GENETIC CONNECTIONS

for Early Intervention Providers

GENETIC CONNECTIONS

A Workshop for Early Intervention Providers

Mountain States
REGIONAL GENETICS NETWORK

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.

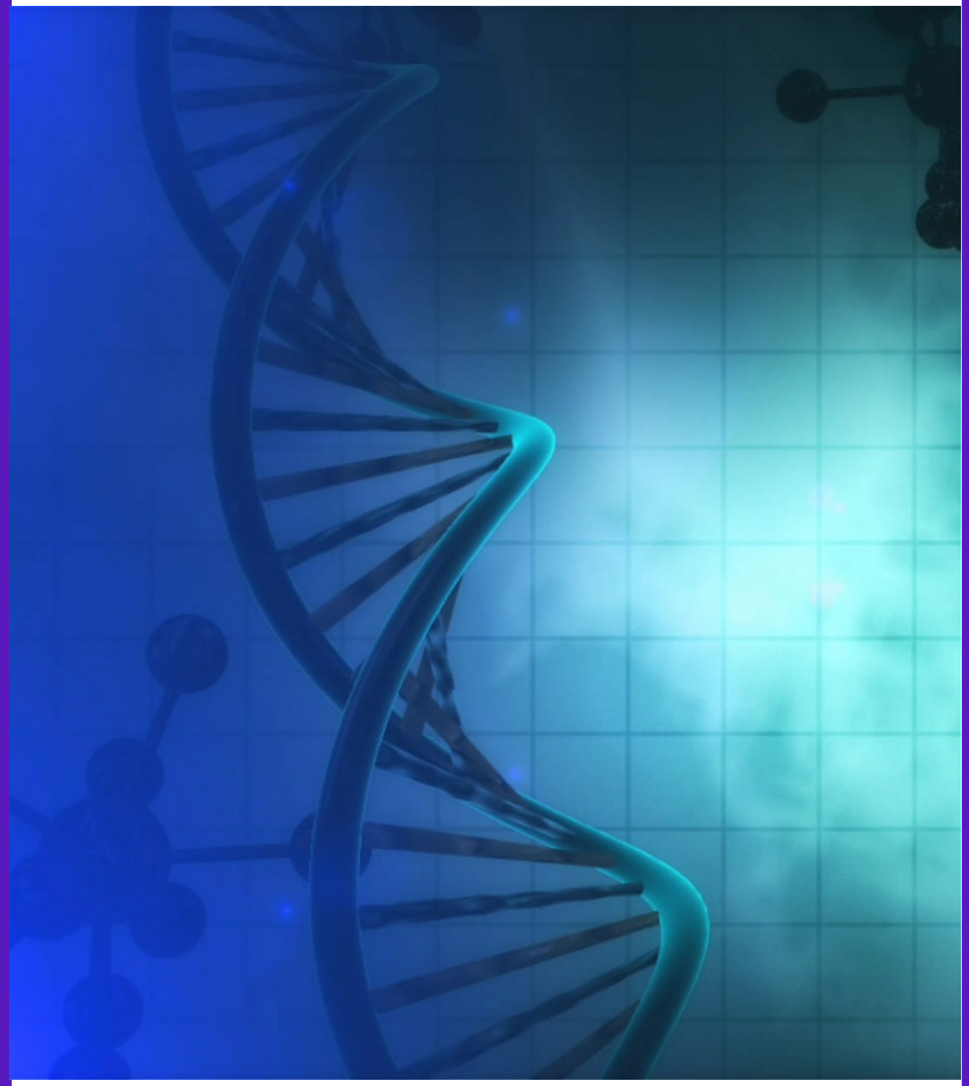
MODULE 3

Four Families

Learning Objectives

The learner will be able to identify example families whom an early intervention provider might refer to MSRGN's Genetic Navigator program.

The learner will be able to show why genetic consultation or testing might be helpful for a family.



