

How NICUs identify infants for rapid genomic testing

Evidence-informed models selected by clinical programs based on patient mix, referral patterns, and available resources.*



Inclusion-based protocol

Clinical features commonly used to guide rapid genomic testing decisions within institutional protocols and clinical practice.

Infants meet at least one of the following clinical presentations:

- ✓ Hypotonia
- ✓ Seizure activity of unknown origin
- ✓ Congenital anomalies, including but not limited to:
 - Orofacial clefts (e.g., lip/palate)
 - Musculoskeletal anomalies (e.g., clubfoot, scoliosis)
 - Abdominal wall defects (e.g. gastroschisis)
- ✓ Cardiac symptoms with unknown cause
- ✓ Unexplained respiratory failure requiring ventilation
- ✓ Immunodeficiency
- ✓ Dysmorphic features not consistent with a known syndrome
 - Craniofacial features
 - Eye anomalies
 - Limb anomalies
- ✓ Severe metabolic acidosis (pH <7.2 without clear cause)
- ✓ Disorders of sex development
- ✓ Suspicion of mitochondrial disease
 - Lethargy
 - Ocular findings
 - Biochemical abnormalities

Broad access to rapid genomic sequencing reduces missed diagnoses in the NICU

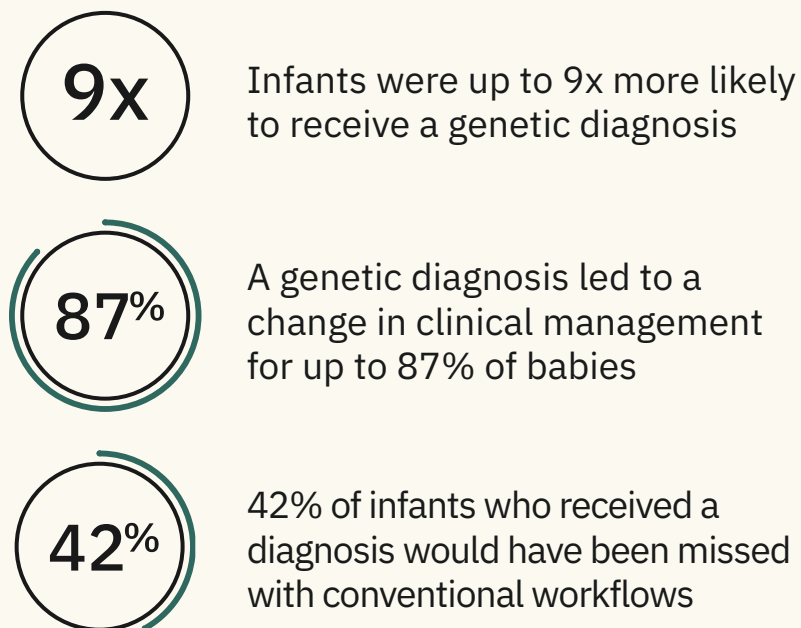
In the SeqFirst-neo study¹, an exclusion-based eligibility model expanded access to rapid genome sequencing in the NICU. This model resulted in more infants receiving testing and genetic diagnoses compared with conventional workflows.

Exclusion-based protocol

Rapid genome sequencing performed on all infants <6 months old, whose clinical findings could not be fully explained by:

- ✓ Trauma
- ✓ Infection
- ✓ Prematurity
- ✓ Pre-existing genetic diagnosis

Results showed:



Selecting the right pathway is unique to each NICU

Let's explore which model aligns with your patient population, referral patterns, and clinical workflow.



To learn more, scan the QR code or visit genedx.co/rapid

*Evidence-informed models derived from peer-reviewed literature^{1,2}, institutional clinical protocols, and payer coverage policies related to rapid genomic sequencing in NICU populations.

References: 1. Wenger TL, Scott A, Kruidenier L, et al. *American Journal of Human Genetics*. 2025. 2. Gubbels CS, VanNoy GE, Madden JA, et al. *Genet Med*. 2020;22(4):736-744.

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