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CLINICAL WHOLE GENOME SEQUENCING ON MGI DNBSEQ-T7 AND DNBSEQ-G400 PLATFORMS: LOW-PASS, STANDARD, AND DEEP WGS APPLICATIONS

Review article

Budhyanto V.[1], Jurgens S.[2], Cicilia C.O.[3,] *
[1] ORCID : 0009-0000-2176-6417;
[3] ORCID : 0009-0003-3231-4629;
[1, 2, 3] Satriabudi Dharma Setia Foundation, Jakarta, Indonesia

* Corresponding author (cindy.o.cicilia[at]dharma.or.id)

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Abstract

Background. Whole genome sequencing (WGS) has evolved into a comprehensive diagnostic approach capable of supporting clinical needs spanning reproductive medicine, pediatrics, oncology, infectious disease and population genomics. Although Illumina platforms still dominate clinical sequencing, the DNBSEQ instruments from MGI Tech which employ DNA nanoball (DNB) technology and combinatorial probe-anchor synthesis (cPAS) chemistry have obtained regulatory approval in several countries and are increasingly used in large-scale clinical sequencing programs.

Scope. This review evaluates the clinical application of WGS on the DNBSEQ-T7 and DNBSEQ-G400 platforms across three coverage-depth tiers: low-pass (~0.1×–5×), standard (~30×), and deep (>60×–100×). Evidence was compiled from indexed scientific publications, manufacturer technical documentation and independent benchmarking studies published between 2018 and 2026.

Key findings. Comparative studies using NIST/Genome in a Bottle (GIAB) reference materials and clinical specimens show that DNBSEQ-T7 and DNBSEQ-G400 deliver single-nucleotide variant (SNV) and small-indel detection performance comparable to the Illumina NovaSeq 6000. Reported concordance exceeds 0.99 for germline SNVs and 0.80 for structural variants when equivalent bioinformatics pipelines are used. Validated clinical applications include cfDNA-based non-invasive prenatal testing (NIPT), preimplantation genetic testing (PGT), rapid WGS in critically ill pediatric patients, tumour-normal analysis for somatic variant detection and homologous recombination deficiency (HRD), and large-scale population genomics. Residual performance gaps concentrate in high-GC regions, homopolymers ≥10 bp and dinucleotide repeats, and are substantially closed by deep-learning callers retrained on DNBSEQ chemistry.

Conclusion. DNBSEQ-T7 and DNBSEQ-G400 are viable short-read WGS platforms for clinical use across a range of sequencing depths, with the strongest validation in reproductive medicine, rare-disease diagnosis and oncology. Routine implementation requires bioinformatics validation tailored to platform-specific characteristics and compliance with internationally recognized laboratory accreditation standards.

Keywords: whole genome sequencing, DNBSEQ-T7, DNBSEQ-G400, MGI Tech, precision oncology.

КЛИНИЧЕСКОЕ ПОЛНОГЕНОМНОЕ СЕКВЕНИРОВАНИЕ НА ПЛАТФОРМАХ MGI DNBSEQ-T7 И DNBSEQ-G400: ПРИМЕНЕНИЕ МЕТОДОВ НИЗКОПОКРЫВНОГО (LOW-PASS), СТАНДАРТНОГО И ГЛУБОКОГО (DEEP) ПОЛНОГЕНОМНОГО СЕКВЕНИРОВАНИЯ

Обзор

Будьянто В.[1], Юргенс С.[2], Сисилия С.О.[3,] *
[1] ORCID : 0009-0000-2176-6417;
[3] ORCID : 0009-0003-3231-4629;
[1, 2, 3] Фонд Сатриабуди Дхарма Сетиа, Джакарта, Индонезия

* Корреспондирующий автор (cindy.o.cicilia[at]dharma.or.id)

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Аннотация

Актуальность. Полногеномное секвенирование (WGS) превратилось в комплексный диагностический метод, отвечающий потребностям таких областей, как репродуктивная медицина, педиатрия, онкология, инфекционные заболевания и популяционная геномика. Хотя платформы Illumina по-прежнему занимают доминирующее положение в сфере клинического секвенирования, системы DNBSEQ от компании MGI Tech, использующие технологию ДНК-наношаров (DNB) и химию комбинаторного синтеза с использованием зондов и якорей (cPAS), получили регуляторное одобрение в ряде стран и все чаще применяются в масштабных программах клинического секвенирования.

Обзор. В данном обзоре оценивается клиническое применение WGS на платформах DNBSEQ-T7 и DNBSEQ-G400 при трех уровнях глубины покрытия: низком (~0,1×–5×), стандартном (~30×) и высоком (>60×–100×). Материалы для обзора были собраны на основе индексированных научных публикаций, технической документации производителя и независимых сравнительных исследований, опубликованных в период с 2018 по 2026 год.

Основные результаты. Сравнительные исследования с использованием референсных материалов NIST/Genome in a Bottle (GIAB) и клинических образцов показывают, что платформы DNBSEQ-T7 и DNBSEQ-G400 обеспечивают

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точность выявления однонуклеотидных вариантов (SNV) и малых инсерций/делеций (indel), сопоставимую с показателями системы Illumina NovaSeq 6000. При использовании эквивалентных биоинформатических алгоритмов обработки данных (пайплайнов) показатель конкордантности превышает 0,99 для герминальных SNV и 0,80 для структурных вариантов. К числу валидированных областей клинического применения относятся: неинвазивное пренатальное тестирование (NIPT) на основе внеклеточной ДНК (cfDNA), преимплантационное генетическое тестирование (PGT), экспресс-WGS у детей в критическом состоянии, анализ «опухоль-норма» для выявления соматических вариантов и дефицита гомологичной рекомбинации (HRD), а также масштабные исследования в области популяционной геномики. Сохраняющиеся различия в качестве данных наблюдаются преимущественно в участках с высоким содержанием GC-пар, гомополимерных последовательностях длиной ≥10 п.н. и динуклеотидных повторах; однако эти расхождения в значительной степени устраняются при использовании алгоритмов вызова вариантов на основе глубокого обучения, переобученных с учетом специфики химии DNBSEQ.

Заключение. Платформы DNBSEQ-T7 и DNBSEQ-G400 являются эффективными системами для WGS с использованием коротких прочтений в клинической практике при различных уровнях глубины секвенирования; наиболее надежные данные об их эффективности получены в таких сферах, как репродуктивная медицина, диагностика редких заболеваний и онкология. Внедрение этих систем в рутинную практику требует валидации биоинформатических методов с учетом специфических характеристик платформ, а также соблюдения международно признанных стандартов аккредитации лабораторий.

Ключевые слова: полногеномное секвенирование, DNBSEQ-T7, DNBSEQ-G400, MGI Tech, прецизионная онкология.

