AUTISM CONNECT 2026

April 7th

Thank You!

SCSM
SOUTH CAROLINA STATE MUSEUM

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ABS Kids
Reaching Milestones. Realizing Potential.

ABLE
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The Nuts and Bolts of a Genetic Evaluation for Autism Spectrum Disorder

Wesley G. Patterson, PhD, MSPA, PA-C, CAQ-Peds

Genetics Physician Assistant/Associate

AutismConnect 2026

April 7, 2026

Disclosures

- I have no financial disclosure

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Learning Objectives

OBJECTIVE #1

- Describe the services provided by the Greenwood Genetic Center

OBJECTIVE #2

- Review the recommendations, purpose, and methods of a genetics evaluation for autism spectrum disorder

OBJECTIVE #3

- Describe common genetic testing options and possible outcomes and implications of genetic testing

Objective #1

Greenwood Genetic Center: Who we are and what we do

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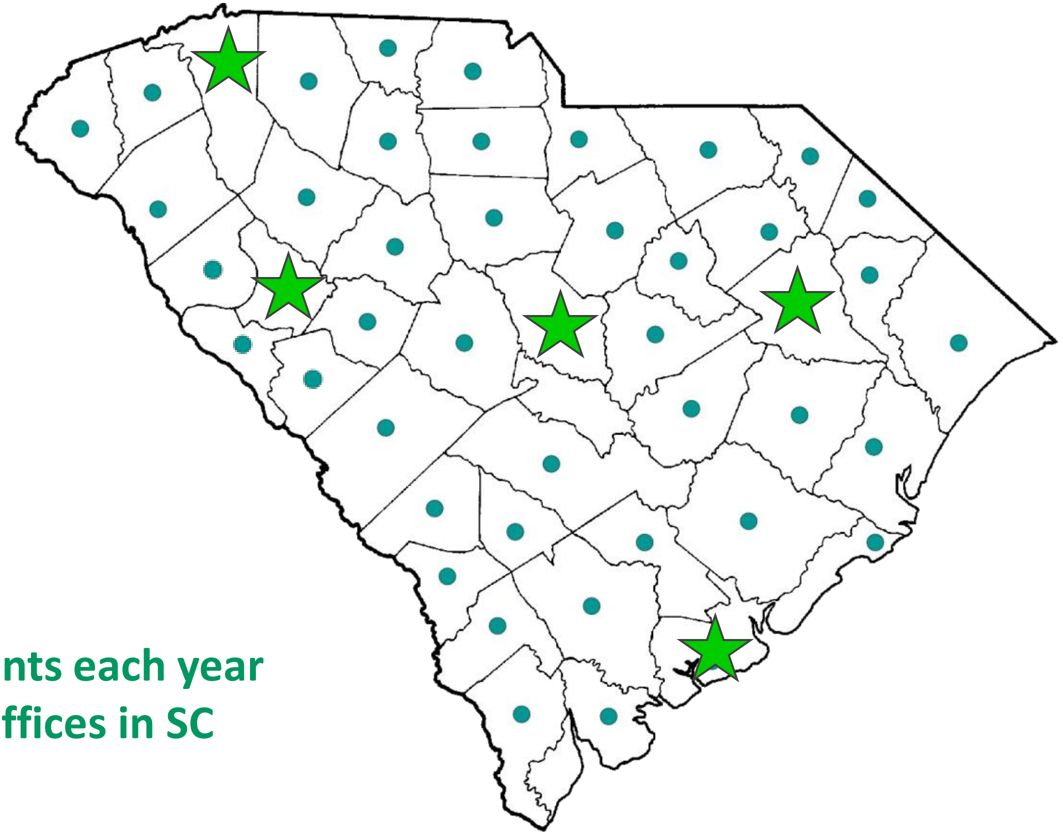
Greenwood Genetic Center

About GGC

- Founded in 1974 through support from the SC Department of Disability and Special Needs (DDSN) and the Self Family Foundation
- Private, 501c3 nonprofit medical center
- Main campus: Greenwood
    - Greenville
    - Columbia
    - Charleston
    - Florence
- MUSC Affiliation

GGC State-Wide Reach



5,300+ patients each year
across 5 offices in SC

Mission and Vision

- Mission
  - GGC is a nonprofit institute organized to provide **clinical genetic services**, **diagnostic laboratory testing**, **educational programs and resources**, and **research** in the field of medical genetics.
- Vision
  - GGC will be a Center of Excellence in Medical Genetics, serving as a resource for **all persons who need genetic services** and working to reduce the prevalence and impact of genetic disorders.

Fabulous Folate
Oh Baby, It's Good for YOU!

