

GENETIC CONNECTIONS

for Early Intervention Providers



A Workshop for Early Intervention Providers



A Note from our Funders

This program is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling \$600,000.00 with 0 percent financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit [HRSA.gov](https://www.hrsa.gov).

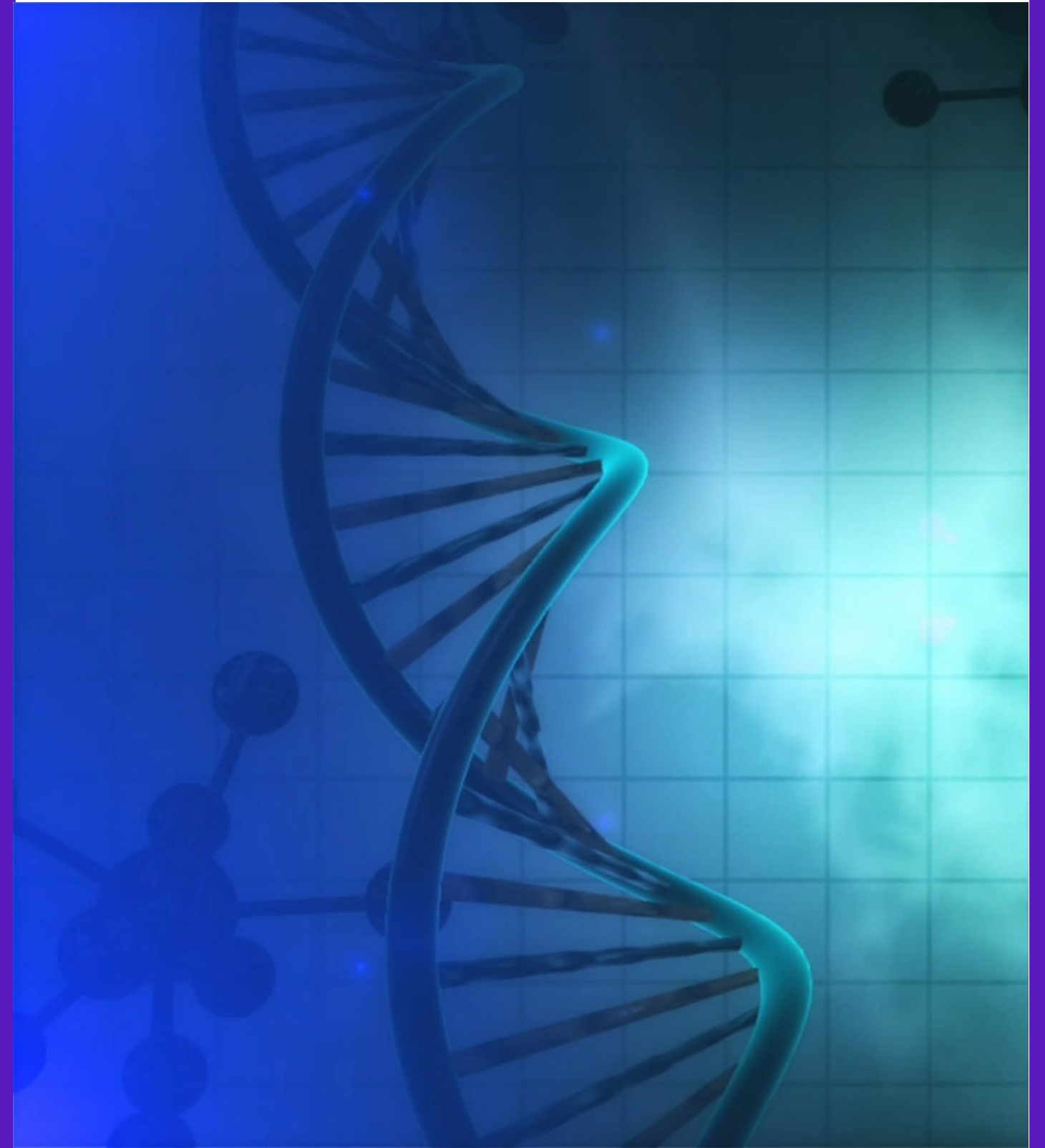
MODULE 1

Who and Why?