Introduction

Whole genome sequencing has evolved from a purely research method into a clinical-grade assay that unifies the analysis of SNVs, small indels, copy-number variants (CNVs), structural variants (SVs), mitochondrial variants and increasingly pharmacogenomics and repeat expansions within a single test [10]. Compared with whole-exome sequencing and disease-targeted panels, which will be addressed in a separate review, WGS offers higher and more uniform coverage of clinically relevant genes, the ability to detect deep-intronic, regulatory and structural variants, and the opportunity for reanalysis as knowledge accumulates. Across published reports of rapid WGS in the NICU, 36–73% of infants tested received a molecular diagnosis, and management changes occurred in up to 72% of diagnosed patients [11]. Petrikin et al. (NSIGHT1) reported a median time to diagnosis of 13 days (range 1–84) with rapid WGS, compared with 107 days (range 21–429) using standard testing in critically ill infants [12], [13].

Over the past two decades, clinical short-read sequencing has been dominated by Illumina's sequencing-by-synthesis (SBS) chemistry. MGI Tech, a Shenzhen-based subsidiary of the BGI Group, has emerged as the most prominent alternative high-quality short-read manufacturer, leveraging the DNB and cPAS technology inherited from Complete Genomics. In its September 2025 T7+ launch release, MGI stated that more than 400 DNBSEQ-T7 units have been installed at over 200 leading institutions worldwide, including the Genomics Thailand Initiative and the largest genomic sequencing project in Brazil [14]. The DNBSEQ-T7+ can now extend ultra-high throughput to more than 14 Tb per 24 hours with a claimed Q40 base accuracy according to vendor data [14]; these are manufacturer-stated maxima obtained with fully loaded flow cells and internal standard libraries, not routine clinical yields. Independent benchmark studies consistently show concordance with the Illumina NovaSeq 6000 within clinically acceptable limits for most variant classes [1], [2], [3], [4], although a 2024 Illumina white paper stated that the NovaSeq X with DRAGEN v4.4 produced 8–12 times fewer SNV/indel errors than DNBSEQ-T7 with MegaBOLT v2.4 when tested against the NIST v4.2.1 truth set, particularly in difficult regions [15].

This review is intended to assist clinicians, clinical molecular geneticists and laboratory directors who need to evaluate platform choices and coverage-depth strategies. It covers sequencing strategies that use the entire human genome or, in the context of liquid biopsy and NIPT, the entire pool of cell-free DNA as the substrate for library preparation, without prior selection of specific genomic regions. Three coverage-depth tiers are examined:

Low-pass WGS (~0.1×–~5×): for calling chromosome- and segmental-level aneuploidy and CNVs, and for genotype imputation against a reference panel.

Standard human WGS (~30×): for germline and tumour/normal variant discovery in constitutional disease and many cancer types.

Deep WGS (>60×, generally 100× and above, sometimes >200×): for tumour heterogeneity analysis, subclonal variant calling, ctDNA detection, mosaicism and minimal residual disease (MRD).

After establishing the technical fundamentals and the concordance evidence base, each tier is reviewed with the depth of discussion calibrated to the strength of the published evidence for the MGI/DNBSEQ platforms specifically. Cross-cutting topics, bioinformatics, ACMG/AMP interpretation, the regulatory landscape and ethical, legal and social issues (ELSI) are addressed thereafter.

Methods

This is a narrative, platform-focused critical review. It was not designed as a systematic review and no meta-analysis was attempted; nevertheless, to make the evidence base reproducible and to limit selective citation, the search strategy, the inclusion and exclusion criteria and the rules used to choose between competing studies are stated explicitly below. Evidence was compiled from indexed scientific publications, independent benchmarking studies, manufacturer technical documentation and a small number of clearly flagged preprints published between 2018 and 2026.

***Search strategy*.** Literature searches were performed in PubMed/MEDLINE, Scopus and Google Scholar, with bioRxiv and medRxiv searched separately for preprints, for material published from January 2018 onward (last search May 2026). Platform terms ("DNBSEQ", "DNBSEQ-T7", "DNBSEQ-G400", "DNBSEQ-G99", "MGISEQ", "BGISEQ", "MGI Tech",

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"Complete Genomics", "DNA nanoball", "cPAS") were combined with application terms ("whole genome sequencing", "WGS", "low-pass", "low-coverage", "non-invasive prenatal testing", "NIPT", "preimplantation genetic testing", "rapid genome sequencing", "tumour", "ctDNA", "minimal residual disease", "metagenomic", "population genomics") and with comparator terms ("Illumina", "NovaSeq", "HiSeq", "Ultima", "benchmark", "concordance", "Genome in a Bottle"). Reference lists of included articles and of recent reviews were hand-searched, and the regulatory-news archive of GenomeWeb and the websites of MGI Tech, Complete Genomics, Illumina and Ultima Genomics were searched for instrument specifications, regulatory notices and white papers.

Inclusion and exclusion criteria. Included were:

a) primary studies, benchmarks or clinical validation reports in which at least one MGI/BGI DNB-based instrument (BGISEQ-500, MGISEQ-2000, DNBSEQ-G400, DNBSEQ-G99, DNBSEQ-T7/T7+) was used for human whole-genome, cell-free DNA or shotgun metagenomic sequencing without prior target enrichment;

b) cross-platform comparisons that included at least one such instrument together with an Illumina or Ultima comparator;

c) landmark clinical studies performed on other platforms (for example NSIGHT1, the 100,000 Genomes pilot, PCAWG and the Mackie NIPT meta-analysis) where they establish the clinical benchmark against which DNBSEQ data are judged, in which case the platform used is stated explicitly;

d) regulatory, specification and cost information from manufacturers or regulators, reported as vendor-stated.

Foundational papers published before 2018 (Drmanac et al. 2010; Liao et al. 2014; Zhang et al. 2015; Griffith et al. 2015; Mackie et al. 2017) were retained where they describe the underlying chemistry or remain the accepted clinical benchmark. Excluded were studies of targeted panels and whole-exome sequencing (addressed in a separate review), non-human applications, RNA-seq and single-cell studies except where they inform cross-platform equivalence, conference abstracts without accessible data, and articles not available in English.

Study selection and weighting. Where several studies addressed the same question, preference was given, in order, to studies with an independent truth set (NIST/GIAB or orthogonal clinical confirmation), the largest cohort, the most recent instrument generation, and studies in which identical bioinformatics were applied to every platform compared. Studies were omitted, rather than cited selectively, when the sequencing platform could not be identified from the methods, when the comparison relied on different pipelines for different platforms without a common-pipeline arm (the two vendor white papers are the deliberate exception and are discussed critically in Section 3.3), or when the results were superseded by a later report from the same group. No formal risk-of-bias scoring was applied; instead, commercial authorship and preprint status are flagged at the point of citation. Sources were weighted in the following order:

1) peer-reviewed primary studies and independent cross-platform benchmarks;

2) preprints, treated as provisional and identified as such at the point of citation;

3) vendor-authored comparative benchmarking from either MGI/Complete Genomics or Illumina, used only where the underlying data and pipeline are specified, with commercial interest explicitly acknowledged [15], [26];

4) product specifications, press releases and regulatory news, reported as vendor- or reporter-stated and not independently validated.