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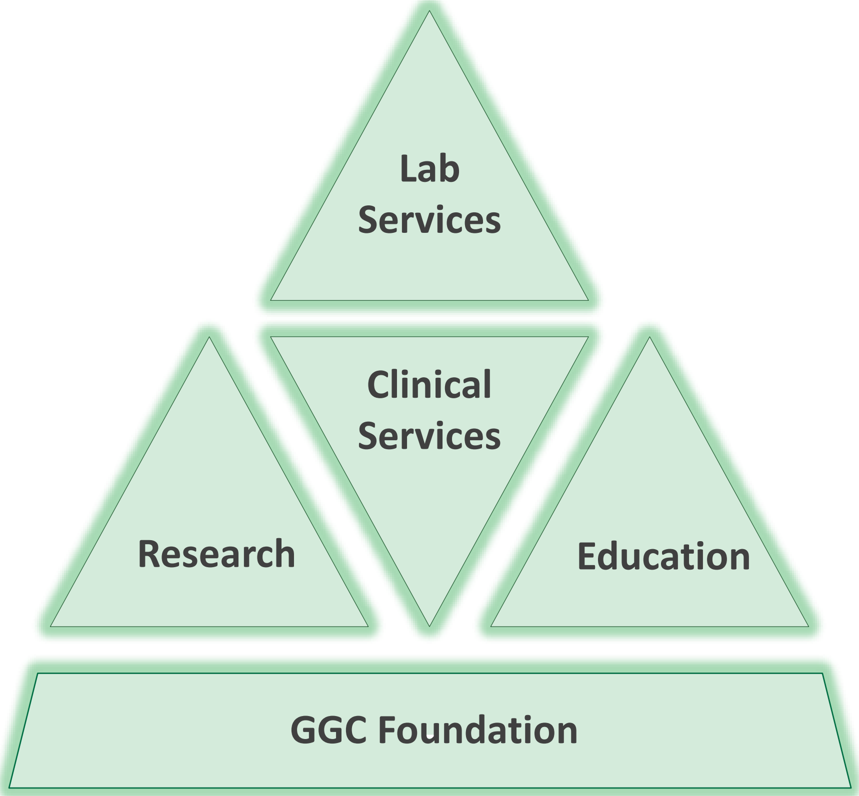
GGC Divisions

- **Clinic**: sees patients
- **Laboratories**: perform testing
- **Research**: conducts experiments to solve problems and give answers
- **Education**: training and outreach
- **Foundation**: fundraising, philanthropy



Also: Administration, Human Resources, Information Technology

Reasons to see Genetics

We accept referrals for many reasons:

- Developmental delay
- Intellectual disability
- Autism spectrum disorder
- Seizures/epilepsy
- Multiple congenital anomalies
- Hearing loss
- Vision loss and genetic eye disorders
- Growth issues
- Abnormal prenatal screening results
- Metabolic disorders
- Personal or family history of cancer
- Genetic cardiac conditions (cardiomyopathy, arrhythmia)
- Suspected or known genetic condition

GGC and Autism

- GGC has a long-standing interest in identifying the underlying genetic causes and contributing factors related to an individual developing autism
  - Many medical journal publications related to gene discoveries
  - Collaborations in SC, nationally, and internationally
- We perform genetic evaluations for children and adults who have a confirmed or suspected diagnosis of autism
- We do not:
  - Diagnose or un-diagnose autism through our evaluations → **there is no genetic test that can diagnose autism**
  - Provide treatment (e.g., therapies, prescribe medication)

Race the Helix

Objective #2

The recommendations, purpose, and methods of a genetics evaluation for autism spectrum disorder

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Genetics Evaluation – Autism

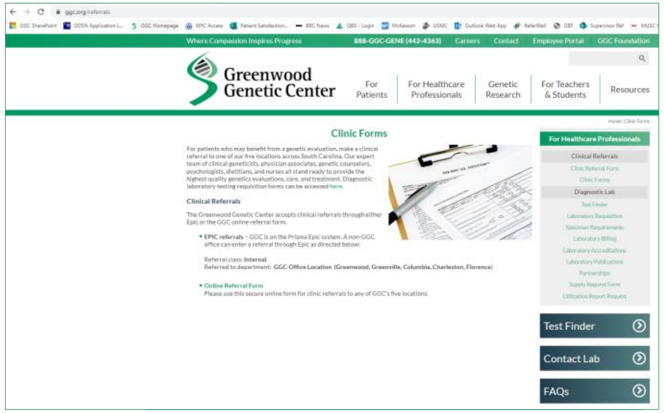
- There are genetic and environmental factors that play a role in an individual developing autism
- A genetics evaluation and testing is recommended and supported by multiple medical organizations, including the American College of Medical Genetics and Genomics and the American Academy of Pediatrics
- Autism has “high heritability” → high level of genetic contributions related to the risk for developing autism
  - Twin studies: If one identical twin has autism, there is a very high probability that the other twin will also have autism
  - If one sibling has autism, there is a higher chance for that child’s sibling to have autism compared to a child in the general population

Genetics Evaluation – Autism

What is the purpose of the evaluation?

- Many genetic conditions include autism as a feature
    - Confirm or rule-out a syndromic cause of autism
- A diagnosis of a specific condition can provide an explanation/answer for the patient and family
- May change future medical management and open opportunities for treatment
- Allows the family to access condition-specific resources and advocacy groups
- May clarify the current and future recurrence risk for the patient and family (*i.e.*, the chance for another family member to have the same condition)

The GGC Evaluation Process



Clinic Forms

For patients who may benefit from a genetic evaluation, make a clinical referral to one of our five locations across South Carolina. Our expert team of clinical geneticists, physician associates, genetic counselors, psychologists, dietitians, and nurses all stand ready to provide the highest quality genetics evaluations, care, and treatment. Diagnostic laboratory testing requisition forms can be accessed here.

Clinical Referrals

The Greenwood Genetic Center accepts clinical referrals through either Epic or the GGC online referral form.

- **EPIC referrals** – GGC is on the Prisma Epic system. A non-GGC office can enter a referral through Epic as directed below:

  Referral class: **Internal**
  Referred to department: **GGC-Office Location (Greenwood, Greenville, Columbia, Charleston, Florence)**

- Online Referral Form
  Please use this secure online form for clinic referrals to any of GGC's five locations.

