



# Genetics and Health

How CHWs can facilitate access to genetic services for NJ families



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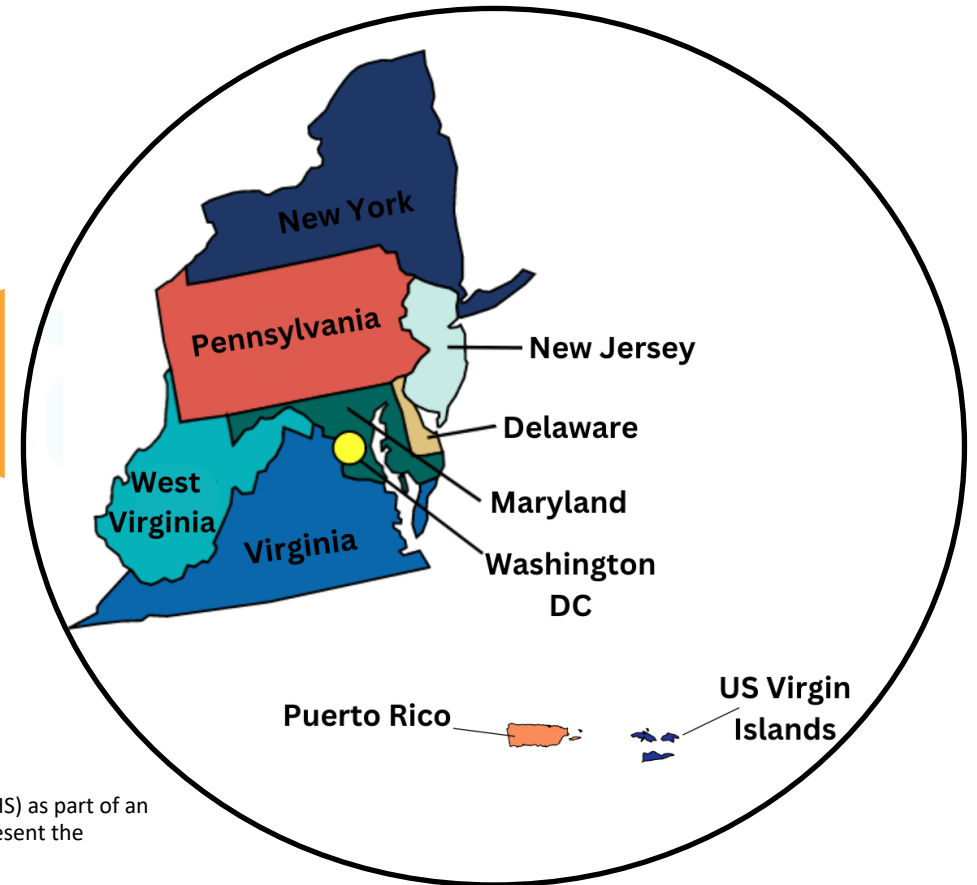
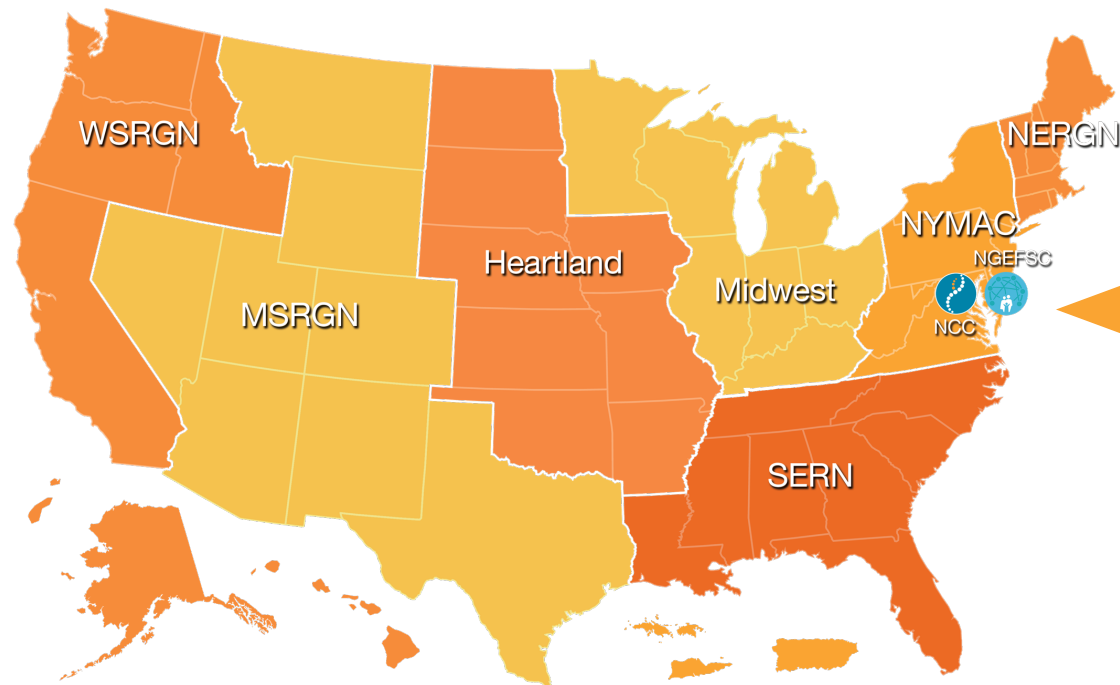
Certified Genetic Counselor



NYMAC Regional Genetics Network

# Who is NYMAC?

- One of seven **Regional Genetics Networks** (RGNs) supported by federal grant funding from **The Health Resources and Services Administration** (HRSA)
- The NYMAC region includes **10 diverse jurisdictions** and **62 million people**.
- Our goal is to **improve access to genetic services** to underserved populations





**New Jersey Chapter**

INCORPORATED IN NEW JERSEY

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®




# NYMAC TEAM NJ: A Collaborative Effort



Genetic Counseling  
Graduate Program

*forward-thinking supportive diverse*





# Learning Objectives

Describe why genetics is important to the health of NJ families

Define basic genetic concepts

Identify clues to genetic conditions

Explain how genetic services can benefit families

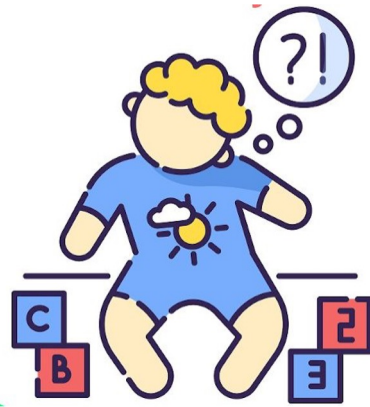
Outline how a CHW can help improve access to genetics

