance at better outcomes.

Behind the Registry

Meet Members of the INTO-HLH Registry Abstraction Team

TAYLORANNE KAUFMANN

As a Registered Nurse, I have always been drawn to work that allows me to truly make a difference in the lives of patients and their families. Being a part of the INTO-HLH Registry has allowed me to do just that. It is incredibly meaningful to contribute to research that not only advances our understanding of HLH but also works towards improving patient outcomes. Working on this registry has strengthened my passion for research, evidence-based medicine, and rare disease advocacy. It has inspired me to continue my education so I can make an even bigger impact on patient outcomes. My goal is to continue contributing to advancements in treatment and patient care through both nursing and clinical research.

I would like families affected by HLH to know they are not alone. I have seen how overwhelming an HLH diagnosis can be, but there is a strong community of doctors, nurses, researchers, and families walking alongside you. There is hope, and you have a team fighting with you and for you.

Outside of work, I enjoy spending time outdoors, traveling, and reading. I also recently bought my first house, so a lot of time has been spent on house projects and remodeling.

JACKALYN WANDSTRAT

The INTO-HLH webpage drew me into this work. I was deeply intrigued as I scrolled through the different sections. I thoroughly enjoyed reading all that has been discovered about the rare disease that is HLH. I was excited to aid in the process of solidifying our current knowledge and uncovering more. The Patient and Families tab on the webpage specifically keeps me motivated. Reading about some of the patients and their families' fights is truly inspiring. Their perseverance and resilience remind me to stay committed to accomplishing the Registry's goals. There are many things that continue to give me hope for the future of HLH. Families are more than willing to be a part of this study, knowing that there are no direct benefits for them. They always seem eager to help future families affected by HLH through participation in the Registry. The families I am lucky enough to meet make the work we do much more meaningful. This work has provided me with valuable experiences that I will carry with me throughout my career. It has truly deepened my compassion for people who experience these life-altering medical conditions and has given me a profound gratitude for life itself.

I would like families affected by HLH to know that their resiliency and bravery in the face of HLH is incredibly inspiring. It inspires the team to consistently put in the hard work required to create a better understanding of HLH in order to help them win their fight and future family's fights.

Outside of work, I am a mom to my 2 ½-year-old, Jaxson. We spend lots of time outside at the pool and park lately. We love to hang out with family and friends, and we are deeply grateful for the time we get to spend together.

Registry Roundup

Progress Updates from the INTO-HLH Registry Team

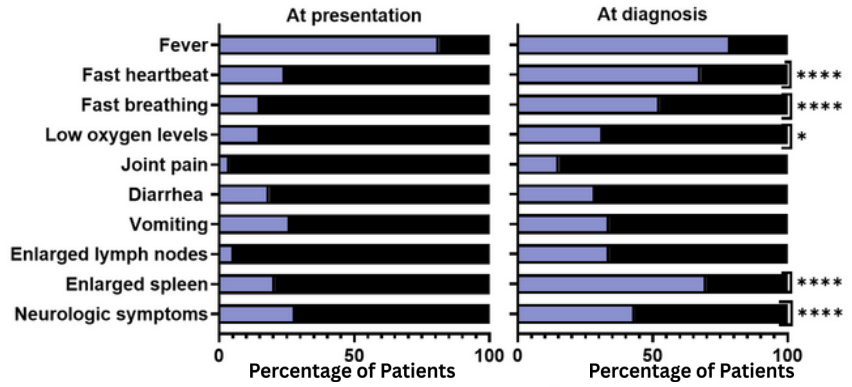
126 Active Participants

Goal: 200 Active Participants

Symptoms at Presentation vs. Diagnosis

How symptoms change from the start of illness to HLH diagnosis:

This chart shows how common certain symptoms are at a patient's initial presentation — when the patient first came to the hospital or general practitioner with HLH symptoms (left bars) — compared to when HLH was officially diagnosed (right bars). Each bar shows the percentage of patients with each issue at these two time points. The asterisks (*) show the significant changes — more stars mean a stronger difference.



What we learn from this chart:

Symptoms such as fast heartbeat, fast breathing, and low oxygen levels in the blood were more common at the time of HLH diagnosis than at the patient's initial presentation. Neurological symptoms (such as confusion, seizures, or difficulty moving) and enlarged spleen (splenomegaly) also became more frequent as the illness progressed. This suggests that these symptoms tend to appear or get worse over time if HLH isn't recognized and treated early.

Why it matters:

Understanding how HLH symptoms develop can help doctors and caregivers know what to watch for. Recognizing that serious symptoms like trouble breathing or neurological changes often come later can lead to earlier diagnosis and faster treatment, which may improve patient outcomes.

CALL TO ACTION

The INTO-HLH Registry is currently primarily composed of participants with Familial HLH. If you or your loved one have secondary HLH, MAS, Cytokine Release Syndrome, or other forms of HLH, please visit our website to learn more more about joining the Registry and how your story can help drive progress.