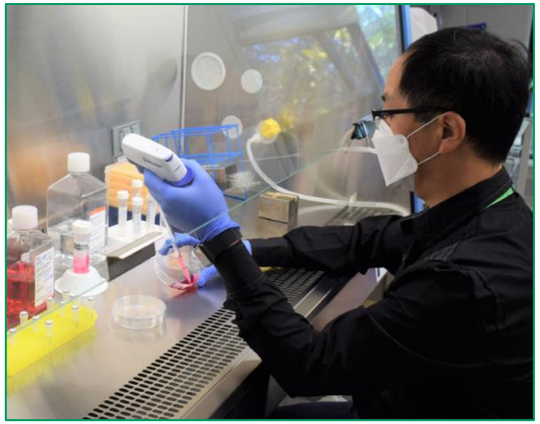
Referral & Scheduling

Receive a referral from BabyNet, DDSN/OIDD, pediatrician, or other specialty medical provider

***NEW – self referral for a know diagnosis of autism**

Evaluation

The patient is seen through one of our clinics – eVisit, virtual visit (telemedicine), or in-person



Testing

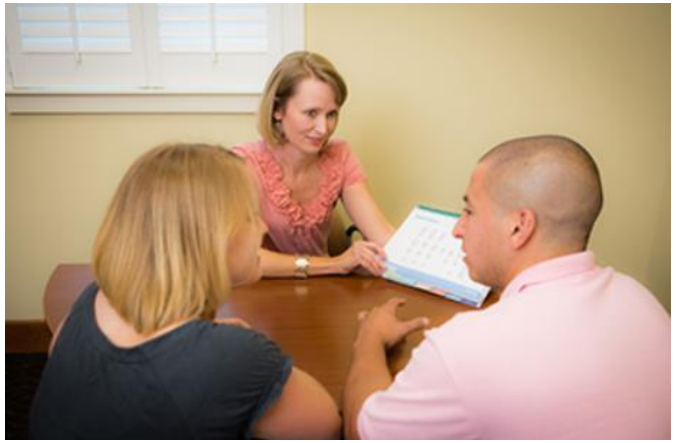
Genetic testing is performed if the family is interested and provides informed consent

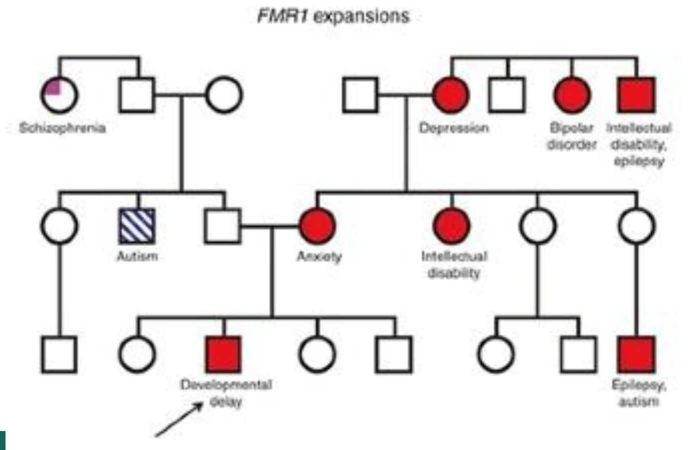
Follow-up

The genetics team communicates results to the family and helps to coordinate management recommendations and provides and identifies support

What does the evaluation entail?

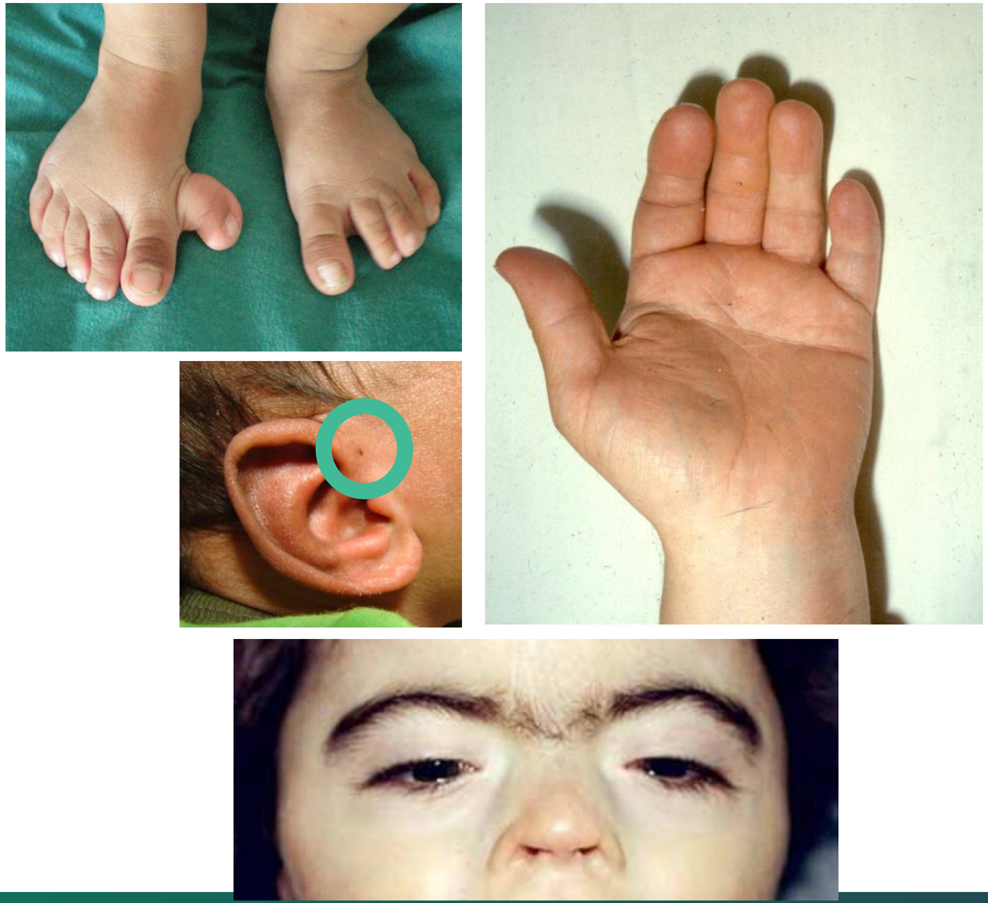
- Contracting: greeting the patient and companions, describing the flow of the appointment, answer questions
- Obtain measurements (height, weight, head circumference, sometimes others like ear size or arm span)
- Genetic counselor will take prenatal history, medical history, review of systems, developmental history, and family medical history
    - Family history: identify other relatives with autism and other neurodevelopmental diagnoses
- Physical exam with a doctor, physician associate, or nurse practitioner (*sometimes challenging for individuals with autism)
- Discuss genetic testing process: benefits, limitations, risks, possible outcomes of testing. Sample collection. Insurance and billing.
- Review the follow-up plan for results disclosure and a follow-up visit in the Genetics clinic (usually between 1-5 years)





