Mountain States
REGIONAL GENETICS NETWORK

Time 4 Genetics

August 2023-November 2023
Topic #8:
Seizures and Genetics

It's TIME to KICK-off Topic #8:

Seizures

For the next 4 months we will explore the topic of NBS in various ways:

August: Newsletter (you are reading that now!)
September: Webinar (**coming up on Sept. 26th, register HERE)**
October: Check your Snail Mailbox for some MAIL!
November: Family Centered Perspectives & Conversation

Please take ONE hour (or more!) to review the resources in this newsletter and please **TAKE THE SURVEY** below!

We thank you for taking TIME 4 GENETICS!
You can access the T4G Library Archives here.

Register NOW!

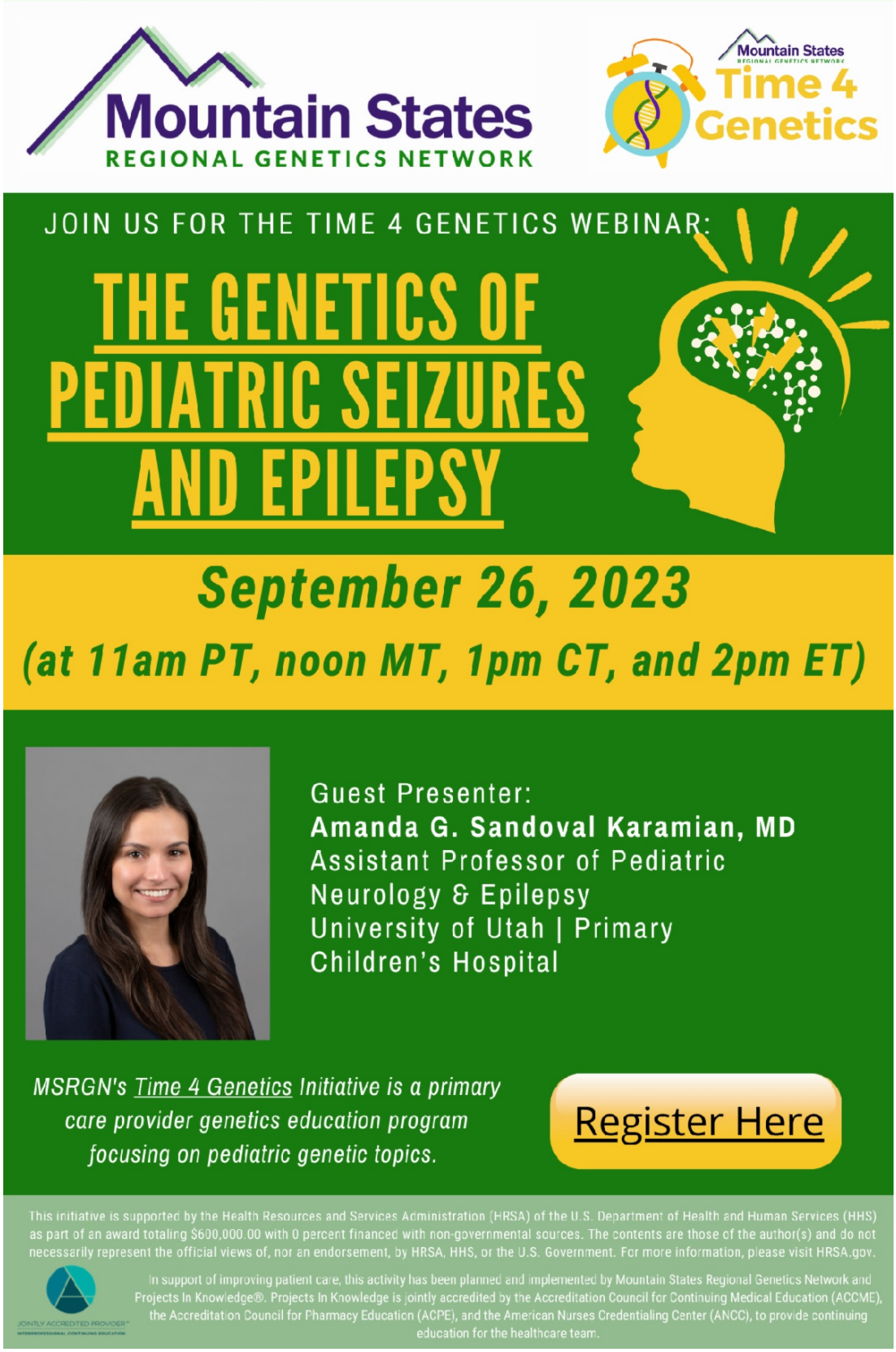
September 26, 2023

(at 11am PT, noon MT, 1pm CT, and 2pm ET)

Webinar:

The Genetics of Pediatric Seizures and Epilepsy

CME and CEU is available



The Genetics of Pediatric Seizures and Epilepsy

September 26, 2023

(at 11am PT, noon MT, 1pm CT, and 2pm ET)

featuring Guest Presenter:

Amanda G. Sandoval Karamian, MD

Assistant Professor of Pediatric Neurology & Epilepsy
University of Utah Primary Children's Hospital

To register please click: here

For a clickable and ready to share PDF version of this flyer please download: here

Register Here

Resources for TOPIC #8: Seizures

We encourage you to take an hour (or more!) to read the information below. Feel free to forward and share this newsletter with your staff and colleagues.