Module 3: Four Families

A G E N D A

Topics Covered

Family #1: Genetic Diagnosis at Birth

Family #2: Undiagnosed- Developmental Delays

Family #3: Developmental Regression

Family #4- Hearing Loss

Resource: Genetic Navigators

Diagnosed at Birth with a Genetic Condition

FAMILY STORY 1

Mountain States
REGIONAL GENETICS NETWORK

Meet the Winn Family

Family #1:
Diagnosed a birth with Down Syndrome

Meet the Winn Family

1. Mom knew she was having a baby with a diagnosis of down syndrome during her 20 week ultrasound while pregnant
2. Child, Eric, referred to Early Intervention at a year old because of developmental concerns.
3. Mom feels like there is something else going on in addition to DS diagnosis because of waxing and waning of milestones. Mom curious about further genetic exploration. EI Occupational Therapist documents milestone changes.
4. Early intervention OT connects mom to MSRGN Genetic Navigator

Genetic Navigator Connection

The Winn Family

Outcome: EI OT connects Mom Winn with MSRGN Genetic Navigator who helps mom identify that her #1 concern is ruling out if something else genetic/metabolic is going on for her son. GN helps mom prepare for a conversation with her PCP about her concerns. This PCP conversation results in referral to genetics clinic for additional evaluation. Genetic/Metabolic testing reveals the child also has a treatable metabolic/genetic condition that is responsive to nutritional treatment.

Undiagnosed Developmental Delays

FAMILY STORY 2

Mountain States
REGIONAL GENETICS NETWORK

Meet the Abibi Family

Family #2:
Developmental Delays

Meet the Abibi Family

1. Pregnancy and Birth normal.
2. Child, Sahil, referred to Early Intervention at a year old because of developmental concerns.
3. EI PT notices skeletal deformities and gross motor delays that have not been previously noted or followed up on by pediatrician. EI PT suspects genetic condition, but feels hesitant to step on anyones toes by suggesting further follow up.
4. EI PT connects mom to MSRGN Genetic Navigator

Genetic Navigator Connection

The Abibi Family

Outcome: Mom Abibi reaches out to the Genetic Navigator and they have a call. GN encourages mom to have EI PT document in writing her concerns about skeletal deformaties and for mom to take that to Sahil's upcoming 18 month year well visit. GN and Mom Abibi prepare for that appointment by coming up with Mrs. Abibi's top 3 concerns/questions and request for further evaluation. PCP confirms PT's findings for skeletal abnormalities upon exam and refers to genetics for further evaluation where testing reveals a genetic diagnosis of MPS1 with several therapies available.

Developmental Regression

FAMILY STORY 3

Mountain States
REGIONAL GENETICS NETWORK

Meet the Xavier Family

Family #3:
Developmental Regression

Meet the Xavier Family

1. Family recently moved to the state for dad's job. The family's 16 month old toddler girl, Alexa, has been self referred by parents to EI because she is not walking yet.
2. Mom shares concerns at the 18 month well visit. Pediatrician has told mom not to worry because sister was a later walker also. Mom still concerned and mentions it to EI SLP.
3. A few days after 18 month well check, Alexa, stops crawling & babbling, and is having jerky movements. Service coordinator and SLP who are in the home a week later and are alarmed by the developmental regression that has lasted for over a week and encourage family to contact pediatrician ASAP.
4. Early intervention service coordinator connects mom and dad to MSRGN Genetic Navigator due to Red Flags 4 Genetics handout.

Genetic Navigator Connection

The XavierFamily

Outcome: The EI staff had encouraged dad to call the pediatricians office to let them know about this sudden developmental regression. Dad Xavier did so that night and the after hour provider recommends they go to the ER to rule out a seizure. When dad speaks to GN, they have just gotten home from the ER and seizures were ruled out, but some of the blood work that was taken in the ER has come back concerning for a metabolic/genetic condition and they are being referred to genetics for an ASAP appointment. The GN helps dad prioritize his concerns, questions, and sends dad a video of what to expect at a genetics consultation to help the family prepare for this appointment. In Genetics, Alexa is diagnosed with a rare genetic condition that can cause autism. With early intervention and intensive therapy, she regains her lost milestones.

