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ORIGINAL RESEARCH

Diagnostic Yield and Variant Spectrum of Whole Exome Sequencing in Neonates with Suspected Genetic Disorders: A Prospective Observational Study From a Tertiary Care Neonatal Intensive Care Unit in South India

P. V. Bharadwaj Reddy [1*], **J. Ashok Raja** [2], **Senthil Kumar** [3], **S. Thangadurai** [4], **C. Mary Reena** [5]

[1] *DM Postgraduate, Department of Neonatology, Madurai Medical College, Madurai, Tamil Nadu, India*
[2] *Professor and Head, Department of Neonatology, Madurai Medical College, Madurai, Tamil Nadu, India*
[3] *Associate Professor, Department of Neonatology, Madurai Medical College, Madurai, Tamil Nadu, India*
[4] *Assistant Professor, Department of Neonatology, Madurai Medical College, Madurai, Tamil Nadu, India*
[5] *Assistant Professor, Department of Neonatology, Madurai Medical College, Madurai, Tamil Nadu, India*

*Correspondence: P. V. Bharadwaj Reddy <bharadwajreddypv@gmail.com>

ABSTRACT

Background: Genetic disorders are an important cause of morbidity and mortality in neonatal intensive care units (NICUs), but nonspecific presentations make clinical diagnosis difficult, and Indian data on whole-exome sequencing (WES) are limited. We evaluated the diagnostic yield, variant spectrum and clinical utility of WES in neonates with suspected genetic disorders. **Methods:** This prospective observational study was conducted in the NICU of Madurai Medical College, Madurai, India, from September 2024 to August 2025. Fifty-two neonates with a suspected genetic disorder (syndromic/dysmorphic features, unexplained encephalopathy, multisystem involvement, suspected inborn error of metabolism or atypical chronic illness) underwent WES. Variants were classified using the 2015 ACMG-AMP guidelines. Diagnostic yield was defined as the proportion of neonates with a phenotype-concordant pathogenic or likely pathogenic (P/LP) variant. **Results:** Median age at sampling was 19 days (IQR 10.5–28) and median turnaround time was 21 days (IQR 20–24.5). P/LP variants were identified in 25 of 52 neonates (diagnostic yield 48.1%; 95% CI 35.1–61.3). Autosomal recessive inheritance (12/25, 48.0%) and homozygous variants (13/25, 52.0%) predominated. Yield was highest for multisystem involvement (8/10, 80.0%) and syndromic/dysmorphic presentations (10/15, 66.7%) and lowest for unexplained encephalopathy (2/12, 16.7%). Two pathogenic copy-number deletions were detected. Among diagnosed neonates, WES results led to treatment modification in 12 (48.0%), modified follow-up or prognostication in 7 (28.0%) and genetic counselling in 6 (24.0%). **Conclusions:** WES diagnosed nearly half of neonates with suspected genetic disorders and frequently influenced management. Phenotype-driven selection, particularly of infants with multisystem or syndromic presentations, maximises diagnostic yield.

Keywords: Exome sequencing; Genetic diseases, Inborn; Infant; Newborn; Intensive care units, Neonatal; Precision medicine

Introduction

Genetic disorders are increasingly recognised as a major cause of neonatal morbidity and mortality, particularly among critically ill infants admitted to neonatal intensive care units (NICUs). A substantial proportion of severe neonatal illness has a genetic basis, especially in infants with multisystem involvement, metabolic derangement, congenital anomalies or unexplained neurological dysfunction [1]. When routine clinical and laboratory evaluation fails to establish a diagnosis, timely genomic evaluation is therefore needed [2]. However, the early features of neonatal genetic disease are often subtle, nonspecific and overlapping. Respiratory distress, seizures, hypotonia, feeding difficulty, encephalopathy, recurrent infections, metabolic crises and syndromic features can readily be mistaken for sepsis, perinatal asphyxia or neuromuscular disease. Conventional investigations such as biochemical assays, imaging, karyotyping and targeted gene testing are time-consuming and frequently fail to identify the underlying cause of complex presentations [3]. Mechanisms such as somatic mosaicism and disruption of neurodevelopmental genes further complicate clinical interpretation [4,5].

