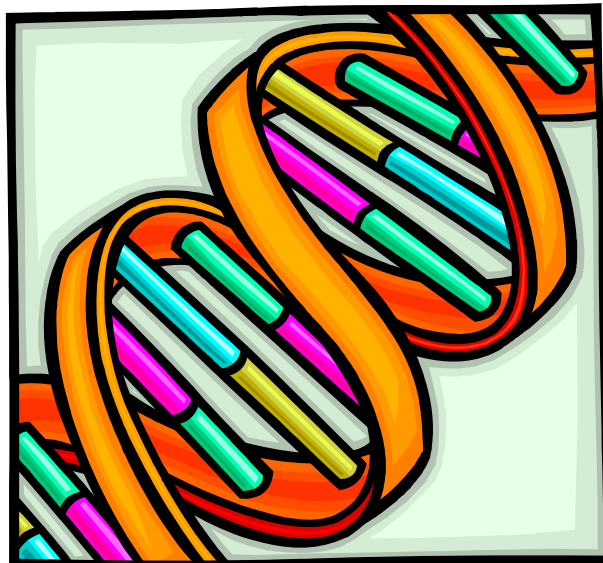


North Carolina State Genetic Services: A Guide for Healthcare Professionals

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*North Carolina Department of Health and Human Services
Division of Public Health
Children and Youth Branch
Genetics and Newborn Screening Unit*

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Regional Genetic Counselors

The definition of genetic counseling as stated by the National Society of Genetic Counselors (NSGC) is *the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.*

This process integrates:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources, and research.
- Counseling to promote informed choices and adaptation to the risk or condition.

There are four regional [genetic counselors](#) that serve the state of North Carolina.

By region they are:

Region 1 – Kristen Wood, M.S., C.G.C.

Region 2 – Ashley Noack, M.S.

Region 3 – Gail Marcus, M.Ed., C.G.C.

Region 4 – Ginny Vickery, M.S., C.G.C.

The regional genetic counselors serve North Carolina residents and medical community in multiple ways. Services to families are provided through the medical centers' [satellite clinics](#) and private [patient consultations](#) with the genetic counselors. [Professional consultations](#) and [educational trainings](#) are available for the medical community.

Satellite Clinics

The regional genetic counselors provide a genetic evaluation with geneticists from the different [medical centers](#) across the state at satellite clinics. A genetic evaluation at one of these sites often includes a review of the medical and family histories, and a physical examination of the patient. The geneticist and genetic counselor will also discuss any possible testing and results, answer questions, review risk to other family members or future children, and make appropriate referrals.

[To refer a patient](#) to be seen at one of the medical centers' satellite clinics please contact the medical center's genetics office.

Region 1 (Kristen Wood)

- Fullerton Genetics Center
 - Murphy – Peachtree Pediatrics (6 clinics per year)
 - Sylva – Sylva Pediatrics (4-6 clinics per year)
 - Macon – Macon Co. Health Department (2 clinics per year)
 - Spruce Pine – Blue Ridge Regional Hospital (2-3 clinics per year)
 - Morganton – Morganton/Hickory CDSA (4-6 clinics per year)
 - Rutherfordton – Rutherford Pediatrics (2 clinics per year)

Region 2 (Ashley Noack)

- Wake Forest University Genetics
 - Hickory – Fairbrook Medical Clinic (8 clinics per year)
 - Statesville – Brenner's Children's Clinic West (4 clinics per year)
 - Greensboro – BCH Greensboro Clinic (10-12 Clinics per year)
 - * Staffed by WFU genetic counselors
 - North Wilksboro – CDSA (6-7 clinics per year)
 - Boone – CDSA (6-7 clinics per year)
- Carolinas Medical Center
 - Shelby – CDSA (clinics are as needed)
 - Concord – CDSA (clinics are as needed)

Region 3 (Gail Marcus)

- East Carolina University Genetics
 - Wilmington – CDSA (number of clinics per year varies)
 - New Bern – CDSA (number of clinics per year varies)

Region 4 (Ginny Vickery)

- University of North Carolina Genetics
 - Raleigh - Rex Hospital (4-5 clinics per year)
 - * Staffed by UNC Genetic Counselors
- East Carolina University Genetics
 - Rocky Mount – CDSA (4-5 clinics per year)
 - Roanoke Rapids – CDSA (limited number)
 - Windsor – Bertie Co Heath Department (limited number)

How to Refer a Patient for A Genetic Evaluation

How do I know if I should refer a patient for a genetic evaluation?

- Please see our Genetics brochure [insert](#) for a list of appropriate reasons to refer a child for a genetic evaluation. If you have questions about the appropriateness of a referral, please contact your [regional genetic counselor](#).

How do I make the referral?

- Once you have confirmed the need to refer a patient for a genetic evaluation, please contact the [medical genetics center](#) nearest to you. The genetics center will either take the referral information from you over the phone, or fax you a referral form to fill out and fax back to them. Depending on the distance the patient lives from the medical center and any difficulties the family may have with travelling, you may request for the patient to be seen at a [satellite clinic](#) that is closer and more convenient. The genetics center will then schedule the patient to be seen at either a satellite clinic or the genetics center, as appropriate. Satellite clinics are usually reserved for pediatric patients. Patients scheduled for satellite clinics will be seen by a medical geneticist from one of the genetics center, along with the state genetic counselor for that region.