Learning Objective

The learner will be able to identify what MSRGN does and what the Genetic Navigators do.

The learner will be able to recognize why genetics matters during the early intervention timeframe.



Module 1: Who and Why?

A G E N D A

Topics Covered

Who: Me, MSRGN & Genetic Navigators

Why: Your Why, Genetics & Stats

What: a Survey

Resource: Red Flags 4 Genetics

Who...



Who am I?

I am a Genetic Navigator
with MSRGN



Who is MSRGN?

Mountain States Regional
Genetics Network
a HRSA funded Regional
Genetics Network



Who are the Genetic Navigators?

Individuals who have
navigated genetics for their
own loved one in one of the
8 Mountain States

W H O A M I ?



A little about me...

- How I got connected to genetics

- How I became a Genetic Navigator

WHO IS

MSRGN ?

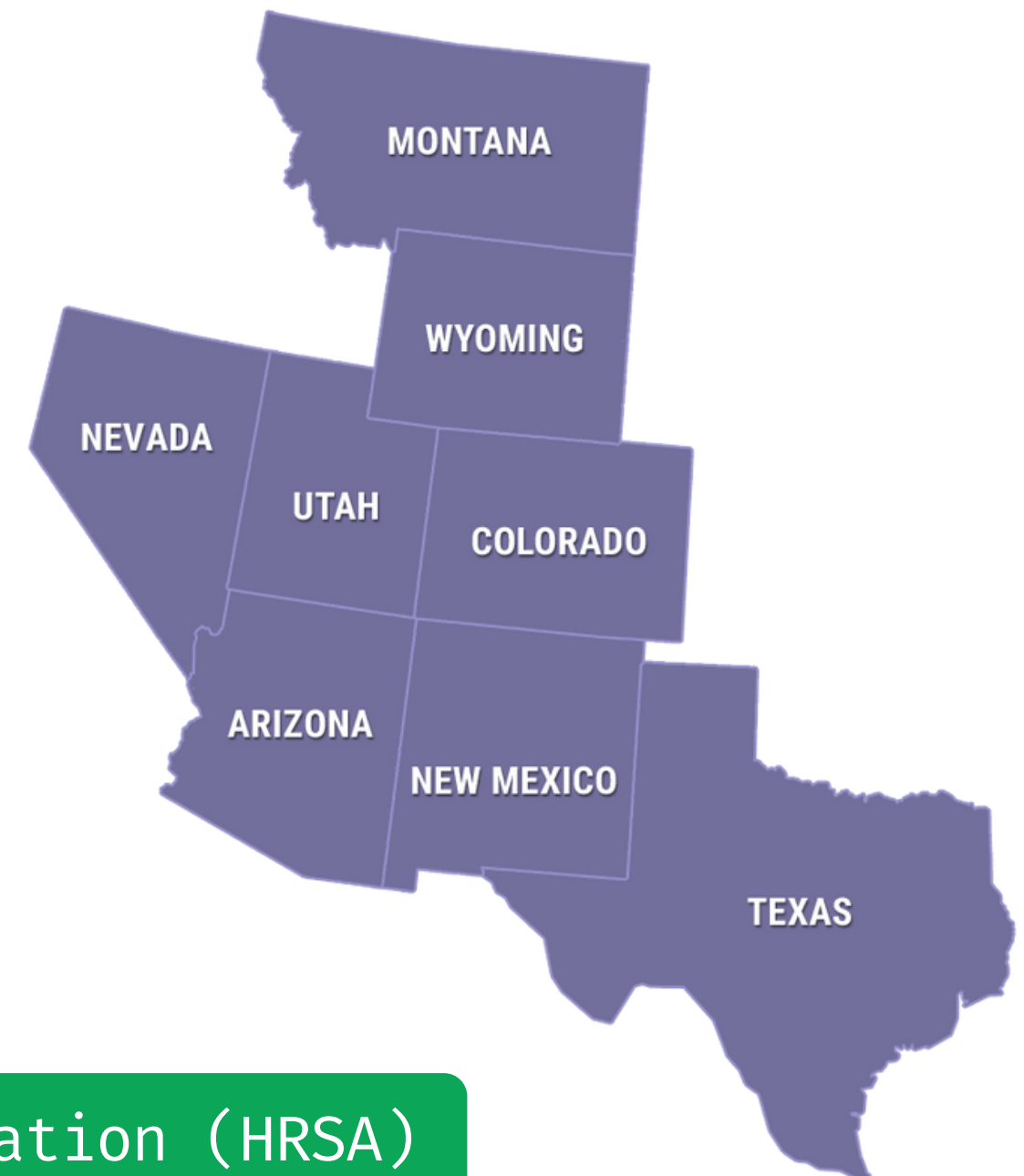


MSRGN: Mountain States Regional Genetics Network

8 States: AZ, CO, MT, NV, NM, TX, UT, WY

Funded Federally by Health Resources Service Administration (HRSA)

