Rare Diseases in Vermont

Department of Health

Required pursuant to Act 164 (2024)

VERMONT

DEPARTMENT OF HEALTH

Act 164 (2024) Requirements

Act 164 (2024) requires the Department of Health to:

1) Describe the public health impact of rare diseases in Vermont and
2) Describe the Department’s role in addressing rare diseases statewide

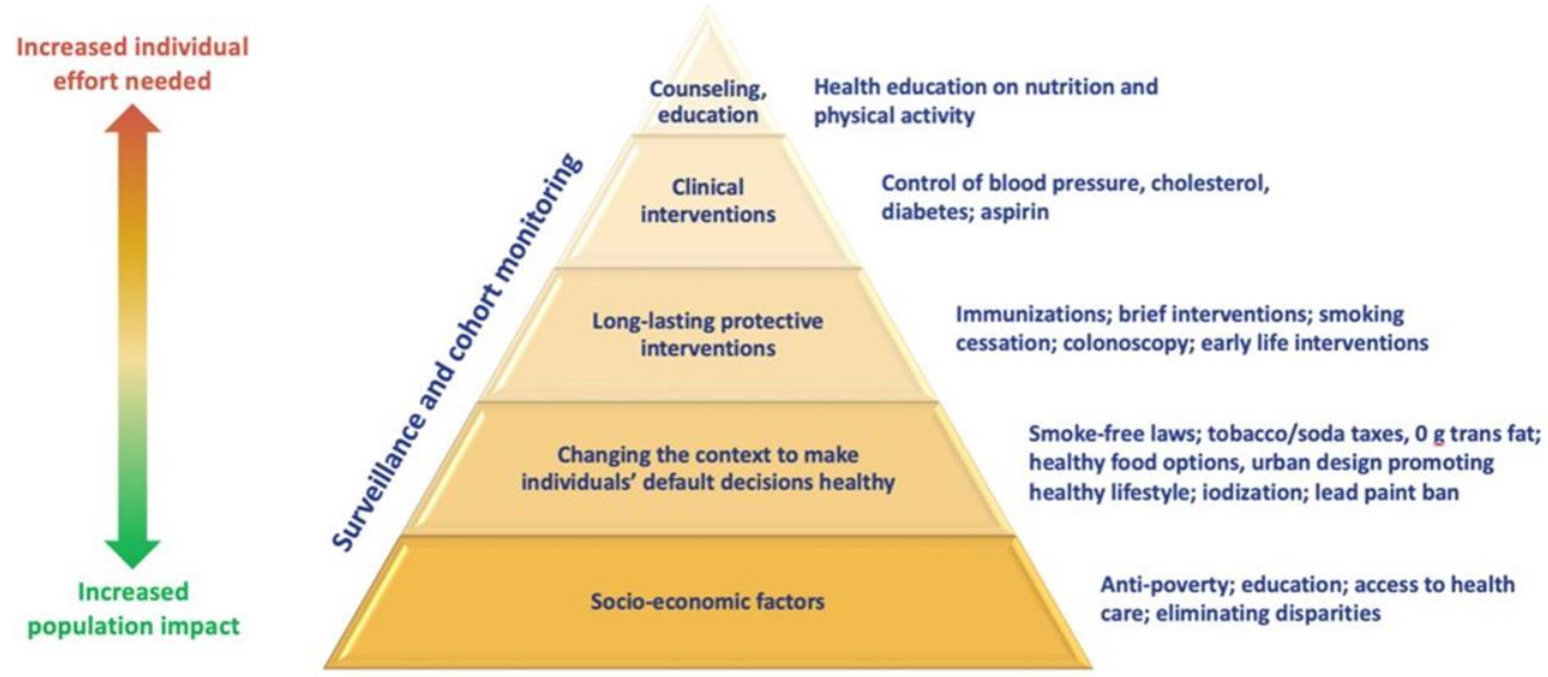
Rare Diseases

National Organization for Rare Disorders**: A rare disease is a disease, disorder, illness or condition that affects fewer than 200,000 Americans. There are over 10,000 rare diseases that together affect more than 30 million Americans.**

Rare Diseases Include:

- Many types of cancers
- Many types of birth defects
- Amyotrophic Lateral Sclerosis
- Many other diseases, anomalies and conditions

Public Health and Medical Model Pyramid



Frieden T. American Journal of Public Health. 2010;100(4):590-595.

