



Each year, the last day of February is dedicated to raising awareness about rare diseases, defined as a condition that affects fewer than 200,000 people in the U.S. Despite the fact that many rare diseases go undiagnosed and no cure exists for a majority of rare diseases, progress is being made to ensure that children and families are receiving the individualized support and care needed to reach their optimal health. Keep scrolling to learn how you can participate in Rare Disease Day with NICHQ!

## Improve Care for Patients

1 in 20 people will live with a rare disease at some point in their life. Read the articles below for strategies that address the needs of those living with rare diseases.

**Partner with Families and Caregivers:** There are a variety of miles to cover and turns to take on the journey of effective engagement and inclusion of a Patient/Family Partner (PFP) onto a work group, advisory council, Quality Improvement (QI) team, or other clinical or health systems initiatives. Pre-planning is likely needed in advance of inviting PFPs to join in on the journey. This roadmap provides a framework for effective engagement and inclusion of a Patient/Family Partner (PFP) onto a work group, advisory council, Quality Improvement (QI) team, or other clinical or health systems initiatives.

[Download the Roadmap](#)

**Stay Informed on Important Research:** ECHO cohorts have produced over 1000 papers published or in press. 71 were ECHO-wide, meaning one or more ECHO cohort was involved, and DINE cohort data contributes to many of these publications. The de-identified data from the ECHO Program are now available as a resource for the entire scientific community. This first public-use version of ECHO-wide Cohort data covers information that our 69 cohort studies collected from 41,299 participants.

[View ECHO Publications](#)

**Improving Care for People Living with SCD:** A model protocol with best practices for caring for people living with sickle cell disease (SCD) and a helpful compendium of tools and resources for providers, patients, and caregivers were developed to accompany the [2017-2021 SCDRCTDP Report to Congress](#). *Funding for this report was provided by the Health Resources and Services Administration.*

[Download Compendium of Tools and Resources](#)

**Reducing Missed Clinic Appointments:** People with rare diseases, such as sickle cell disease, experience unique challenges, such as maintaining scheduled appointments. [Watch a series of helpful webinars](#) about reducing missed appointments, and download our patient/caregiver infographic that suggests some steps to find solutions and help make getting to appointments easier. *This project was funded through the Patient-Centered Outcomes Research Institute (PCORI).*



## Diseases

[Content Development for Newborn](#)

**Screening Clearinghouse:** To increase awareness, knowledge, and understanding of newborn screening and genetic conditions, NICHQ partnered with Genetic Alliance to develop, revise and deliver general, state-specific and condition-specific newborn screening website content.

### **Environmental Influences on Child Health Outcomes: Developmental Impact of NICU**

**Exposures (ECHO DINE):** ECHO DINE is a research study on the long-term impact of environmental exposures in neonatal intensive care units (NICUs). NICHQ serves as a coordinating center for 8 DINE sites participating in ECHO. In this role, NICHQ provides technical assistance, hosts monthly webinars for study sites and quarterly training for study coordinators, and supports and maintains communication and coordination among DINE sites and between DINE sites-ECHO.

**Improving Care for Children with Special Healthcare Needs:** NICHQ is leading a learning and action network (LAN) for seven SNAQ teams to support a high-quality system of care in Florida that serves all children and youth with special healthcare needs, regardless of insurance status and location.

**Improving Sickle Cell Disease Care - Hemoglobinopathies Coordinating Center:** NICHQ, with partner organization Abt Associates, is supporting a Hemoglobinopathies National Coordinating Center (NCC) to help SCD treatment demonstration program grantees — including community and federally qualified health centers — address structural and systemic barriers in their regions and implement evidence-based SCD care.

[View NICHQ's Current Initiatives](#)

**I SUPPORT  
RARE DISEASE DAY  
28 FEBRUARY 2023  
[#RAREDISEASEDAY](#) [RAREDISEASEDAY.ORG](#)**



## Spread the Word about Rare Disease Day

Download this online collection of social media posts, posters, infopacks, tool kits, fact sheets, infographics, and more, sharing key statistics and facts that illustrate the impact rare diseases have on more than 25 million American families.

### [Download Promotional Resources](#)

*Interested in learning more about the Rare Disease Day movement?*

*Visit the official [Rare Disease Day](#) campaign page for more resources and ideas that support those living with rare diseases.*



**MANY STRONG PROUD**