

Abstract

Introduction

Newborn screening (NBS) is a multi-component, multi-stakeholder system that begins with prenatal education, neonatal screening in birthing hospitals and state-based NBS laboratories, diagnosis and clinical care, and longitudinal medical management. All components of this system are necessary to ensure newborns with genetic conditions achieve the best possible outcome. The majority of the 12,500 newborns diagnosed each year with a genetic condition through NBS require lifelong care and management. While recent additions to NBS panels involve a wide range of specialists from immunology to neurology, medical geneticists continue to play a dominant role in diagnosing, treating, and managing most cases. The involvement of medical geneticists coupled with the delivery of care across the lifespan offers a unique opportunity to advance understanding of genetic disease while improving health outcomes. We describe a new initiative to capitalize on medical geneticists' efforts to deliver long-term follow-up (LTFU) care of thousands of newborns each year.

Methods

For over a decade, the American College of Medical Genetics and Genomics (ACMG) has led two networks that advance NBS: the National Coordinating Center (NCC) for the seven Regional Genetics Networks (RGNs) and a Newborn Screening Translational Research Network (NBSTRN). The NCC promotes the translation of genetic medicine into public health and health care services, while the NBSTRN develops tools and resources that support discovering new technologies to improve and expand NBS. Both NCC and NBSTRN are well-known in the NBS community and involve the medical genetics community of clinicians, genetic counselors, and researchers. We sought to use NCC and NBSTRN established networks of families, clinicians, state NBS programs, and state public health agencies to advance genetic disease understanding and improve health outcomes in newborns diagnosed with a genetic condition through NBS. This effort utilizes the NBSTRN-developed Longitudinal Pediatric Data Resource (LPDR), a repository currently housing longitudinal health information deposited by families, community-based organizations, clinical investigators, and state NBS programs. We expanded the LPDR's functionality to include a longitudinal care, data tracking, and reporting system to enable state public health agencies, clinicians, and families to enroll, track, and report newborns and children identified with a condition through NBS. The system will feature disease-specific LTFU care algorithms (LTFU-Cares) that list recommended therapies, interventions, and management approaches for each stage of life and disease. The content of the LTFU algorithms will be generated by subject matter experts, designed for parents, clinicians, and state NBS programs, and incorporate a web-based tracking checklist (LTFU-Check) for state public health agencies to monitor LTFU at both the case- and state-levels. The collection of longitudinal data can facilitate care coordination through a medical home, inform and improve treatments based on evidence, facilitate improvement of the NBS system over time, and advance understanding of NBS conditions, comorbidities, and health outcomes. We used an innovative approach, computational social choice engagement, to enable parents, families, clinicians, and state NBS programs to prioritize services and outcome measures over time for each condition. The initiative will deliver web surveys at defined time points to a variety of NBS stakeholders and facilitate electronic data tracking, reporting, and sharing between all stakeholders. We used best practices in data governance and role-based access to ensure that data privacy was protected.