The Agency of Human Services and Rare Diseases

- The Department of Health
  - The Vermont Birth Information Network
  - Children with Special Health Needs
  - Newborn Screening and Hearing Screening programs
  - Cancer Registry
  - ALS Registry

- The Department of Vermont Health Access
  - Provider Resources and Clinical Programs

- The Department of Disability, Aging and Independent Living
  - Programs and services

Department of Health's role in addressing rare diseases

1. **Screening & Data Collection: Screening for diseases, collecting diagnosis reports, and reporting instances of rare diseases.**
2. **Partner Collaboration: Connecting families with supports and services across Vermont to optimize quality of life.**
3. **Disease Prevention & Health Promotion: Most rare diseases are not preventable and are based on genetics, but there are some areas where we can promote healthy choices to lower the risks.**

Data Collection, Screening and Surveillance

Collecting data on diseases and other health risks and behaviors is a critical part of public health. The following screening programs and registries help us understand the impact of rare diseases in Vermont.

Newborn Screening Program	**Birth Information Network**	**Cancer Registry**	**ALS Registry**
Finds rare health problems early so newborns, and their families, can get the support and care they need. Dried blood spot from newborn is tested for 33 conditions.	Registry of selected rare diseases (birth defects and other congenital conditions). Established in 18 V.S.A. § 5087, includes medical claims data, newborn screenings, and Vital Record information.	Registry of cancer diagnoses of people in Vermont. Health care facilities report per 18 V.S.A. Chapter 4.	Registry of ALS diagnoses of people in Vermont. Health care facilities report per 18 V.S.A. chapter 4A.

Newborn Screening Program

- Newborn Screening is a public health program that screens babies for diseases and conditions.
- The goal is to find diseases and conditions early so newborns, and their families, can get the support and care they need.
- This is a comprehensive system, not just testing, this includes follow-up to make sure all babies are screened, babies get prompt referral to specialists, education, and quality assurance.
- The Recommended Uniform Screening Panel (RUSP) is determined by a group of national experts called the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) based on evidence review. **Federal Update:** Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) has been disbanded

Birth Information Network

- Tasked to conduct statewide, population-level surveillance of selected birth defects and other congenital conditions.

- Evaluate efforts to **prevent health problems.**

- In partnership with Environmental Health staff, explore possible associations between **environmental and chemical exposure with health conditions** of Vermont's infants and children.

- Share data with local and national partners.
  - Vermont and National Environmental Public Health Tracking (EPHT) programs.
  - National Birth Defect Prevention Network (NBDPN).

Overlap between the Newborn Screening Program, Birth Information Network, and Rare Diseases

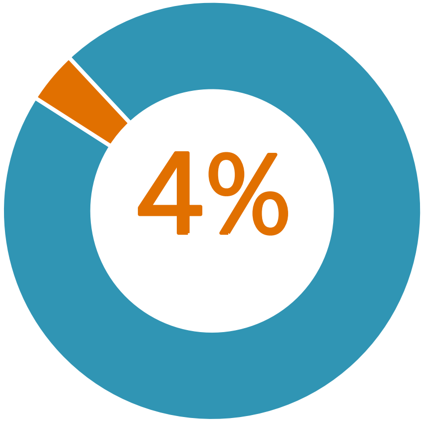
- The Department collects data on approximately 115 conditions and diseases through the Newborn Screening Program (NBS) and Birth Information Network (BIN)
- While some of the diseases and conditions screened for and collected in the NBS and BIN are rare diseases, not all are considered rare diseases.
- It is **not** the purpose of the NBS and BIN to be a global screening and repository for all rare diseases.
- The goal of the NBS and BIN is to find diseases and conditions early so newborns, and their families, can get the support and care they need. This is a comprehensive system, not just testing, this includes follow-up to make sure all babies are screened, babies get prompt referral to specialists, education, and quality assurance.

Examples of Birth Information Network Data

- Of the 26,610 infants born to Vermont residents from 2018 through 2022, 1,049 (4%) had one or more conditions monitored by the BIN.