MSRGN's Fiduciary Agent: Texas Health Institute (THI) Austin, TX

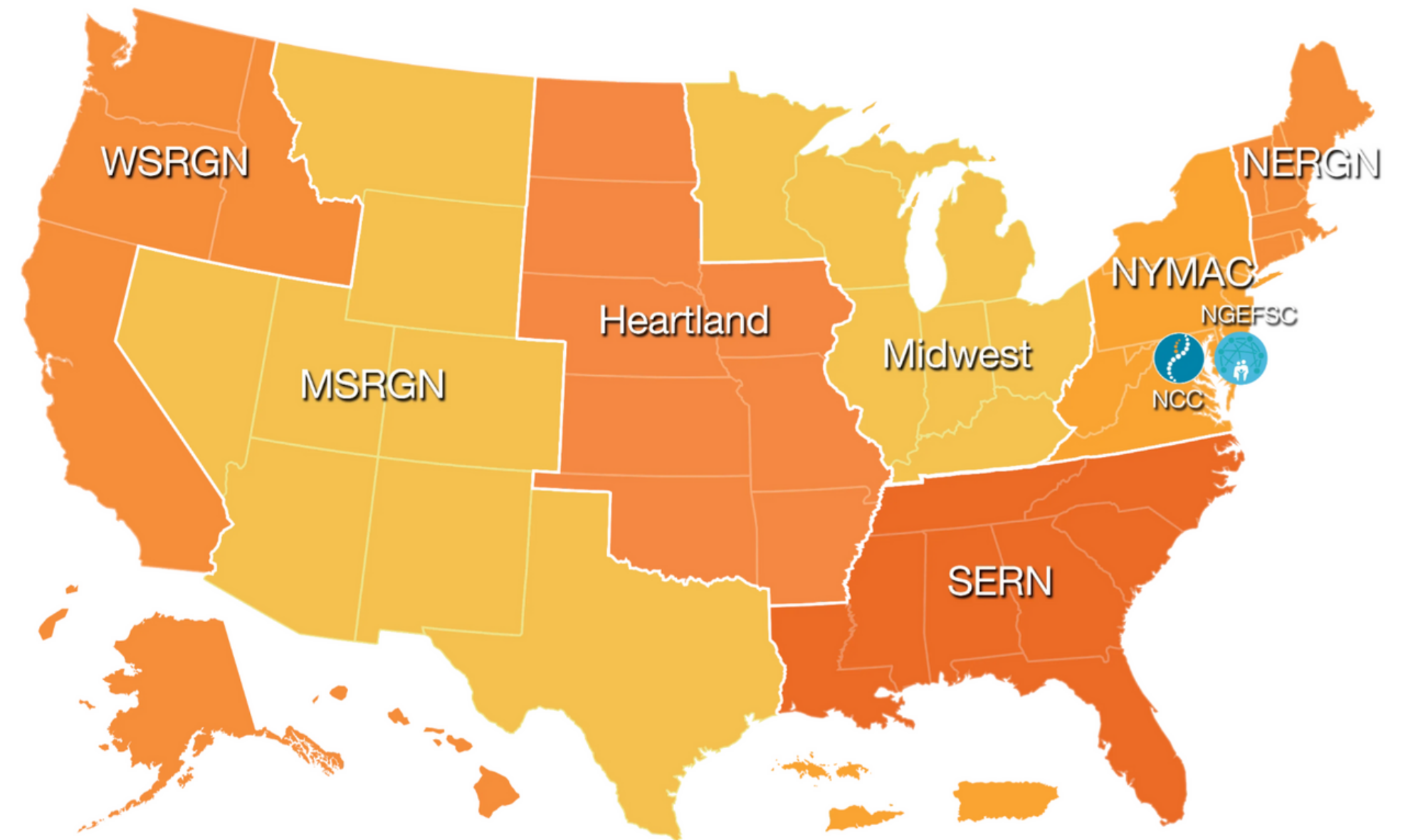


Part of a Larger National Network

- MSRGN is one of 7 Regional Genetics Networks

- National Coordinating Center (NCC)

- National Genetics Education and Family Support Center (Family Center)



Genetic Navigators: Who we are?

Individuals or family members who have navigated genetics in one of the 8 Mountain States

Selected through an application process

Completed 5 hours of Genetic Navigator training from MSRGN in addition to monthly continuing education touchpoints

Compensated by MSRGN stipend

Genetic Navigators: What we do?

Meet with families at any point along the genetics journey (concerns, red flags, pre and post diagnosis, support, odyssey, etc)

Discuss next steps with a family who has concerns.

Prepare families to have a conversation with their PCP.

Provide resources to a family who has encountered a barrier to accessing genetic services.

Connect families who have received a genetic diagnosis to support organizations.

Meet with families who want to get more involved as an advocate in the genetic system.

Write your

W

H

Y

?

Why do you think a family **MAY benefit** from exploring genetics in the Early Intervention Period of Development (birth to 3yr)?

To follow up
on a
concerning
symptom

To help a
family
find answers

To explore
treatments and
therapies for
concerning
symptoms

It may help
connect
symptoms
that seem
unrelated

Seeing pattern of
more than one child
in a family impacted
by similar symptoms

Why Genetics? Brainstorm

Why a Family might pursue a Genetic Consultation or Testing

To explore a
family history
of parent or
close relative
of a genetic
condition and
if child is
impacted

Concerning
symptoms
from birth

Regression of
