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# Illness Identity as an Emerging Concept in Adult Congenital Heart Disease

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Due to the numerous advances in the worlds of pediatric cardiology and cardiothoracic surgery, there are now more adults than children living with Congenital Heart Disease. Survival into adulthood is as high as 97%, with over 75% of those surviving into adulthood reaching 60 years of age.[1] However, living with Congenital Heart Disease (CHD) or Adult Congenital Heart Disease (ACHD), especially through middle age and beyond, comes with numerous emotional and psychosocial challenges.[2] Recently, researchers have begun exploring these challenges through prospective studies, seeking to understand the nature of these challenges and their impact on quality of life (QoL).[2-4]

Data have primarily been mixed comparing QoL between patients living with ACHD and the general public. On the one hand, some studies have found that people with ACHD report higher degrees of life satisfaction than the general population, perhaps because of overcoming challenges related to their illness.[4] On the other hand, some studies have shown a lower self-reported QoL in people with ACHD, regardless of disease severity.[4] For adolescents and young adults ACHD may hinder social acceptance as it limits their participation in various school activities; for those seeking employment, ACHD may have implications for employability; and, for those who wish to have biological children, ACHD often influences decision-making surrounding pregnancy.[3,5] Lastly, many people with ACHD report significant anxiety as it relates to disease progression and prognosis, post-traumatic stress related to hospitalizations and procedures, and survivor's guilt resulting in hesitancy to engage in peer groups.[3]

Most adults living with Congenital Heart Disease were diagnosed with their condition at or near birth, though occasionally a congenital heart lesion may be found in adulthood. Either way, these conditions can be repaired or palliated but never truly cured, sometimes due to the late sequelae of interventions. Even the simple, transcatheter closure of an atrial septal defect requires routine follow-up to monitor for potential complications.[6] As a result, ACHD is a chronic, lifelong illness, and it behooves us as clinicians to help patients face the unique challenges that this illness can bring.

## What is Illness Identity?

The impact of chronic illness on a person's identity has been deeply explored by sociologists such as Dr. Kathy Charmaz, who coined the term illness identity as, "the degree to which a chronic illness becomes integrated into a person's identity." This encompasses four dimensions: engulfment, rejection, acceptance, and enrichment[7,8] (**Figure 1**). Engulfment refers to how much one's identity is dominated by their illness, influencing their daily life and impacting all of their decisions. Rejection refers to how much one refuses to incorporate their illness into their identity, sometimes avoiding treatment and other behaviors that remind them of their illness. Acceptance refers to a balanced incorporation of one's illness into their identity while still maintaining other aspects of their identity. And finally, enrichment refers to how much one recognizes positive life changes as a result of their condition.[8]

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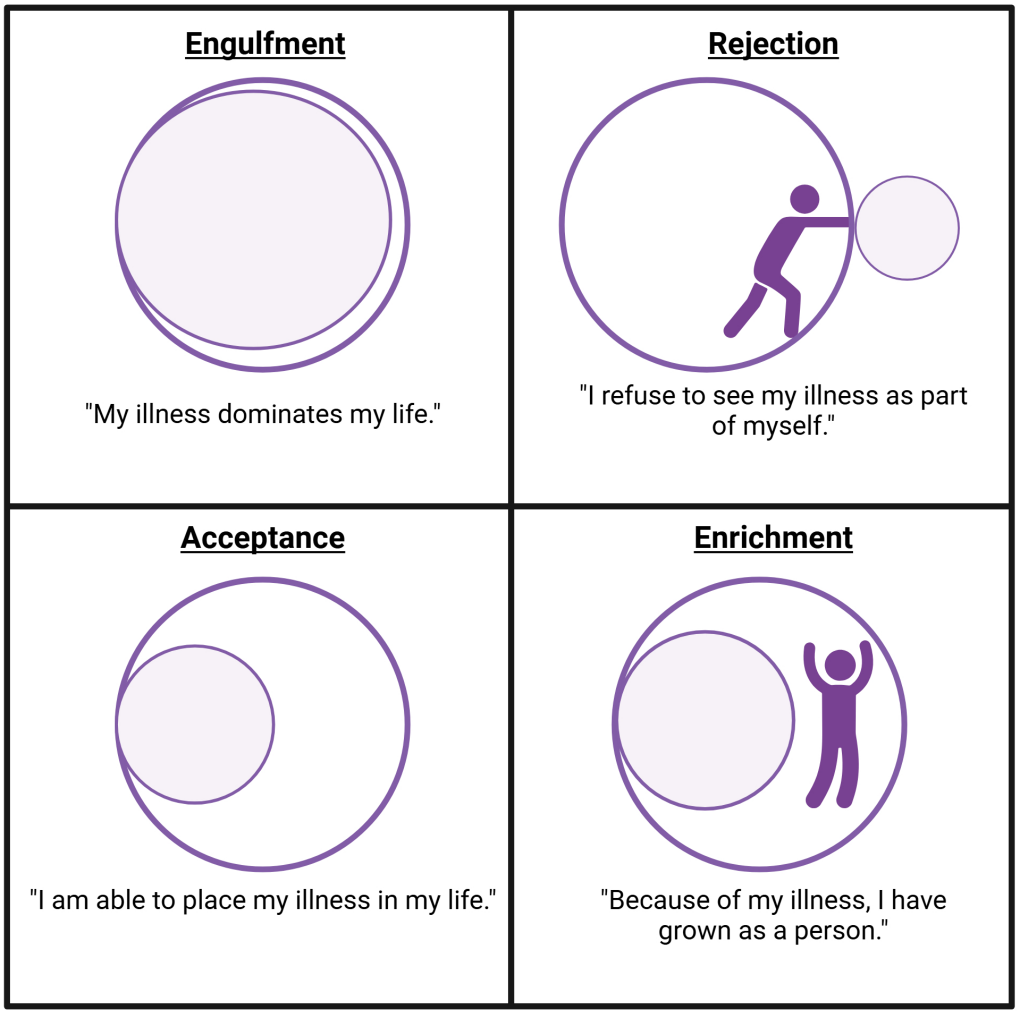
FIGURE 1 *Adapted from Van Bulck et al[8]*