Clinical applications are organized by coverage-depth tier, and within each tier the strength of the DNBSEQ-specific evidence is distinguished from evidence generated on other platforms for which only analytical equivalence has been demonstrated.

Results and Discussion

3.1. DNB and cPAS chemistry

DNBSEQ chemistry differs from Illumina SBS in two principal respects. First, clonal template formation uses rolling-circle replication (RCR) of single-stranded circular library molecules to generate DNA nanoballs, each containing ~300–500 tandem copies of the original insert without PCR cluster generation by bridge amplification. Because all RCR copies derive directly from the original template rather than from copies of copies, the accumulation of inter-copy errors is suppressed, and there is no clonal PCR error on the formed array. Second, sequencing uses combinatorial probe–anchor synthesis (cPAS): fluorescently labelled, nucleotide-conjugated probes with reversible termination anchored near the read position, rather than direct enzymatic extension of a primer. DNBs are loaded onto a patterned silicon flow cell with sub-pixel registration, yielding high signal density and low index hopping [5], [16], [63].

The practical advantages of this architecture are a low duplication rate, absence of index swapping, and stable base quality throughout the run. The principal drawbacks are a shorter maximum read length (PE150 on the T7/G400) and an error spectrum that differs from SBS, with vendor-specific bias in certain homopolymer and GC-extreme contexts [2].

Library preparation, input quality and the DNB workflow. The sequencer is only one determinant of DNBSEQ performance; the route by which a clinical sample becomes a DNA nanoball matters as much, and the consequences are most visible in low-input and degraded specimens. DNBSEQ libraries must be denatured and circularized into single-stranded circles before rolling-circle replication, which adds a circularization and exonuclease-digestion step to any adapter-ligated library and makes the workflow sensitive to adapter-dimer and short-fragment carry-over. The single-stranded circular (ssCir) library is then quantified and a defined molar amount is converted to DNBs, which are loaded onto the patterned array at a fixed target occupancy. Because the array positions are fixed, both under-loading (empty spots and lost output) and over-loading (signal interference between adjacent spots) reduce usable yield, so accurate quantification of the ssCir library and of every sample within a pool is required for balanced demultiplexing. MGI's own application data report per-sample split-rate deviations of about 1% when eight libraries were pooled without balancing on the DNBSEQ-G99 [58], but larger imbalances can leave individual samples below the depth required for reporting and are a common cause of re-runs in clinical laboratories.

Two library routes are available. PCR-free kits (for example the MGIEasy FS PCR-Free and DNBSEQ Fast PCR-FREE FS sets) require approximately 25–900 ng of intact genomic DNA and, combined with the PCR-free clonal amplification of

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RCR, give a workflow with no amplification step at all [61]; this is the route used in the vendor benchmarks discussed in Section 3.3 and in most 30× germline WGS, and it is the configuration that delivers the low duplication rate, low GC bias and favourable indel accuracy associated with DNBSEQ [26], [61]. PCR-amplified kits (for example the MGIEasy Fast FS set V2.0) accept 1 ng–1 µg and are the only option for FFPE tissue, low-input biopsies, whole-genome-amplified PGT biopsies and most cell-free DNA [62]; they reintroduce polymerase errors, PCR duplicates and GC-dependent amplification bias, so that the low duplication rate quoted for DNBSEQ then applies only to the sequencing step and not to the library. The magnitude of the effect is illustrated by the Illumina white paper itself, in which TruSeq PCR-free libraries converted for DNBSEQ with an adapter-conversion PCR produced about five-fold more indel errors and two-fold more total errors than native PCR-free DNBSEQ libraries run on the same instrument [15]. FFPE-derived DNA adds fragmentation and cytosine-deamination (C>T) artefacts that are platform-independent but are amplified by additional PCR cycles and require uracil-DNA-glycosylase treatment and variant-allele-frequency thresholds to be set during validation; the DNBSEQ-T7 has been shown to give mtDNA variant results equivalent to the NovaSeq 6000 from FFPE as well as blood and saliva [4], but systematic nuclear-genome FFPE comparisons between DNBSEQ and Illumina at whole-genome scale remain sparse. For cell-free DNA, the short (~167 bp) fragments are well suited to PE100 circularized libraries, and the absence of index hopping on DNB arrays [63] is an advantage when hundreds of NIPT or ctDNA libraries are multiplexed; however, the circularization efficiency of very short or damaged fragments and the input-dependent duplicate rate should be established during validation for each specimen type, as the AMP/CAP guidelines recommend [65]. In practical terms, laboratories should validate each combination of specimen type, library kit and sequencer as a distinct assay and should not extrapolate PCR-free germline performance to PCR-amplified FFPE or liquid-biopsy workflows.

3.2. The DNBSEQ instrument family and comparator platforms

***The DNBSEQ family: T7, G400 and G99*.** The three DNBSEQ instruments relevant to clinical laboratories occupy distinct throughput tiers (Table 2). The DNBSEQ-T7 (four flow cells, ~5,800 million reads per flow cell, up to ~7 Tb per run) is the production-scale instrument for 30× and deeper human WGS, and the DNBSEQ-G400 (two flow cells, ~55 Gb–1.4 Tb per run depending on flow cell and read length) is the mid-throughput workhorse for NIPT, PGT, products-of-conception analysis, exomes and smaller WGS batches [58]. The DNBSEQ-G99, launched in September 2022, is a benchtop (~140 kg) mid-to-low-throughput instrument with two independently operable flow cells, 40–200 million reads per flow cell and 8–240 Gb per run depending on the FCS, FCL or FCU flow cell and the read length; its defining feature is speed, with PE150 completed in about 12 hours and SE100/PE50 in about 5 hours on an FCL flow cell, and with Q40 quality claimed for ≥85% of bases using StandardMPS 2.0 reagents [58]. The manufacturer positions it for targeted oncology panels, infectious-disease and metagenomic sequencing, methylation panels, small genomes, 16S and low-pass WGS, and recommends approximately 8–20 NIPT or PGS samples per flow cell at ~10 million SE50 reads each [58]. It obtained NMPA medical-device registration in September 2023, permitting clinical use in China [59], and carries a CE mark under the EU IVDR as a self-declared Class A (low-risk) instrument, a route that involves no notified body and that does not itself certify any clinical assay run on the instrument [60]. For the purposes of this review the G99 is therefore relevant to the low-pass tier (Section 3.4) and to metagenomic NGS (Section 3.5): even the largest FCU flow cell at PE150 (60–120 Gb) yields at most one 30× human genome, so standard-depth and deep WGS remain the domain of the G400 and T7. Its rapid turnaround and small footprint make it the natural DNBSEQ entry point for hospital molecular laboratories that currently run PCR-based tests and wish to add sequencing-based NIPT, PGT-A, mNGS or tumour panels without the batching constraints of a high-throughput instrument; the CE-IVD somatic HRD workflow being developed by MGI with SeqOne and Agilent is designed for this platform (Section 3.5) [40]. It should be stressed that, unlike the T7 and G400, the G99 has no peer-reviewed clinical WGS validation literature yet; all performance figures quoted here are manufacturer-stated.

