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 study[1], 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:

9x — Infants were up to 9x more likely to receive a genetic diagnosis

87% — A genetic diagnosis led to a change in clinical management for up to 87% of babies

42% — 42% of infants who received a diagnosis would have been missed with conventional workflows

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.* 220;22(4):736-744.