skills with no
clear cause

To rule in/out
a genetic
condition

To help the
family find
support

Following up
on a therapist
or PCP
concerns

Write your

W

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?

Why do you think a family **MAY NOT benefit** from exploring genetics in the Early Intervention Period of Development (birth to 3yr)?

Some genetic conditions have no treatments

Too costly

Too mysterious
family doesn't understand it

Family does not have financial resources

Testing is culturally inappropriate with family's beliefs

Why NOT Genetics Brainstorm

Insurance won't cover it

Genetics Clinic is too far away

What are reasons a family may not benefit from pursuing a Genetic Consultation or Testing

Too invasive of a test

Takes too long to get results

Nothing you can do if the test is positive

Family is not interested in testing

Will the information change anything?

Myths about Genetics

All genetic conditions are caught at birth through Newborn Screening.

FALSE

Every childhood condition is caused by genetics.

FALSE

All genetic conditions are untreatable or incurable.

FALSE

A genetic diagnosis cannot help a child thrive.

FALSE

All genetic changes are harmful and cause disease.

FALSE

Genetic testing is VERY expensive.

FALSE

Why some families find Genetics helpful...



Seemingly unrelated symptoms may be connected by a genetic condition

3 or more systems?



Genetic Information can help a family plan and prepare for care

Not just for family planning but care planning



A genetic diagnosis can help a family find support

Even some of the most rare conditions provide a community of not being alone on the journey.



A genetic test can "rule out" a number of diagnoses.

A negative test allows doctors and families to cross possibilities off the list.