Illness identity dimensions can be divided into those that are more integrative (acceptance and enrichment) and those that are non-integrative (rejection and engulfment).[9] In general, this distinction has been found to parallel adaptive and maladaptive psychosocial functioning as it relates to mental health, psychological QoL, and other patient-reported outcomes (PROs).[9-14] Engulfment, in particular, has been consistently shown to correlate strongly with depressive symptoms, anxiety, physical symptoms, and even absence from work or school.[9-15] Engulfment has also been explored as a novel predictor of healthcare use, as the more engulfed a person is by their illness, the more likely this person is to be either hospitalized or see a healthcare provider.[16] The relationship between the other illness identity dimensions and PROs is less strong, though rejection appears to be related to anxiety, while the integrative illness identity dimensions are at least somewhat correlated with higher QoL, less depressive symptoms, less anxiety, and more life satisfaction.[9-15]

## What is Known About Illness Identity in Chronic Disease?

The Illness Identity Questionnaire (IIQ), created by Dr. Leen Oris while at KU Leuven, allows for illness identity to be studied more systematically.[9] The IIQ consists of 25 statements that the reader rates on a five-point scale from one (strongly disagree) to five (strongly agree). Example statements include, "I refuse to see my illness as part of myself," and, "Because of my illness, I have grown as a person," each corresponding to one of the four illness identity dimensions. This questionnaire has since been validated in several chronic disease populations including adults with type 1 diabetes mellitus (T1DM),[9] mixed connective tissue disorders,[10] inflammatory bowel disease,[11] celiac disease,[12] refractory epilepsy,[13] as well as ACHD.[10]

Many of the existing illness identity studies are cross-sectional, so the degree to which illness identity affects PROs and vice versa cannot be ascertained. Indeed, several theories are described in these reports regarding the bidirectionality of a person's experience and their illness identity. For example, those with the most severe disease may feel engulfed by their condition because of how often they are hospitalized, which may reinforce depressive thoughts, making them less likely to engage in health-related behaviors, ultimately worsening their overall health and making them feel more engulfed by their condition. To address these concerns, several studies have emerged from the original KU Leuven group to study illness identity more longitudinally.[17-19]

In the first study, Rassart et al followed adolescents with T1DM prospectively over three years, and found that illness identity correlated with hemoglobin A1c values, treatment adherence, and diabetes-specific distress.[17] More specifically, high rejection and engulfment predicted diabetes-specific distress, which in turn predicted further rejection and engulfment, while high enrichment predicted treatment adherence, which in turn predicted further enrichment.[17] Interestingly, none of the illness identity dimensions predicted changes in the hemoglobin A1c, but a high A1c predicted increases in engulfment, suggesting that poorly controlled disease may limit illness identity integration.[17]

Two subsequent studies then followed patients with ACHD to identify longitudinal trajectories of illness identity, and the relationship between illness identity and well-being.[18,19] Overall, illness identity was found to have relationships with age, ACHD complexity, PROs, and healthcare use.[19] Notably, ACHD complexity was found to be strongly associated with both engulfment and enrichment, suggesting that engulfment and enrichment may be responses to severe illness.[19] Regarding illness identity and well-being, integrative dimensions correlated with improved well-being (and non-integrative dimensions with worse well-being), but only acceptance and engulfment emerged as impacting well-being over time when directionality was examined.[18]

In all three studies, illness identity was found to be stable over the study period (roughly three years), and the most recent study in ACHD described three "classes" of illness identity groups that emerged in the analysis.[17-19] These classes were characterized by clusters of scores across the illness identity dimensions: class one had high enrichment and acceptance but low rejection and engulfment; class two had high rejection with low scores in the other three dimensions; and class three had low acceptance with high scores in the other three dimensions.[18] Interestingly, classes one and two had similarly high QoL with low depressive symptoms, but class three had low QoL with high depressive symptoms suggesting a potential role of clustering illness identity dimensions, as well as confirming the strong correlation seen between QoL and depressive symptoms with engulfment.[18]

## Future Directions for Illness Identity in Adult Congenital Heart Disease

Initial work demonstrates the applicability and implications of illness identity in a number of chronic disease populations, and has primarily focused on its relationship with QoL and PROs. However, there remains significant work to be done, particularly regarding its clinical significance in ACHD.

To further study the clinical significance of illness identity in ACHD, it must first be distinguished from illness severity. Much of the initial work done suggests that illness severity is not clearly associated with illness identity, though illness severity was only partially encompassed in these studies by surrogates such as ACHD anatomic complexity by the Bethesda classification, hypoxemia, and New York Heart Association functional class. Recently, ACHD guidelines have clarified how to define illness severity in terms of physiology in the new ACHD AP classification system.[20,21] Therefore, a plausible next step would be to distinguish illness identity from illness severity as defined by the ACHD AP system, as well as other objective measures of illness severity such as peak oxygen consumption and levels of circulating biomarkers, which is currently being studied here at New York University ACHD. We hypothesize that those with the most severe illnesses are more likely to be either engulfed or enriched by their illness as suggested by prior studies, but that illness severity alone will be insufficient to predict illness identity integration or non-integration. Further studies may then explore whether illness identity integration is prognostic for clinical outcomes, other factors predictive of illness identity integration vs non-integration, and whether illness identity may be an intervenable target to improve the well-being of our ACHD patients.

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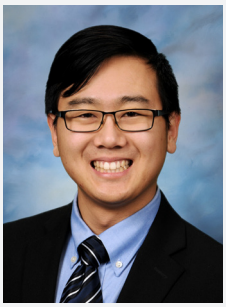

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# The CHD Data Desert: Why We Still Do Not Have Enough Real-World Heart Data—and How AI Can Help Fill the Gaps

*Stuart Long*

For millions of patients living with Congenital Heart Defects (CHDs), initial pediatric interventions are just the beginning of their cardiac journey.

