Pre-Webinar Survey

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Genetic Contributions to the Developmental Journey

Part I

A conversation with the NYMAC Genetics Team and Virginia Genetic Navigators

2

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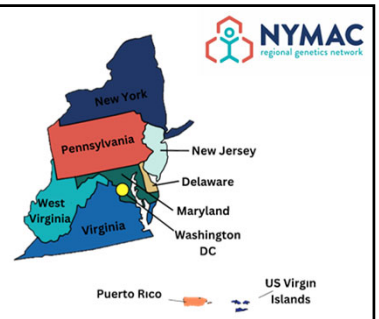
Who are we? What is NYMAC?



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Our Collaborators in Virginia

NYMAC has a local team of diverse stakeholders in each of our locations to shape local plans and priorities.

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Pre-Webinar Survey

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Pre-Webinar Survey

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AGENDA

Part 1

- Genetics 101 review
- Why does genetics matter for EI?
- "Red Flags" for genetics in children

Part 2

- Who is NYMAC and what do we do in VA?
- Who are the Virginia Genetic Navigators (GNS) and what do they do?
- How can NYMAC and VA's GNs be a resource for EI professionals?

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Why is genetics important?

Because some health challenges can be caused by changes in our genes.

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Genetics and EI by the numbers….

1500 genes → developmental challenges

Autism	Hearing loss in babies	Retinitis Pigmentosa *
25%	50-60%	50-60%

* a common cause of vision loss

% of clinical diagnoses that are caused by a known genetic cause

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Genetics 101

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What words come to mind when you hear the word **GENETICS**?

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GENETIC ANALOGY: COOKING

See handout for more info!

DNA Alphabet = Letters used to write out the recipe

AT
CG

Gene = Recipe

Chromosome = Recipe Book

Genome = Set of Recipe Books

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DNA = Deoxyribonucleic Acid

Alphabet used to write our body's instructions

DNA Alphabet → **A, T, C, G**

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GENES

Sections of the DNA that contains instructions ("recipe") to make proteins, enzymes, and other products in the cells of the body.

Typically, one copy of each gene is inherited from each parent.

Gene = Recipe

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CHROMOSOME

DNA is packaged into chromosomes (recipe book).
Each chromosome holds many genes.

Cells typically each have 46 chromosomes (23 pairs).
The first 22 pairs are numbered 1-22.
The 23rd pair is the sex chromosomes (X and Y).

Chromosome = Recipe Book

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GENOME

The genome is the entire set of genetic instructions found in a cell.

Genome = Set of Recipe Books

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GENETIC ANALOGY: COOKING

DNA Alphabet = Letters used to write out the recipe

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How can genetic changes lead to health challenges and disability?

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GENETIC CHANGES

…do not always cause health challenges

Mutations/pathogenic variants = changes in genes that can cause needed products to form and/or function differently

Can have very big changes or very small changes

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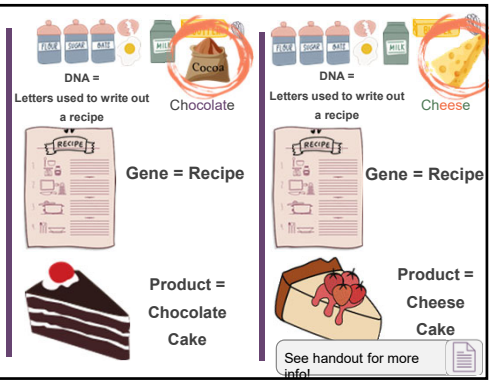
GENETIC CHANGE: COOKING

How can a change in a recipe (or gene) change the final product (the person)?



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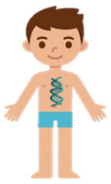
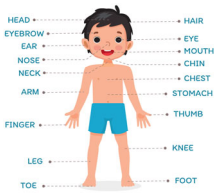
GENETIC CHANGES



Genetic change

Causes a change in a protein

(Amount, Shape, Format)



Protein change can impact any part of the body

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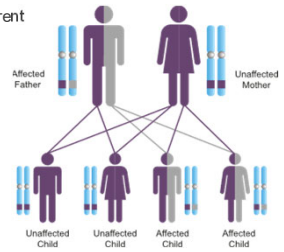
How can genetic changes run in families?