***Illumina sequencing-by-synthesis*.** Illumina relies on bridge amplification of adapter-ligated library molecules on a patterned (NovaSeq) or random (older HiSeq) flow cell, followed by SBS with cyclic reversible terminators. The NovaSeq X, launched in 2023, uses XLEAP-SBS chemistry delivering up to ~16 Tb per run with a dual flow cell and supporting PE150 at Q40-grade base quality per vendor metrics. The NovaSeq 6000, now categorized as legacy, delivers up to ~6 Tb per run in a dual-flow-cell configuration. Illumina's principal strengths for clinical use are a mature regulatory footprint (the NovaSeq 6000Dx and NextSeq 550Dx hold CE-IVD/FDA certification in their respective configurations), a broad ecosystem of validated library kits, third-party variant callers and supporting software, and well-documented error and indel characteristics in difficult regions, with DRAGEN v4.4 and graph-based references improving callability [15], [17].

***Ultima Genomics 100*.** The UG 100 uses a silicon-wafer-based open-fluidics chemistry termed mostly natural sequencing-by-synthesis (mnSBS), in which a single nucleotide type is flowed per cycle with a small labelled fraction. Base identity at each flow is unambiguous, but homopolymer length must be inferred from incorporation-signal magnitude. Reads are single-end with an effective length of ~300 bp. The platform's principal appeal is cost: ~US$80–100 per 30× genome for reagents, with a capacity of ~10 billion reads per run [9], [18]. Independent benchmarks on NIST GIAB samples show SNV precision and recall comparable to the NovaSeq 6000 in non-masked regions. However, accuracy in homopolymer regions declines below 90% beyond 8 bp and drops further in the 11–20 bp range, a limitation with clinical impact, given that many medically relevant loci contain long homopolymer tracts [9]. Paired plus-minus sequencing (ppmSeq), which processes both original DNA strands simultaneously, achieves raw accuracy up to ~Q60 and enables whole-genome MRD applications (Labcorp Plasma Detect Genome MRD) with a reported limit of detection below 3 ppm [19], [20]. To date, UG 100 deployment in clinical settings remains limited and largely investigational, including proof-of-concept work for CSF metagenomics [21].

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Table 1 - Specifications and clinical attributes of current short-read WGS platforms

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Attribute	DNBSEQ-T7 (V3.0)	DNBSEQ-T7+ (2025)	DNBSEQ-G400	Illumina NovaSeq X Plus	Illumina NovaSeq 6000	Ultima UG 100
Chemistry	DNB + cPAS	DNB + cPAS (SM2.0)	DNB + cPAS	SBS (XLEAP)	SBS	Flow-based mnSBS, single-end
Maximum read length	PE150	PE150	PE150 (FCL); PE300 (specific kits)	PE150	PE150	~300 bp single-end
Maximum output / 24 h	Up to 7 Tb	>14 Tb	~1.08 Tb / run (~38 h)	Up to ~16 Tb / 48 h	Up to ~6 Tb / 44 h	~6 Tb / run (≤20 h)
30× genome capacity per year	~28,000	~33,600 (300-day model)	Mid-throughput, daily operation	20,000+	~7,500	Thousands
Run time (PE150)	22–24 h	<24 h	37–88 h	17–48 h	24–44 h	~20 h
Q30 (vendor data)	≥85% (typically >90%)	Q40 base accuracy	≥85%	Q40 level (XLEAP)	≥85%	Q≥60 (ppmSeq); varies in homopolymers
Cost per 30× WGS (reagents, vendor data)	~US$150	~US$100 ($1/Gb)	Higher	Vendor target ~US$200	Higher	~US$80–100
SNV F1 (GIAB, independent studies)	0.995–0.999 (equivalent pipeline)	n/a (new)	0.995–0.999	~0.999	~0.999	~0.998 (non-masked regions)
Indel F1 on long homopolymers	High (PE150)	Improving	High	High	High	<0.90 above 8 bp
Regulatory status (selected)	NMPA Class III (2020); CE-IVD (2022)	CE-IVD in process	NMPA; CE-IVD (2022)	FDA Dx in process	6000Dx: CE-IVD/FDA cleared	RUO; clinical pilot

Note: combined manufacturer data and independent reports; figures are approximate and depend on instrument version; throughput, run time and Q30/Q40 values are manufacturer-stated maxima for fully loaded flow cells and internal standard libraries; cost figures are vendor list or target prices for sequencing reagents only and exclude library preparation, instrument depreciation, service, labour, QC failures and re-runs, compute, storage and interpretation (see text); the DNBSEQ-G99 is not designed for standard-depth human WGS and is compared separately in Table 2

The throughput and cost figures in Table 1 require careful interpretation. Quoted maximum outputs (for example 7 Tb per T7 run or >14 Tb per day for the T7+) describe fully loaded flow cells sequenced with internal standard libraries; MGI's own specifications state that effective read numbers depend on sample type and library preparation and that Q30 performance is affected by library quality and insert size [58]. In routine clinical practice the deliverable yield per run is reduced by adapter and low-quality read filtering, duplicate removal, under-loading when clinical batch sizes do not fill a flow cell, library failures that must be repeated and occasional run failures; laboratories should therefore plan capacity on validated in-house yields rather than on nameplate output. Similarly, the cost figures quoted in Table 1 are vendor-stated reagent list or target prices per 30× genome (~US$1 per Gb for the T7+ [14]). They exclude DNA extraction and library preparation, instrument purchase or depreciation, service contracts, labour, quality control and re-run costs, compute, storage, variant interpretation and reporting, and the fixed costs of validation and accreditation. A microcosting study of Illumina-based clinical WGS in a UK National Health Service laboratory processing ~400 samples per year estimated £6,841 per cancer case (tumour plus germline) and £7,050 per rare-disease trio, with sequencing consumables accounting for 68–72% of the total and the remainder arising from staff, equipment and bioinformatics; the authors concluded that the cost of genome sequencing is underestimated if only sequencing costs are considered [57]. Comparable fully loaded costs for DNBSEQ workflows have not been published

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independently. Vendor per-genome figures should therefore be regarded as a lower bound on the consumables component rather than as the cost of a clinical test, and the cost advantage of DNBSEQ relative to Illumina is most pronounced precisely in high-volume, fully loaded settings such as NIPT and population programs and narrows at low sample volumes.

