



January 2021

MSRGN Project Directors' Message



Happy New Year!

2021 is finally here! We hope your holidays, while possibly quite different from years past, were still filled with joy and peace. As we look forward to the last 5 months of this grant year, we eagerly anticipate MANY new surveys, applications, and initiatives launching! It was so great to share those with those of you who joined us for the virtual winter meeting this week! Check out all the links below and please share with a friend or colleague!

MSRGN January Highlights:

- MSRGN Virtual Winter Meeting took place on January 28th with a great turnout and recaps from all of the state teams on their projects as well as an MSRGN Grant overview, HRSA overview,

First Baby Identified with GAMT by NBS in Utah

In December 2020, Utah's Newborn Screening Program identified the first patient with Guanidinoacetate methyltransferase (GAMT) deficiency solely through newborn screening. GAMT deficiency is an inherited condition that affects the body's ability to produce creatine. Without creatine, the body is unable to use and store energy resulting in severe neurological problems including intellectual disability, limited speech development, recurrent seizures, autistic-like behavior, and involuntary movements. Because early diagnosis and treatment can lead to improved health and development in children, GAMT deficiency was added to Utah's newborn screening panel in 2015.

Links Below:

- [Utah Department of Health Press Release](#)
- [Association of Creatine Disorders Press Release](#)
- [ARUP's Press Release](#)
- [News video featuring Heidi Wallis, UT State Team Co-lead](#)

Evaluation overview, and ReRe demonstration. The slides and our collective Genetic GASP are recorded [here](#).

- Our Genetic Navigator Application launched. Apply [here](#).
- Our **Genetic Ambassador's** Red Flags for Genetics Survey launched. Click [here](#) for the English & Spanish version and share far and wide!
- Our Genetics Explainer Video survey launched for providers. Learn more about it [here](#).



2021 Genetic Pop-Ups Announced!

Genetic Pop-Ups will be held near Rare Disease Day in February and March.

Congratulations to:

Heidi Klomhaus (AZ)
Becki Coleman (AZ)
Jamie Stefanski (CO)
Maria and Jesus Castillo (CO)
Emma Burt (CO)
Jennifer Banna (MT)
Hannah Barnhart (NV)
Lourdes Torres (TX)

Genetic Navigator Program Launches with Application!

The MSRGN Genetic Navigator program is an initiative to help families navigate the genetic services system in their state of residence. Eight genetic navigators will be selected through an **application process**, 1 per each of the **8 states** in the MSRGN region, and will complete 8 training modules on various aspects of genetic navigation.

NERGN Genetic Education Series with Weitzman Institute

In partnership with the New England Regional Genetics Network, the Weitzman Institute is excited to announce our 2021 genetics webinars.

Continuing education credits are available. A survey will be sent to all attendees of the webinar and certificate will be sent one week after completion of the survey. This webinar will be recorded and sent to all registrants. You can also view past recordings [here](#).

The topics are noted below - join one or join them all!

Session 1: Jan 11, 12pm-1pm ET
Topic: Red Flags in Metabolic disorders Part II (recording available [here](#))

Session 2: Feb 8, 12pm-1pm ET
Topic: Skin Findings in Genetic Disorders

Session 3: March 8, 12pm-1pm ET
Topic: Genetic metabolic cases

Session 4: April 12, 12pm-1pm ET
Topic: An Atypical Pathway to Clinical Genetics

Session 5: May 10, 12pm-1pm ET
Topic: Genetics reporting

[Register Here](#)

Article:
Early Intervention and Newborn Screening Parallel Roads or Divergent Highways?

Upon completion of this training, these 8 genetic navigators will be available by email to connect with families and assist them on their genetics journey. The genetics navigator role is a stipend funded opportunity for individuals or their family members impacted by genetic conditions who have navigated the genetic services system in their home state. Currently, this program is only open to individuals in the Mountain States Region States of Arizona, Colorado, Montana, Nevada, New Mexico, Texas, Utah, and Wyoming.

Application Deadline March 1, 2021.

[Apply Here](#)

"Both early intervention (EI) programs for infants and toddlers with disabilities and newborn screening (NBS) programs to identify specific disorders shortly after birth rest on the assumption that the best way to prevent or lessen the impact of a disorder is to provide treatment as early as possible. Despite this shared vision, the two programs differ in substantial ways, including historical beginnings and subsequent evolution; program entry and eligibility; service models; evidence, efficacy, and outcomes; roles and responsibilities vis-à-vis families; and financing."

[Read Article Here](#)

Save the Date!

May 24-28, 2021 ~Public Health Genetics Week~ www.phgw.org



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The MSRGN Management Team

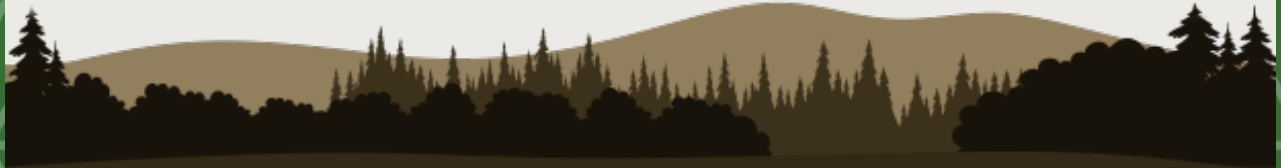
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