Results

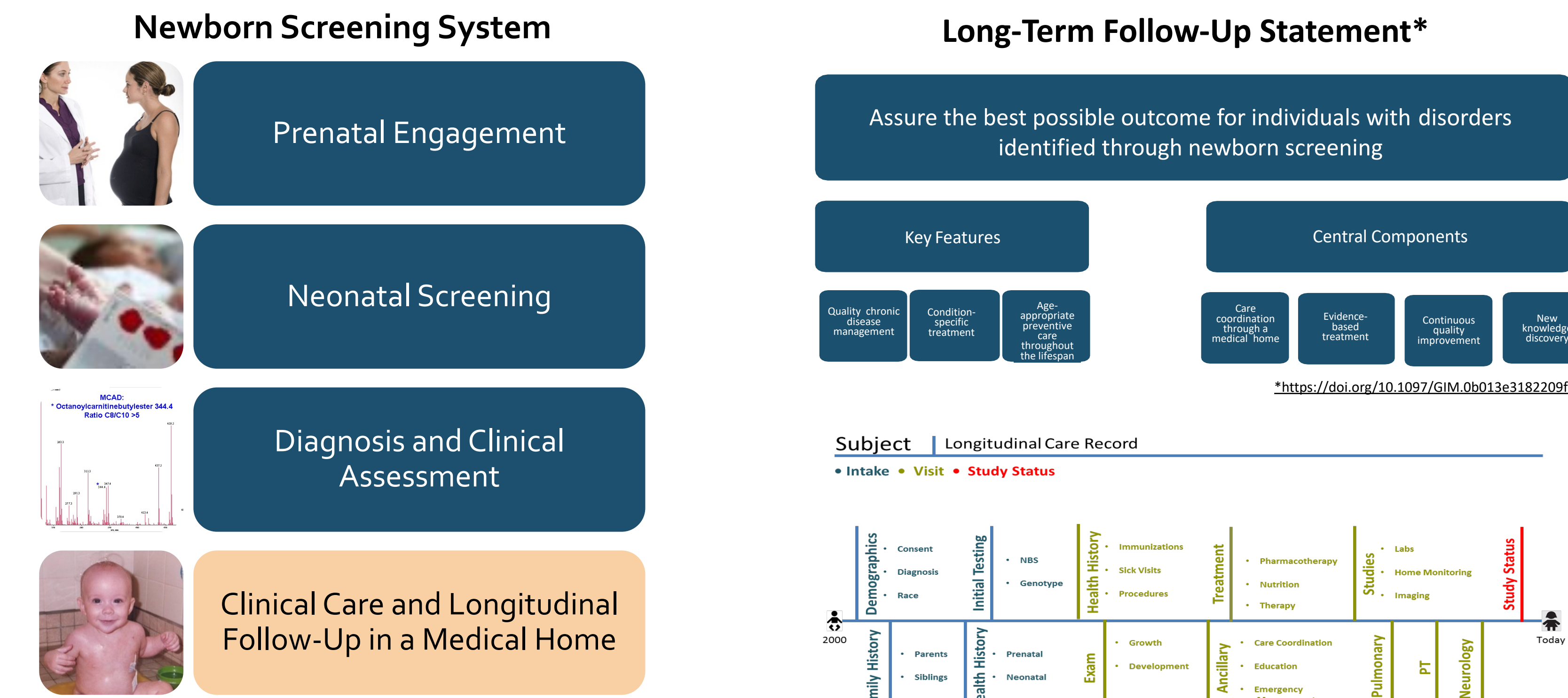
We worked with subject matter experts to develop common data elements (CDEs) that addressed the four components of LTFU care and outcomes reporting: care coordination, evidence-based treatment, continuous quality improvement, and new knowledge discovery. The LTFU-Care and LTFU-Check will enable the reporting of critical metrics including the percent of families reporting they are currently receiving specialty care for the condition; percent of patients with a medical home; the number of formal and informal partnerships developed with primary and specialty clinicians, state NBS programs, state public health and maternal and child health programs, parents/families, condition-specific organizations, laboratorians, researchers, educators, and relevant community services; percent of families who report their needs are being met; and percent of infants lost to follow-up.