CHD patients can encounter additional risk factors, considerations, and complications—often making for a complex and nuanced care journey. Unfortunately, healthcare providers are often forced to navigate this complex landscape without a map. According to reports, nearly 80% of healthcare data remains unstructured and inaccessible,[1] limiting its use for insightful decision-making. This affects all areas of healthcare, including cardiology, especially when it comes to the treatment of CHDs.

In the realm of CHD, real-world cardiac data remains even more scarce, fragmented, and inconsistent, creating what many experts call a "data desert."

Despite being the most common birth defect globally,[2] CHD is often misunderstood in real-world clinical contexts. Read on to learn how AI has the potential to address the CHD "data desert" by unleashing an abundance of insight, enabling more effective clinical oversight and more proactive interventions to flourish.

## The Clinical Significance of High-Acuity CHD Data

Advances in surgical and medical management have transformed CHD survival rates over the past several decades, but for the 2.4 million Americans currently living with CHD,[3] gaps in long-term, real-world cardiac data continue to create blind spots in patient care, risk assessment, and treatment optimization.

The consequences of this data deficiency are profound. Without a continuous stream of high-acuity cardiac data, healthcare providers may struggle to accurately track disease progression, anticipate complications, and tailor interventions to individual patients. Research tells us that CHD patients face a dramatically higher risk of heart failure—220 times higher for children (infants through 17 years old)[4] and 8.7 times higher for adults compared to their peers.[5]

But heart failure most often develops gradually. With more complete cardiac monitoring data, clinicians can identify subclinical warning signs or early-stage heart dysfunction, such as subtle ischemic changes, depolarization/repolarization abnormalities, and arrhythmia precursors. AI-driven pattern recognition, risk stratification, and alerts can enhance clinicians' ability to intervene before early-stage cardiac dysfunction progresses to irreversible structural damage to the heart and, ultimately, progressive heart failure.

But the impact of high-quality CHD data extends far beyond preventing individual cases of heart failure. Collectively, these datasets can improve our understanding of CHD's lifelong progression, refine predictive models, and enable more proactive risk management.

CHD is vitally important, both for individual patients and for the entire CHD community. Why, then, is high-quality CHD data in such scarcity? We will explore the challenges behind the CHD data desert—and how we can overcome them.

## Key Challenges Leading to CHD's Data Deficiency

Understanding the barriers to data collection and integration is essential for identifying solutions. The following challenges highlight why CHD remains a data desert, preventing patients, clinicians, and researchers from making data-driven progress in this critical disease state.

## Challenge 1: Transition Gaps Between Pediatric and Adult CHD Care

CHD is a lifelong condition, yet patient records infrequently follow individuals from infancy through adulthood. Unlike some other chronic conditions with robust, centralized registries tracking long-term outcomes, CHD data is highly fragmented.

Even though 40,000 US infants are born with CHD each year,[6] there's no centralized registry tracking lifelong outcomes. While some CHD-specific registries exist, they are not universally integrated across pediatric and adult care, leaving patients without comprehensive, lifelong tracking. And given that many CHD cases require multiple interventions, including staged surgical repairs, a lack of centralized tracking creates challenges in maintaining long-term cardiovascular health. This is concerning, considering that 30% of cases require staged-repairs, and another 15% will require subsequent re-operations.[7]

As CHD patients age, they must transition from pediatric to adult cardiology—yet this transition is often marked by changes and gaps in care. One study reveals 42% of adults with CHD have gaps in care longer than three years, and 8% have gaps longer than a decade[8] —which leaves many patients vulnerable to undiagnosed complications, hospitalizations, and poorer long-term outcomes. The same study also found the mean age at the first gap in care was 19.9 years,[9] suggesting the transition from pediatric to adult care is a critical period for maintaining follow-up.

One barrier to seamless care transitions is poor interoperability between pediatric and adult cardiology systems.[10] Different EHRs create fragmented medical histories, making it difficult for adult cardiologists to track a patient's surgical history, medications, and diagnostics. Without shared data, patients must repeatedly relay their history, increasing the risk of misdiagnosis, redundant testing, and improper treatment.

Compounding this issue is the lack of CHD-specific expertise among adult cardiologists.[11] Many general cardiologists specialize in acquired heart disease (e.g. coronary artery disease or hypertension) and may have limited training in congenital conditions. CHD patients transitioning to adult care may find

that their new providers lack experience with complex congenital heart anatomy and long-term complications, leading to suboptimal or delayed interventions.

Even when a patient finds a specialist, clinician shortages, distance, and expense often lengthen the time between in-office visits, where the vast majority of monitoring tests occur. These gaps leave long periods of 'data darkness' in which key clinical changes may go unnoticed until they cause acute complications.

## Challenge 2: Lack of Longitudinal Data Tracking

One of the most significant challenges in treating CHD lies in how cardiac performance is monitored. Cardiac assessments are typically episodic, relying on periodic office visits or short-term tests that provide just a snapshot of a patient's heart function. Standard ECG tests may last five to 15 minutes, but they only capture seconds of cardiac data at a time,[12] making it easy to miss transient but serious arrhythmias. Holter monitors capture 24 to 48 hours of activity, but they also fail to provide the long-term data needed to track CHD progression.[13]

While these brief observations can reveal acute issues, they are unable to fully capture the nuances of cardiac performance longitudinally. Indeed, CHD is not solely about what happens during a single doctor's visit or over a few hours on a Holter monitor; it is about how a patient's heart—which is structurally anomalous—responds to the realities of living. Without a continuous, real-world longitudinal dataset gathered in the context of daily life, clinicians are left with an incomplete picture, making it harder to detect the earliest, most nuanced signs of worsening function.