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Whole-exome sequencing (WES) and rapid genomic sequencing have become valuable tools for identifying disease-causing variants in critically ill infants and children. By interrogating the coding regions of thousands of genes simultaneously, WES is more comprehensive than sequential single-gene testing and is particularly suited to neonates with suspected monogenic disorders. Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing has been associated with a high diagnostic yield [6], and newborn sequencing programmes have highlighted the need for careful interpretation of results [7]. An early genomic diagnosis can guide treatment, inform prognosis, support decisions on escalation or redirection of care, avoid unnecessary investigations and enable recurrence-risk counselling. Randomised and observational studies have demonstrated clinically meaningful changes in management following rapid sequencing in seriously ill infants and children [8,9], and rapid whole-genome sequencing (rWGS) has been associated with shorter diagnostic pathways, lower resource use, reduced morbidity and lower hospitalisation costs [10,11].

Genome sequencing has influenced clinical decision-making in acutely ill infants with suspected genetic disease, and both targeted neonatal gene panels and rWGS have been shown to aid diagnosis in this population [12,13]. rWGS has also proved clinically useful in highly selected groups such as neonates receiving extracorporeal membrane oxygenation [14], and Helm et al. have proposed standardised criteria to guide patient selection for genomic testing in the NICU [15]. Despite this growing international evidence, data from Indian NICUs remain scarce. Diagnostic yield and variant spectrum may differ between populations because of consanguinity, regional genetic variation and a higher frequency of autosomal recessive disorders. Variant interpretation in rare disease often requires multidisciplinary input and, at times, functional validation [16], and recent advances in trio-exome sequencing and precision medicine underline the need for population-specific data on genotype–phenotype correlation and clinical utility [17,18].

We therefore aimed to determine the diagnostic yield of WES in neonates with suspected genetic disorders admitted to a tertiary care NICU in South India, and to describe the spectrum and classification of detected variants, genotype–phenotype correlations and the impact of WES on clinical management and counselling.

Methods

Study design and setting

This prospective observational study was conducted in the NICU of the Department of Neonatology, Madurai Medical College, Madurai, Tamil Nadu, India, over 12 months from September 2024 to August 2025. Neonates were enrolled by convenience sampling on the basis of clinical suspicion of an underlying genetic disorder, and institutional clinical protocols were followed throughout.

Participants

Neonates admitted to the NICU who underwent WES for a strong clinical suspicion of a genetic disorder were eligible. Indications for WES were syndromic or dysmorphic features, unexplained neurological dysfunction or encephalopathy, multisystem involvement, suspected inborn error of metabolism (IEM), and atypical chronic illness not explained by conventional investigations. Neonates in whom routine evaluation established a non-genetic aetiology, those with chromosomal abnormalities identified by conventional cytogenetic testing (karyotyping or fluorescence in situ hybridisation), and those whose parents did not consent to genomic testing were excluded. All enrolled neonates underwent a detailed clinical assessment before genomic testing.

Ethics

The study was approved by the Institutional Ethics Committee of Madurai Medical College (R. No. 003772/IEC/2026-09). Written informed consent was obtained from the parents or legal guardians of all participants before genomic testing.

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Clinical evaluation and sample collection

All neonates underwent detailed clinical evaluation and routine laboratory and radiological investigations to exclude common non-genetic causes. Once a genetic aetiology was suspected, approximately 2 mL of peripheral venous blood was collected aseptically in an EDTA tube and sent to an accredited reference laboratory for sequencing and analysis. Phenotypic details, family history and parental consanguinity were recorded and submitted with each sample.

Phenotyping and variant interpretation

Detailed phenotypic information was coded using Human Phenotype Ontology (HPO) terms to enable phenotype-driven interpretation [19]. Variants were classified as pathogenic (P), likely pathogenic (LP), variant of uncertain significance (VUS) or benign/likely benign according to the 2015 American College of Medical Genetics and Genomics–Association for Molecular Pathology (ACMG-AMP) guidelines [20]; tests with no reportable variant were recorded as negative. Only P/LP variants with good genotype–phenotype concordance were considered diagnostic. Where variants were reported in more than one gene, only phenotype-concordant P/LP variants were regarded as diagnostic, and secondary variants and VUS were considered non-diagnostic.

Genetic and genotype–phenotype analysis

For each diagnostic variant, zygosity (homozygous, heterozygous, compound heterozygous or copy-number variant [CNV] deletion) and mode of inheritance (autosomal recessive, autosomal dominant, X-linked or multilocus/combined) were recorded. Genotype–phenotype correlation was assessed by the extent to which the identified variant(s) explained the neonate's clinical presentation.