Table 2 - Clinical positioning of the DNBSEQ-G99 relative to the DNBSEQ-G400 and DNBSEQ-T7

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Attribute	NBSEQ-G99	DNBSEQ-G400	DNBSEQ-T7
Format	Benchtop (~140 kg); 2 independent flow cells	Floor-standing; 2 flow cells	Floor-standing; 4 flow cells
Reads per flow cell (vendor)	40 M (FCS), 80 M (FCL), 200 M (FCU)	300–1,800 M	~5,800 M
Output per run (vendor)	8–240 Gb	55–1,440 Gb	1–7 Tb
PE150 run time	~12 h (FCL); ~16 h (FCU)	37–88 h	22–24 h
Fastest mode	SE100/PE50 in ~5 h (FCL)	n/s	n/s
Maximum read length	PE300 (App-D FCL/FCU); SE400	PE150 (FCL); PE300 (specific kits)	PE150
Approx. 30× human genomes per flow cell (derived)	≤1 (FCU PE150)	~5 (FCL PE150)	~15–17
Regulatory status (selected)	NMPA registration (Sept 2023); CE mark under IVDR, Class A self-declared (2023)	NMPA; CE-IVD (2022)	NMPA Class III (2020); CE-IVD (2022); no FDA clearance
Vendor-recommended clinical applications	Targeted oncology panels; mNGS/pathogen detection; methylation panels; NIPT/PGS (~8–20 samples per flow cell); 16S; small genomes	NIPT (96–384 per flow cell); PGT; POC; WES; small-batch 30× WGS	30× and deep WGS at scale; population programs; high-volume NIPT
Role in this review's coverage tiers	Low-pass tier and mNGS only	Low-pass and standard tiers	Standard and deep tiers

Note: manufacturer-stated specifications taken from the MGI G99 brochure (version 20251201) [58] and the regulatory notices cited in the text [59], [60]; all caveats on nameplate throughput stated above apply. n/s = not stated in the source used; genome counts assume ~100 Gb of raw PE150 data per 30× genome and are derived from vendor read counts, not independently measured; G99 regulatory dates are from GenomeWeb reports

3.3. Concordance and validation: DNBSEQ versus Illumina

Multiple peer-reviewed studies demonstrate that DNBSEQ-platform WGS produces variant calls clinically equivalent to Illumina for most variant classes.

Korostin et al. (2020) compared the MGISEQ-2000 with the Illumina HiSeq 2500 for human WGS, finding read quality, mapping rate and concordance equivalent >99% for SNVs and >97% for indels, when the same pipeline was applied to both [22].

Patch, Nones, Kazakoff et al. (2018) performed cancer tumour/normal WGS on the BGISEQ-500 and Illumina HiSeq X Ten for three mesothelioma trios. Germline SNV concordance with SNP arrays exceeded 99% on both platforms; 86% of germline and 72% of somatic SNVs were identified by both platforms, with only 0.36% of SNV calls unique to the BGISEQ-500 [23].

Jeon et al. (2021) compared the NovaSeq 6000, MGISEQ-2000 and DNBSEQ-T7 on paired tumour/normal samples. The MGISEQ-2000 was most concordant with the NovaSeq 6000 for germline SNVs/indels, whereas the DNBSEQ-T7 was most concordant for somatic SNVs/indels [1].

Kim et al. (2021) compared seven short-read platforms such as BGISEQ-500, DNBSEQ-T7, HiSeq 2000/2500/4000/X10 and NovaSeq 6000 using the Korean Reference Genome. The MGI platforms showed higher SNP-array concordance than the HiSeq 2000/4000 and were equivalent to the NovaSeq 6000 on nearly all quality metrics [2].

Senabouth et al. (2020) showed that the MGISEQ-2000 and NovaSeq 6000 provide equivalent cell, UMI and gene detection in single-cell RNA-seq [24].

Feng et al. (2024) found equivalent mtDNA variant detection between the DNBSEQ-T7 and NovaSeq 6000 for blood, FFPE and saliva samples; for plasma cf-mtDNA, the NovaSeq 6000 captured a broader fragment-size range [4].

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Rao et al. (2025) applied 40 SV-calling tools across eight DNBSEQ and two Illumina WGS datasets. Inter-platform correlation reached >0.80 for SV count, size, precision and sensitivity, while DNBSEQ showed superior performance for small CNVs [3].

Conversely, the Illumina white paper (2024), comparing the NovaSeq X with DRAGEN v4.4 against the DNBSEQ-T7 with MegaBOLT v2.4, reported 8–12 times more SNV/indel errors in the MGI workflow against the NIST v4.2.1 benchmark, arising primarily from high-GC regions, homopolymers ≥10 bp and dinucleotide repeats [15]. This claim was contested by MGI/Complete Genomics, which subsequently released a cross-platform benchmark of the DNBSEQ-T7, T7+ and T1+ against PCR-free HG002 datasets from the Illumina NovaSeq 6000, Element AVITI and PacBio Onso, all subsampled to 30× effective depth and aligned with MegaBOLT (BWA-MEM2), and reported higher SNP and indel F-measures for all three DNBSEQ instruments than for the comparators (SNP F-measure 99.80% versus 99.57% for the NovaSeq 6000 against NIST v4.2.1) [26]. Both documents are vendor-authored; neither is decisive absent independent replication, and neither isolates the sequencing chemistry from the analysis pipeline, as discussed next.

Why neither vendor benchmark isolates the sequencing chemistry. Neither document compares the sequencers alone. In the Illumina study the Illumina arm was analyzed with DRAGEN v4.4, a proprietary hardware-accelerated pipeline with machine-learning recalibration and graph-reference options, whereas the MGI arm was analyzed with MegaBOLT v2.4.0 running either a GATK 4.1.8.1 HaplotypeCaller module or a DeepVariant module, with the MGI libraries prepared at a third-party core laboratory and all data down-sampled to 35× before duplicate removal [15]. In the MGI study the DNBSEQ arm was analyzed with MGI's proprietary pangenome-based PanVariants pipeline, while every comparator platform was analyzed with open-source DeepVariant v1.9.0 at 30× effective depth [26]. Each vendor therefore paired its own instrument with its own optimized secondary analysis and the competitor's instrument with a different caller, so the reported error differences conflate sequencing chemistry, library preparation, pipeline and caller training data, and cannot be attributed to the platforms themselves. The two studies are also not comparable with each other: they examined different Illumina instruments (the NovaSeq X versus the legacy NovaSeq 6000), used different depth conventions and, in the Illumina study, included a library-conversion arm that is not representative of native DNBSEQ workflows. A benchmark intended to characterize the platforms rather than the software would run an identical baseline pipeline (for example BWA-MEM2 with GATK HaplotypeCaller at default settings, or a single DeepVariant release with one model) on libraries prepared by each vendor's recommended PCR-free protocol from the same reference DNA, at matched effective depth, with results stratified by the GIAB genomic-context regions, and would report the vendor-optimized pipelines only as a secondary analysis; ideally it would be performed by an independent laboratory. The independent studies summarized above [1], [2], [22], [23] approximate this design because they applied the same pipeline to every platform, which is why their conclusion, namely equivalence within clinically acceptable limits for most variant classes with residual DNBSEQ weaknesses in GC-extreme and long-homopolymer contexts, is more credible than either vendor claim. A further caveat applies to deep-learning callers: DeepVariant's standard short-read models were trained on Illumina data, so out-of-the-box performance on DNBSEQ reads partly reflects training-set mismatch rather than sequencing error, and platform-specific retraining (Sentieon DNAscope for MGI, MegaBOLT-DV and DNBSEQ-specific DeepVariant models) recovers much of the difference [27]. For the same reason, a DeepVariant-versus-PanVariants comparison is no more informative about chemistry than a DRAGEN-versus-MegaBOLT one.