Summarize frequently asked questions about genetics

# The impact of genetics on health

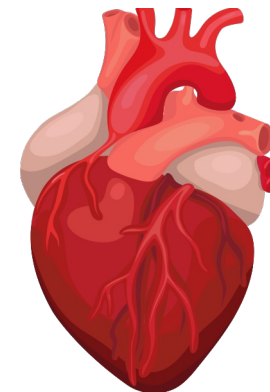
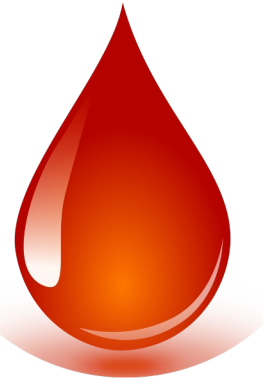






## Why is genetics important?

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





# Genetics by the numbers...

Autism

25%

Hearing loss  
in babies

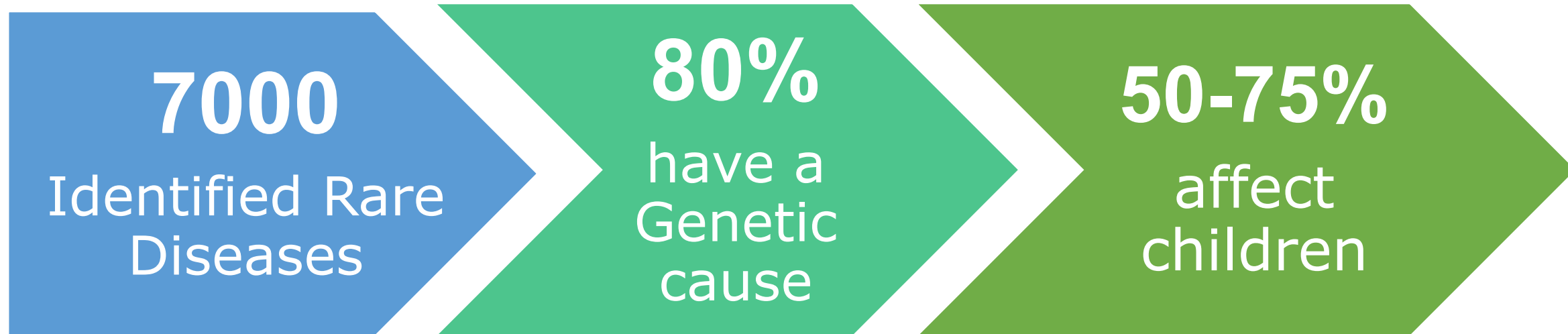
50-60%

Cancer

~10%

*Many families are unaware that they have a genetic condition or are at risk for one!*

# Rare Disease & Genetics by the Numbers



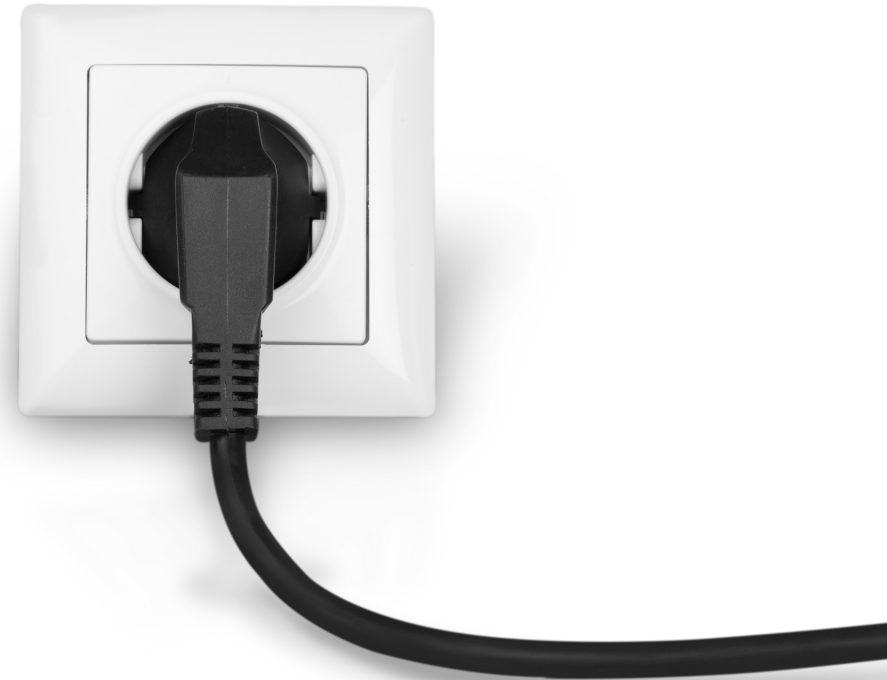
**Rare diseases are INDIVIDUALLY RARE but COLLECTIVELY COMMON**

The National Organization for Rare Disorders (NORD) supports families, builds community, advocates for policies, and advances research for rare disorders

A hand is shown holding a glowing blue DNA double helix structure. The background is dark and slightly blurred, suggesting a laboratory or medical setting. The text is overlaid on the image in a bold, white font.

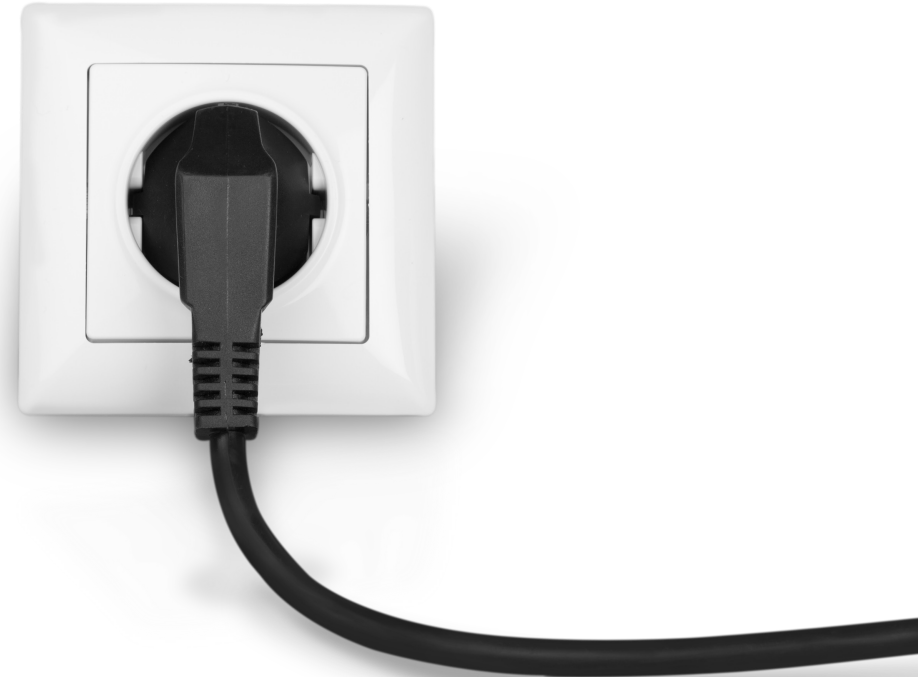
**What does genetics have  
to do with Community  
Health Workers?**

What does this have to do with  
your work with families?



