

## Updates from the UDN



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## Managing the Odyssey

**By Kelly Schoch, genetic counselor at Duke**

The search to find the cause of an undiagnosed disorder is often called a “diagnostic odyssey”. Most of the participants in the UDN are quite familiar with this journey, which often stirs a roller-coaster of feelings including fear, disappointment, stress, and frustration, but also hope. To learn more about the emotional impact of parenting an undiagnosed child, researchers at the Duke Clinical Site asked parent participants to share information about their family’s experience. We learned a lot from parents as they told us the stories of their journey and the love and concerns they have for their children. We also found that many parents (40%) were experiencing significant levels of anxiety and depression. However, despite the challenges, parents of children in the UDN remain actively engaged in their child’s health care and are able to effectively manage the uncertainty that having an undiagnosed disorder brings. YOU are effective advocates for your children and yourselves!



The results from our work suggest that more needs to be done to help families manage the natural feelings that often come from parenting a child with an undiagnosed disorder. More work also

needs to be done to help individuals with undiagnosed disorders manage the natural feelings of living with an undiagnosed disorder. Involvement in support groups, such as the National Organization for Rare Disorders (NORD), Syndromes Without A Name (SWAN), or Rare & Undiagnosed Network (RUN), can help by connecting families facing similar challenges searching for a diagnosis. With the same goal in mind, the UDN PEER created a closed Facebook group solely for families who have completed the UDN evaluation process and would like to communicate with each other in a protected environment. [Click here](#) or search “UDN PEER” on Facebook to find the group. Finally, although connecting with people undergoing similar journeys is often helpful, our study results show that sometimes professional counseling is a valuable tool for coping with these difficult circumstances.

We would love to hear about YOUR experiences regarding what has worked well for you as you navigate life with an undiagnosed disorder or care for a child with an undiagnosed disorder. We invite you to email us at [PEERudn@gmail.com](mailto:PEERudn@gmail.com) or join the UDN Facebook group if you would like to share.

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## “We are not alone” – Thad’s story

**By Liz Worthington**

What does it look like to be undiagnosed? To what extent do we really want answers? How do we best take care of our children and ourselves? We are the Worthingtons, and our son Thad is undiagnosed.



Thad

Our family already had two beautiful children (with congenital heart defects and developmental delays) when we found out we were pregnant with a third child. Our children’s health was challenging but still manageable. We dreamed of having a healthy and uncomplicated baby. After Thad was born, however, we discovered he had the same congenital heart defect as his siblings. Unfortunately, Thad’s defect required heart surgery, which led to significant complications in

surgery. A routine procedure turned into a long hospital stay, followed by a complicated homecoming – with medical equipment, prescriptions, and complicated instructions. This time, it was different; the dream was shattered.

When you have a child with special needs, your world changes. At first, we relied on supportive friends who reached out to us in our time of need. When it became obvious that this was not a short-term situation, however, our friends became less available. Having to work our social schedule around therapies, appointments, and hospital visits became a challenge. Slowly, we found ourselves isolated on a medical island and less able to attend social functions with our friends. Slowly, our circle shrunk.

When you have a child who qualifies for home health care, it's not unreasonable to assume you have additional/unmet needs. Shortly after qualifying for home health, for example, I received a request to meet a social worker. My initial thought was, "That sounds like someone getting in my business and picking my life apart with a fine-tooth comb!" Previous experiences had predisposed me to believe social workers only helped people in crisis. I almost declined her visit, but I'm glad that I met her. Her first statement was, "Tell me about some of your struggles and what makes your day difficult." With her compassionate care/advocacy, we have received accommodations and equipment that met our needs, including a bigger stroller, a safety bed, and a security system for the house (all items necessary for a non-verbal child who likes to wander off). In addition, she helped me contact our insurance company and get a personalized case worker assigned to the needs and care of my son, something I didn't realize was even a possibility beforehand. Without knowing these resources were available to us, I was struggling to survive. By grudgingly accepting help, I was able to dramatically change the care I provide to my son. Instead of feeling isolated, now I have people helping me work for things that Thad needs.



Thad and his siblings, Christmas 2017

After joining the UDN and being introduced to other families with similar struggles, we have found that we are not alone. We have more resources and help available to us than we realized. We may be undiagnosed, but we are a part of something bigger/great, we are a part of the UDN.

## The Coordinating Center at the UDN

By Emily Glanton, Kim LeBlanc, and Cecilia Esteves

The Undiagnosed Diseases Network (UDN) is a group of health care providers and researchers across the United States working together to find answers for patients with symptoms that have been difficult to diagnose. On any given day in the UDN, providers are reviewing applications and evaluating patients, researchers are investigating the underlying causes of patient conditions, and clinical site coordinators are working to make sure that patients move through this process smoothly. Coordinating these and other network activities is a full-time job for the UDN Coordinating Center, located within the Department of Biomedical Informatics at Harvard Medical School. The Coordinating Center, led by Alexa McCray, PhD and Isaac Kohane, MD, PhD, is made up of a team of project managers, doctors, researchers, genetic counselors, technology developers, and research assistants. This team is responsible for providing support for patients and referring health care providers through the UDN Helpdesk, managing the UDN database and protocols, and sharing the work of the UDN with the broader community.



The Coordinating Center team

UDN participants may interact with the Coordinating Center at many different points throughout the study process. The UDN Helpdesk in particular is a resource available to answer questions and help patients navigate the application, evaluation, sequencing, and research process.

Since it is important to the UDN to check in with participants over time, the Coordinating Center also follows up with every participant after their in-person clinical site visit is complete. While the clinical site coordinators remain the main point of contact for participants, the Coordinating Center contacts all participants on an ongoing basis to collect updates.

Another important role of the Coordinating Center is to manage information that is shared within and outside of the UDN. UDN health care providers and researchers need to easily and securely share information about participants in order to make diagnoses. The Coordinating Center team created a database specifically for this purpose and manages the data that is entered by the various UDN members.

The team shares updates about the UDN with families, healthcare providers, and researchers on its [Twitter](#) and [Facebook](#) page.

Members of the Coordinating Center team travel to conferences and meetings a few times a year to share the work of the UDN with others.



Paul, Kim, and Cecilia at the National Society of Genetic Counselors annual conference in 2016

One project that has kept the Coordinating Center busy lately is creating participant pages on the UDN website ([click here to view](#)). If a participant is interested in having a webpage created to connect with similar patients, the Coordinating Center works with the participant and clinical site to create this page after their in-person clinical site visit. If the Coordinating Center receives an inquiry about one of these pages, the Coordinating Center lets the UDN clinical site coordinator know. This project has helped the UDN connect patients around the world with the same rare conditions.

Coordinating a network as big as the UDN keeps the Coordinating Center quite busy. With that said, being able to provide support to patients, health care providers, and researchers in the rare and undiagnosed disease community provides constant motivation to continue this meaningful work.

**Contact the UDN Coordinating Center at:**

Phone: 1-844-RING-UDN (1-844-746-4836)

Email: [udn@hms.harvard.edu](mailto:udn@hms.harvard.edu)

*Hours: Monday through Friday from 9 a.m. – 5 p.m. Eastern Time*

## Connect with a UDN PEER member

Please contact us at [PEERudn@gmail.com](mailto:PEERudn@gmail.com) to connect with other individuals who have been through the UDN.  
We would love to connect with you!

**Chad “Chad the Dad” Smith**



**Luke Shantz**



**Debbie Krieger**



**Sarah Marshall**



**Susan “Liz” Worthington**



**Edward “Jamie” Mills and Liz Aronin**



**Teresa Locklear**



**Gina Szajnuik**

