Two Families.

Two Genetic Syndromes.

One Bond.

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2025 National EHDI Conference

Personal Disclaimer

Personal choices to pursue genetic testing don't indicate a desire to change the outcome, but a desire to better understand how to manage the diagnosis and prepare for the future

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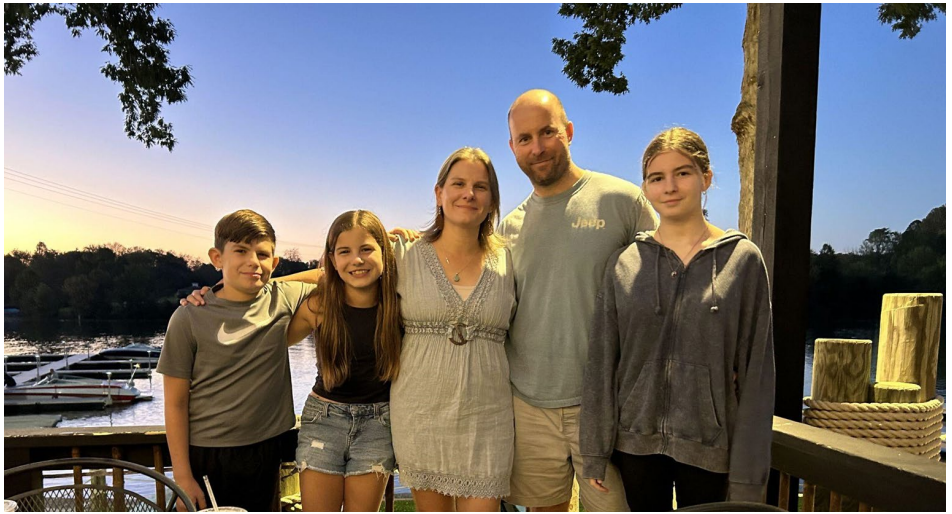
Teri Urban

- Parent of a deaf teenager
- Virginia Hands & Voices
- Supporting Success for Children with Hearing Loss
- CHAMPS-DHH Program



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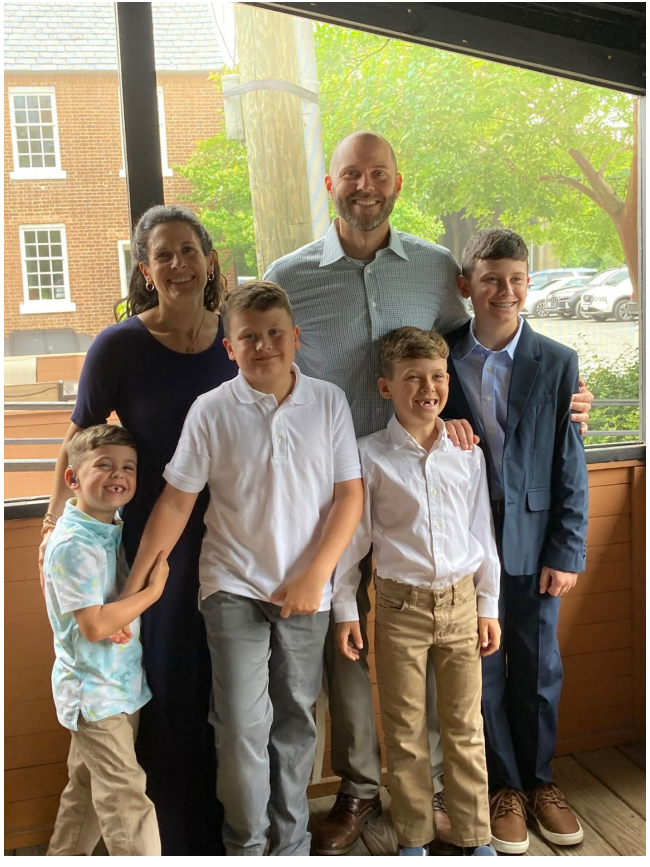
Alicia Rodriguez

Alicia Rodriguez

- Parent to four boys
- Virginia Hands & Voices
- Garden Educator
- Yoga and Book Lover



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Learning Objectives

Understand genetic testing from a family perspective

Identify three benefits of genetic testing and how it can impact a family's journey

Recognize the lived experiences of raising a child with Connexin 26 and Pendred Syndrome

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Benefits of Genetic Testing

- Recognize medical needs
- Parental guilt
- Prepare for the future
- Non-invasive procedure
- Support siblings
- Understand diagnosis

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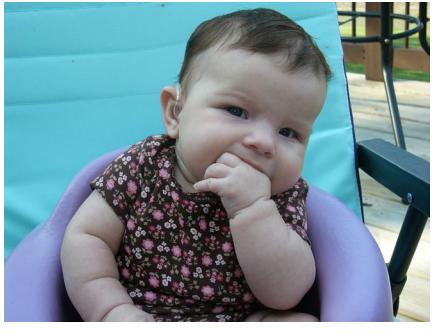
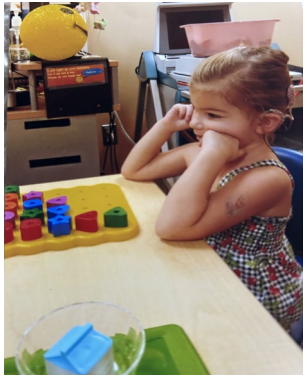
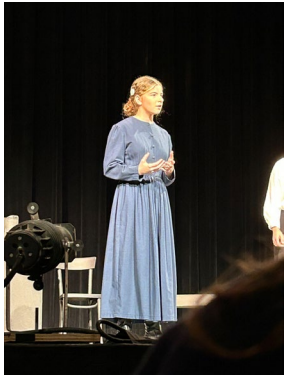
GJB2 / Connexin 26