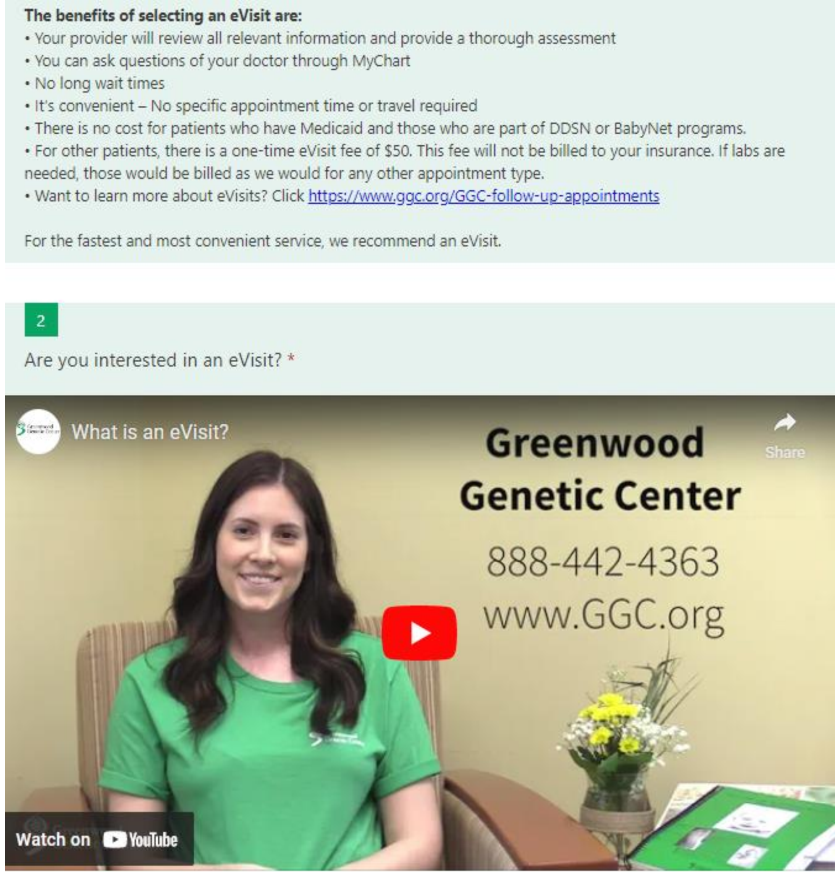
Physical Exam

- Our doctors, PAs, and NP are trained in a specific type of medical practice called “dysmorphology”
    - Know the “typical” way all the structures of the body are formed and be able to note and describe differences
        - Ear shape and placement, facial structure, birthmarks, and much more
    - There are guidelines about how to describe the way body structures look if they are different than “typical”
    - Consider: familial features, ancestry/ethnicity
    - We explain that there is nothing inherently wrong with having dysmorphic features; they can be clues to help us identify a specific genetic condition
- We can refer to pictures, if exam is challenging



Types of Appointments

- Traditional appointment
  - In-person visit in one of our offices
- Virtual visit via telemedicine
  - Patient/family, genetic counselor, and MD/PA/NP are all logged into the same video appointment together. Physical exam is virtual.



The benefits of selecting an eVisit are:

- Your provider will review all relevant information and provide a thorough assessment
- You can ask questions of your doctor through MyChart
- No long wait times
- It's convenient – No specific appointment time or travel required
- There is no cost for patients who have Medicaid and those who are part of DDSN or BabyNet programs.
- For other patients, there is a one-time eVisit fee of $50. This fee will not be billed to your insurance. If labs are needed, those would be billed as we would for any other appointment type.
- Want to learn more about eVisits? Click https://www.ggc.org/GGC-follow-up-appointments

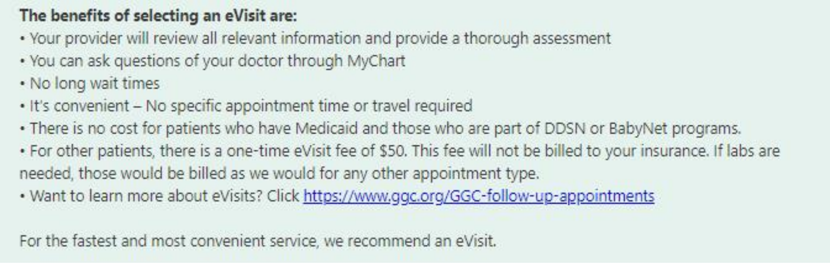
For the fastest and most convenient service, we recommend an eVisit.