In addition, a lack of comparative data makes it challenging to measure how an individual's heart function is changing in relation to previous baselines. When a clinician sees a CHD patient for the first time in adulthood, there may be no continuous dataset to compare against—only fragmented medical records and single-moment assessments. This makes it difficult to distinguish whether a patient's cardiac performance is stable, declining, or compensating in ways that may not be immediately apparent.

To improve CHD management in adults, the shift toward high-acuity longitudinal monitoring is critical. By capturing continuous cardiac data outside of the clinic, providers can better understand how a patient's heart performs over time—not just during clinical events but during their daily routine. This approach not only helps identify progression earlier, but also enables more personalized, data-driven care that adapts to the evolving nature of CHD, and ultimately, improves long-term outcomes.

## Challenge 3: Underrepresentation in Clinical Research and Outsized Economic Impact

CHD's complexity and heterogeneity make it one of the most underrepresented conditions in cardiovascular research.[14] Unlike common acquired heart diseases, CHD presents with wide anatomical variations,[15] differing surgical histories, and long-term complications that vary significantly across patient populations. These factors make it difficult to create standardized research cohorts, leading to critical knowledge gaps in understanding long-term risks, emerging treatments, and preventative care strategies.

Additionally, the majority of CHD research focuses on early childhood outcomes, leaving limited data on long-term complications like arrhythmias, HF, and stroke risk. Furthermore, CHD also carries a substantial financial burden. CHD-related hospital costs exceed $9.8 billion annually,[16] yet most data-driven cost reduction strategies focus on adult cardiovascular disease, not CHD. The high cost of care stems from multiple factors like: complex surgeries and extended hospital stays for newborns and children, frequent specialist visits, imaging, medications throughout life, and repeat hospitalizations due to undiagnosed complications, missed follow-ups, or disease progression.

Addressing these collective challenges requires a smarter, more efficient approach to cardiac monitoring that harnesses advanced technology to deepen and automate insight. AI-enabled remote cardiac monitoring is one technology that can bridge the gaps, reshaping the future of CHD management.

## An Impactful Solution: How AI-Enabled Remote Cardiac Monitoring Creates a Bounty of Insight

Remote cardiac monitoring can transform CHD care by generating near real-time data and using AI to analyze it instantly—without adding to clinicians' workloads. By continuously tracking subtle cardiac changes, AI-driven monitoring fosters greater continuity between pediatric and adult care, enables proactive interventions, and enhances efficiency at scale.

Let's look at how AI-enabled remote cardiac monitoring can transform the CHD data desert into a bounty of insight, helping raise the standard of care.

## Solution 1: Bridges the Gap Between Childhood and Adulthood

AI-enabled remote cardiac monitoring can help bridge the disconnect that often occurs when CHD patients transition to adulthood. By extending powerful data visibility into adulthood and ensuring data is readily available as a patient moves between providers, this technology enables new continuity of care for patients living with CHD. AI can also alleviate challenges related to poor interoperability and lack of data continuity across providers. In fact, one study on AI-powered data integration in healthcare claims processing highlights how AI systems can automatically convert and standardize data across formats.[17]

Finally, AI-enabled cardiac monitoring addresses specialist shortages by ensuring cardiologists can monitor patients remotely and with ease, receiving near real-time insights and alerts about potential complications before they escalate. As a result, patients distributed across geographies have the potential to identify specialists for their specific congenital heart defect, then engage with them from afar—without expensive, time-consuming travel. Patients living with CHD in remote areas are similarly empowered to access higher quality care. Over time, this can incentivize cardiologists to research and specialize in niche defects by letting them engage with patients from a much larger area.

## Solution 2: Unlocks Longitudinal CHD Insights

AI-enabled remote cardiac monitoring is transforming CHD management by shifting from episodic testing to continuous tracking. Traditional cardiac assessments capture only brief snapshots, often missing early warning signs of complications. In contrast, AI-powered monitoring surfaces meaningful patterns over time—offering deeper insights into the early stages of arrhythmias, heart failure, and other cardiovascular risks.

For example, subtle fluctuations in heart rate trends over days or weeks may signal early-stage deterioration, even in the absence of acute symptoms during an office visit. Similarly, changes in autonomic regulation, nocturnal heart rate variability, or response to daily activities can indicate heart failure progression before it becomes clinically

evident. These insights, often imperceptible in traditional assessments, become clear with long-term, AI-enhanced monitoring.

Additionally, AI-powered analytics enable clinicians to compare new data against a patient's historical baseline, rather than relying on isolated ECG readings. This is especially critical for CHD patients, whose cardiac function may evolve unpredictably over time. Research suggests that AI-driven CHD-specific learning algorithms could improve imaging efficiency, enhance diagnostic accuracy, and provide early warnings of structural or functional deterioration.[18] When paired with continuous monitoring, AI helps detect subtle but meaningful shifts, allowing for earlier, more precise interventions.

By expanding care beyond episodic assessments, AI-driven monitoring helps shift CHD management from reactive to proactive data-driven care. This continuous visibility enables more personalized treatment strategies, improving long-term outcomes for patients.

## Solution 3: Scales High Quality Insight in the CHD Community

The wide range of anatomical variations and diverse surgical interventions in CHD has historically made it difficult to conduct large-scale, standardized studies that yield meaningful insights across patient populations. Traditional CHD research is often constrained by small sample sizes and fragmented datasets, making it challenging to draw definitive conclusions about long-term outcomes and treatment efficacy.

AI-enabled remote cardiac monitoring helps address these constraints by supporting scalable, high-quality data collection across entire populations of patients with specific congenital defects. AI-powered analytics can stratify patients based on an infinite number of clinical variables, allowing researchers to analyze the nuances of disease progression, surgical outcomes, and intervention effectiveness in ways that were previously difficult.