Different genetic conditions have different forms of inheritance:

- Autosomal dominant
- Autosomal recessive
- X-linked dominant
- X-linked recessive
- Mitochondrial
- De Novo
- Somatic



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Why might a family want a genetics appointment?

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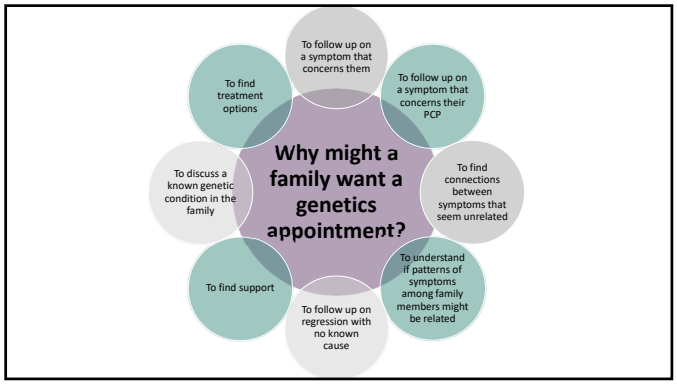
Why might a family want a genetics appointment?
To follow up on a symptom that concerns them
To follow up on a symptom that concerns their PCP
To find connections between symptoms that seem unrelated
To understand if patterns of symptoms among family members might be related
To follow up on regression with no known cause
To find support
To find treatment options
To discuss a known genetic condition in the family

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Genetics Poll:
Select All That Are True

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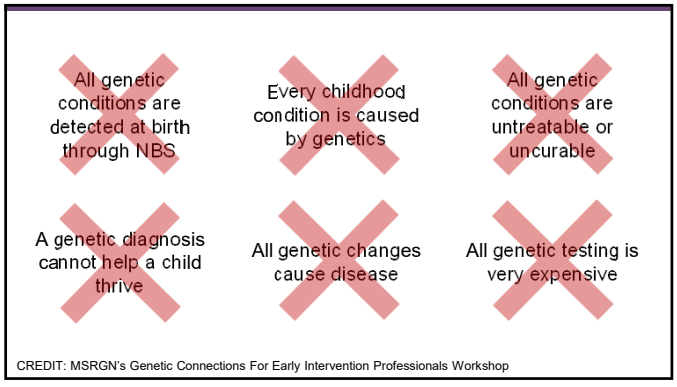
All genetic conditions are detected at birth through NBS
Every childhood condition is caused by genetics
All genetic conditions are untreatable or uncurable
A genetic diagnosis cannot help a child thrive
All genetic changes cause disease
All genetic testing is very expensive
CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop

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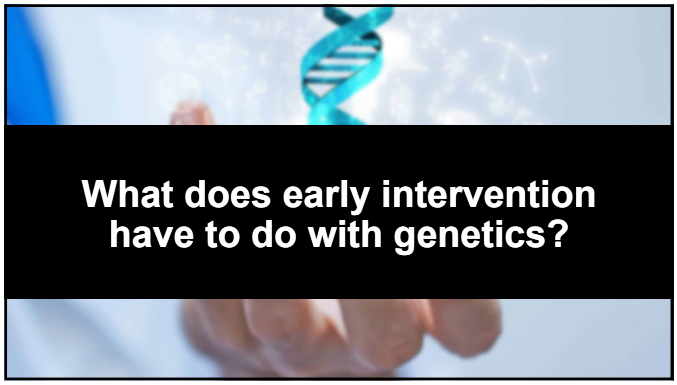
What does early intervention have to do with genetics?