You can be a

Detector & Connector



# Community Health Workers can increase community **AWARENESS** of and **ACCESS** to genetic services



Help families understand the life-long importance of genetics



Reduce the guilt, shame, secrecy and silence about genetic conditions in the family

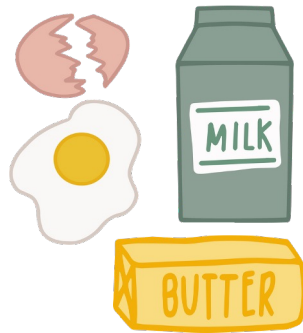


Help families overcome barriers to obtaining a genetics appointment

# Genetics concepts



# GENETIC ANALOGY: COOKING



DNA Alphabet =  
Letters used to write out the  
recipe

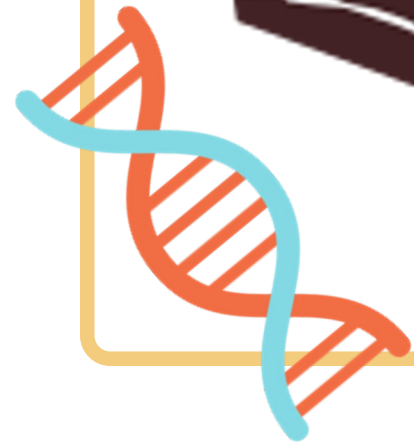
AT  
CG

Gene = Recipe



Chromosome = Recipe Book

Genome = Set of Recipe Books





# DNA = Deoxyribonucleic Acid



*Alphabet used to write our body's instructions*

**DNA  
Alphabet**



**A, T, C,  
G**

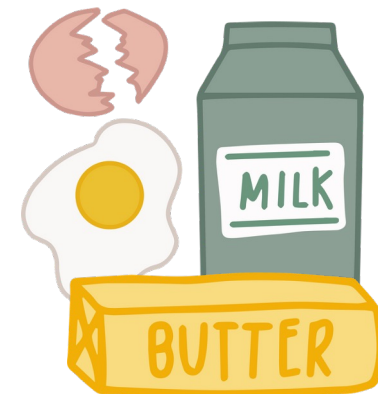
for more info: [genome.gov/genetics-glossary/Deoxyribonucleic-Acid](https://www.genome.gov/genetics-glossary/Deoxyribonucleic-Acid)

# 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



# 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**



# GENOME

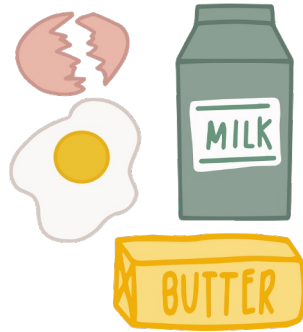
The genome is the entire set of genetic instructions found in a cell.



Genome = Set of Recipe Books



# GENETIC ANALOGY: COOKING



DNA Alphabet =  
Letters used to write  
out the recipe

AT  
CG

Gene = Recipe



Chromosome = Recipe Book

Genome = Set of Recipe Books



How can genetic changes lead to health challenges and disability?



# GENETIC CHANGES

*...do not always cause health challenges*



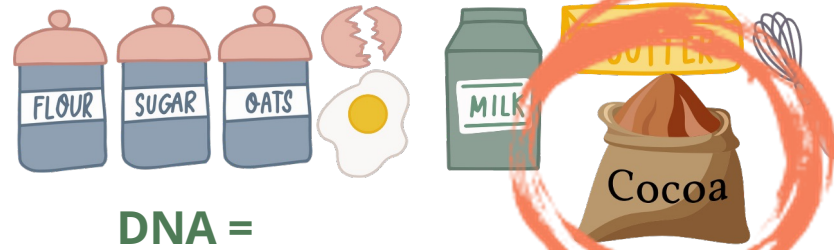
We all have “typos” or changes in genes. These changes are sometimes called mutations or variants and can have a big, small or no impact on the development of the body.

They can be advantageous or neutral (non-pathogenic), such as differences in eye color.

Or they can cause needed products to not form and/or function properly. These changes can cause health challenges.