Video from:
The Epilepsy Foundation

Epilepsy and Genetics: The Basics

Topics

The basics of genes and genetic testing
What do we know about the genetics of epilepsy?
What are the benefits and limitations of testing?
What genetic tests are available for people with epilepsy?
How do I go about getting testing and insurance coverage?

Genetic Testing and Counseling for the Unexplained Epilepsies: An Evidence-based Guideline of the National Society of Genetic Counselors

"Epilepsy, defined by the occurrence of two or more unprovoked seizures or one unprovoked seizure with a propensity for others, affects 0.64%

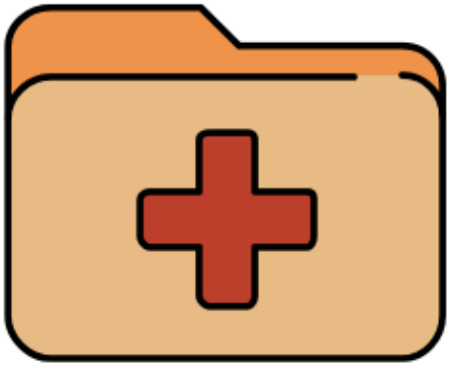
Guest Speaker
Beth Rosen Sheidley, MS, CGC
Co-Director, Epilepsy Genetics Program
Senior Genetic Counseling Program Manager
Division of Epilepsy, Department of Neurology
Boston Children's Hospital
Boston, MA

Recorded: January 22, 2020

Watch Video Here

of the population and can lead to significant morbidity and mortality. A majority of unexplained epilepsy (seizures not attributed to an acquired etiology, such as trauma or infection) is estimated to have an underlying genetic etiology."

Read Paper Here



Genetic Testing and Seizures Resource and Video to share with Patients and Families

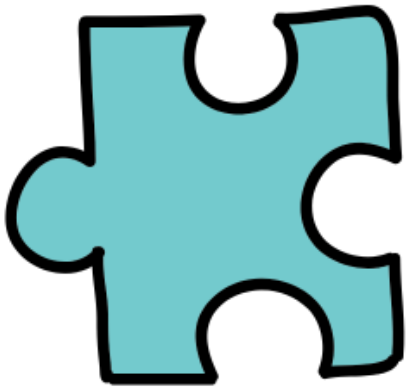
What Kinds Of Results Can I Expect From Genetic Testing?

Positive Test Result

- For a person with epilepsy, a positive result means that a genetic change has been identified as the cause of his or her epilepsy diagnosis.
- A positive result in an otherwise healthy individual could indicate that person is at risk for epilepsy.
- Genetic testing may also give information about reproductive risks and can help clarify if and how epilepsy is inherited in a family.

Negative Test Result

- A negative test result in a person with epilepsy means



News Article:

Genetic Testing Provides a Missing Piece:

Two Colorado sisters had dozens of seizures every day.

Then a genetic test revealed they needed a vitamin.

Read Article Here

that a genetic cause of epilepsy was not identified using the test that was performed.
- A negative result does not completely exclude the possibility of a genetic diagnosis.
- In some cases, more testing may be considered.

Uncertain Test Result

- It is also possible to receive uncertain results from a genetic test.
- An uncertain result means that a genetic finding was detected, but the meaning of the finding is not known.
- These results are referred to as genetic **variants of uncertain significance (VUS)**. A VUS result may be re-interpreted at a later date when more information becomes available and could end up being re-classified as either benign (negative) or pathogenic (disease causing/positive).

(excerpt from: **https://www.epilepsy.com/causes/genetic/testing**)

Video for families about Variants of Uncertain Significance (VUS)

Don't forget to REGISTER for the MSRGN Genetics Summit 2023

October 11-12, 2023

A virtual summit to educate, engage, and connect families, providers, and public health professionals around contemporary topics in genetics.

Register Here

DATA TIME!

Don't Forget!

Data from Topic #7 (Newborn Screening) is DUE NOW! (by 9/6/23)

We are all done with topic #6! We hoped you enjoyed the **Newborn Screening** topic area.

Please **email Kristi** any zip codes **(download tool here)** of those

Download the Zip Code Collection Tool Here

Take Topic #7 Survey on Newborn Screening Here

patients with whom you utilize any of the Time 4 Genetics information, resources, and tools! You can also include any zip code of patients you used Topic #1 (Red Flags), Topic #2 (Developmental Delays), Topic #3 (EDS), Topic #4 (Genetic Testing), Topic #5 (Autism),or Topic #6 (Cerebral Palsy) info for as well!

Email Kristi with questions or zip codes

If you have not yet had a chance to provide feedback on topic #7 (NBS) through the **evaluation/survey**, please do so. **WE HAVE GREATLY SHORTENED THE SURVEY** and it should only take you about 2-4 minutes to complete!

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