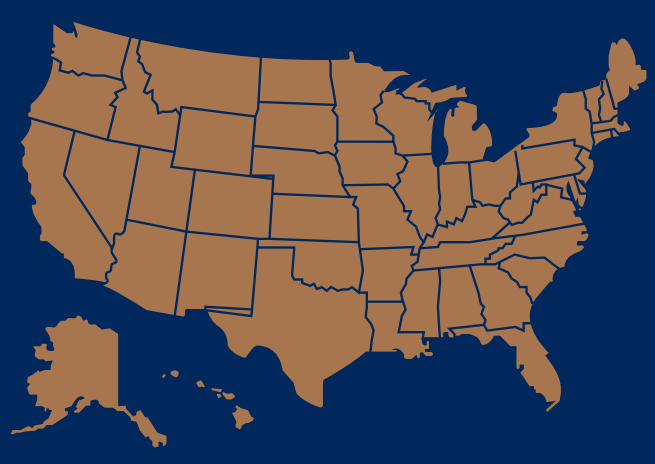
NEWBORN SCREENING

For Rare Disease Day, we're highlighting newborn screening and the importance of early diagnoses to improve long-term health.

Newborn screening is a nationwide practice to test babies for conditions and rare diseases a few hours after they're born. Newborn screening is different in each state, as each state chooses what diseases to test for and conducts tests in their own labs. These tests analyze a blood sample taken from a prick of a baby's heel.

CF SCREENING NATIONWIDE

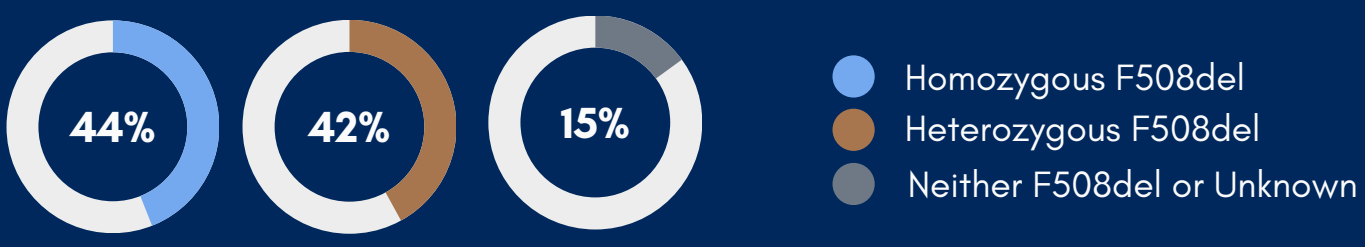
Cystic fibrosis is one of the dozens of conditions that all fifty states screen for. To screen for CF at birth, tests use heel blood to measure immunoreactive trypsinogen (IRT), a marker of pancreatic inflammation. A DNA panel is then used to confirm the CFTR gene variant present. The amount and type of CFTR gene variants tested differ by state. Lastly, a sweat test is performed to determine a diagnosis of CF.



EARLY DIAGNOSES SAVE LIVES

Early CF diagnosis is associated with better health outcomes, improved nutritional status, a more rapid increase in lung function, and delayed chronic P aeruginosa infections. However, there are still disparities in diagnoses of CF, particularly among babies who have rarer mutations. Some states test only for the most common mutations, whereas others test for hundreds. This means that some babies do not receive a timely diagnosis and face worse health outcomes later in life because they have a less common mutation. This disparity disproportionately affects babies who are less likely to have the most common CFTR gene, F508del, and not of European ancestry.

F508DEL VARIANT PREVALENCE IN THE US



2023 CFF Patient Registry

BEF advocates for fast and comprehensive screening for all newborns born with CF and the adoption of recommendations made by the Cystic Fibrosis Foundation, as well as continuing robust education programs for primary care providers to recognize CF symptoms and make timely diagnoses.

You can read the Cystic Fibrosis Foundation's latest update on newborn screening here and learn more about newborn screening for rare diseases here.