# GENETIC CHANGE: COOKING

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



DNA =

Letters used to write out a recipe

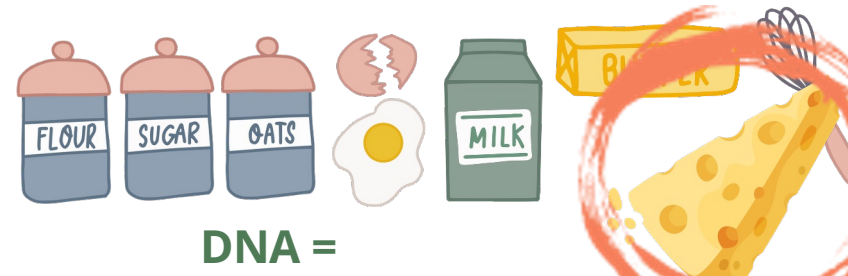
Chocolate



Gene = Recipe



Product =  
Chocolate  
Cake



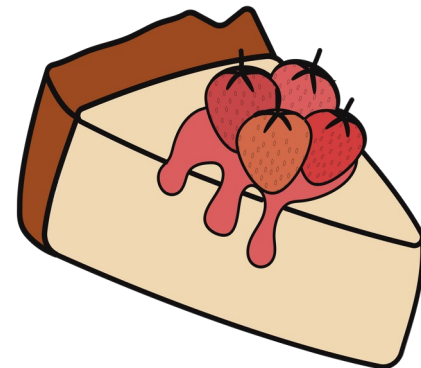
DNA =

Letters used to write out a recipe

Cheese



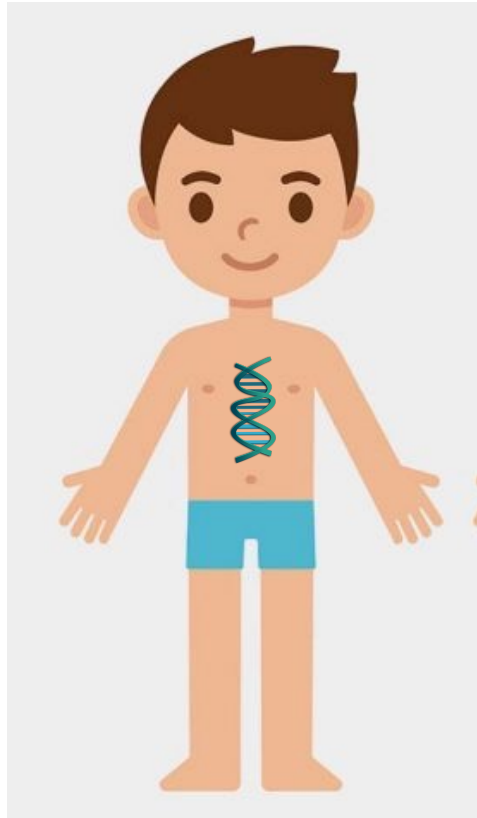
Gene = Recipe



Product =  
Cheese  
Cake

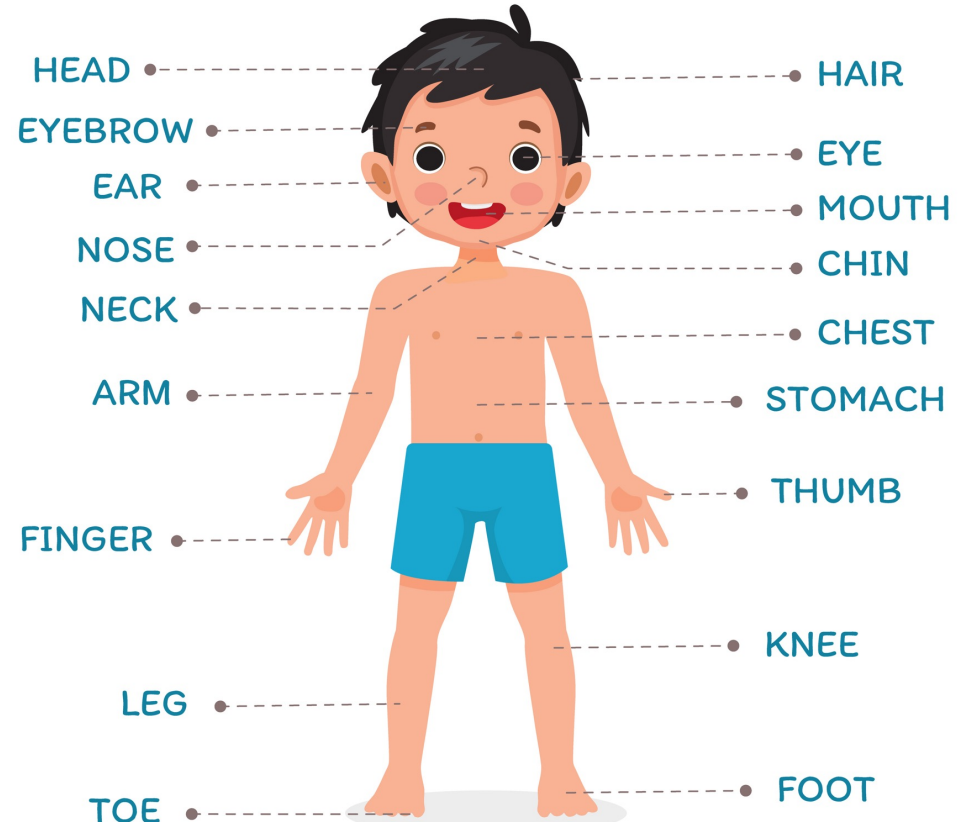


# GENETIC CHANGES



Genetic change

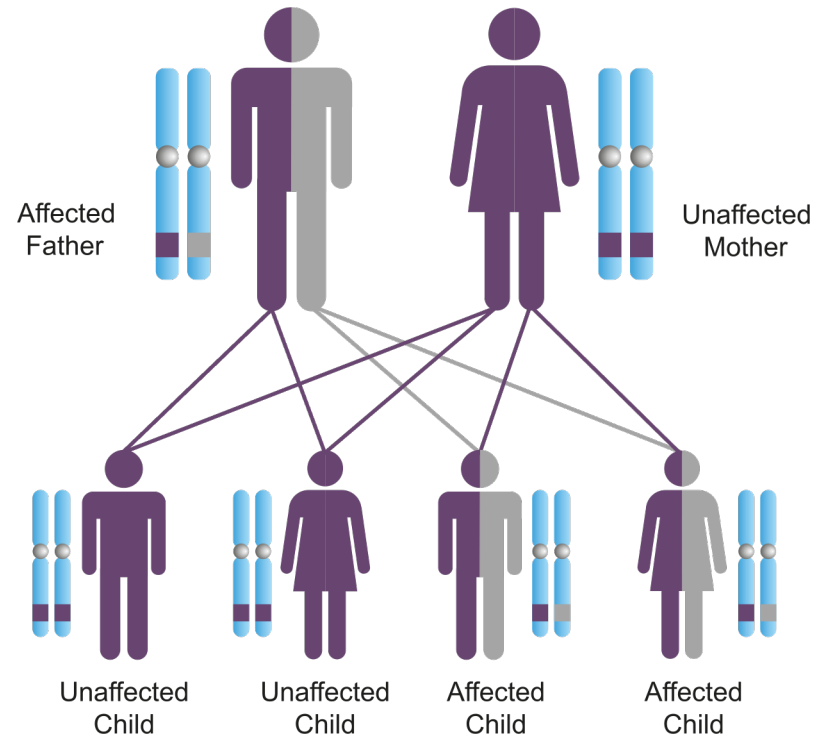
Causes a  
change in a  
protein  
→  
(Amount,  
Shape, Format)



Protein change can impact the body

# How can genetic changes run in families?

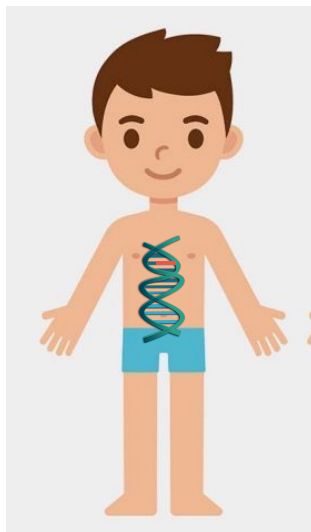
Different genetic conditions have different forms of inheritance:



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



# GENETIC CHANGES

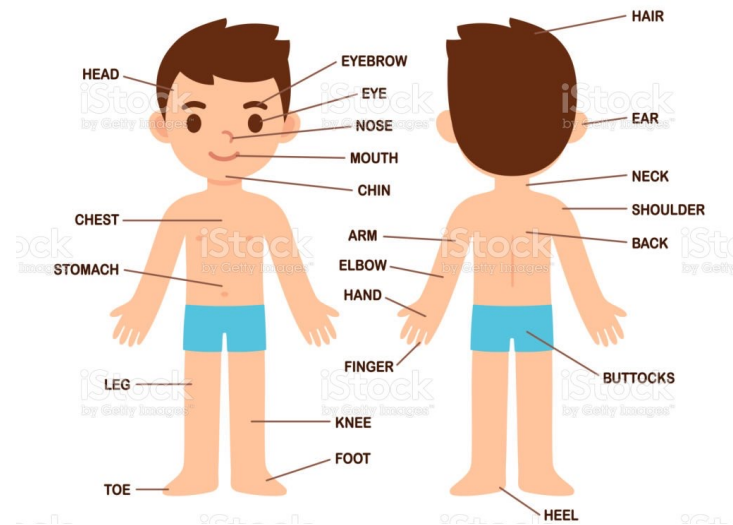


Genetic change

Causes a change in a protein



(Amount, Shape, Format)



Protein change can impact the body



Community Health Worker

**Signs a  
Genetic  
Condition  
may be  
present**





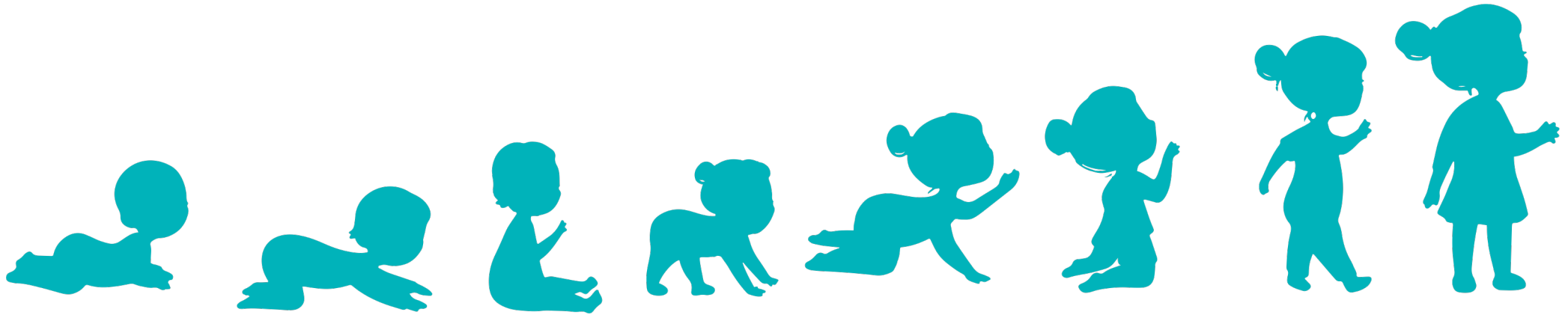
**Signs, Symptoms and Clues that a family member could have a genetic condition**



# During the Prenatal/ Newborn Period

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- There is an unusual finding during an ultrasound
- Screening bloodwork comes back out of the expected range
- Either partner has a child or family member with a medical issue that is known or suspected to be genetic
- Either partner has been told they have a higher chance to have a child with a health issue
- A newborn has a newborn screening blood test, hearing test, or cardiac test with results outside of the expected range
- A newborn baby has unexplained medical challenges after birth
- Either partner believes their family's medical issues could have a genetic cause



# During early childhood

**If you have concerns about a child's development or**

- Speech
- Learning or development
- Movement
- Growth
- Physical features
- Digestion or feeding issues
- Behavior
- Vision
- Hearing
- Heart defects
- Complicated medical issues

# What are some 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**



# What are some examples of **RED FLAGS** for **SPEECH**?

Delayed or Hard  
to Understand  
Speech

Not babbling

Not responding  
to name

**Regression**

Loss of any  
milestone

# What are some examples of RED FLAGS for BEHAVIOR?

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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

# What are some examples of RED FLAGS for DIGESTIVE/FEEDING ISSUES?

Difficulty swallowing

Extreme struggles with latching and breastfeeding

Feeding tube

Constantly hungry

Poor appetite and/or extreme pickiness

Persistent Vomiting

Gagging on food

**Regression**  
Loss of any milestone

# What are some examples of RED FLAGS for GROWTH?

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Slow growth

Slow weight gain

Uneven growth

Growing too fast/too tall

Not thriving

Fatigues quickly when active

“Growing pains” and muscle aches

# What are some examples of RED FLAGS for PHYSICAL FEATURES?

Birth defects

Extra fingers or toes

Head Size that's unusually large or small for age (macrocephaly/microcephaly)

Unusually short or tall height for age (stature)

Cleft lip and/or palate

Facial feature differences (small chin, wide forehead)

Frequent joint dislocations (hip dysplasia)

Spinal and skeletal differences

# What are some examples of COMPLICATED MEDICAL ISSUES that could be RED FLAGS?

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Regression/Loss of skills

Symptoms with no known cause

Children with multiple symptoms

Seizures

Neurological concerns

Pulmonary/cardiac concerns

Bruising

Immune issues

# During Adulthood

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- Cancer at a young age or unusual cancer
- Heart Disease at a young age
- Sudden Cardiac Death
- Certain neurological conditions
- Some Kidney diseases
- Seizure Conditions
- Early onset vision or hearing loss



# Clues from Family History

If multiple members of the family have....

Unusual Cancer History

Heart Disease

Sudden Cardiac Death

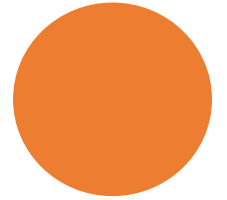
Pregnancy Loss

Birth Defects

Known Genetic Conditions

Anemias

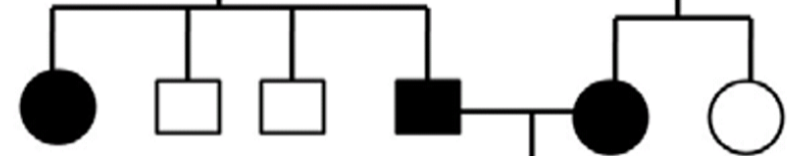
Certain Neurological Conditions



First generation



Second generation



Third generation





# Other Red Flags from the Medical History:

## 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

# Red Flags Rules



EVERY PERSON WITH A  
GENETIC CONDITION  
HAS **RED FLAGS**...

