

Benefits and Downsides of Genetic Testing for Families

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Five Generations
of Deaf and Hard
of Hearing Family
Members

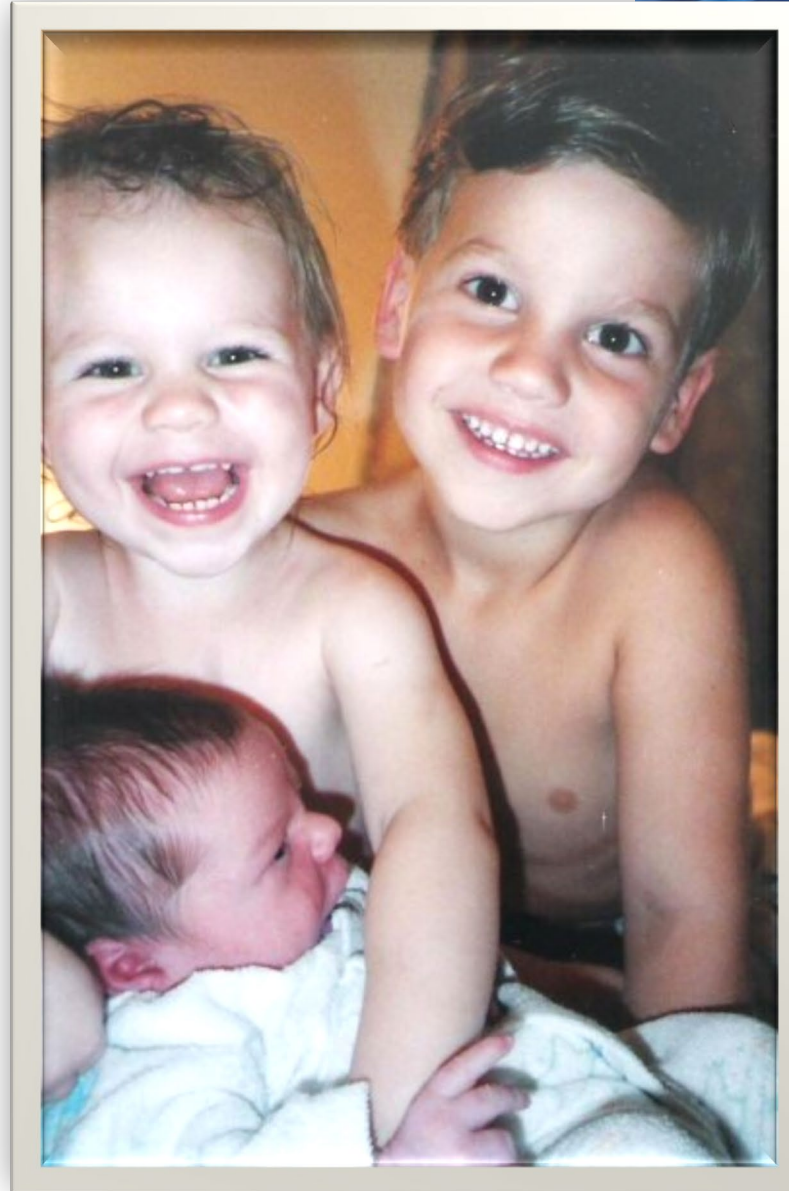


The Early Days

We said “no” to genetic testing

Did not feel a need to know “why”

It was not covered by insurance



The World Wide Web Influence

“It’s probably Connexin, that’s the most common genetic reason.”

“I’m pretty sure you have Enlarged Vestibular Aquaduct Syndrome due to hearing loss from hitting your head.”

“I’ve never heard of a family like yours!”