2

Are you interested in an eVisit? *

Types of Appointments

- eVisits (BabyNet referrals)
  - Newer type of visit to help children under 3-years-old with developmental delay, many with a concern for autism, obtain quicker access to a “asynchronous” genetic evaluation and testing
  - The family/guardian fills out a questionnaire and provides patient photos. A genetic assistant (GA) reviews available medical records. A MD/PA/NP reviews the information. If testing is recommended, the GA will coordinate insurance preauthorization and sample coordination. A genetic counselor will assist to help discuss abnormal test results or consent for further genetic testing.
  - Quickest time from referral to recommendations. Goals are to increase accessibility, flexibility, convenience, efficiency.



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2

Are you interested in an eVisit? *

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Types of Appointments

- eVisits (confirmed diagnosis of autism)
  - **Newer type of visit to help individuals with autism obtain quicker access to a “asynchronous” genetic evaluation and testing**
  - The family/guardian fills out a questionnaire and provides patient photos. A genetic assistant (GA) reviews available medical records. A MD/PA/NP reviews the information. If testing is recommended, the GA will coordinate insurance preauthorization and sample coordination. A genetic counselor will assist to help discuss abnormal test results or consent for further genetic testing.
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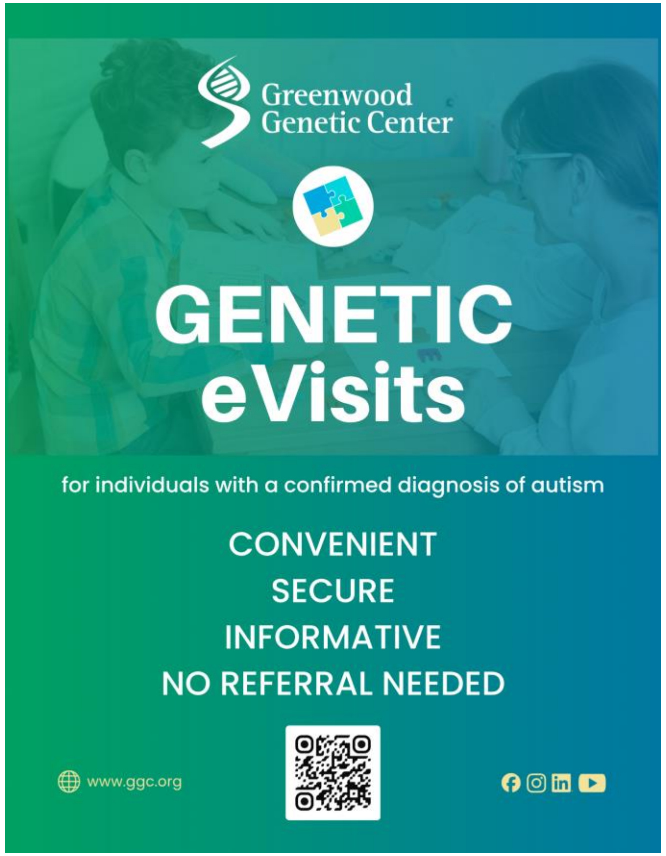

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GENETIC eVisits

for individuals with a confirmed diagnosis of autism

CONVENIENT
SECURE
INFORMATIVE
NO REFERRAL NEEDED

www.ggc.org

Objective #3

Common genetic testing options and the possible outcomes and implications of genetic testing

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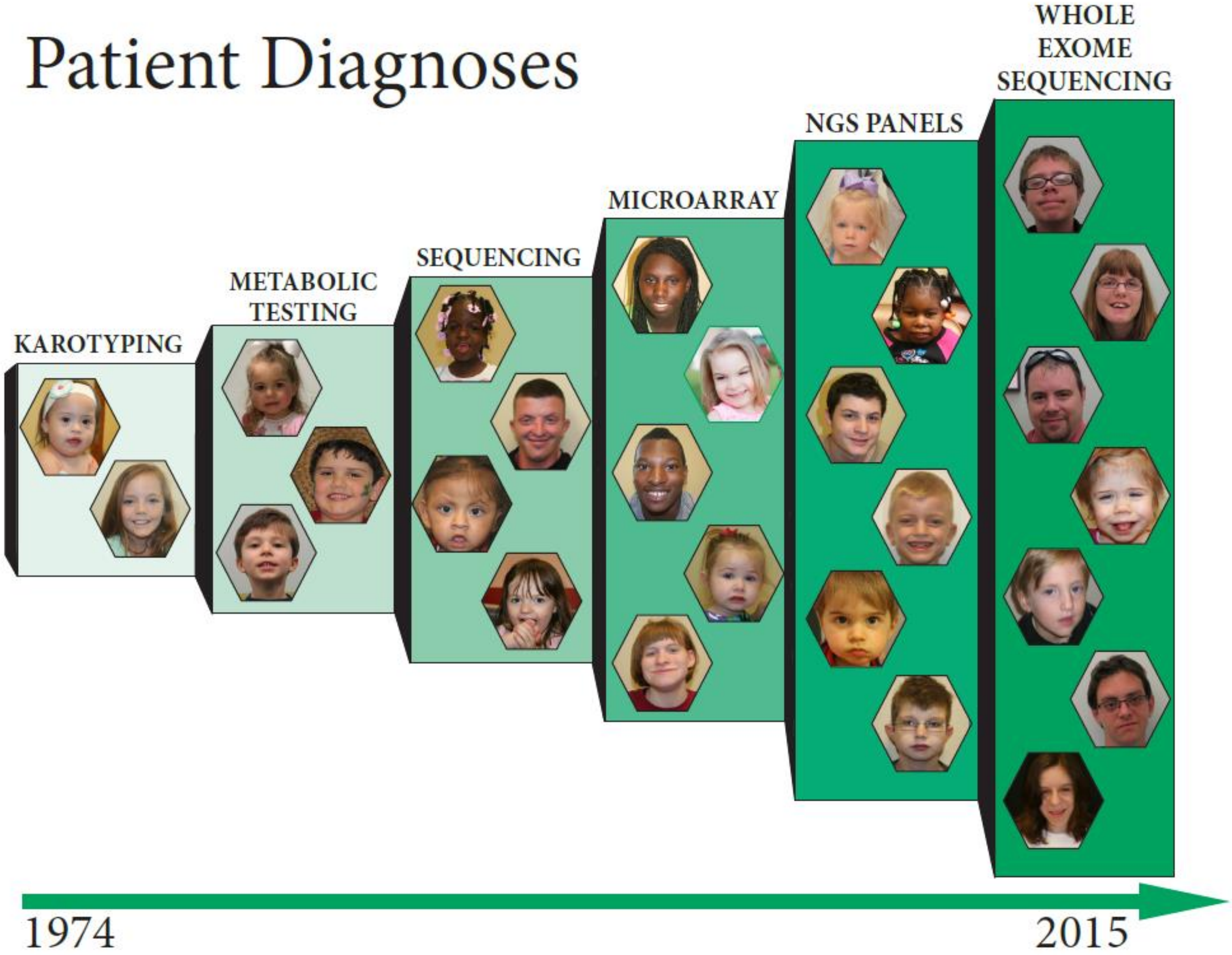