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



# Benefits of Genetics Services



The background is a solid teal color. It features several decorative elements: a cluster of white dots in the top-left corner, a larger, irregularly shaped area of white dots in the top-center, a solid teal shape on the right side, a solid teal shape on the left side, and a cluster of white dots in the bottom-left corner.

# One Family's Story



# CHW Core Competencies

## 1. Effective Communication

Plain language communication that is blame free and understandable

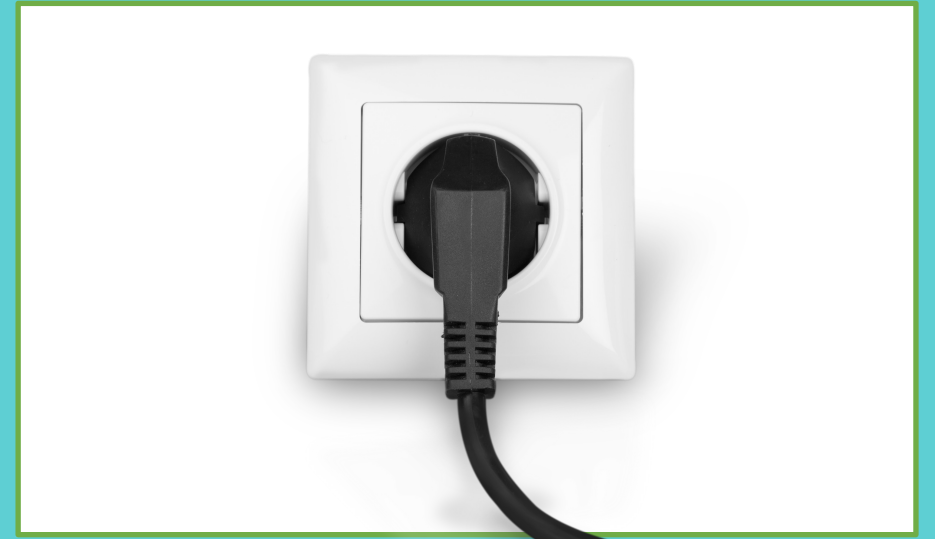
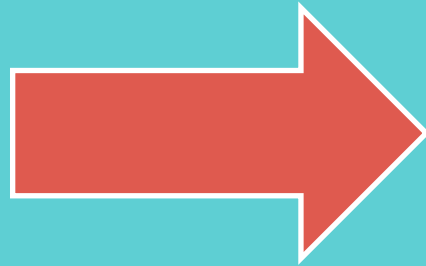
## 2. Education to Promote Healthy Behaviors

Medical screening, testing, potential dietary changes and treatment options

## 3. Care Coordination & Systems Navigation

Understanding the value of and locating a medical home

# Cindy's Role as a Community Health Worker



*How could working with a CHW have impacted  
Cindy's story?*

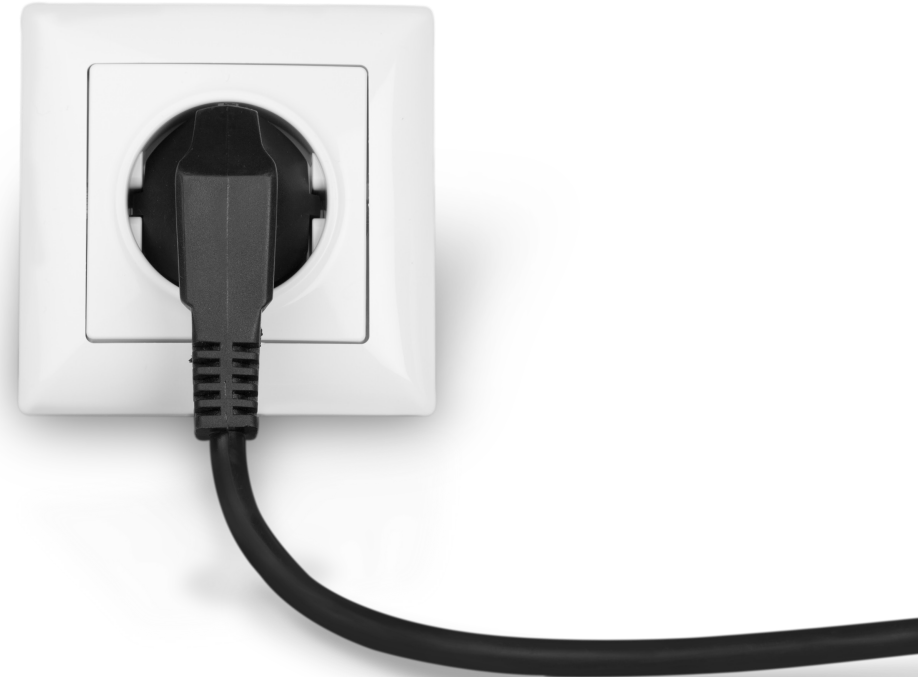
**How  
CHWs can  
help NJ  
families  
access  
Genetics  
Services**





You can be a

Detector & Connector



# Community Health Workers could....



Educate pregnant women how genetics can impact them and their baby



Help families understand the importance of newborn screening



Identify children whose developmental issues could be caused by genetic changes



Recognize patterns of cancer in families that could be caused by genetics

# Community Health Workers can increase community **AWARENESS** of and **ACCESS** to genetic services



Help families understand the life-long importance of genetics

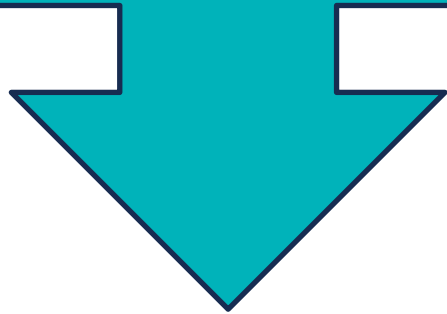


Reduce the guilt, shame, secrecy and silence about genetic conditions in the family



Help families overcome barriers to obtaining a genetics appointment

What should you do if you believe a family might benefit from a genetics referral?



Use the  
**Medical Home  
Model of Care**



# CHWs can help EMPOWER the family to talk to their PCP

💡 "You are the expert on your family"

💡 Teach the family tools to advocate.

💡 The provider may have the scientific knowledge but the family has the personal history and day to day experiences



# How CHWs can start a conversation about genetics

“I notice you/your child has seen several specialists. Has anyone ever talked to you about a genetics referral?”

“Have you ever told your doctors that there are several people in your family with the same symptoms as you?”

# What tips can CHWs give a family about talking with their PCP about genetics concerns?

Prioritize top 3 concerns

Bring data about your concerns

Bringing a trusted person with you to the appointment

Schedule a follow-up appointment if there is not time to address all of your concerns.