In practical terms, the lesson for laboratories is unchanged. For clinical laboratories adopting DNBSEQ, validation against an internal orthogonal truth set and careful attention to the pipeline used remain essential. Deep-learning callers trained specifically on DNBSEQ chemistry, Sentieon DNAscope and MegaBOLT-DV close most of the remaining gap [27].

3.4. Low-pass WGS (~0.1×–5×): reproductive applications

Non-invasive prenatal testing like cfDNA-based NIPT is the most widespread low-pass WGS application worldwide and simultaneously the most commercially successful clinical use of MGI-platform sequencing. The technical principle is straightforward: whole-genome sequencing of maternal plasma cfDNA at ~0.1–0.3× depth, after which read counts per bin are normalized and compared with a euploid reference to detect imbalance at the chromosomal and, increasingly, sub-chromosomal level. DNBSEQ chemistry has an advantage here because of its low GC bias, GC-correlated bin variance being a major source of NIPT false positives [28].

Zhang et al. (2015) validated BGI low-coverage WGS NIPT across 146,958 pregnancies throughout China. Sensitivity was 99.17%/98.24%/100% and specificity >99.95% for T21/T18/T13, with equivalent performance in both low- and high-risk women [29].

Li et al. (2019) published the first clinical evaluation of BGISEQ-500 NIPT (cPAS chemistry) specifically for sex chromosome aneuploidy in 570 pregnancies, with no false negatives for 45,X, 47,XXY, 47,XXX or 47,XYY and an overall PPV of 60.5% [30].

Lee et al. (2019) validated the BGI/NIFTY workflow in 1,055 Korean pregnancies. Sensitivity/specificity was 100%/99.9% for T21, 92.9%/100% for T18 and 100%/99.9% for T13, with per-trisomy PPV of 90–100% [31].

Zhen et al. (2025) reported the largest single-centre genome-wide BGISEQ-500 NIPT cohort published to date: 59,877 pregnancies enrolled, of which 59,771 were successfully tested, at ~0.17× coverage. Sensitivity exceeded 97% for fetal aneuploidy and 63.6% for CNVs ≥5 Mb. PPVs were 83.1% (T21), 25.8% (T18), 10.3% (T13), 51.9% (SCA), 2.0% (rare autosomal aneuploidy) and 33.9% (CNV ≥5 Mb) [32].

A cross-platform meta-analysis by Mackie et al. (2017) pooled 117 studies and reported, for trisomy 21, a sensitivity of 99.4% (95% CI 98.3–99.8%) and specificity of 99.9% (95% CI 99.9–100%) across 148,344 tests, a clinical benchmark from which DNBSEQ-based workflows show no meaningful deviation [33]. In routine practice, DNBSEQ-G400 or DNBSEQ-T7 workflows can multiplex 96–384 NIPT samples per flow cell and produce a report within 5–7 working days; for lower-volume hospital laboratories the benchtop DNBSEQ-G99 offers a same-day sequencing alternative at a vendor-recommended 8–20 NIPT samples per flow cell (~10 million SE50 reads per sample) [58], although, to our knowledge, no peer-reviewed clinical NIPT validation specific to the G99 had been published at the time of writing.

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Preimplantation genetic testing (PGT-A) that previously using FISH or array-CGH and now dominated by NGS uses ultra-low-pass WGS (~0.002×–0.1×) to call 24-chromosome and segmental aneuploidy from trophectoderm biopsies, typically 5–10 cells with whole-genome amplification. Commercial PGT-A kits such as the Revvity PG-Seq Rapid Kit v2 are designed and validated for the Illumina, Element and MGI platforms [34], while the DNBSEQ-G400 with a PE150 FCS flow cell is the workhorse of choice in many Asian PGT laboratories. Recent publications show a shift from PGT-A alone toward comprehensive PGT on a single low-pass run: Chen et al. (2025) presented KaryoSeq, integrating PGT-A, PGT-M and PGT-SR at ~2× sequencing per embryo (about 20 million raw reads), achieving 100% concordance with conventional PGT while additionally detecting triploidy, uniparental disomy, the parental origin of CNVs and maternal cell contamination [35]. Earlier work showed comprehensive PGT can be performed reliably via 10× parental and 4× embryonic sequencing with haplarithmisis-based haplotyping [36]. PGT workflows on the DNBSEQ-G400 typically generate 200,000–1,000,000 PE100 reads per embryo from a multiplexed flow cell, with sequencing turnaround under 24 hours and per-embryo sequencing-reagent cost under US$100 (vendor-stated, excluding biopsy, whole-genome amplification, library preparation and labour). The DNBSEQ-G99's 12-hour PE150 and 5-hour SE100 runs make it suitable for small PGT batches in which embryo-transfer timing is critical, again on vendor rather than peer-reviewed evidence [58].

Besides, products of conception and carrier screening which from low-pass WGS (≤1×) has largely replaced karyotyping and CMA for POC analysis in many laboratories, yielding aneuploidy results together with high-resolution CNVs and avoiding culture failure, which can derail 10–40% of karyotyping attempts. DNBSEQ-G400-based POC workflows have been applied at large scale by BGI Genomics and partner laboratories in China. For expanded carrier screening, combining low-pass WGS with reference-panel imputation is technically feasible on the DNBSEQ-G400 but not yet widely offered commercially, most clinical carrier panels still use targeted capture.

3.5. Standard 30× WGS: diagnostics, oncology and population genomics

***Rare disease and paediatric diagnostics*.** 30× short-read WGS is the mainstay for clinical diagnosis of rare Mendelian disease, with diagnostic yield of 25–55% in pediatric populations and a clear advantage over WES [10], [11], [12], [13]. The clinical argument for WGS over WES rests on more uniform coverage of clinically relevant exons, detection of deep-intronic, regulatory and splice-altering variants, simultaneous detection of SVs and mitochondrial variants, and the opportunity for reanalysis as gene–disease evidence accumulates.

For DNBSEQ, the most rigorous published clinical evidence comes from the China Neonatal Genomes Project and partner studies. Wu et al. (2021) reported the largest prospective trio rapid-WGS study on DNBSEQ, covering 202 critically ill infants across 13 hospitals in 10 Chinese provinces. Trio-r WGS achieved a diagnostic yield of 36.6% (95% CI 30.1–43.7%) compared with 20.3% for proband-only clinical exome ($p = 0.0004$), with a median turnaround of 7 days versus 20 days ($p < 2.2 \times 10^{-16}$) [37]. Wang et al. (2020) validated Optimized Trio Genome Sequencing on DNBSEQ for 130 NICU/PICU patients at Fudan Children's Hospital (proband 40–50×, parental 8–10×), yielding a diagnostic rate of 47.7% (62/130) with a median turnaround of 90 hours [38].