Diagnostic Rate for Autism

Microarray = 10-15%

Whole exome sequencing = 30% for all patients

It is more difficult to find a specific genetic cause of autism for individuals who have a normal physical exam, normal IQ, and no/few medical issues

Genetic Testing for Autism

- When genetic testing is offered, it is **always optional and can be declined**
- **Genetic testing cannot diagnose autism!**
- If genetic testing is normal/negative, it does not mean:
  1) the patient does not have autism
  2) there are not genetic factors that contributed to the diagnosis of autism
  3) that relatives are not at-risk to have or be diagnosed with autism
- We discuss the benefits, risks, limitations, and possible outcomes of genetic testing with the patient and family/guardian
- Sample types: saliva swab, blood, rarely other types (e.g., skin biopsy)
- We obtain insurance approval. We are in-network with many versions of SC Medicaid and insurances to get reimbursed for testing. We do not "balance bill" patients who receive BabyNet/DDSN/OIDD services. The GGC Cares Fund provides financial assistance to families who need it.

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Genetic Testing for Autism

Common Tests:

- Fragile X syndrome (FXS) testing
- Whole genome chromosomal microarray
- Rett syndrome testing
- NGS syndromic autism panel (83 genes)
- Whole exome sequencing (WES)
- Whole genome sequencing (WGS)

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Fragile X and Rett syndromes

FRAGILE X SYNDROME

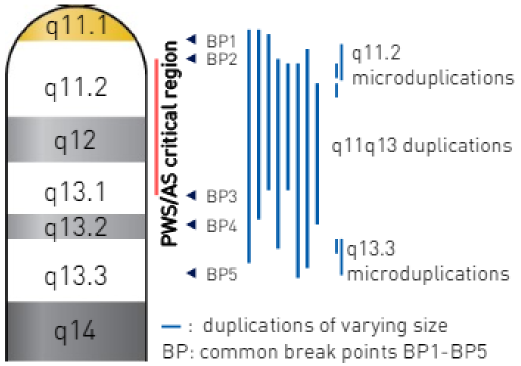
- Most common inherited/familial cause of intellectual disability and/or autism
- *FMR1* gene: over 200 repeats of the CGG DNA pattern in a part of gene that will shut the *FMR1* gene off, or a pathogenic (harmful) sequence variant
- Symptoms are more common and apparent in males than females
- Features: delay, intellectual disability, behavior and attention issues, seizures, cardiac issues, scoliosis, characteristic facial features
- Implications for *FMR1* carriers

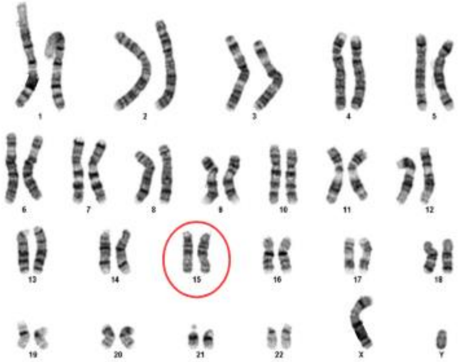
CLASSIC RETT SYNDROME

- Usually not inherited
- *MECP2* gene: pathogenic variant causes the condition
- Many more females than males are diagnosed; males can pass in infancy
- Features: Normal development, then regression by 3-years-old; autism, slow growth and small head size (microcephaly), seizures, scoliosis, breathing issues, sleep problems

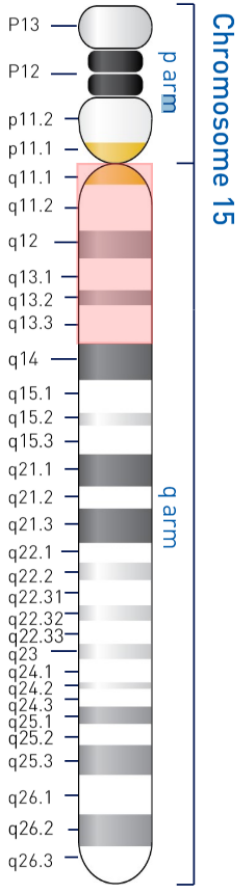
Chromosomal Microarray (CMA)