Speak in your native language & ask for an interpreter to help you explain your concerns.

# How families can start discussions about genetics with their PCP...

I am concerned about \_\_\_\_\_

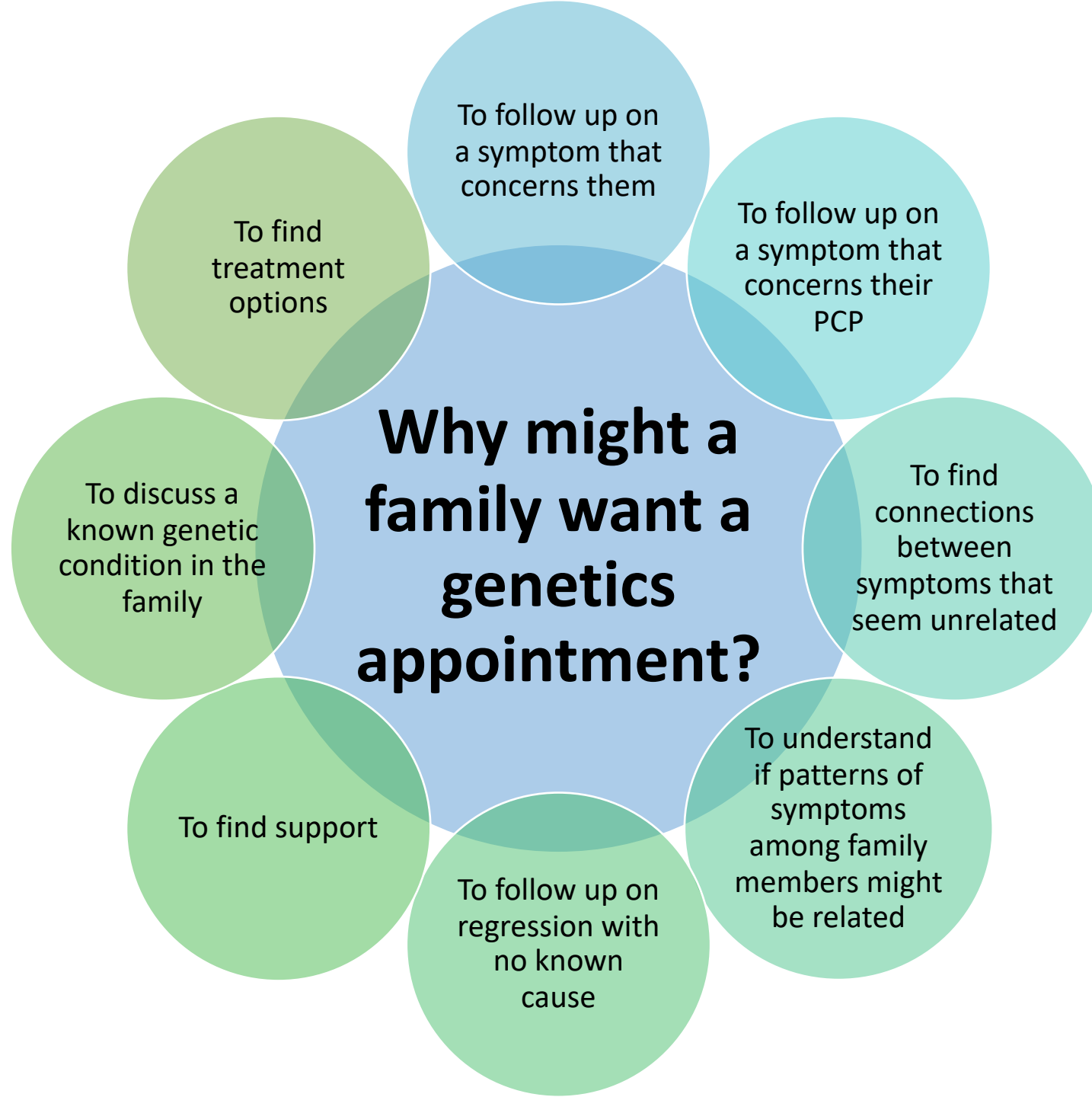
My concern of \_\_\_\_\_ from our last visit has not resolved (worsened), what can be done to explore further?

In talking with my family, I learned we have a history of \_\_\_\_\_, is it possible my concern of \_\_\_\_\_ is genetic?





Why might a family want a genetics appointment?



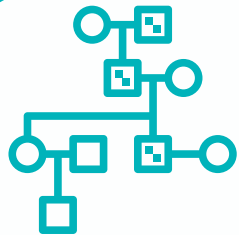
# How can a genetic specialist help a family?



Look for a cause for the concerns you are observing in your patient



Explain what genetic testing options could help make/confirm a diagnosis



Review the family history for any clues that suggest a possible genetic condition