These yields and turnaround times are comparable to landmark rapid-WGS studies on Illumina platforms. Petrikin et al. (NSIGHT1, a randomized controlled trial) reported a median time to diagnosis of 13 days versus 107 days for standard testing ($p = 0.04$), and Farnaes et al. showed a change in clinical management in 31% (13/42) of infants undergoing rWGS versus 2% (1/42) of controls ($p = 0.0015$) [11], [12], [13]. Operationally, a single DNBSEQ-T7 PE150 flow cell (~5,800 million reads, ~1.7 Tb) generates data sufficient for ~15–17 human 30× genomes, or roughly 60 per fully loaded four-flow-cell run (derived from vendor read counts [58]); paired with the FPGA-accelerated MegaBOLT germline pipeline (1.5 hours per 30× genome to VCF) [27], an end-to-end turnaround under 72 hours is achievable with good coordination between library preparation and analysis prioritization.

Germline cancer predisposition. Beyond targeted hereditary cancer panels, 30× WGS can detect deep-intronic and structural variants in BRCA1/2, PALB2, RAD51C/D, mismatch-repair genes and others that frequently escape capture-based assays; large rearrangements in BRCA1 and Alu-mediated intronic deletions are well-documented WGS-only findings. No large prospective study has yet demonstrated an added diagnostic yield specific to DNBSEQ-platform germline cancer-predisposition WGS relative to Illumina, and both platforms show comparable performance in head-to-head SV analysis [3]. Laboratories adopting DNBSEQ for this indication should validate SV calling (Manta, Delly, GRIDSS) on representative reference samples and compare against MLPA as an orthogonal method.

Somatic tumour/normal WGS. Paired tumour WGS (30–60×) and normal-tissue WGS (~30×) is increasingly used in oncology programs, enabling simultaneous detection of SNVs, indels, CNVs, SVs/fusions, mutational signatures, tumour mutational burden, microsatellite instability, HLA typing, and HRD. Assaad, Hadi, Levine et al. (2025) identified HRD in 21%, 20%, 17%, 9%, and 2% of breast, pancreatobiliary, gynaecologic, prostate, and upper-gastrointestinal cancers, respectively; 24% of HRD cases were BRCA1/2 wild-type, with deleterious SVs in FANCC, FANCF, XRCC2, and RAD51B that may be missed by panel assays [39]. Although the cohort used Illumina sequencing, the reported somatic concordance between DNBSEQ-T7 and NovaSeq 6000 supports technical feasibility on MGI platforms [1] but not clinical validation. Under EU IVDR, FDA, and NMPA frameworks, diagnostic performance applies to the specific combination of specimen, library chemistry, instrument, software, and reporting pipeline; therefore, an HRD classifier validated on Illumina data must be independently re-established on DNBSEQ. This requires analytical validation across relevant variant classes, specimen types (including fresh-frozen and FFPE), and tumour-purity ranges, following the ACMG technical standard for clinical NGS [64] and the AMP/CAP consensus recommendations for the validation of NGS-based oncology assays [65], with the bioinformatics pipeline, including the HRD classifier itself, validated as a distinct component according to the AMP/CAP pipeline-validation guideline [66]. In the United States, such an assay is regulated either as a laboratory-developed test under the FDA framework in force [67] or, if marketed as an in vitro diagnostic, against the FDA guidance on analytical validation of NGS-based IVDs, which although written for germline diagnostics, sets out the accuracy, precision, and limit-of-detection expectations that a somatic HRD assay would also need to meet [68]; under the EU IVDR, Annex XIII requires a performance evaluation

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demonstrating scientific validity, analytical performance, and clinical performance for the specific device [69]. In every setting, thresholds should be verified against an orthogonal HRD assay and where result guide treatment, supported by clinical outcome or bridging data. The emerging MGI-SeqOne (somaHRD) and MGI-Agilent CE-IVD HRD programme for DNBSEQ-G99 [40] provides a relevant platform-specific precedent, although it concerns a panel-based rather than WGS assay.

Population genomics and pharmacogenomics. Population-scale programs benefit substantially from DNBSEQ-T7 throughput economies. The China Metabolic Analytics Project (ChinaMAP) sequenced 10,588 individuals at ~40× depth using DNBSEQ-category instruments, generating reference allele frequencies for eight Chinese ethnic groups (Han, Zhuang, Hui, Manchu, Miao, Yi, Tibetan, Mongolian) and identifying Mendelian, pharmacogenomic (CYP2C19, CYP3A5, NAT2, SLCO1B1, UGT1A1) and complex-trait variants previously underrepresented in European-oriented databases [41]. A subsequent ChinaMAP analysis extracted virome data identifying EBV, HBV and 12 other viruses from the WGS data of 10,585 individuals, a concrete illustration that WGS provides clinically relevant additional yield relative to targeted panels [42]. For pharmacogenomics, standard 30× WGS already phases most CPIC tier-1 actionable variants in CYP genes; however, CYP2D6 and HLA haplotype resolution remains incomplete with short reads alone and should be complemented with long-read or other orthogonal methods [43].

Infectious disease and metagenomic NGS. Clinical mNGS of cerebrospinal fluid, respiratory specimens, and plasma has demonstrated strong diagnostic performance. A 2022 systematic review and meta-analysis of 12 studies reported pooled sensitivity of 77% (95% CI 70–82%) and specificity of 96% (95% CI 93–98%) for CNS infections, with individual studies reporting sensitivity up to 92% in selected cohorts [44], [45]. Although most clinical evidence is based on Illumina platforms, the BGI PMseq assay on DNBSEQ-T7 received NMPA emergency approval for SARS-CoV-2 and broader pathogen detection during the COVID-19 pandemic, supporting clinical feasibility of DNBSEQ-based mNGS [7]. Reported limits of detection range from 0.2–313 genome copies or CFU/mL, although performance is affected by hemolysis and high host nucleic-acid backgrounds [44]. The DNBSEQ-G400 can support 24–48-hour clinical workflows, while the DNBSEQ-G99 offers approximately 5-hour SE50/SE100 sequencing runs and vendor-recommended ~20 million reads per sample, potentially enabling faster turnaround for urgent CSF and respiratory specimens [58].

3.6. Deep WGS (>60×–100×+): oncology and low-level variant detection

Tumour heterogeneity and subclonal architecture. Standard 30× WGS calls clonal variants at a variant allele frequency (VAF) of ~10–20% well but is less reliable for subclonal variants below 5% VAF in heterogeneous, low-purity or aneuploid tumours. Griffith et al. (2015) demonstrated, via ~312× WGS supplemented with ~433× exome capture on a primary–relapse–normal acute myeloid leukaemia trio, that conventional 30–50× WGS is inadequate for tumours with even moderate heterogeneity, purity or aneuploidy, and that deep sequencing provides clear improvement in variant discovery across a wide VAF range while supporting a more convincing model of clonal architecture [46]. These findings are supported by the PCAWG consortium's analysis of 2,658 tumours, which revealed extensive intra-tumour heterogeneity and subclonal driver mutations [47]. The DNBSEQ-T7's economies of scale enable 100–200× tumour WGS at a relatively small incremental reagent cost over 30× (Section 3.2) which is a meaningful advantage for hematologic malignancies (AML, MDS, ALL, lymphoma) and low-purity solid tumours.

ctDNA and liquid biopsy. Plasma cfDNA WGS at ~60–100× depth can detect circulating tumour DNA using tumour-naive or tumour-informed approaches. Ganesamoorthy et al. (2022) sequenced plasma cfDNA (~100×), paired tumour (~60×), and germline (~30×) from breast and benign-tumour cases, demonstrating broader detection of tumour-derived alterations than low-coverage WGS or targeted methods in low-tumour-fraction samples [48]. Although performed on Illumina HiSeq X Ten, concordance studies support the technical transferability of plasma WGS to DNBSEQ-T7 [1], [4]; however, this does not replace platform-specific analytical and clinical validation. Any DNBSEQ-based ctDNA assay must establish its limit of detection using contrived and clinical samples across the intended tumour-fraction range [65]. The DNB platform's absence of on-array amplification and index hopping may reduce duplicate and mis-assigned reads relevant to ultra-low-VAF detection [26], [63], but cfDNA workflows generally still require library PCR. Therefore, PCR-induced duplicates and sequencing errors must be characterized during validation.

