In the NICU setting, birth defects or genetic conditions contribute to approximately 30% of admissions and nearly 40% of deaths. Many acutely ill infants do not present with stereotypic phenotypes specific to their condition, making a hypothesis-driven approach to diagnostic testing difficult. Additionally, the clinical picture of many acutely ill infants is complicated by many comorbidities (i.e., prematurity, birth trauma, and sepsis) making the development of an accurate phenotype-based differential diagnosis even more challenging.

With an increase in implementation of rapid WES/WGS as a first-tier test in the NICU/PICU setting, supporting evidence shows this diagnostic and prognostic tool offers dramatic increases in speed and efficiency, allowing for proper medical management of critically ill neonate patients.