April 2010	May 2010	November 2010	June 2013	September 2015	March 2017
Failed Newborn Hearing Screen	ABR Mild/Moderate	Received Genetic Results	Sedated ABR Severe/Profound	Cochlear Implant Left Ear	Cochlear Implant Right Ear







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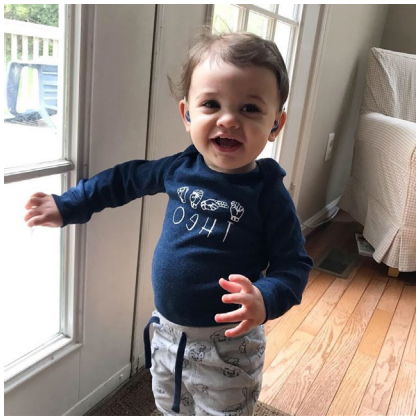
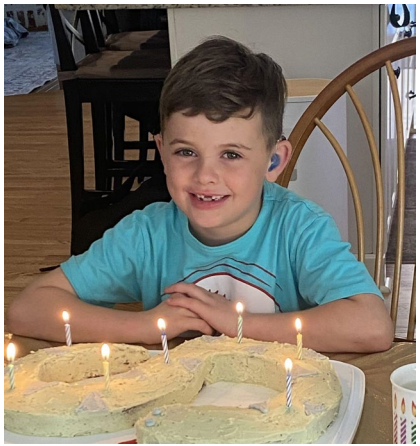
Pendred Syndrome / SLC26A4





What is Pendred Syndrome?

- Caused by lack of protein “Pendrin”
- Possibly affects hearing, thyroid and kidney function
- All our children had 1 in 4 chance of having the diagnosis
- Only 1 in 10,000 are diagnosed with Pendred Syndrome

Theo’s Journey

- March 2018 - ENT appointment
- October 2018 - Geneticist appointment
- November 2018 - Follow-Up Call and Diagnosis
- Theo’s journey with EVA - Progressive and fluctuating

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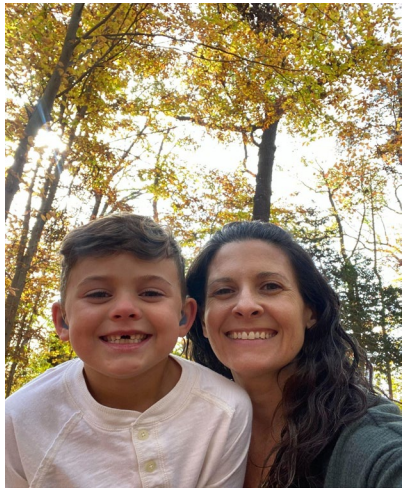
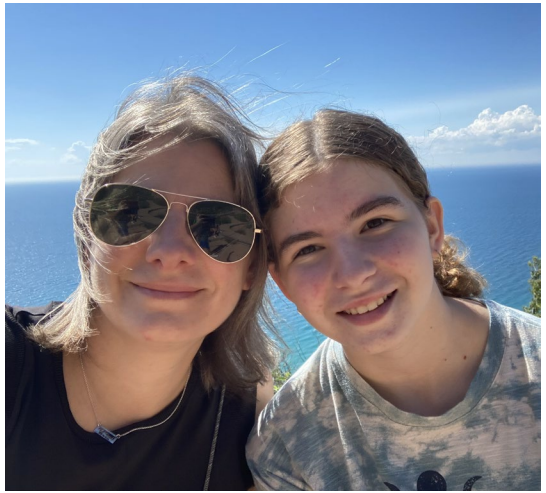
Navigating Results





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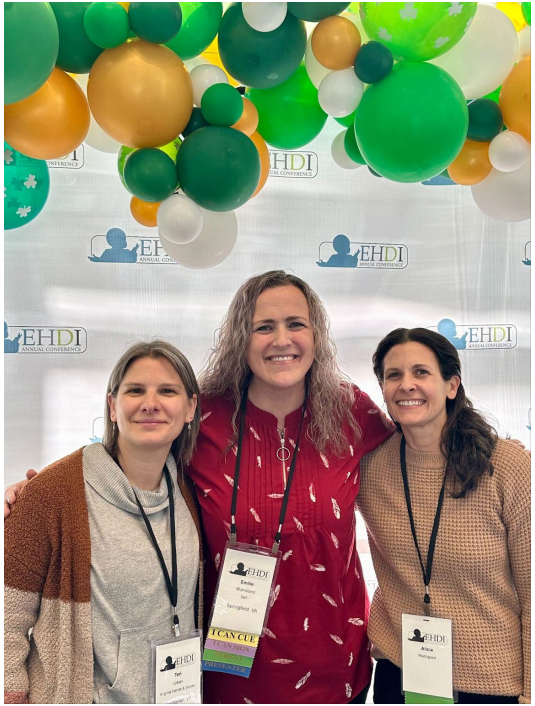

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