Outcomes

The primary outcome was diagnostic yield, defined as the number of neonates with a diagnostic P/LP variant divided by the total number sequenced. Secondary outcomes were the variant spectrum, inheritance and zygosity patterns, diagnostic yield by clinical phenotype, and clinical utility. A result was considered clinically actionable if it directly led to a change in treatment, clarified prognosis or altered follow-up planning, or informed genetic counselling regarding recurrence risk and future pregnancies.

Statistical analysis

Demographic data, phenotype categories, genes involved, variant classification, inheritance pattern and zygosity were extracted from genomic reports and clinical records. Data were analysed using descriptive statistics [IBM SPSS Statistics version 26.0]. Categorical variables are presented as frequencies and percentages and continuous variables as medians with interquartile ranges (IQR). The 95% confidence interval (CI) for the overall diagnostic yield was calculated using the Wilson score method.

Results

Baseline characteristics

Fifty-two neonates with suspected genetic disorders underwent WES during the study period (26 male, 26 female). The median age at sampling was 19 days (IQR 10.5–28) and the median turnaround time for WES was 21 days (IQR 20–24.5). The most frequent indications were syndromic/dysmorphic presentation (15, 28.8%), unexplained encephalopathy (12, 23.1%) and multisystem involvement (10, 19.2%) (Table 1).

Diagnostic yield and variant classification

P/LP variants were identified in 25 of 52 neonates, giving an overall diagnostic yield of 48.1% (95% CI 35.1–61.3). Pathogenic variants were found in 18 (34.6%) and likely pathogenic variants in 7 (13.5%) neonates. The remaining 27 (51.9%) had non-diagnostic results: a VUS in 13 (25.0%) and a negative result in 14 (26.9%) (Figure 1).

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Table 1. Baseline characteristics and clinical indications of the study population (N = 52)

Characteristic	Value
Sex, n (%)	
Male	26 (50.0)
Female	26 (50.0)
Age at sampling, days, median (IQR)	19 (10.5–28)
WES turnaround time, days, median (IQR)	21 (20–24.5)
Clinical indication for WES, n (%)	
Syndromic/dysmorphic presentation	15 (28.8)
Unexplained encephalopathy	12 (23.1)
Multisystem involvement	10 (19.2)
Suspected metabolic disorder/IEM	8 (15.4)
Atypical chronic illness	7 (13.5)

IEM, inborn error of metabolism; IQR, interquartile range; WES, whole-exome sequencing.