Hearing Loss

FAMILY STORY 4

Mountain States
REGIONAL GENETICS NETWORK

Meet the Yen Family

Family #4:
Hearing Loss

Meet the Yen Family

1. Family with 4 children, youngest child has a diagnosis of hearing loss after confirmation of NBS. Family seeks genetic testing . The youngest child is diagnosed with "connexin 26" (Cx26, GJB2) (~50% of non-syndromic hearing loss)
2. Family's 2.5 year old child (sibling) is in EI for developmental and speech concerns.
3. Mom shares speech delay concerns with pediatrician for 2.5 yr old. Despite the youngest's genetic diagnosis, the 2.5 yr olds pediatrician has said it cannot be hearing loss because the 2.5 yr old's NBS was normal.
4. Early intervention service coordinator connects mom and dad to MSRGN Genetic Navigator for help advocating for mom's concerns.

Genetic Navigator Connection

TheYen Family

Outcome: Mom Yen is introduced to the Genetic Navigator by the service coordinator via email. GN and Mom Yen correspond via email and GN encourages mom to share information about Connexin 26 with pediatrician, highlighting that this genetic change can cause both hearing loss at birth as well as progressive hearing loss in late childhood. GN also encourages mom to ask Pediatrician if audiological testing can be done.

Mom Yen meets with pediatrician and advocates (sharing the genetic information for younger sibling) for audiological testing and Pediatrician agrees for referral.

The male sibling was diagnosed with significant hearing loss at 3yr old.

Genetic Testing can be powerful for health of siblings based on another child in the family's genetic testing diagnosis.

Mountain States
REGIONAL GENETICS NETWORK

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.

Developmental Delay

- 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)

Physical Features

- 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)

Feeding Issues

- Difficulty swallowing
- Struggles with breastfeeding
- Poor latching
- Feeding tube dependent
- Never feeling full
- Poor appetite
- Vomiting
- Gagging on food

Behavior

- 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

Failure To Thrive

- 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.

Strength & Tone

- 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)

Other Red Flags

- Seizures
- Sleep concerns
- Fatigue
- Lethargy
- Hearing/vision concerns
- Immune issues
- Headaches/migraines
- Neurological concerns
- Pulmonary/cardiac concerns
- GI issues (reflux/bowel concerns)
- Bruising

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

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

A RESOURCE

Resource:

GENETIC NAVIGATORS

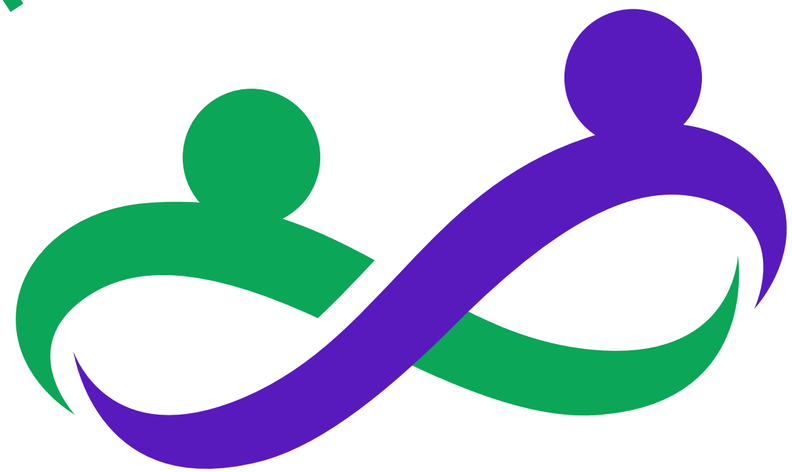
How to Connect with a MSRGN

Genetic Navigator

You can email them directly, or email introduce the family (with family permission), or give email address to the family to reach out when they are ready:

Arizona ~ arizonagenetic@gmail.com
Colorado ~ coloradogenetic@gmail.com
Montana ~ montanagenetic@gmail.com
Nevada ~ nevadagenetic@gmail.com
New Mexico ~ newmexicogenetic@gmail.com
Texas ~ texasgenetic@gmail.com
Utah ~ utahgenetic@gmail.com
Wyoming ~ wyominggenetic@gmail.com

GENETIC CONNECTIONS



WHAT'S DNA HAVE TO DO WITH IT?

DON'T NAVIGATE ALONE

DO NAVIGATE ALONGSIDE

THANK YOU

Join us for Module 4:
Next Steps