Conclusion

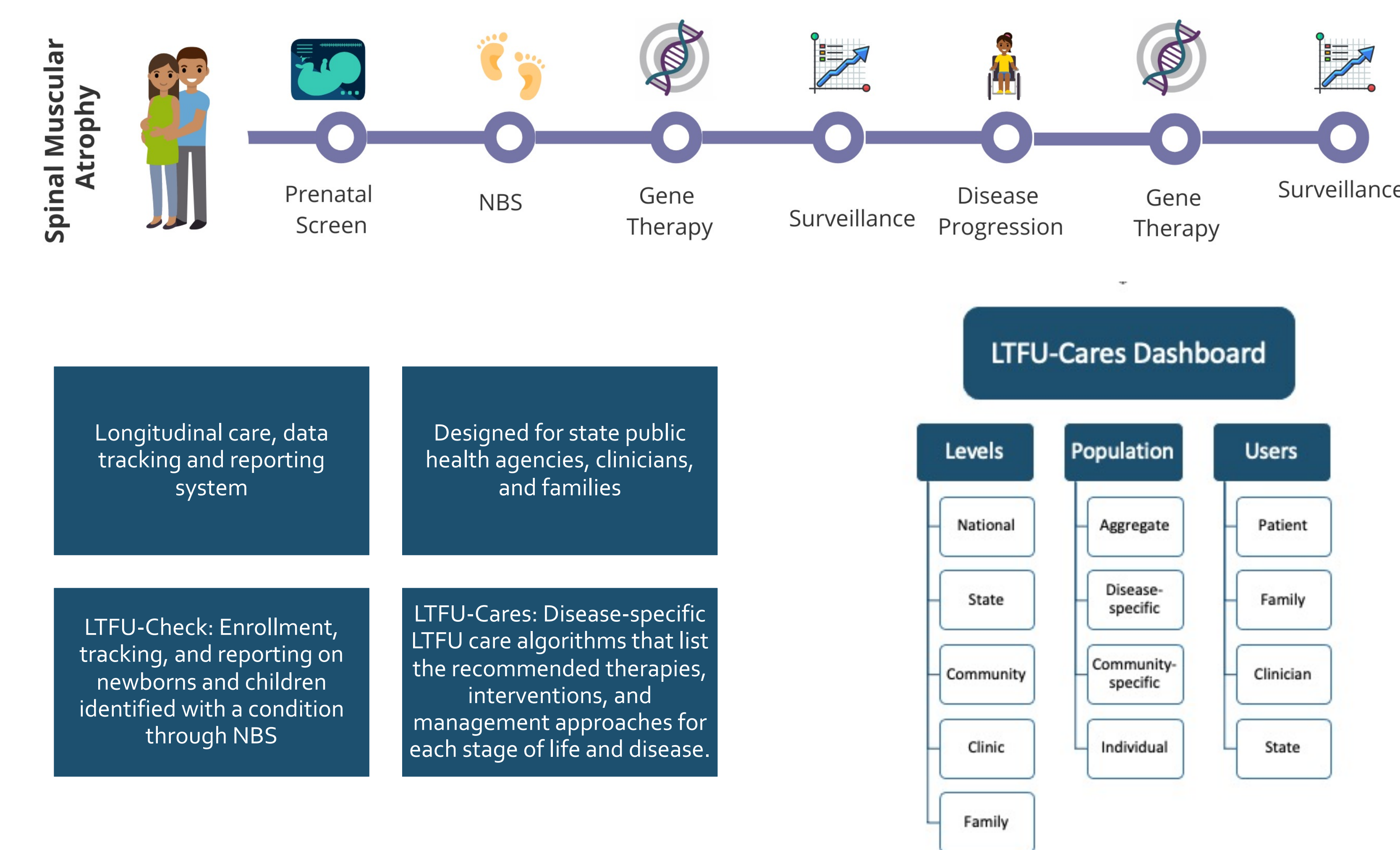
Newborn screening is an important public health program that aims to ensure the best outcomes for newborns identified with disorders that benefit from early identification and intervention. This public health program saves thousands of newborns in the United States every year, and the role of medical genetics and genomics is vital. The potential to leverage the NBS system of neonatal identification followed by timely diagnosis, care, and lifelong management to advance the important goals of improving understanding of the genetic disease, informing and developing best practices in clinical care and treatment is unprecedented. The LTFU-Cares and LTFU-Check Initiative provide the tools and resources to realize these goals.

The Importance of Long-Term Follow-Up in Newborn Screening

The majority of newborn screened conditions require life-long care and management through a medical home. This offers an opportunity to conduct long-term follow-up data collection to advance disease understanding and improve health outcomes.