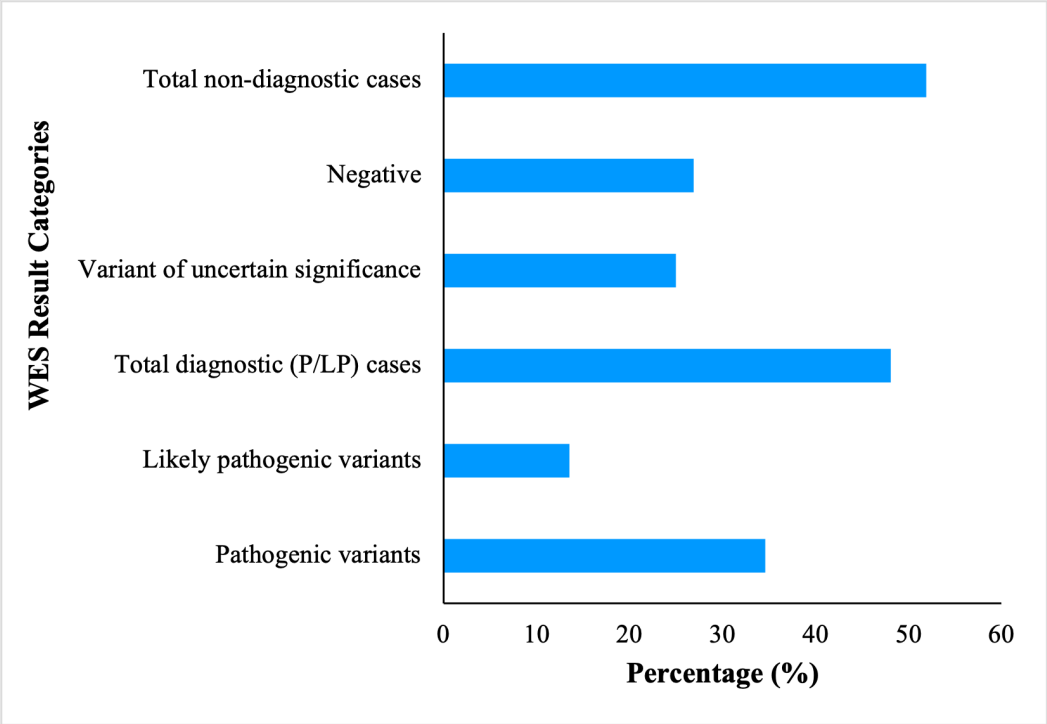


Figure 4. *FDistribution of whole-exome sequencing result categories among neonates with suspected genetic disorders (N = 52).*

Inheritance pattern and zygosity

Among the 25 diagnosed neonates, autosomal recessive inheritance was most common (12, 48.0%), followed by autosomal dominant (7, 28.0%) and multilocus/combined inheritance (5, 20.0%); one neonate (4.0%) had an X-linked disorder. Homozygous variants predominated (13, 52.0%), followed by heterozygous variants (10, 40.0%). Pathogenic CNV deletions were found in two neonates (8.0%), and no compound heterozygous variants were detected (**Table 2**).

Table 2. Mode of inheritance and zygosity among neonates with a diagnostic result (n = 25)

Parameter	n	%
Mode of inheritance		
Autosomal recessive	12	48.0

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Parameter	n	%
Autosomal dominant	7	28.0
Multilocus/combined	5	20.0
X-linked	1	4.0
Zygosity		
Homozygous	13	52.0
Heterozygous	10	40.0
CNV deletion	2	8.0
Compound heterozygous	0	0

CNV, copy-number variant.

Diagnostic yield by clinical phenotype

Diagnostic yield varied by clinical presentation (**Table 3**; **Figure 2**). It was highest in neonates with multisystem involvement (8/10, 80.0%) and syndromic/dysmorphic presentations (10/15, 66.7%), intermediate in suspected metabolic disorders/IEM (3/8, 37.5%) and atypical chronic illness (2/7, 28.6%), and lowest in unexplained encephalopathy (2/12, 16.7%).

Table 3. Diagnostic yield of WES by clinical phenotype

Phenotype category	Neonates, n	P/LP, n	Diagnostic yield, %
Multisystem involvement	10	8	80.0
Syndromic/dysmorphic presentation	15	10	66.7
Suspected metabolic disorder/IEM	8	3	37.5
Atypical chronic illness	7	2	28.6
Unexplained encephalopathy	12	2	16.7
Total	52	25	48.1

IEM, inborn error of metabolism; P/LP, pathogenic/likely pathogenic; WES, whole-exome sequencing.

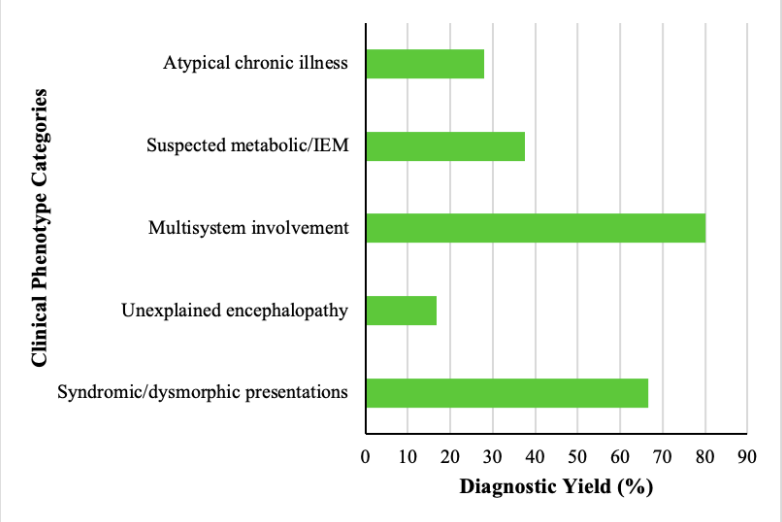


Figure 2. *Diagnostic yield of whole-exome sequencing by clinical phenotype category.*

Clinical utility

WES results had a direct clinical impact in all 25 diagnosed neonates (**Table 4**; **Figure 3**). Treatment was modified on the basis of the genetic diagnosis in 12 (48.0%), prognostic clarification led to modified follow-up in 7 (28.0%), and genetic

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counselling regarding recurrence risk was the principal impact in 6 (24.0%). No immediate clinical impact was recorded among the 27 neonates with non-diagnostic results.