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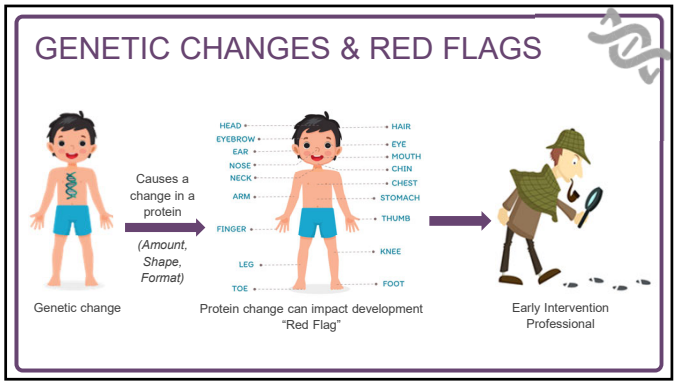
GENETIC CHANGES & RED FLAGS
Causes a change in a protein
(Amount, Shape, Format)
HEAD
EYEBROW
EAR
NOSE
NECK
ARM
FINGER
LEG
TOE
HAIR
EYE
MOUTH
CHIN
CHEST
STOMACH
THUMB
KNEE
FOOT
Genetic change
Protein change can impact development "Red Flag"
Early Intervention Professional

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Developmental Milestones and "Red Flags"

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Genetics & Development

What are some genetic conditions that the children you've worked with have had?

Has anyone on the call today ever suspected that a health condition in a family that you're working with might run in their family (but there's no diagnosis yet)?

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Overlap Between Developmental Red Flags And Genetics Red Flags

Example: Developmental delay and hypotonia may be a "red flag" for conditions such as autism or various genetic conditions

https://nymacgenetics.org/providers/when-to-refer-patient-to-genetics/pediatric-genetics-referrals/

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A visit to a genetics specialist can be helpful if you are concerned about your child's...

or if...

- you or your partner have been told you have a higher chance to have a child with a health issue
- you have a family member with a genetic condition
- you believe your family's medical issues could have a genetic explanation

CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop;

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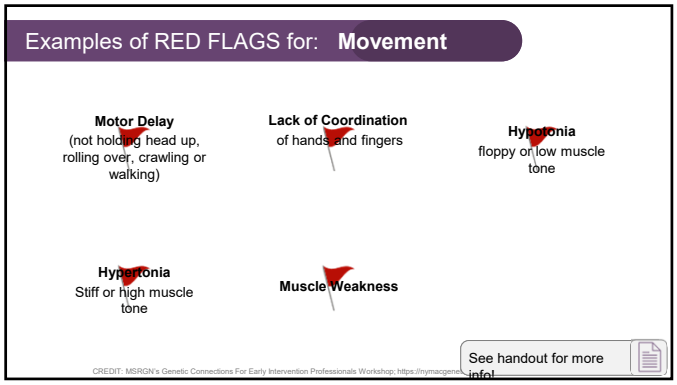
Examples of RED FLAGS for: Movement
Motor Delay
(not holding head up, rolling over, crawling or walking)
Lack of Coordination
of hands and fingers
Hypotonia
floppy or low muscle tone
Hypertonia
Stiff or high muscle tone
Muscle Weakness
See handout for more info!
CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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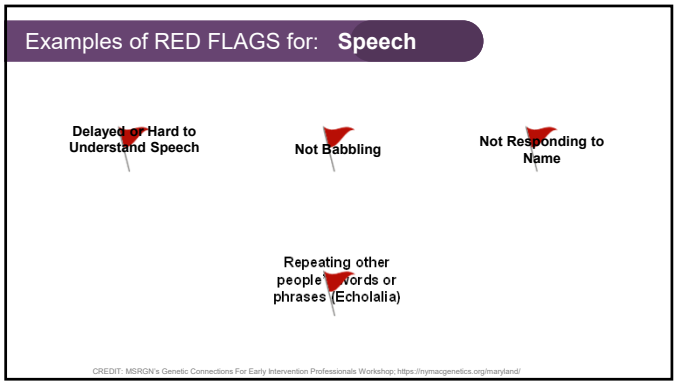
Examples of RED FLAGS for: Speech
Delayed or Hard to Understand Speech
Not Babbling
Not Responding to Name
Repeating other people's words or phrases (Echolalia)
CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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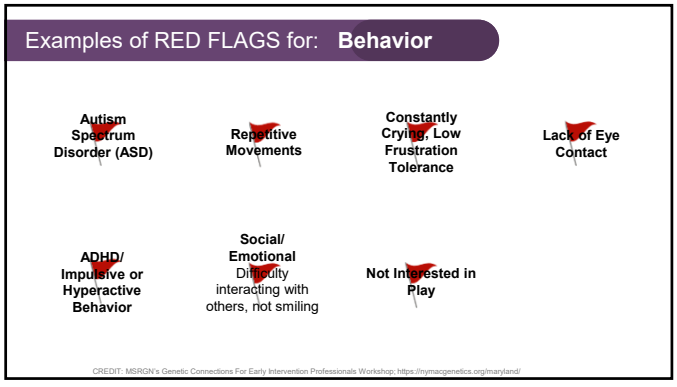
Examples of RED FLAGS for: Behavior
Autism Spectrum Disorder (ASD)
Repetitive Movements
Constantly Crying, Low Frustration Tolerance
Lack of Eye Contact
ADHD/ Impulsive or Hyperactive Behavior
Social/ Emotional
Difficulty interacting with others, not smiling
Not Interested in Play
CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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Examples of RED FLAGS for: **Digestive/Feeding**