- Chromosome – Package of DNA that includes genes and proteins. Humans typically have 23 pairs of chromosomes, for a total of 46. We get half of our chromosomes from each biological parent. Numbered from 1-22. X and Y are the sex chromosomes.
- CMA is a test that looks for losses (deletions) and gains (duplications, triplications, etc.) of the chromosomes
- There are chromosome conditions that can predispose an individual to have autism
    - 15q11q13 duplication syndrome: developmental delay, intellectual disability, autism, hypotonia (low muscle tone), seizures, hearing loss, vision/eye issues, scoliosis, obesity





Chromosomes pairs 1-22, X and Y (male)
Chromosome pair 15 is circled in red



Whole Exome/Genome Sequencing (WES/WGS)

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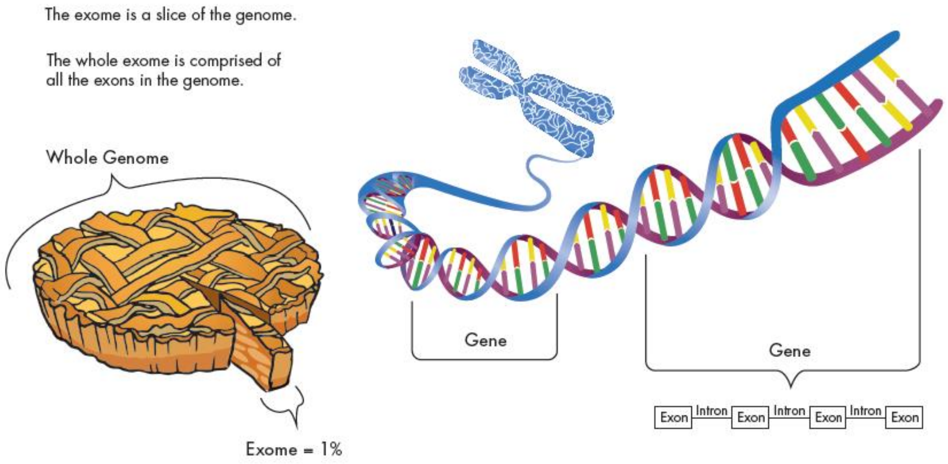
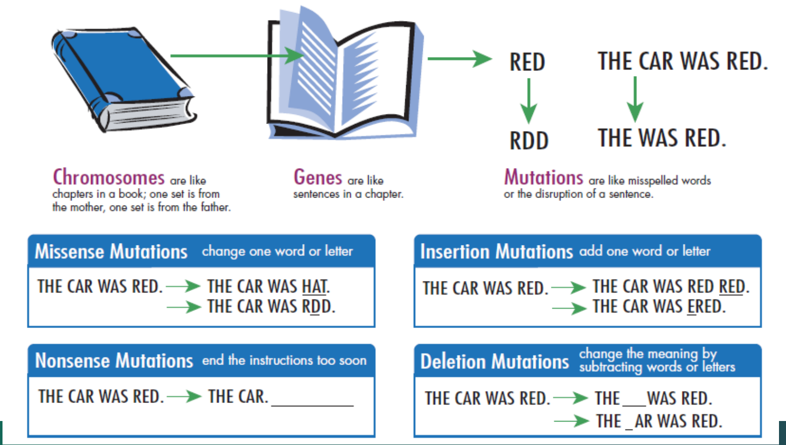
- Gene sequencing of the most important parts (the exons) for the 20,000+ known human genes
  - Gene sequencing reads through the letters of DNA (A, C, T, G) within the gene to make sure it is in the right order → functional gene
  - Exome is 1-2% of an individual's total genetic information
  - Multiple types of variants can be identified
- WGS can also pick up intronic variants, copy number variants (gains and losses), and mitochondrial DNA variants
- Patients and families have the option to opt-in to receiving secondary findings and have GeneMatcher findings reported
- Best if the testing is run as a trio – with both biological parents
- Test turn-around-time = 8-10 weeks



The exome is a slice of the genome.

The whole exome is comprised of all the exons in the genome.



Chromosomes are like chapters in a book; one set is from the mother, one set is from the father.

Genes are like sentences in a chapter.

Mutations are like misspelled words or the disruption of a sentence.

Missense Mutations change one word or letter

THE CAR WAS RED. → THE CAR WAS HAT.
→ THE CAR WAS RDD.

Insertion Mutations add one word or letter

THE CAR WAS RED. → THE CAR WAS RED RED.
→ THE CAR WAS ERED.

Nonsense Mutations end the instructions too soon

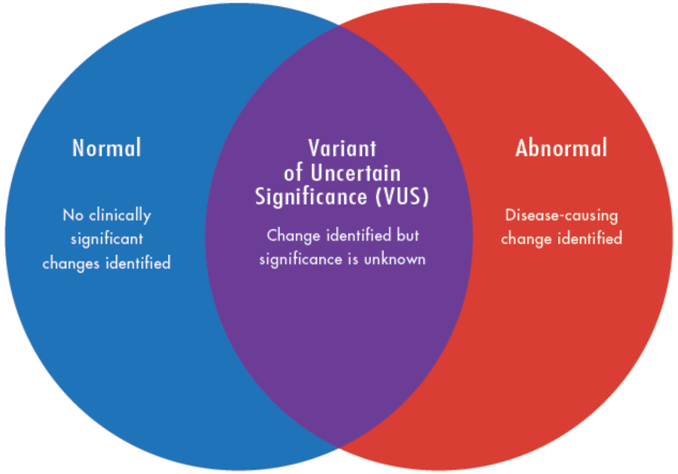
THE CAR WAS RED. → THE CAR. ________

Deletion Mutations change the meaning by subtracting words or letters

THE CAR WAS RED. → THE ___ WAS RED.
→ THE _AR WAS RED.