Table 4. Clinical utility of WES among neonates with a diagnostic result (n = 25)

Primary clinical impact	n	%
Direct treatment modification	12	48.0
Follow-up/prognosis modification	7	28.0
Genetic counselling provided	6	24.0

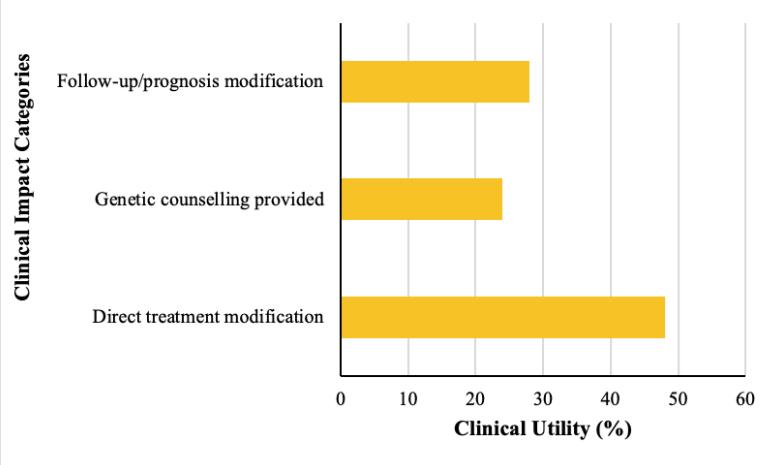


Figure 3. *Primary clinical impact of whole-exome sequencing among neonates with a diagnostic result (n = 25).*

Notable genomic findings

Three neonates had variants reported in more than one gene; in these cases, only the phenotype-concordant P/LP variant was considered diagnostic. The two clinically significant CNV deletions comprised a deletion associated with severe combined immunodeficiency and a deletion of the Prader–Willi/Angelman critical region at 15q11–q13. One critically ill neonate was found to have a homozygous pathogenic *CFTR* variant, confirming cystic fibrosis, with good genotype–phenotype correlation.

Discussion

In this prospective study of neonates with suspected genetic disorders in a South Indian tertiary NICU, WES established a molecular diagnosis in nearly half (48.1%) of those sequenced. Autosomal recessive inheritance and homozygous variants predominated, diagnostic yield was highest in infants with multisystem or syndromic presentations, and a genetic diagnosis altered treatment, follow-up or counselling in every diagnosed neonate.

Our diagnostic yield is comparable with previously published neonatal studies. Zhang et al. reported a yield of 52.9% with first-tier WES in NICU patients [21], and Olde Keizer et al. demonstrated high diagnostic utility of rapid exome sequencing as a first-tier test in neonates with suspected genetic disorders [22]. Wells et al. showed that rapid exome sequencing could be successfully implemented in routine care of critically ill infants [23]. Together, these findings support phenotype-directed genomic sequencing in the NICU.

Autosomal recessive inheritance (48%) and homozygous variants (52%) were the most common patterns in our cohort. This probably reflects population-specific genetic architecture and the contribution of consanguinity. A high prevalence of recessive disorders has also been reported in a large exome-sequencing cohort of neonates with suspected inherited disease [24], underscoring the importance of recessive monogenic disorders in neonatal illness in populations with high rates of consanguinity.

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Diagnostic performance varied considerably by phenotype, being highest for multisystem involvement and syndromic/dysmorphic presentations and lowest for unexplained encephalopathy. Robust clinical phenotyping and recognisable syndromic features probably strengthen genotype–phenotype correlation and improve the likelihood of identifying a causal variant, as observed in other sequencing studies of critically ill neonates and early infant deaths [25]. The low yield in encephalopathy may partly reflect non-genetic causes, such as hypoxic–ischaemic injury, that were not excluded with certainty, as well as genetic mechanisms that exome sequencing captures poorly. The small numbers within each phenotype category, however, mean that these subgroup estimates are imprecise.