Rare Diseases & Genetics

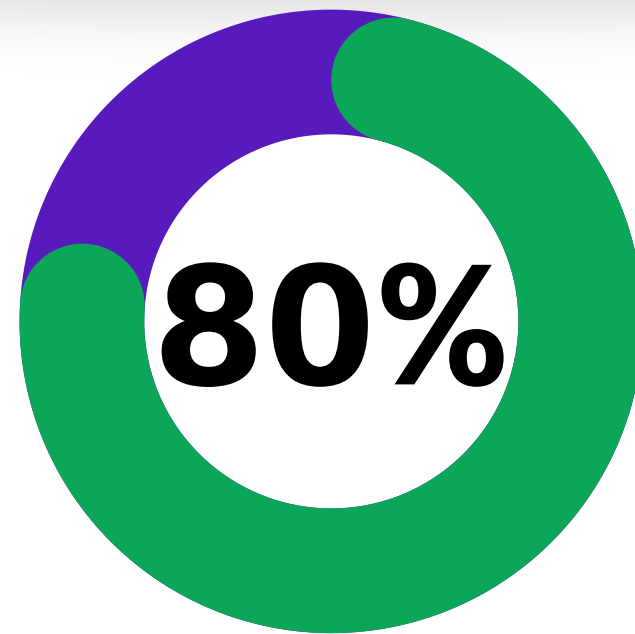
By the Numbers...

7000

identified
Rare Diseases*



*A rare disease is any disease or condition which affects or directly impacts less than 200,000 people in the U.S.



of Rare Diseases are
thought to have a
Genetic Cause
(5600)

50-75%

of Rare Diseases
(2800-4200)
Impact children



Source: <https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2022/04/nrg.2017.116.pdf>

doi:10.1038/nrg.2017.116 Published online 5 Feb 2018

More Stats...

35%

of deaths in first year of
life are caused by rare
diseases

33%

of children born with a
rare disease will not live
to see their 5th birthday

1500

genes have been shown
to cause
developmental
disorders

116

inborn errors of
metabolism which are
causally related to
Intellectual Disability
and amenable to therapy.
(Treatable-id.org)

Source: <https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2022/04/nrg.2017.116.pdf>

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Here's where Early Childhood Intervention comes in... 3 Formative Years