Professional Genetic Consultations

What is a professional genetic consultation?

- A professional genetic consultation is a consultation between a regional genetic counselor and any health care or early intervention provider. The regional genetic counselor may be consulted for many reasons, including but not limited to:
 - Gathering genetic condition information including symptoms, testing and management guidelines
 - Review of patient materials for accuracy regarding genetic conditions/testing
 - Assessment of possible genetic referrals

Who do I contact for a professional genetic consultation?

- Contact your regional genetic counselor. To identify and get contact information for your regional genetic counselor, please see the regional [genetic counselor](#) map.
- All consultation services are **free**.

Patient Genetic Consultations

What is a patient genetic consultation or “Counseling Only” appointment?

- Health Care Providers may refer a patient for genetic counseling and education through a patient genetic consultation. The purpose of a “Counseling Only” appointment is to discuss genetic issues that may be affecting the patient, but do not require the services of a doctor. **A doctor will not be present**, and no testing can be performed during the meeting with the genetic counselor. However, the genetic counselor can make testing recommendations that the referring physician may choose to order.

What happens during a “Counseling Only” appointment?

- The genetic counselor will meet with the patient or family at their local health department, CDSA, or referring provider’s office. The counselor will gather information from the patient, provide explanations and information as requested, offer relevant support information, and possibly make testing recommendations. **No diagnoses will be made.** A report summarizing the information discussed and gathered during the appointment will be sent to the referring physician and the patient.

What kinds of patients are appropriate for “Counseling Only” appointments?

- *Any patients or families that are seeking information about a previously diagnosed genetic condition. Information provided may include:*
 - Condition symptoms and associated health concerns
 - Explanation of genetic causes
 - Recurrence risks
 - Testing options
 - Current research in condition
 - Support services
- *An individual or couple who is seeking information about family planning or a current pregnancy based on:*
 - Advanced maternal age
 - Teratogenic exposure
 - Family history of genetic conditions, birth defects, or mental retardation
- *An individual, couple, or family where an individual has been identified as a carrier of a genetic disease based on newborn metabolic screening or prenatal carrier testing.*

What kinds of patients are NOT appropriate for “Counseling Only” appointments?

- Any patient who requires the services of a doctor. The genetic counselor will not perform a physical exam, provide testing, or write prescriptions.
- Prenatal patients with abnormal prenatal screens or ultrasound findings. These patients should be referred directly to a medical center that can provide Level II ultrasounds and diagnostic testing.

How do I make a “Counseling Only” appointment?

- Contact your regional genetic counselor. To identify and get contact information for your regional genetic counselor, please see the regional [genetic counselor](#) map.
- The genetic counselor will contact the referring provider to arrange a date and time to meet with the patient at their local health department, CDSA, or referring provider’s office. Counseling sessions will last about 1 hour and require an office or exam room where counseling can occur comfortably. *This service is free.*

Available Genetics Trainings

Please contact your regional [genetic counselor](#) to schedule training for your health department or CDSA.

Presentation Title	Description	Length (in hours)	Nursing Credit	Infant-Toddler Credit
Prenatal Diagnosis	Reviews enhanced MSAFP/quad screening, 1 st trimester, genetic screening, ultrasound accuracy, CVS, and genetic amniocentesis	1.5	✓	✓
Newborn Screening	Explains the newborn screening process and conditions detected in North Carolina	1.5	✓	✓
46 Chromosomes: More or Less	Discusses cytogenetic testing techniques and reviews chromosomal disorders such as Down syndrome, Turner syndrome, and Williams syndrome	1.0	✓	✓
It's in the Genes	Reviews single gene disorders such as Marfan syndrome, cystic fibrosis, and Fragile X syndrome with emphasis on inheritance patterns	1.25	✓	✓
Genetic Evaluation Process	Reviews reasons for referral to genetics clinics and explains the steps of a genetic evaluation	1.25	✓	✓
Teratogens	Discusses the risks to a fetus of exposures to medications, chemicals, alcohol, drugs, and maternal health conditions	1.25	✓	✓
Genetics of Hearing Loss	Reviews the syndromes and genes associated with hearing loss	1.25	✓	✓
The Family Pedigree	Explains how to take a family history and what red flags to look for	1.25	✓	✓

Updates in Genetic Testing	Reviews the different types of genetic testing currently available	1.25		✓
Hereditary Breast and Ovarian Cancer	Reviews basic cancer genetics, focusing on the genes involved in breast and ovarian cancers	1.5	✓	
Hereditary Colon Cancer	Reviews basic cancer genetics, focusing on the genes involved in colon cancer	1.25	✓	
X-linked Mental Retardation Syndromes	Reviews X-linked inheritance using four X-linked mental retardation syndromes as examples	1.25		✓
Cystic Fibrosis	Review of symptoms, inheritance, and testing for cystic fibrosis, including an overview of NC newborn screening protocol	1.0	✓	✓
Hemochromatosis	Review of symptoms, inheritance, and testing for hemochromatosis	1.5		
Craniosynostosis syndromes	Review of symptoms, inheritance, and testing of 3 FGFR related and 4 non-FGFR related craniosynostosis syndromes	1.5		✓