WES also had substantial clinical utility. Nearly half of the diagnosed neonates had their treatment modified, and others benefited from prognostic clarification or genetic counselling. Similar effects of molecular diagnosis on management and individualised care planning have been reported in critically ill children [24,26], and early genomic diagnosis has been shown to aid decision-making in fetal and neonatal genetic disorders by allowing targeted interventions early in the disease course [27]. The median turnaround time of 21 days in our setting, however, is longer than that of rapid sequencing programmes, and shorter reporting times would be needed for results to inform acute decisions in the sickest infants.

The prospective design and phenotype-driven approach, including HPO-based variant interpretation and systematic assessment of genotype–phenotype correlation, are strengths of this study. The detection of clinically significant CNV deletions and monogenic disorders such as cystic fibrosis illustrates the breadth of conditions that WES can identify. The study also has limitations. It was conducted at a single centre with a small sample selected by convenience sampling, which limits generalisability and may have introduced selection bias. Trio sequencing was not available for all neonates, which may have limited the interpretation of VUS. Standard WES may not detect structural rearrangements, repeat expansions, mitochondrial DNA variants, epigenetic abnormalities or deep intronic variants, which may partly explain the proportion of non-diagnostic results. Finally, clinical utility was assessed in the short term only, and longer-term outcomes were not evaluated.

Larger multicentre studies incorporating rapid trio-based sequencing are needed to refine estimates of diagnostic yield and clinical utility in Indian NICUs. Reanalysis of non-diagnostic exomes, whole-genome sequencing, transcriptomics and functional validation may help resolve VUS and identify novel disease genes, and standardised criteria for genomic testing in the NICU could support wider implementation of precision neonatal medicine.

Conclusion

WES provided a molecular diagnosis in nearly half of neonates with suspected genetic disorders in a tertiary care NICU, with autosomal recessive and homozygous variants predominating. Diagnostic yield was highest in infants with multisystem involvement or syndromic presentations, and results frequently informed treatment, prognosis, follow-up and genetic counselling. WES should be considered early in the evaluation of neonates with a strong clinical suspicion of a genetic disorder.

References

1. Kingsmore SF, Henderson A, Owen MJ, Clark MM, Hansen C, Dimmock D, et al. Measurement of genetic diseases as a cause of mortality in infants receiving whole genome sequencing. NPJ Genom Med. 2020;5:49.
2. Kingsmore SF, Cole FS. The role of genome sequencing in neonatal intensive care units. Annu Rev Genomics Hum Genet. 2022;23:427–48.
3. Chad L, Anderson J, Cagliero D, Hayeems RZ, Ly LG, Szuto A. Rapid genetic testing in pediatric and neonatal critical care: a scoping review of emerging ethical issues. Hosp Pediatr. 2022;12(10):e347–59.
4. D'Gama AM, Walsh CA. Somatic mosaicism and neurodevelopmental disease. Nat Neurosci. 2018;21(11):1504–14.
5. Guo H, Li Y, Shen L, Wang T, Jia X, Liu L, et al. Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Sci Adv. 2019;5(9):eaax2166.
6. Gubbels CS, VanNoy GE, Madden JA, Copenheaver D, Yang S, Wojcik MH, et al. Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genet Med. 2020;22(4):736–44.

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7. Ceyhan-Birsoy O, Murry JB, Machini K, Lebo MS, Yu TW, Fayer S, et al. Interpretation of genomic sequencing results in healthy and ill newborns: results from the BabySeq Project. Am J Hum Genet. 2019;104(1):76–93.

8. Dimmock DP, Clark MM, Gaughran M, Cakici JA, Caylor SA, Clarke C, et al. An RCT of rapid genomic sequencing among seriously ill infants results in high clinical utility, changes in management, and low perceived harm. Am J Hum Genet. 2020;107(5):942–52.

9. Freed AS, Candadai SVC, Sikes MC, Thies J, Byers HM, Dines JN, et al. The impact of rapid exome sequencing on medical management of critically ill children. J Pediatr. 2020;226:202–12.

10. Diaby V, Babcock A, Huang Y, Moussa RK, Espinal PS, Janvier M, et al. Real-world economic evaluation of prospective rapid whole-genome sequencing compared to a matched retrospective cohort of critically ill pediatric patients in the United States. Pharmacogenomics J. 2022;22(4):223–9.