- Structural and chromosomal birth defects were the most common type 726 infants (2.7% of births).

- For more information, please see the [Vermont Birth Defect Data Brief 2018-2022]() and visit the [Department's Birth Information Network webpage]().



Cancer Registry

- Some cancers are considered a rare disease.
- The Department of Health operates a Cancer Registry
- The Vermont Cancer Registry tracks every case of cancer among Vermont residents and any case diagnosed in Vermont, including rare cancers.
- 18 V.S.A. §§ 151-157 requires all health care facilities diagnosing or treating cancer or benign brain-related tumors in the State of Vermont to report information to the Vermont Cancer Registry.
- For more information, see the Cancer in Vermont data pages

ALS Registry

- ALS is considered a rare disease.
- Vermont Department of Health operates a statewide ALS Registry
- Act 149 (2022) requires healthcare providers who diagnose or treat ALS patients to report all individual cases to the Vermont Department of Health.
- Amyotrophic Lateral Sclerosis (ALS) Registry 2025 Report to the Legislature
- Ten health care providers reported that 27 Vermonters received an ALS diagnosis in 2023.

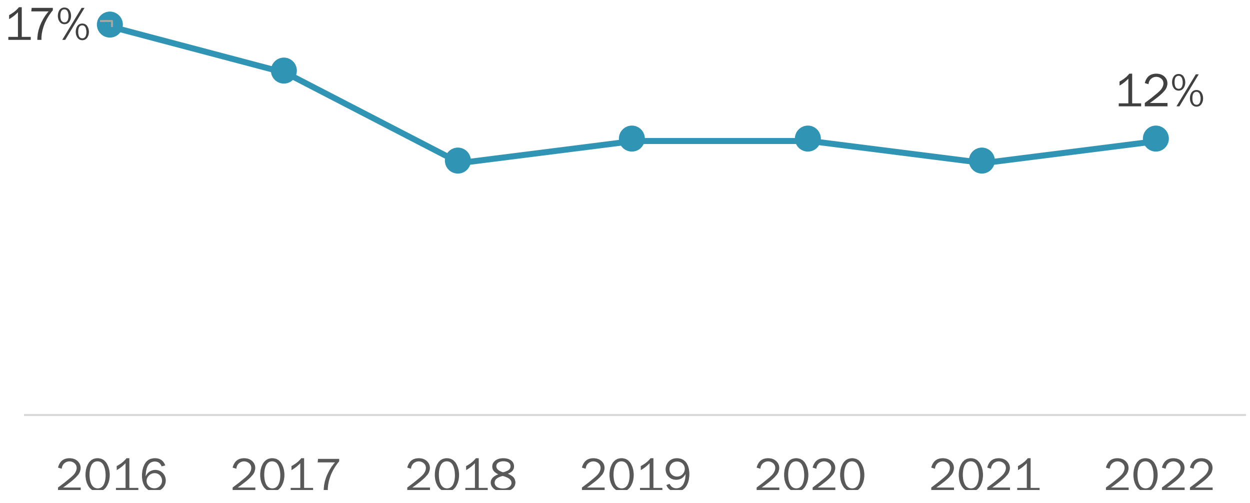
Partner Collaboration: Connecting Families to Partners who Provide Care

- Children with Special Health Needs (CSHN) supports Vermont children and youth with complex, chronic health conditions, and/or developmental needs through Medicaid administered programs and community support.
- Once an infant is found to have a BIN condition, a letter is sent to the parents which includes information about CSHN.
- Support from birth to age 21
- A team of social workers, nurses, and specialty staff with experience supporting children with special health needs, their families, and providers.

Disease Prevention & Health Promotion

We know that alcohol use during pregnancy can lead to birth defects (some being rare diseases). Prevention efforts have helped decrease the rates of drinking during pregnancy in Vermont.

Alcohol use during pregnancy is decreasing in Vermont.



Pregnancy Risk Assessment Monitoring System, 2016-2022

Resources

- The National Birth Defect Prevention Network: https://nbdpn.org/
- Baby's First Test: https://babysfirsttest.org
- Health Resources and Services Administration (HRSA): https://newbornscreening.hrsa.gov/newborn-screening-process

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Thank you!