Traditional CHD studies are often limited by the episodic nature of data collection, where only isolated points in time are available for analysis. AI-enabled monitoring aggregates real-world cardiac data continuously, uncovering previously hidden trends to inform predictive models and lead to more effective, evidence-based treatment strategies. With AI's ability to identify patterns across diverse patient groups, researchers and clinicians can more clearly understand which treatments and interventions contribute to better long-term outcomes.

Beyond research, AI-driven monitoring has the potential to improve long-term CHD management while reducing costs. By identifying patients at the highest risk for deterioration earlier, AI enables more timely intervention and prevents unnecessary hospitalizations. A recent study of infants later diagnosed with CHD found that late detection was significantly associated with 52% more hospital admissions, 18% more hospitalized days, and 35% higher inpatient costs.[19] Expanding access to AI-driven monitoring could help mitigate this, ensuring patients receive earlier, more effective care.

## Turning the CHD Data Desert into a Data-Driven Future

For too long, CHD care has been hindered by fragmented data, gaps in long-term monitoring, and a lack of comprehensive research. The consequences—missed diagnoses, preventable hospitalizations, and an unclear understanding of long-term outcomes—underscore the urgent need for a more connected, data-driven approach to CHD management.

AI-powered remote cardiac monitoring is driving advancements in CHD care by enabling continuous, scalable, and high-quality data collection that transforms how CHD is studied and managed. With deeper insights, earlier interventions, and better long-term tracking, the future of CHD care is no longer defined by gaps and guesswork—it's being rewritten by data. For patients living with CHD, the forecast is changing. The data desert is no more.

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# The SickKids Advanced Cardiology Education (ACE) Program

*Alyssa Gumapac, BHSc, Communications Specialist, SickKids Learning Institute*


As one of the world's leading heart centres, we here at the Hospital for Sick Children (SickKids) have an inherent responsibility to drive innovation in the treatment and management of children with Congenital and acquired Heart Disease. Through transformative research, education, international collaboration, and patient-centered care, we strive to shape the future of cardiac clinical care.

While it's easy to focus on the latest technological advancements and novel techniques, it's equally important to reflect on the historical breakthroughs that have shaped modern cardiac care. By sharing knowledge and embracing diverse perspectives, we can continue to optimize outcomes for children with heart disease—principles that lie at the heart of SickKids Advanced Cardiology Education (ACE) Program.

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## What to Expect

Throughout the program, you will explore cutting-edge technologies such as Ventricular Assist Devices (VADs) which play a crucial role in bridging care for patients awaiting other interventions, including heart transplants. Additionally, Dr. Luc Mertens, Section Head of Echocardiography and Co-Director of the Pulmonary Hypertension Program at SickKids, will review echocardiograms of single ventricular physiology—an incredibly complex subspecialty in which new surgical interventions have significantly improved infant survival rates over the last 50 years.

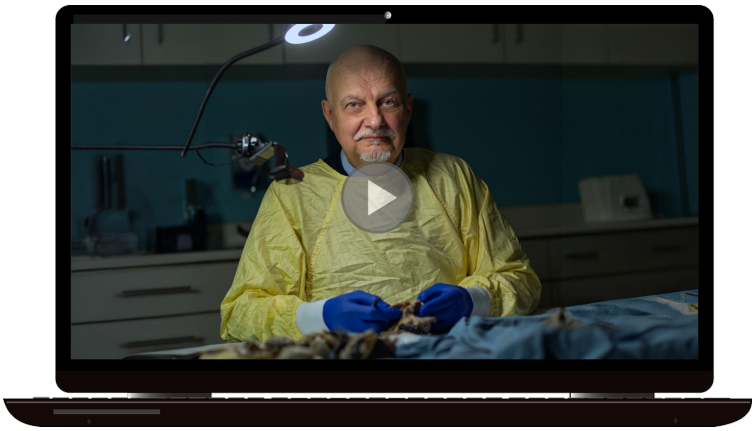
While these advancements have transformed outcomes for children with Congenital Heart Disease, early diagnosis remains one of the most powerful tools in optimizing care. Sessions such as *Fetal and Neonatal Heart and Lungs: Embryology, Anatomy, and Physiology* by Dr. Davide Marini will provide a foundational understanding of cardiac development, while Dr. Lindsay Freud's discussion on *Fetal Intervention in the Catheterization Lab*, highlights the growing role of prenatal diagnostics. Additionally, Dr. Israel Valverde, will discuss *3D Printed Models and Holograms* demonstrating how emerging technologies enhance visualization and preoperative planning.

Dr. Mike Seed, Division Head of Cardiology at SickKids, will delve into the critical role of early interventions and their lasting impact on neurodevelopment, in his talk *Fetal Hemodynamics in Congenital Heart Disease and Impacts on Brain Development*. In doing so, Dr. Seed will also reflect on key historical innovations that paved the way for modern advancements, such as the Mustard Operation (a procedure developed by Dr. William Mustard in 1966 at SickKids). By examining both past and present breakthroughs, we gain a deeper understanding of how far the field has come and how continued innovation can further improve care of children with Congenital Heart Disease.

## Strong Foundation

Just as balancing historical perspective with cutting-edge advancements is essential to progressing clinical care, a strong grasp of fundamental principles is equally as critical as exploring rare and complex conditions. At the start of the program, you can expect to review assessment skills such as heart sounds and murmurs and gain diagnostic expertise in interpreting: 15-lead electrocardiograms, cardiac MRIs and CTs, angiograms, chest radiographs and more.

Additionally, SickKids houses one of the world's largest collections of congenital heart pathology specimens. Dr. David Chiasson, Senior Staff Pathologist at SickKids, will lead a seven-part pathology series, beginning with a session on normal cardiac anatomy, laying the foundation for understanding the complexities of congenital heart disease. Building on this, you



will explore various types of septal defects, including ASD, VSD, and AVSD. From there, you will navigate the challenges of right ventricular outflow tract obstruction lesions such as PS, PA, and TOF. You will also explore left ventricular outflow tract obstruction lesions such as CoA and IAA.

