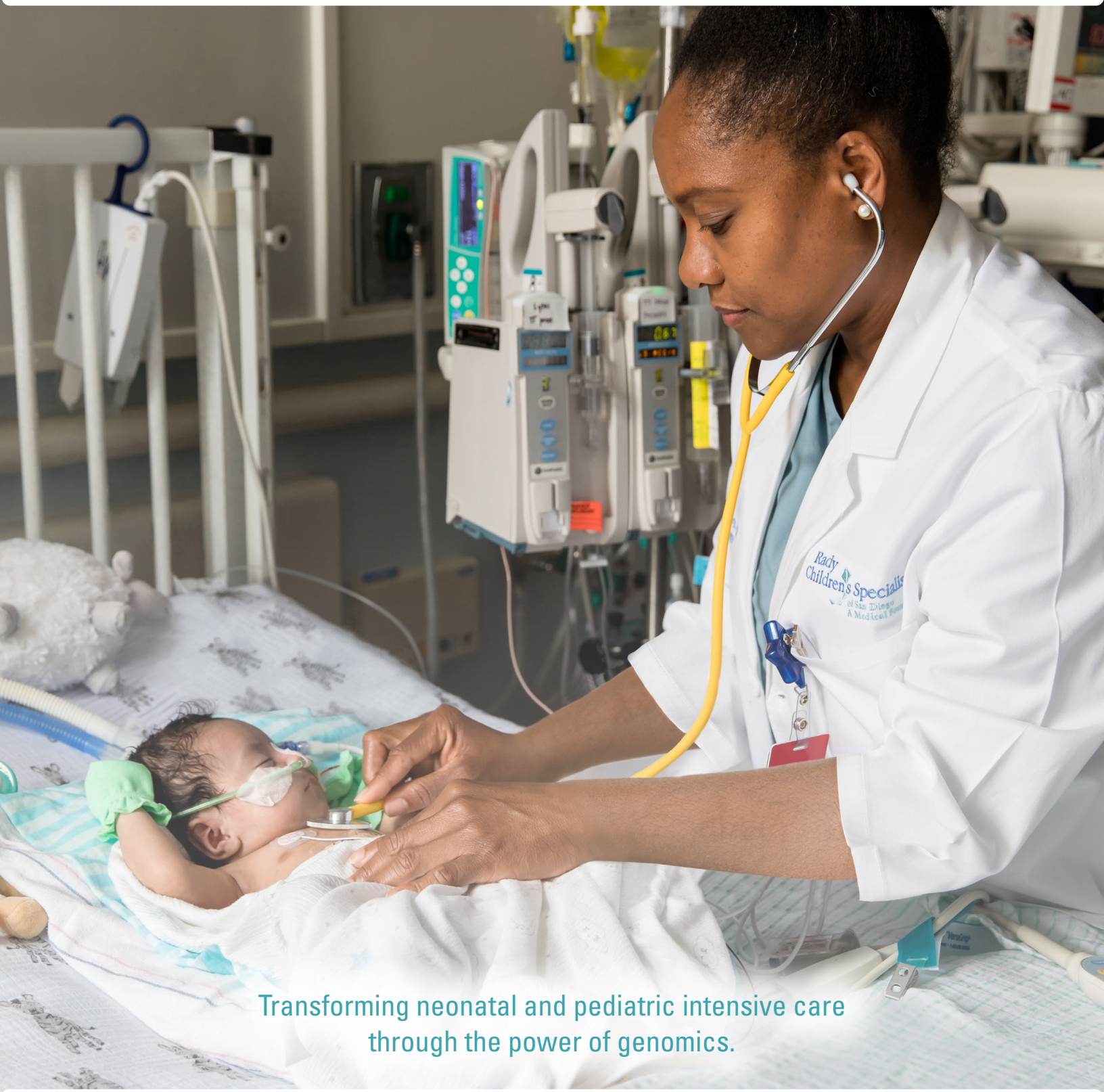
Rady Children's Institute
Genomic Medicine

Genomic Diagnostic Solutions to Accelerate Rapid Precision Medicine



Transforming neonatal and pediatric intensive care through the power of genomics.