11. Farnaes L, Hildreth A, Sweeney NM, Clark MM, Chowdhury S, Nahas S, et al. Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. NPJ Genom Med. 2018;3:10.

12. NICUSeq Study Group; Krantz ID, Medne L, Weatherly JM, Wild KT, Biswas S, et al. Effect of whole-genome sequencing on the clinical management of acutely ill infants with suspected genetic disease: a randomized clinical trial. JAMA Pediatr. 2021;175(12):1218–26.

13. Maron JL, Kingsmore S, Gelb BD, Vockley J, Wigby K, Bragg J, et al. Rapid whole-genomic sequencing and a targeted neonatal gene panel in infants with a suspected genetic disorder. JAMA. 2023;330(2):161–9.

14. Amin MD, Wigby K, Suttner D, Niemi AK, Guidugli L, Carroll J. Clinical utility of rapid whole genome sequencing in neonatal patients receiving extracorporeal membrane oxygenation (ECMO). J Perinatol. 2025;45(4):495–9.

15. Helm BM, Hays M, Swaggart KA, Parker CB, Thomas V, Ware SM, et al. Standardized criteria for genomic testing in the NICU. Pediatrics. 2025;156(1):e2024069591.

16. Baldridge D, Wangler MF, Bowman AN, Yamamoto S, Schedl T, Pak SC, et al. Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. Orphanet J Rare Dis. 2021;16(1):206.

17. Lan X, Tang X, Weng W, Xu W, Song X, Yang Y, et al. Diagnostic utility of trio-exome sequencing for children with neurodevelopmental disorders. JAMA Netw Open. 2025;8(3):e251807.

18. Kansal R. Rapid whole-genome sequencing in critically ill infants and children with suspected, undiagnosed genetic diseases: evolution to a first-tier clinical laboratory test in the era of precision medicine. Children (Basel). 2025;12(4):429.

19. Köhler S, Gargano M, Matentzoglu N, Carmody LC, Lewis-Smith D, Vasilevsky NA, et al. The Human Phenotype Ontology in 2021. Nucleic Acids Res. 2021;49(D1):D1207–17.

20. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015;17(5):405–24.

21. Zhang R, Cui X, Zhang Y, Ma H, Gao J, Zhang Y, et al. Whole-exome sequencing as the first-tier test for patients in neonatal intensive care unit: a Chinese single-center study. BMC Pediatr. 2024;24(1):351.

22. Olde Keizer RA, Marouane A, Kerstjens-Frederikse WS, Deden AC, Lichtenbelt KD, Jonckers T, et al. Rapid exome sequencing as a first-tier test in neonates with suspected genetic disorder: results of a prospective multicenter clinical utility study in the Netherlands. Eur J Pediatr. 2023;182(6):2683–92.

23. Wells CF, Boursier G, Yauy K, Ruiz-Pallares N, Mechin D, Ruault V, et al. Rapid exome sequencing in critically ill infants: implementation in routine care from French regional hospital's perspective. Eur J Hum Genet. 2022;30(9):1076–82.

24. Yang L, Wei Z, Chen X, Hu L, Peng X, Wang J, et al. Use of medical exome sequencing for identification of underlying genetic defects in NICU: experience in a cohort of 2303 neonates in China. Clin Genet. 2022;101(1):101–9.

25. Yang L, Liu X, Li Z, Zhang P, Wu B, Wang H, et al. Genetic aetiology of early infant deaths in a neonatal intensive care unit. J Med Genet. 2020;57(3):169–77.

26. Sanford EF, Clark MM, Farnaes L, Williams MR, Perry JC, Ingulli EG, et al. Rapid whole genome sequencing has clinical utility in children in the PICU. Pediatr Crit Care Med. 2019;20(11):1007–20.

27. Wojcik MH, Reimers R, Poorvu T, Agrawal PB. Genetic diagnosis in the fetus. J Perinatol. 2020;40(7):997–1006. Figure Legends Figure 1. Distribution of whole-exome sequencing result categories among neonates with suspected genetic disorders (N = 52). Figure 2. Diagnostic yield of whole-exome sequencing by clinical phenotype category Figure 3. Primary clinical impact of whole-exome sequencing among neonates with a diagnostic result (n = 25).

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