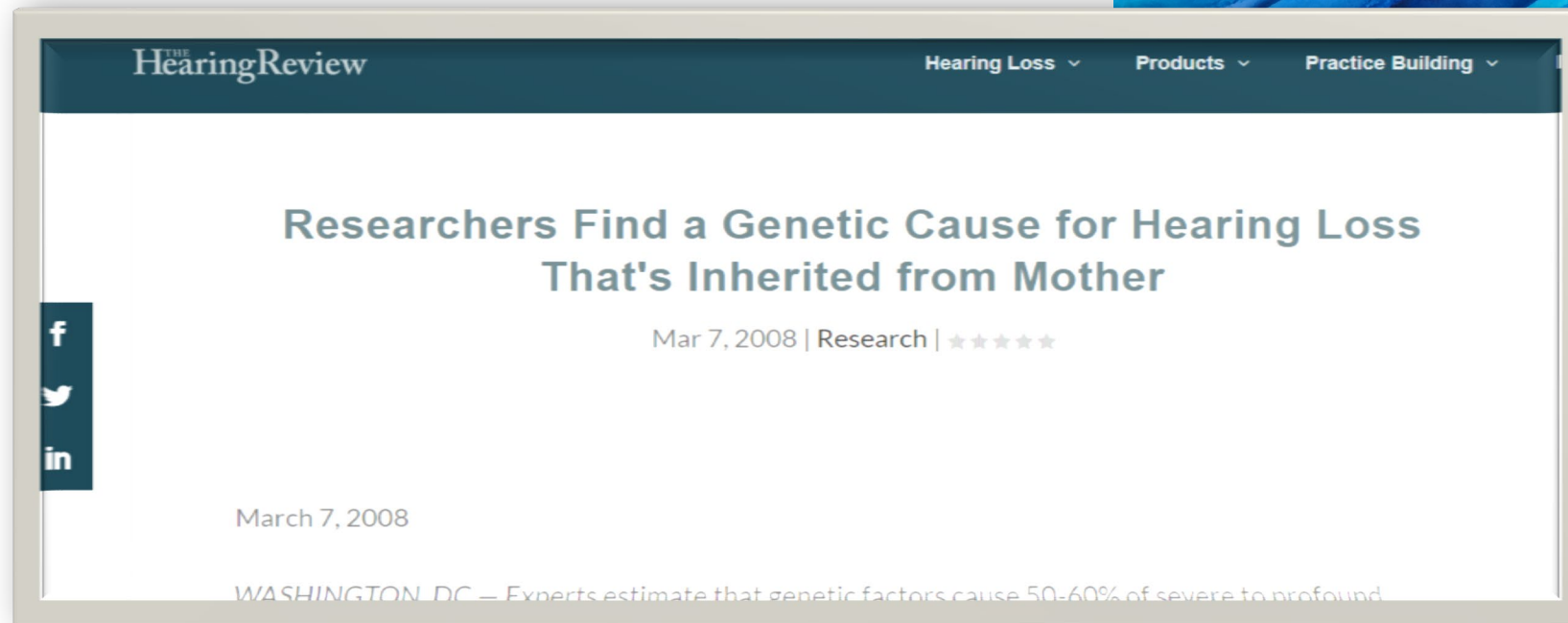


Getting Answers

Mutation: 7510T>C of the tRNA^{Ser(UCN)} gene

One of THREE families in the world (now four)

Passed on through mothers 100%



The screenshot shows a news article on a website. The header includes the logo 'THE HearingReview' and navigation links for 'Hearing Loss', 'Products', and 'Practice Building'. The main headline reads 'Researchers Find a Genetic Cause for Hearing Loss That's Inherited from Mother'. Below the headline, it says 'Mar 7, 2008 | Research | ★★★★★'. On the left side, there are social media icons for Facebook, Twitter, and LinkedIn. The date 'March 7, 2008' is displayed below the icons. At the bottom, the start of the article text is visible: 'WASHINGTON, DC — Experts estimate that genetic factors cause 50-60% of severe to profound'.

THE HearingReview

Hearing Loss ▾ Products ▾ Practice Building ▾

Researchers Find a Genetic Cause for Hearing Loss That's Inherited from Mother

Mar 7, 2008 | Research | ★★★★★

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March 7, 2008

WASHINGTON, DC — Experts estimate that genetic factors cause 50-60% of severe to profound

We were not prepared for the
EMOTIONAL
impact of genetic testing





The Benefits of Genetic Testing

Getting answers to the questions on your mind

Having a “diagnosis” or explanation

Better understanding of how the gene moves through your family

Can pave a path forward for the family

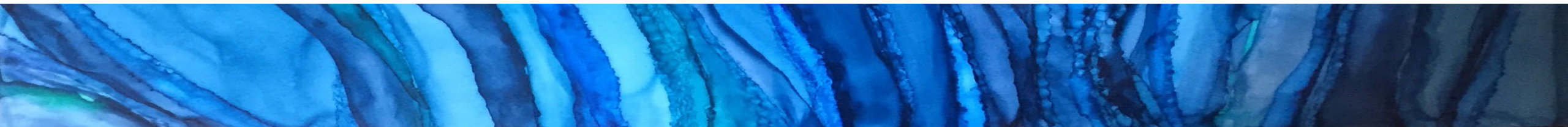
Becomes part of the family’s story

With More Information... Comes More Questions

Do families WANT to know answers?

Do families NEED to know answers?

HOW will families use the information?



The Most Important Question:

**How will I FEEL after getting
this new information?**



From a Genetic Counselor Erica Wright:



“There’s no ‘right’ time for genetic testing.”

“Your genes don’t change.”

“Testing later may bring more advanced information.”



When Genetic Testing Reveals “Even More”

Families seek answers to find out the “why” of hearing loss

Specialist uncovers genetic information that reveals a syndrome or additional information



The Emotional Impact of Genetic Testing

- The information is lifelong
- There's no going back
- Can impact future generations
- Brings on “Labeling”



Should ALL Families Test for Genetics?



Write out the pros and cons

Does the family need/want more information?

Consider the emotional impact

Work with a Genetic Counselor before and after testing

A New Family Member to Add to the Mix



Misdiagnosed with EVA

Benefitted from our genetic testing

Provided answers to questions he had wondered about for years



What Scientists call
a “Mutation”

I call a
“Beautiful Journey”

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