The pathology series will continue with in-depth explorations of complex congenital heart defects, including D-TGA with IVS and CC-TGA. You will also examine abnormalities in pulmonary venous drainage, such as PAPVD and TAPVD. Finally, you will venture into univentricular connections, such as HLHS. With each installment, you will gain a deeper understanding of the resilience of the pediatric heart.

Dr. Amshu Shakya, Pediatric Cardiologist Shahid Gangalal National Heart Centre in Nepal says: "The morphology sessions took me back to med school days, the museum of heart collection with varied pathologies was a visual retreat."

**View all sessions:**
**https://cvent.me/PmvYeD?RefId=Sessions**

### Whole Hearts

While this course prides itself on delivering both breadth and depth in the care of children with heart disease, it is crucial to remember that technological advancements, historical insights, and comprehensive management tools must always be complemented by a holistic, patient-centered approach. This is why the SickKids ACE Program includes topics on affective considerations such as family counselling and ethical principles and dilemmas in pediatric cardiology. Empathy and compassion for the patient and their family are at the core of effective care, ensuring that treatment plans are developed through shared decision-making.

Lastly, the SickKids ACE Program brings together participants from across the globe. By incorporating international perspectives and case-based learning, we can enrich our understanding and bring diverse cultural considerations back to practice and tailor care to local contexts. By integrating clinical expertise with a deep understanding of the emotional and psychological needs of families, we can provide care that is truly transformative, not only for the child but for their loved ones as well.

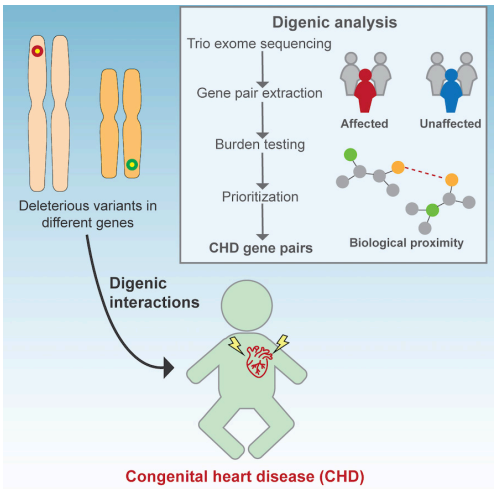
- Register now: **https://cvent.me/5Y8kQE**
- Early bird deadline (up to $400 off): July 18th, 2025
- Group rates available: **https://cvent.me/OxzNZB?RefId=Fees**
- Semester 1 begins on September 5th, 2025
- For inquiries, email: **ace.program@sickkids.ca**

# Hidden Genetic Causes of Congenital Heart Disease Identified

## *New Approach Uncovers Genetic Interactions Contributing to Heart Defects*

Scientists at the Icahn School of Medicine at Mount Sinai and collaborators have identified novel genetic interactions that may contribute to Congenital Heart Disease (CHD), a common birth defect. Details on their findings were reported in the February 20th online issue of The American Journal of Human Genetics [DOI: 10.1016/j.ajhg.2025.01.024].

"Our research reveals the potential for digenic inheritance—where two genes work together to cause disease—expanding our understanding of the genetic underpinnings of congenital heart disease," says co-corresponding senior author Yuval Itan, PhD, Associate Professor of Genetics and Genomic Sciences, a core member of The Charles Bronfman Institute for Personalized Medicine, and a member of The Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai. He co-supervised the study with Bruce Gelb, MD, Gogel Family Professor and Director of The Mindich Institute. "By identifying these gene pairs and their combined effects, we uncover previously hidden genetic risks, which could improve diagnostic precision and open new avenues for personalized treatment strategies."




*A new study from Mount Sinai presents a novel methodology for investigating digenic inheritance in human disease, identifying gene pairs involved in congenital heart disease. Credit: Kars ME, Stein D, Stenson PD, Cooper DN, Chung WK, Gruber PJ, Seidman CE, Shen Y, Tristani-Firouzi M, Gelb BD, Itan Y. Deciphering the digenic architecture of congenital heart disease using trio exome sequencing data. Am J Hum Genet, 2025; (DOI: 10.1016/j.ajhg.2025.01.024).*

Congenital Heart Disease is the most common congenital anomaly, affecting millions worldwide. Despite decades of research, more than half of CHD cases still lack a molecular diagnosis. By analyzing trio exome sequencing data from affected and unaffected children in the Pediatric Genomic Consortium (PCGC), the team identified 10 novel gene pairs potentially linked to the development of CHD.

"Our work demonstrates that genetic interactions, rather than single-gene causes alone, could play a significant role in Congenital Heart Disease. By developing a method to uncover these interactions, we are broadening the scope of genetic research, which could lead to improved diagnosis, enhanced risk assessment, and more informed genetic counseling," says first author Meltem Ece Kars, MD, PhD, a postdoctoral fellow in The Bronfman Institute. "As clinical genetic testing advances, integrating digenic models could significantly improve diagnostic yield, offering patients and their families greater clarity about their condition and guiding the development of targeted therapies and interventions."

The research team used a robust computational method to identify gene pairs that may act together to cause CHD. This innovative approach could transform how genetic studies are conducted for complex diseases, providing deeper insights into the role of genetics in disease development, say the investigators.

The study also paves the way for advancing genetic diagnoses in other complex disorders. "With the tools we've developed, our research provides a framework for future studies into genetic interactions that could affect a wide range of human diseases," says Dr. Itan.

Next, the researchers plan to apply the digenic approach to other disease groups that have traditionally been studied using the monogenic model, potentially explaining some of the missing heritability in these disorders. Ultimately, they aim to extend the digenic approach into a robust polygenic framework capable of identifying multiple disease-causing variants and genes in patients.

"Our findings hold promise for improving genetic diagnoses, offering better risk assessments, and ultimately guiding more personalized treatments for individuals with congenital heart disease," says Dr. Kars.