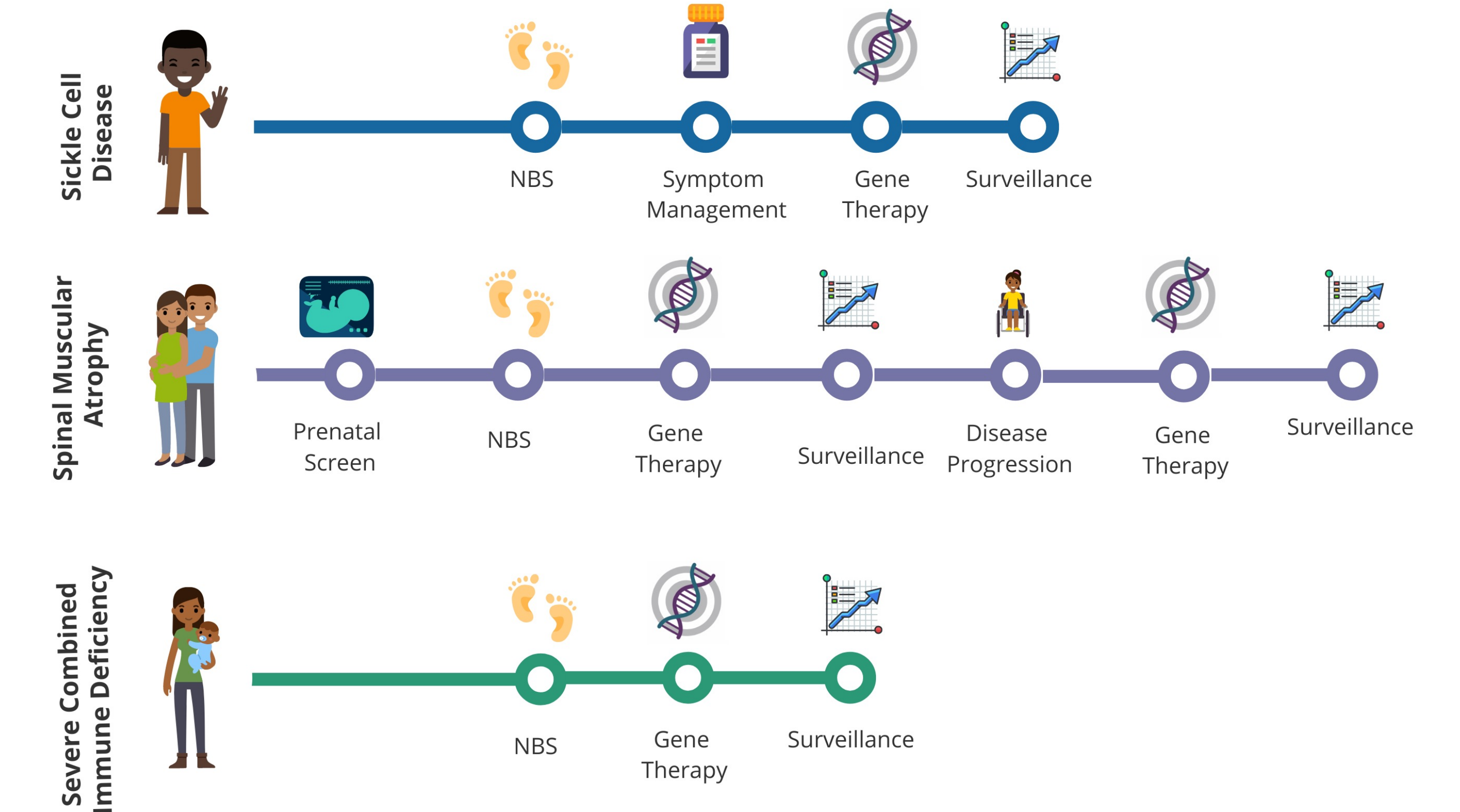


Case Study



LTFU-Cares and LTFU-Check Initiative

A longitudinal care, data tracking and reporting system designed by and for all NBS stakeholders



Next Steps

Engage stakeholders to conduct long-term follow-up data collection within the medical home to assess health outcomes and inform future efforts.

- LTFU-Cares**
 - LTFU Data Collection for Families, Patients, Clinicians
- LTFU-Check**
 - LTFU Monitoring, Assessment, and Reporting for States
- LTFU-Cares Dashboard**
 - LTFU Data Visualization for Families, Patients, States, Clinicians