Birth

Newborn
Screening &
Prenatal
Diagnosis



1 year

Vision,
Hearing,
Feeding and
Early Milestone
Challenges



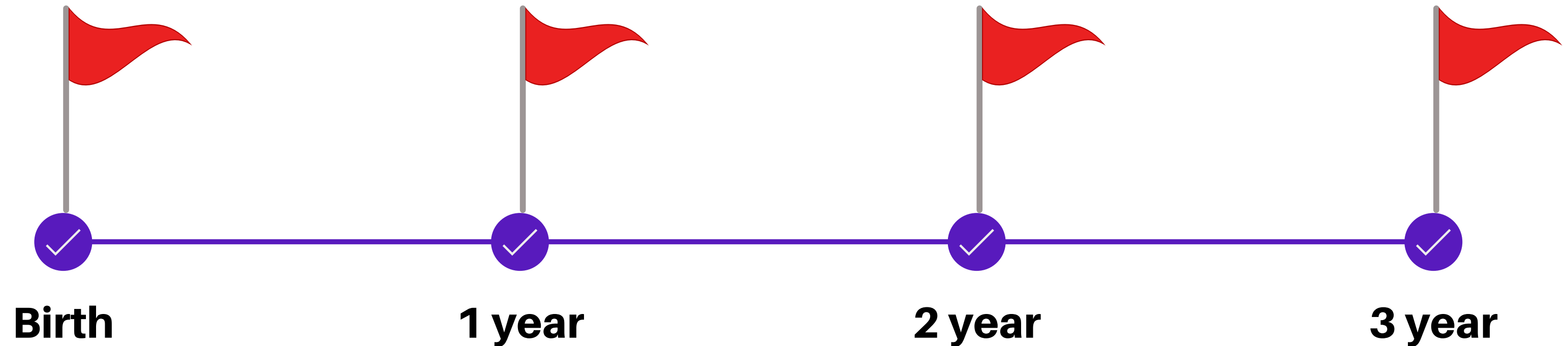
2 year

Developmental
Milestones
& Social
Emotional
Milestones

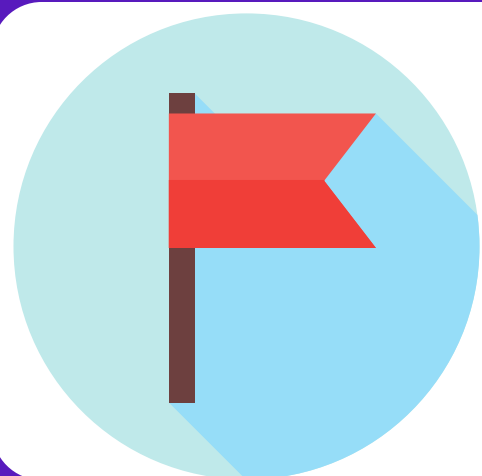


3 year

A Child ages-
out of EI at 3
but may continue
in school based
interventions.



**As Early Intervention Professionals you are
trained to notice RED FLAGS
during this early period of development.
Some RED FLAGS you observe may also be
RED FLAGS 4 GENETICS.**



A Survey...

In 2021, MSRGN asked families to tell us about the **Red Flags for Genetics** (indicators, symptoms, warning signs) they noticed. Below is some of what we learned:

6 Years

On average, families reported it took 6 years to receive a genetic diagnosis after noticing their first Red Flag indicator.

Who Made the Diagnosis

61% Geneticist
37% SubSpecialist
9% Primary Care Provider

Before the age of 4

89% of reported Red Flags were noted before 4 years of age.

almost 500 Red Flags...

114 families shared
479 red flags as part of this Survey

A R E S O U R C E

Red Flags 4 Genetics

**A Resource for having a conversation
about Genetics with a Family**

or

**A Family having a conversation about
genetics with a provider**

RED FLAGS 4 GENETICS

Do you have concerns about your child?
Below are **red flags** or warning signs and symptoms that were self-reported by families whose child went on to receive a genetic diagnosis. 89% of reported **red flags** were noted before 4 years of age.

 <h2>Developmental Delay</h2>		<ul style="list-style-type: none"> Physical/motor delay (not rolling over, crawling or walking) Speech (not babbling or responding to name) Adaptive (coordination of hands and fingers) Cognitive (unable to or not interested in play) Social/emotional (difficulty interacting with others, not smiling) Loss of any milestone (regression)
 <h2>Physical Features</h2>	<ul style="list-style-type: none"> Extra fingers or toes Large/small head size (macrocephaly/microcephaly) Short/tall height (stature) Cleft palate Facial feature differences (small chin, wide forehead) Frequent joint dislocations (hip dysplasia) Spinal deformities (scoliosis) 	
 <h2>Feeding Issues</h2>		<ul style="list-style-type: none"> Difficulty swallowing Struggles with breastfeeding Poor latching Feeding tube dependent Never feeling full Poor appetite Vomiting Gagging on food
 <h2>Behavior</h2>	<ul style="list-style-type: none"> Autism Spectrum Disorder (ASD) Repetitive movements Constantly upset or crying Lack of eye contact Difficulty in school or social settings ADHD/impulsive or hyperactive behavior Easily distracted High pitched cry/tone of voice 	
 <h2>Failure To Thrive</h2>		<ul style="list-style-type: none"> Slow growth Slow weight gain Not thriving Fatigues quickly when active Weight or rate of weight gain being much lower than that of other children.
 <h2>Strength & Tone</h2>	<ul style="list-style-type: none"> Floppy or low muscle tone (hypotonia) Stiff or high muscle tone (hypertonia) Baby not attempting to roll over or hold head up when on tummy Muscle weakness Body is floppy Inability to lift or control head (head lag) 	
<h3>Other Red Flags</h3> <ul style="list-style-type: none"> Seizures Sleep concerns Fatigue Lethargy Hearing/vision concerns Immune issues Headaches/migraines Neurological concerns Pulmonary/cardiac concerns GI issues (reflux/bowel concerns) Bruising 		<p>If your child is exhibiting any 'red flags' on this page please talk to your child's pediatrician or primary care physician. For more resources and help having that conversation, go to: www.mountainstatesgenetics.org/redflag</p>  <p>This resource is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling \$600,000.00 with 0 percent financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov</p>

SEÑALES DE ALARMA SOBRE GENÉTICA

¿Tiene preocupaciones sobre su hijo?
Abajo le brindamos señales de alarma o señales de aviso y síntomas las cuales fueron reportadas por familias con hijos que recibieron un diagnóstico genético.. El 89% de las señales de alarma notificadas se notaron antes de los 4 años de edad.