Results of Genetic Testing

Potential Results of Genetic Testing



Normal

No clinically significant changes identified

Variant of Uncertain Significance (VUS)

Change identified but significance is unknown

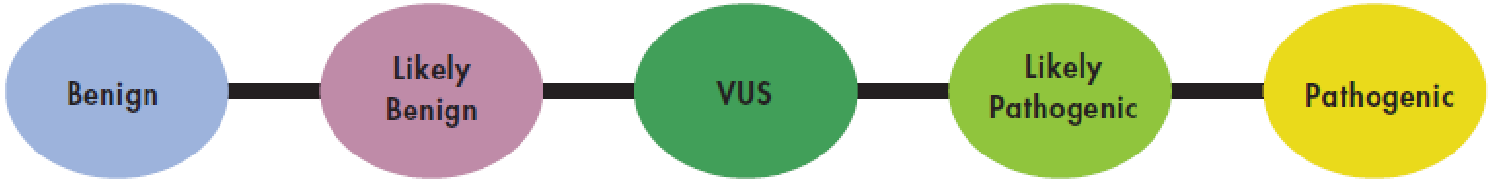
Abnormal

Disease-causing change identified

Supporting Information

- *Functional Studies*
- *Bioinformatics Predictions*
- *Segregation/Family Testing*
- *Other Clinical Data*

Variant Classification



Benign — Likely Benign — VUS — Likely Pathogenic — Pathogenic

- A variant of uncertain significance (VUS) is a change in the DNA/genetic material, that may or may not be related to the patient's concern and reason for testing.
- Testing the biological parents and other relatives for the VUS may help determine its significance for the patient.
- Future reanalysis of data may also clarify the VUS result.
- As more data is collected, a VUS may be reclassified as benign, likely benign, likely pathogenic, or pathogenic.

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Test Results Disclosure

- The plan for results disclosure (e.g., phone call, in-person, normal results letter) is discussed at the time of ordering the testing
- Results are discussed with the patient/guardian in a timely, sensitive, understandable, and inclusive manner
- We discuss the contents of the test results report; all test results have implications, even normal results
- Review the medical management recommendations, prognosis, other issues to monitor for in the future, and recurrence risk
- GC provides the family with informational materials about the genetic condition, as well as research opportunities, possible treatment/therapies, and family advocacy and connections
- Provide acute emotional support related to giving a new diagnosis or confirming a suspected one
- Families are encouraged to ask questions and reach out for help

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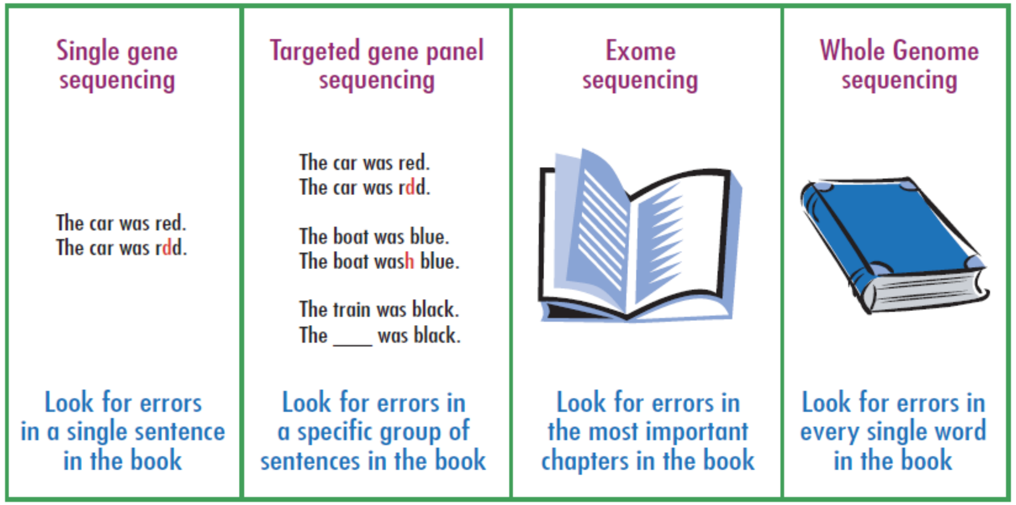
The Future of GGC Genetic Testing

- Long-read Whole genome sequencing (lrWGS) – can detect every type of variant that causes a genetic condition
  - Requires a skilled bioinformatics team to process the patient's genomic data
- GeneMatcher: We are currently able to identify variants in "genes of uncertain significance" that may be associated with autism or other neurodevelopmental differences and can help a patient/family get connected to research that may help confirm the connection between a gene and autism
- Genomics Discovery Program (GDP) – 3 tracks:
  - Discovery
  - Variant Resolution
  - Treatment



Single gene sequencing	Targeted gene panel sequencing	Exome sequencing	Whole Genome sequencing
The car was red. The car was rdd.	The car was red. The car was rdd. The boat was blue. The boat wash blue. The train was black. The ___ was black.		
Look for errors in a single sentence in the book	Look for errors in a specific group of sentences in the book	Look for errors in the most important chapters in the book	Look for errors in every single word in the book

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GGC: Giving Greater Care

GC is committed to helping patients and families, as well as caregivers, the medical community, and the public, understand the genetic factors associated with autism. We also perform genetic evaluations, help families navigate the genetic testing process, and assist with understanding and processing the results.

www.ggc.org

888-GGC-GENE (442-4363)

Thank you!

Wesley Patterson, PA-C

wpatterson@ggc.org

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eVisits offer a convenient, secure way to connect with a genetics provider to explore possible genetic causes of autism.

LEARN MORE

Session Survey

Senate A 1:30-2:30