The paper is titled "Deciphering the digenic architecture of Congenital Heart Disease using trio exome sequencing data."

The remaining authors are David Stein (PhD student at the Icahn School of Medicine at Mount Sinai); Peter D. Stenson, (Cardiff University, UK); David N. Cooper, PhD (Cardiff University, UK); Wendy K. Chung, MD, PhD (Boston Children's Hospital and Harvard Medical School); Peter J. Gruber, MD, PhD (Yale School of Medicine); Christine E. Seidman, MD, (Harvard Medical School, Brigham and Women's Hospital, Howard Hughes Medical Institute); Yufeng Shen, PhD (Columbia University Irving Medical Center); and Martin Tristani-Firouzi, MD (University of Utah School of Medicine).

This research is supported by the National Heart, Lung, and Blood Institute of the National Institutes of Health and the U.S. Department of Health and Human Services through grants UM1HL128711, UM1HL098162, UM1HL098147, UM1HL098123, UM1HL128761, and U01HL131003. Additional support was provided by Clinical and Translational Science Awards (CTSA) grant UL1TR004419 from the National Center for Advancing Translational Sciences.

# Survey Confirms Radiation and Orthopedic Health Hazards in Cardiac Catheterization Laboratories are 'Unacceptable'

*SCAI Leadership Calls for Action to Protect Interventional Cardiologists and Staff*

A survey conducted by the Society for Cardiovascular Angiography and Interventions (SCAI) highlights ongoing radiation and orthopedic hazards faced by interventional cardiologists and cardiac catheterization laboratory ("cath lab" or CCL) staff. The survey revealed that despite technological advancements, significant risks often remain unaddressed despite advances in protective equipment.

"Occupational Health Hazards in the Cardiac Catheterization Laboratory: Results of the 2023 SCAI Survey" highlights alarming trends in radiation exposure and orthopedic injuries for interventional cardiologists and staff. The survey was published in JSCAI with a call to action by SCAI Leadership.

"This study confirms what many of us in the field have long suspected—occupational hazards in the cath lab remain unacceptable and are largely unchanged over the past two decades," said SCAI President James B. Hermiller, MD, MSCAI. "We must act now to implement stronger protections for interventional cardiologists and cath lab staff to ensure their long-term health and safety. Addressing these risks is not just about protecting today's workforce but also about ensuring the sustainability of our profession."

The survey, conducted by SCAI's Professional Well-Being Committee, compared data from 2014 to 2023 and found that although technological advancements in the field have been made, the risks associated with working in the CCL remain unacceptably high. Over 60% of respondents reported experiencing orthopedic injuries, and 6% reported being diagnosed with cancer, far exceeding normal rates and underscoring the urgent need for improved safety measures.

"This data provides an undeniable call to action. The results showed that the preponderance of cancers and other types of radiation injury, such as cataracts, was threefold higher than what is observed in the general population," said coauthor Allison Dupont, MD, FSCAI, chair of SCAI's Professional Well-Being Committee. "Hospitals and healthcare systems need to prioritize investments in advanced protective equipment and safer work environments. We know that new technologies exist that could significantly reduce radiation exposure and orthopedic strain, but we need institutional commitment to make these solutions accessible."

## The Study's Key Findings

Persistent orthopedic injuries: Nearly 60% of respondents reported orthopedic injuries due to wearing lead aprons for hours, with spine injuries being the most common. Such aprons can weigh over 10 pounds. Chronic pain related to these injuries remains a leading cause of career limitations and early retirement among interventional cardiologists.

Radiation exposure concerns: Despite the availability of protective equipment, usage remains inconsistent. Many respondents cited high costs and administrative barriers as obstacles to adopting newer radiation protection technologies. Additionally, 17% of respondents admitted to limiting their time in the cath lab to reduce their radiation exposure, a significant increase from previous surveys.

Concerns for female interventionalists: The survey highlighted the specific needs of female interventional cardiologists, particularly regarding pregnancy and radiation exposure. Among female respondents, 28% reported being discouraged from working in the cath lab due to pregnancy, while 71% expressed a desire to step away during pregnancy, highlighting the need for more flexible policies and better workplace accommodations.

In addition to highlighting existing concerns, the study proposes concrete steps to mitigate these hazards, including the adoption of new shielding technologies, reducing reliance on heavy lead aprons, and expanding formal radiation safety education. The survey also found that despite the availability of various radiation mitigation tools, many are underutilized due to cost concerns and a lack of administrative support, underscoring the need for systemic change.

"We cannot afford to ignore these findings," said coauthor Islam Abudayyeh, MD, FSCAI, co-chair of SCAI's Professional Well-Being Committee. "At a time when recruitment and retention in interventional cardiology are already challenging, we must ensure that our work environments are not only cutting-edge in terms of patient care but also safe and sustainable for providers."

Hermiller called upon hospitals, healthcare administrators, and policymakers to address these critical issues by investing in safer work environments and fostering a culture that prioritizes the well-being of CCL professionals. The Society also plans to use these findings to advocate for legal changes that would promote safer working conditions and increased institutional accountability.