 Retraso del desarrollo		<ul style="list-style-type: none">• Retraso físico/motor (no se da vueltas, no gatea ni camina)• Retraso del habla (no balbucea ni responde a su nombre)• Adaptable (coordinación de manos y dedos)• Cognitivo (no puede o no tiene interés en jugar)• Social/emocional (dificultad para interactuar con los demás, no sonríe)• Pérdida de logros del desarrollo (regresión)
 Características físicas	<ul style="list-style-type: none">• Dedos adicionales en los pies o manos• Tamaño de la cabeza grande/pequeño (macrocefalia/microcefalia)• Estatura baja/alta• Paladar hendido• Diferentes rasgos faciales (mentón pequeño, frente ancha)• Dislocación frecuente de las articulaciones (displasia de la cadera)• Deformidades de la columna vertebral (escoliosis)	
 Problemas de alimentación	 <ul style="list-style-type: none">• Dificultad para pasar los alimentos• Problemas con la lactancia• Mal agarre del pecho• Dependiente de la sonda de alimentación• Nunca se siente saciando• Falta de apetito• Vómitos• Se atraganta con la comida	
 Comportamiento	<ul style="list-style-type: none">• Desorden del Espectro Autista (ASD)• Movimientos repetitivos• Está constantemente molesto o llorando• Falta de contacto visual• Dificultad en la escuela o en entornos sociales• TDAH/Comportamiento impulsivo o hiperactivo• Se distrae fácilmente• Llora o tiene un tono de voz agudo, como un grito	
 Falta de crecimiento	 <ul style="list-style-type: none">• Crecimiento lento• Aumento de peso lento• Falta de desarrollo• Se fatiga rápidamente cuando está activo• El peso o la tasa de aumento de peso es mucho más bajo que el de otros niños	
 Fuerza y tono muscular	<ul style="list-style-type: none">• Tono muscular flácido o bajo (hipotonía)• Tono muscular rígido o alto (hipertonía)• El bebé no intenta darse la vuelta ni mantiene la cabeza eruida al estar boca abajo• Debilidad muscular• Tiene el cuerpo flácido• Incapacidad de levantar o controlar la cabeza (deja caer la cabeza hacia atrás)	



Otras Señales

- Convulsiones
- Falta de sueño
- Fatiga
- Letargo
- Preocupaciones auditivas/visuales
- Problemas inmunológicos
- Dolores de cabeza/migrañas
- Preocupaciones neurológicas
- Preocupaciones pulmonares /cardíacas
- Problemas del sistema digestivo
- Moretones



Si su hijo muestra alguna 'señal de alarma' aquí mencionada, hable con el pediatra o médico de atención primaria de su hijo. Para obtener más recursos y ayuda para tener esa conversación, visite:
www.mountainstatesgenetics.org/redflag

Este proyecto está respaldado por la Administración de Recursos y Servicios de Salud (HRSA) del Departamento de Salud y Servicios Humanos (HHS) de los EE. UU. bajo el número de subvención UH7MC30776, por \$600 000. Esta información o contenido pertenecen al autor y no deben ser considerados una opinión oficial o políticas de la HRSA, HHS ni del gobierno de los EE. UU. ni se debe asumir ningún apoyo por parte de ellos.



WHAT'S DNA HAVE TO DO WITH IT?

DON'T NAVIGATE ALONE

DO NAVIGATE ALONGSIDE

[www. MountainStatesGenetics.org](http://www.MountainStatesGenetics.org)

T H A N K
Y O U

**Join us for Module 2:
The Vocabulary of Genetics**