Unlocking the Complexities of Rare Genetic Disease with...

RAPID RESULTS

Every minute matters when a child is struggling to survive. That's why our workflow is optimized for speed and accuracy. We aim to provide actionable results within a therapeutic window. We detect, analyze and interpret rare genetic disorders from a minimal blood sample, in a matter of days.

UNIQUE EXPERTISE

In every case, deep phenotype-driven analysis and interpretation is conducted by PhD-level genomic analysts and physician-scientists. Clinical lab directors issue reports based on variant classification aligned with guidelines established by the American College of Medical Genetics and Genomics (ACMG).

PERSONAL CONSULTATION

We work closely with ordering physicians throughout the testing and reporting process. Our experts offer rich insight on the results, relevant medical literature and documented disease-specific interventions to help enhance patient care.

CLINICAL GENOME SERVICES

TEST TYPE	TURNAROUND TIME*
Ultra-rapid Whole Genome Sequencing	≤ 3 calendar days (trio testing preferred)
Rapid Whole Genome Sequencing (rWGS)	≤ 5 days
Whole Genome Sequencing	< 30 days

*TAT to clinically actionable report when a positive diagnosis is made

Why Choose Rady Children's Institute for Genomic Medicine?

Led by Dr. Stephen Kingsmore, our team has pioneered the clinical use of ultra-rapid whole genome sequencing for newborns in intensive care and demonstrated the clinical utility of rWGS in published peer-reviewed studies.

Our laboratory is equipped with state-of-the-art Illumina Novaseq instruments and is CAP accredited, CLIA certified and licensed to provide clinical diagnostic testing nationwide.

Embedded within Rady Children's Hospital-San Diego, our multi-disciplinary team includes board certified clinical geneticists, genome analysts, genetic counselors, neonatologists and specialists in pediatric cardiology, oncology, infectious disease, gastroenterology and neurology.

COMPARE OUR COMPREHENSIVE SOLUTION

TEST	TAT	SNVs & INDELS	CNVs (1 kB - ANEUPLOIDY)	UNBALANCED TRANSLOCATIONS	MITOCHONDRIAL	SMA
RCIGM Ultra-rapid WGS	≤3 days*	Yes	Yes	Yes	Yes	SMN1 & SMN2 Copy Number Analysis
RCIGM Rapid WGS	≤5 days*	Yes	Yes	Yes	Yes	SMN1 & SMN2 Copy Number Analysis
Commercial Lab Rapid WGS	1-2 weeks	Yes	Limited	Yes	Limited	No
Commercial Lab WES	8-12 weeks	Yes	Limited	Limited	Limited	No
Chromosomal Microarray	1-2 weeks	No	Limited	Yes	No	No
Targeted Gene Panel	4-6 weeks	Yes	Limited	No	mtDNA panel	SMA panel

*Time to provisional, positive report SNV - single nucleotide variant CNV - copy number variant Indel - small insertion/deletion

When to test

The earlier genomic testing is ordered, the greater the potential patient benefit.

Rapid or ultra-rapid WGS should be considered whenever an acutely ill inpatient is presenting with a disease of unknown etiology.

RCIGM OFFERS:

- Fastest return of results
  <3 days for medically urgent cases
- Enables personalized care
- Improves outcomes
- Decreases cost of care
- Identifies or rules out genetic disease in a single test
- Avoids unnecessary tests, treatments or surgeries

PLACING AN ORDER

1. Contact RCIGM
   RCIGM_rWGS@rchsd.org
   858 / 966-8127
2. Send patient sample to RCIGM via overnight delivery
3. Sample sequenced, results analyzed and interpreted by RCIGM
4. Results that are immediately actionable are promptly communicated