Read the study at: **https://www.jscai.org/article/S2772-9303(24)02182-3/fulltext**

# CAREER OPPORTUNITIES

*Click the position title to view the full job description – page 1 of 2*

**Adult Congenital Heart Disease (ACHD) Cardiologist**
Phoenix Children's
*Phoenix, Arizona*

**Pediatric Cardiologist Heart Transplant and Advanced Heart Failure**
Phoenix Children's
*Phoenix, Arizona*

**Pediatric Cardiologist**
Banner University Medical Center
*Tucson, Arizona*

**Pediatric Cardiologist**
Loma Linda University Children's Hospital
*Loma Linda, California*

**Assist / Assoc Professor of ACHD**
Yale University
Yale New Haven Children's Hospital
*New Haven, Connecticut*

**ACHD Cardiologist**
Nemours Children's Hospital
*Wilmington, Delaware*

**Medical Director**
Nemours Children's Hospital
*Wilmington, Delaware*

**Director of Fetal Cardiology**
Nicklaus Children's Hospital
*Miami, Florida*

**Pediatric Cardiologist, Fetal Specialist**
Nicklaus Children's Hospital
*Miami, Florida*

**Pediatric Cardiac Intensivist**
Nicklaus Children's Hospital
*Miami, Florida*

**ACHD Cardiologists**
University of Chicago Comer Children's Hospital
*Chicago, Illinois*

**ACHD Cardiologist**
Louisiana State University Children's Hospital of New Orleans (CHNOLA)
*New Orleans, Louisiana*

*Click the position title to view the full job description – page 2 of 2*

**Pediatric Cardiologist**
Tulane University
Children's Hospital of New Orleans (CHNOLA)
*New Orleans, Louisiana*

**Pediatric Cardiologist Advanced Imaging with Cross-Sectional Focus**
MaineHealth Maine Medical Center
*Portland, Maine*

**Pediatric Cardiologist (Echocardiographer)**
University of Minnesota
The Pediatric Heart Center
*Minneapolis, Minnesota*

**Pediatric Cardiologist**
Children's Mercy
*Springfield, Missouri*

**ACHD Cardiologist**
Dartmouth School of Medicine
Dartmouth Health Children's
*Manchester, New Hampshire*

**Pediatric Cardiologist**
Dartmouth School of Medicine
Dartmouth Health Children's
*Manchester, New Hampshire*

**Pediatric Electrophysiologist Cardiologist**
UNC Chapel Hill
UNC Health Children's
*Chapel Hill, North Carolina*

**Adult Congenital Cardiologist**
Akron Children's Hospital
*Akron, Ohio*

**Pediatric Cardiologist**
Akron Children's Hospital
*Akron, Ohio*

**Acute Care**
UPMC Children's Hospital of Pittsburgh
University of Pittsburg School of Medicine
*Pittsburgh, Pennsylvania*

**Electrophysiology**
UPMC Children's Hospital of Pittsburgh
University of Pittsburg School of Medicine
*Pittsburgh, Pennsylvania*

# New Treatment Option for Severe Hypertrophic Cardiomyopathy in Children Shows Promise

*Study Finds Trametinib Significantly Reduces Risk of Death, Cardiac Surgery and Transplantation*

Trametinib, a mitogen-activated protein kinase (MEK) inhibitor, reduces mortality and morbidity in children with severe hypertrophic cardiomyopathy (HCM) caused by pathogenic variants in the RAS/MAPK pathway, according to a study published today in *JACC: Basic to Translational Science*. The study provides strong evidence for personalized treatment targeting the underlying genetic causes of RASopathies, a group of rare disorders that often lead to life-threatening cardiac complications.

"Our findings represent a breakthrough in the treatment of HCM in children, particularly those suffering from severe forms of the disease due to genetic variants in the RAS/MAPK pathway," said Gregor Andelfinger, MD, PhD, co-author of the study and a cardiologist at CHU Saint-Justine in Montreal. "The positive results we observed with trametinib are a promising step forward in addressing an urgent medical need for children whose condition has not responded to standard therapies."

HCM, a condition where the heart muscle thickens abnormally, is particularly dangerous in children and can lead to heart failure or premature death. About 20% of patients with RASopathies have HCM; RASopathy-associated HCM is often caused by genetic mutations in the RAS/MAPK signaling pathway, which regulates cell growth and development. It is often a more severe form of HCM and has a higher mortality rate. Until now, treatment options for severe cases of RASopathy-associated HCM (RAS-HCM) in pediatric patients have been limited.

The study, which involved 61 children with severe RAS-HCM, compared 30 children receiving trametinib to 31 children receiving standard care. The results showed a significant reduction in the outcome of death, cardiac transplantation, or the need for cardiac surgery in the trametinib group. No life-threatening adverse events were observed, although dermatologic and mucous membrane side effects were common but manageable.

"This study provides crucial evidence that targeted therapies like trametinib could dramatically improve the outlook for children suffering from severe HCM," Andelfinger said. "It underscores the importance of developing genotype-specific therapies for RASopathies and other rare diseases."

"The paper by Andelfinger and colleagues provides exciting data with respect to treating 'Rasopathies' in children with HCM," said Douglas Mann, MD, FACC, Editor-in-Chief of JACC: Basic to Translational Science. "Rasopathies are a group of rare genetic disorders that are caused by mutations in genes that regulate the Ras/mitogen-activated protein kinase (MAPK) signaling pathway."

"Given the rarity of this condition and the lack of existing therapies for these children, we felt it was important to publish this paper, with the hope that it will advance the field by providing a foundation for future randomized clinical trials to definitively evaluate the safety and efficacy of Trametinib in children with HCM," Mann said.

Study limitations include potential biases due to its design and incomplete data collection for the control group. Additionally, the trametinib group included patients with prior heart surgeries, which could affect the results. The shorter follow-up for the trametinib group may also underestimate long-term side effects. Lastly, the study does not determine the optimal dosing of trametinib for RAS-HCM.

# MAY

## 01ST-03RD

**SCAI 2025 Scientific Sessions**
Washington, DC, USA
https://scai.org/scai-2025-scientific-sessions

## 05TH-06TH

**CARDIO 2025 - 4th CME Cardiologists Conference**
Istanbul, Turkey
https://cardiologists.plenareno.com/

## 15TH-16TH

**International Conference on Pediatrics and Child Health**
Dubai, UAE
https://www.pediatricsummit.scientexconference.com/

# AUGUST

## 25TH-28TH

**PICS 2025**
Chicago, IL, USA
https://www.picsymposium.com/home.html

# SEPTEMBER

## 25TH-28TH

**ASE 2025 36th Annual Scientific Sessions**
Nashville, TN, USA
https://www.asescientificsessions.org/registration/

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