- **Difficulty Swallowing**
- **Extreme Struggles with Latching and Breastfeeding**
- **Use of a Feeding Tube**
- **Constantly Hungry**
- **Poor appetite and/or Extreme Pickiness**
- **Persistent Vomiting**
- **Gagging on Food**

CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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Examples of RED FLAGS for: **Growth**

- **Slow or Uneven Growth**
- **Slow Weight Gain/Failure to Thrive**
- **Growing too Fast/too Tall**
- **Fatigues Quickly When Active**
- **"Growing Pains" and Muscle Aches**

CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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Examples of RED FLAGS for: **Physical Features**

- **Birth Defects** Such as Cleft Lip and/or Palate
- **Macrocephaly/Microcephaly** Head Size that's Unusually Large or Small for Age
- **Stature** Unusually Short or Tall Height for Age
- **Facial Feature Differences (Small Chin, Wide Forehead)**
- **Frequent Joint Dislocations (Hip Dysplasia)**
- **Spinal and Skeletal Differences**

CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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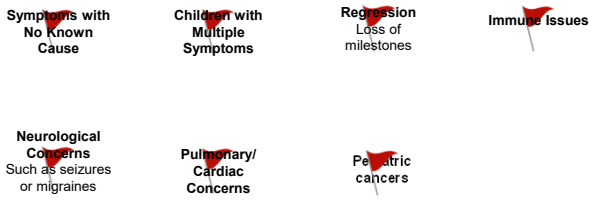
Examples of RED FLAGS for: **Complicated Medical Issues**



CREDIT: MSRGN's Genetic Connections For Early Intervention Professionals Workshop: https://nymacgenetics.org/maryland/

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Other Red Flags
The Rules of "Too" / "Two"

- TOO tall
- TOO short
- TOO early
- TOO many
- TOO young
- TOO different
- TOO many illnesses

- TWO tumors
- TWO generations
- TWO in the family
- TWO birth defects

See handout for more info!

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EVERY CHILD WITH A GENETIC CONDITION HAS RED FLAGS…

BUT EVERY CHILD WITH A RED FLAG **DOES NOT** HAVE A GENETIC CONDITION

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Red flag?

Use the
Medical Home Model of Care

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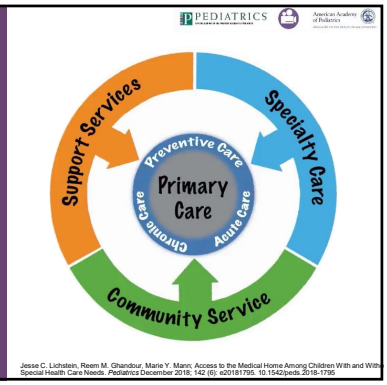
Red flag?

Use the
Medical Home Model of Care



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Thank you!
Questions?

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Post-Webinar Survey

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Post-Webinar Survey

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Genetic Contributions to the Developmental Journey

Part II

- Who is NYMAC and what do we do in VA?
- Who are the Virginia Genetic Navigators and what do they do?
- How can NYMAC and VA's GNs help EI professionals?

October 5th

12:00 – 1:00 PM EST

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Genetic Contributions to the Developmental Journey Part I
Resource Handout | September 2023
Resources

See the Handout for More Resources!

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Check Your Inbox


Take the Survey

Download the Certificate!

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