Vermont Family Network

facts

Why we are who we are and how genetics can help us?

Genetics in Vermont

Understanding genetics and how genetic testing can help diagnose health conditions in infants and children is important to ensure a child receives appropriate early support and intervention. These interventions might include life-saving therapies and/or medications.

What is Genetics?
Genetics is the study of genes and heredity, or how certain features and traits are passed from parents to their children. Genes are made up of DNA, which is packaged into chromosomes. DNA is present in the cells of all living things. It provides the code, or instructions, for proteins and molecules that are essential for growth, development, and health.

Genetics helps to explain what makes each of us unique, why family members may have similar features, and why some diseases or conditions run in families.

A problem may occur when there are changes in a gene. Types of changes include loss or addition of DNA or mixing-up of the genetic code. Changes in genes can be passed on (inherited) or they can happen out of the blue (spontaneously, or de novo). Most gene changes have no effect on our health.

Genetic conditions can be diagnosed before birth, in childhood or adolescence, or even in adulthood. Many conditions are detected by newborn screening.

What is newborn screening and how can it help families?
Newborn Screening is a public health program that began in the 1960s. Dr. Robert Guthrie discovered a way to test for a condition called Phenylketonuria (PKU) using dried blood spots from newborns. Now, many more health conditions can be found through testing dried bloodspots.

Finding rare health conditions early helps families get the support and care that they need for their newborns to start treatment right away.

What may be confusing is that some healthcare providers still refer to newborn screening as "PKU" testing.

Newborn Screening in Vermont
Vermont screens for 35 of the Recommended Uniform Screen Panel conditions (37 total). Thirty-three of the conditions are tested on dry blood spot sample cards while hearing and heart screening are done at the hospital or at home. Vermont's goal is to have all babies screened as a part of routine newborn care unless the parent or guardian does not want it done.

When and how will I get the results of the Newborn Screen?
The test is usually done 24 hours after birth. It takes several days for the lab to perform all the testing. Results are sent to your baby's primary healthcare provider, the hospital where they were born (or midwife if your baby was born at home). If there is an abnormality with a test, the lab will call your baby's healthcare provider or the hospital. Your baby's healthcare provider should review the results of newborn testing. If that doesn't happen, ASK! Hearing and heart testing results can typically be given right away after testing is performed.

What if the results are abnormal?
If the results are abnormal, your baby's healthcare provider will contact you. However, not all abnormal results mean your baby has a health condition. Stress, being sick, prematurity, some medications, and other circumstances can cause false positive results.

It's important to follow up on recommendations from your baby's healthcare provider. Sometimes a repeat of the newborn screen is warranted and other times, further testing might be needed. The screening cannot test for all health conditions.

What if I don't want my baby tested?
It's suggested that you first talk with your baby's healthcare provider. You can also call the Vermont Newborn Screening Program with questions. If you'd like to opt-out of the testing, you will need to sign a form.

What if I have a concern when my child is older?
If you have concerns with your child's development or your child has been diagnosed with health and/or developmental conditions, speak with your child's doctor about whether genetic testing may be helpful. You may also contact Vermont Family Network and ask to speak with their genetic navigator.

Genetic Navigation
Vermont Family Network offers a genetic navigation resource. A genetic navigator (GN) is a person who has lived experience getting a genetic diagnosis. A GN can:

- Provide support throughout the genetic testing process and share personal experiences navigating different systems of care
- Assist with accessing funding for genetic testing and insurance appeals

- Help advocate for genetic testing
- Help providers and families in identifying red flags and determining when a referral for genetic testing may be necessary
- Connect families with resources related to genetic conditions

Genetics and Resources in Vermont
Depending on screening and/or test results, you may be referred to a genetic counselor or a geneticist. A genetic counselor supports families with genetic testing decisions and can help families navigate the world of genetics. A geneticist conducts physical examinations, performs diagnostic tests, and reviews and interprets genetic lab results. Other types of geneticists perform and oversee laboratory research and testing.

Where are genetic services located?
Genetic services in Vermont are currently located at the University of Vermont Medical Center in Burlington.
Dartmouth Hitchcock Medical Center in Lebanon, New Hampshire, has a genetics department that provides counseling and genetic testing.

How do I seek genetic testing for my child in Vermont?
If you have concerns and you are interested in genetic testing or if genetic testing has been recommended, speak with your child's doctor about your concerns and to make sure required referrals and insurance paperwork are completed on time.

Is genetic testing expensive?
The cost of genetic testing has significantly decreased since 2001. Checking in with your insurance company and a genetic counselor can help you have information about what might be covered and what testing might cost before you begin the process.

Is genetic testing covered by insurance?
Prior authorization is often required for genetic testing. Insurance companies want to know information such as why has the test been ordered? Has the family participated in genetic counseling? What clinical features of the condition are present? The test may be denied because of too few symptoms or if the diagnosis can be made simply from the symptoms alone, or for other reasons.

What can be done if genetic testing is denied?
Many insurance companies already have policies on what they will cover.
Common Denials are:
- Not a covered benefit or listed as an exclusion
- Provider is out of network and plan doesn't have an out of network option
- Service considered experimental or investigational
- Treatment not considered medically necessary
- Services no longer appropriate in a specific healthcare setting
- The claim was not filed timely
- Preapproval was required and not submitted

If you receive a denial, make sure that you understand the reason for the denial.

Pay close attention to timelines for appeal submittal. Contact your doctor for support and helpful documentation to include in your appeal letter. After sending the appeal letter, follow up to be sure the insurance company received it.

All Medicaid programs and most private insurance companies have a method for appealing a decision. Many are reviewed internally and can be expedited if your child is in the hospital. Sometimes a peer review is necessary (an MD or other healthcare provider meeting with the insurance company). Typically, this would come through the insurance company, but following up and knowing where things are in the process is important.

If my child is denied genetic testing, what other resources are available?
If your doctor strongly feels that the testing would be helpful, talk with a genetic counselor about working directly with a testing company. A genetic counselor can help you get testing set up with the testing company. Hospitals and testing companies typically have payment plans as well.

Resources

Baby's First Test

Dartmouth Health Children's Medical Genetics Program
603-629-8355

Genetic Connections Part 1
Ashley Michaud- VFN Genetic Navigator

Genetic Connections Part 2
Ashley Michaud- VFN Genetic Navigator

Health Resources and Services Administration (HRSA)

National Genetics Education and Family Support Center
202-966-5557

National Organization for Rare Disorders (NORD)

Newborn Screening & Early Hearing Detection and Intervention Program
Linda A. Hazard, Ed.D. CCC-A, Program Director of Vermont Early Hearing Detection and Intervention Program, and Sydney Williamson-White, BSN, RN, Nurse Program Coordinator of the Vermont Newborn Screening Program

Rare New England
University of Vermont Medical Center – Clinical Genetics
802-847-4310

The Mystery in Our Genes: Why We Are Who We Are
Ed Kloza MS, CGC - Genetics consultant, New England Regional Genetics Network

Vermont Family Network (VFN)
802-876-5315 or info@vtfn.org

Vermont Newborn Screening Program
802-951-5180

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