Discuss potential treatments or therapies



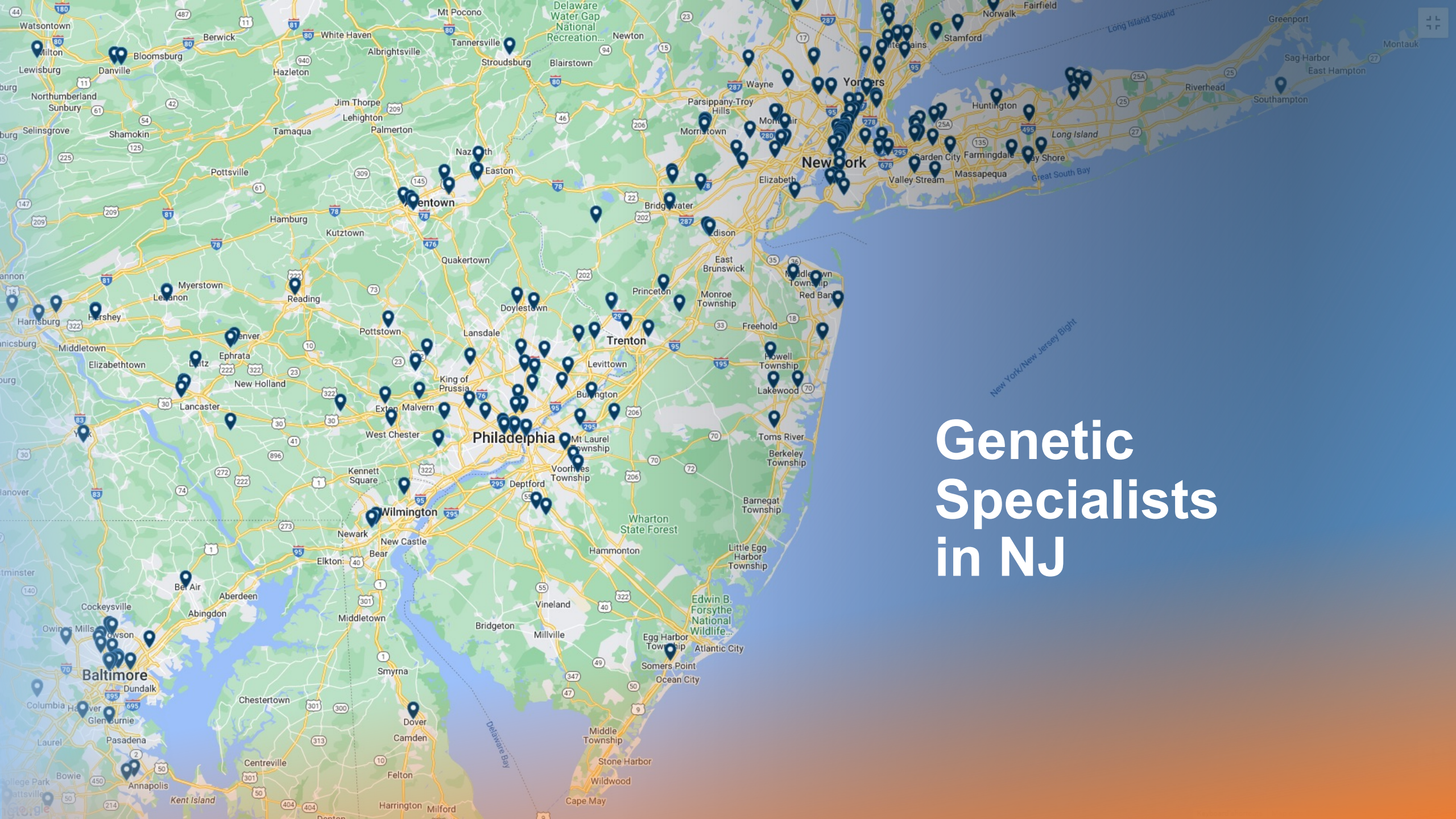
Provide recommendations to other helpful doctors or professionals



Offer connections to other families, support groups, and family organizations

# Frequently Asked Questions





# Genetic Specialists in NJ



**Christina Kresge**

 Certified Genetic Counselor

 Rutgers New Jersey Medical School




**Karen Valdez-Gonzalez**

 Certified Genetic Counselor

 Hackensack University Medical Center



**Christina Botti**

 Certified Genetic Counselor

 Rutgers-Robert Wood Johnson Medical School

# *What happens at a genetics appointment?*



- The patient will often meet with a genetic counselor first and asked detailed questions about:
  - their family history
  - their or their child's developmental and/or medical history
- Sometimes, a geneticist (a doctor) will join the session to do a physical exam and provide an assessment
  - This may include some measurements and possibly photos for documentation
- If appropriate, the patient will be offered genetic testing

***\*\*Every session will be slightly different based on the reason for referral***

# *Does a patient have to do genetic testing?*

- The genetic specialist will
  - offer genetic tests based on the patient's personal, medical and family history
  - discuss the benefits and risks of test
  - explain how the results may affect the patient and their family

***Genetic specialists work with the patient to help them make the best decision for themselves and their family***







Information  
privacy

*Will a patient's  
genetic  
information  
remain private?*

- There are some protections in place, such as the Genetic Information Nondiscrimination Act of 2008 (GINA)
  - Prohibits discrimination on the basis of genetic information with respect to health insurance and employment
- A genetic specialist can help a patient understand the benefits and risks involved with genetic testing

# *What other costs are involved with a genetics appointment?*

- Most insurance plans cover genetics appointments \*
- Medicaid has minimal out of pocket costs
- Medicare covers an evaluation by a geneticist but may not cover an appointment with a genetic counselor
- Other expenses may include
  - Travel costs
  - Missing work

*\* Telehealth may be an option depending on the reason for the appointment and type of insurance plan*





## *How much does it cost to do genetic testing?*

- The cost depends on the patient's:
  - insurance plan
  - type of coverage
  - type of testing
- The genetics team will help find the least costly options
- Some patients may qualify for financial assistance programs

# Resources for Genetics in New Jersey

Home / Resources for Genetics in New Jersey

Approximately 17.4% of children in New Jersey have one or more special health care needs. Sometimes these health challenges can be caused by changes in our genes. Genetics providers help families learn whether genetic changes may explain the health conditions in their family and connect families to appropriate care.



To address the unique needs of families across New Jersey, a diverse group of stakeholders met over the course of a year to share their perspectives and experiences about the barriers families face when trying to access genetic services in the state. The team includes both genetic and non-genetic healthcare professionals, families and public health representatives. Together they identified the key obstacles that are preventing local families from obtaining needed genetic services. Currently, the team is brainstorming ideas on how to address one of these pressing issues.

Visit this webpage for updates and details about their upcoming project and efforts to improve access to genetic services in New Jersey.

## Resources for Children and Families with Genetic Conditions in NJ:

- + [State Resources for Children and Families with Genetic Conditions in New Jersey](#)
- + [Support groups for parents](#)
- + [Specific resources for common health conditions](#)

Meet the NYMAC NJ Team

[LEARN MORE](#)

Genetics Clinics in NJ

[FIND A GENETICS CLINIC](#)

For further information about genetics and local resources, please visit our NYMAC Team New Jersey webpage at:

<https://nymacgenetics.org/new-jersey/>

FIND A GENETICS CLINIC

# For More Information and Resources...

- CDC's Learn the Signs, Act Early info:  
<https://www.cdc.gov/ncbddd/actearly/about.html>
- NYMAC website: <https://nymacgenetics.org/>
- Pediatric Red Flags for Genetics: <https://nymacgenetics.org/providers/when-to-refer-patient-to-genetics/pediatric-genetics-referrals/>
- Genetic Education Materials for School Success (GEMSS):  
<https://www.negenetics.org/genetic-education-materials-school-success-gemss>
- [NHGRI's Online Genetics Education Resources](https://www.genome.gov/10000464/):  
<https://www.genome.gov/10000464/>: The National Human Genome Research Institute has compiled a list of online genetics educational materials.
- [Genetics Home Reference: Your Guide to Understanding Genetic Conditions](https://ghr.nlm.nih.gov/) :  
<https://ghr.nlm.nih.gov/> Genetics Home Reference, a service of the National Library of Medicine, provides consumer-friendly information about the effects of genetic variations on human health. It includes sections on Genetic Conditions, Newborn Screening, Genes, Chromosomes, a Handbook, Glossary and Resources.



**Questions?**