Minimal residual disease monitoring. ctDNA-based MRD detection with WGS is developing rapidly; the tumour-informed approach tracks hundreds to thousands of patient-specific somatic mutations. Recent WGS methods including MRD-EDGE, duplex-sequencing approaches and Labcorp's ppmSeq-based Plasma Detect Genome MRD have achieved a limit of detection of ~10^{-6} to ~3×10^{-6} tumour fraction [19], [49], [50]. Early breast cancer cohort data presented in 2025 showed that WGS-based ctDNA MRD improves detection at baseline and follow-up while extending lead time to clinical relapse compared with panel-based approaches [51]. The DNBSEQ-T7 has begun to be used in clinical MRD pipelines, although most published whole-genome MRD validation to date uses Illumina NovaSeq or Ultima ppmSeq chemistry.

Mosaicism, mitochondrial disease and repeat expansions. Deep WGS enables detection of low-level somatic mosaicism, for example postzygotic PIK3CA, AKT1 and RAS variants in overgrowth syndromes and segmental disorders whose VAF can fall below 5% and thus out of reach of standard 30× sequencing. In mitochondrial disease, quantifying mtDNA heteroplasmy below 5% requires effective mtDNA coverage ≥500×, easily achieved as a by-product of 60–100× nuclear WGS; Feng et al. confirmed that heteroplasmy quantification is equivalent between the DNBSEQ-T7 and NovaSeq 6000 in tissue, plasma and urine [4]. For long pathogenic repeat expansions such as FMR1, C9orf72, DMPK and RFC1, deep short-read WGS retains real limitations; such cases are increasingly handled with long-read sequencing or specialized tools (ExpansionHunter, STRique) that benefit from greater depth but still require orthogonal validation.

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Table 3 - Coverage vs Application Matrix for DNBSEQ-Platform Clinical WGS

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Coverage	NIPT/POC	PGT-A/M/SR	Rare disease (constitutional)	Cancer predisposition (germline)	Solid tumor (somatic)	Hematologic malignancy	ctDNA / MRD	Mosaicism	mNGS	Newborn screening	Population genomics
0.1–1×	✓✓✓	✓✓✓	—	—	—	—	—	—	—	—	—
1–5×	✓✓	✓✓	—	—	—	—	—	—	✓	—	For imputation
5–30×	—	Research/PGT-P	Lower yield	—	—	—	—	—	✓✓	For screening	✓
30×	—	—	✓✓✓ (rWGS)	✓✓	✓ (with paired normal)	✓	—	—	✓✓✓	✓✓ (diagnostic)	✓✓✓
60–100×	—	—	—	—	✓✓✓ (tumor)	✓✓	✓✓	✓	—	—	—
>100×	—	—	—	—	✓✓ (heterogeneity)	✓✓✓	✓✓✓	✓✓✓	—	—	—

Note: symbol key: ✓✓✓ – well-established clinical use with peer-reviewed evidence on DNBSEQ; ✓✓ – clinical use with strong evidence generated on other platforms (usually Illumina), with analytical concordance data suggesting transferability to DNBSEQ; platform-specific local validation is still required before diagnostic reporting (Section 3.7); ✓ – emerging clinical use; — – generally not indicated

3.7. Cross-cutting considerations

***Bioinformatics*.** DNBSEQ data can be analysed with standard alignment pipelines (BWA-MEM, BWA-MEM2) and common variant callers (GATK HaplotypeCaller, DeepVariant, Strelka, Mutect2). Platform-specific enhancements include MegaBOLT, MGI's FPGA-based accelerator providing a ~28× speed-up over GATK for 30× germline WGS (vendor-reported 1.5 hours per genome) and incorporating the MegaBOLT-DV deep-learning caller (99.9% SNV and 99% indel accuracy per vendor data) [27]; Sentieon DNAscope with a DNBSEQ-G400-specific model, which Sentieon reports outperforms GATK HaplotypeCaller for both SNV and indel calling on MGI data [27]; and the PanVariants pipeline used in the 2025 MGI cross-platform benchmark with results comparable to DeepVariant on Illumina data [26]. For SV calling, a multi-tool ensemble (consensus of Manta, Delly, GRIDSS and LUMPY) is preferred; Liu et al. showed high cross-platform consistency with this approach [3]. Clinical laboratories must validate the complete pipeline — basecalling → alignment → variant call → annotation → reporting — on MGI data using a platform-appropriate truth set.

***Variant interpretation*.** Interpretation follows the 2015 ACMG/AMP guidelines and the ACMG/ClinGen framework without distinction by sequencing platform. WGS-specific considerations include stratifying confidence intervals by region (callable, low-mappability, segmental duplication, homopolymer), applying refinements from the ClinGen Sequence Variant Interpretation Working Group, and using SpliceAI and similar tools for non-coding variants. For oncology, the 2017 AMP/ASCO/CAP somatic variant tier classification applies. Secondary and incidental findings should follow institutional lists and ACMG SF v3.x. DNBSEQ-specific error profiles in difficult regions should be flagged within standard operating procedures as a "do not report without orthogonal confirmation" zone, the same approach applied to Illumina data.

***Regulatory landscape and accreditation*.** MGI's regulatory position has expanded substantially since 2018. The MGISEQ-2000/2000 and MGISEQ-2000 obtained NMPA Class III registration in 2018, followed by DNBSEQ-T7 and the PMseq metagenomics kit in 2020 [7], [54]. DNBSEQ-T7, DNBSEQ-G400, and DNBSEQ-G50 received CE-IVD marking in 2022, while DNBSEQ-G99 obtained CE marking under IVDR and NMPA registration in 2023 [8], [59], [60]. No DNBSEQ instrument currently has FDA 510(k) clearance or PMA; US clinical use therefore relies primarily on CLIA-validated laboratory-developed tests, with Complete Genomics beginning US shipment of DNBSEQ-T7+ in February 2026 [55], [67]. In contrast, Illumina NovaSeq 6000Dx and NextSeq 550Dx have both FDA clearance and CE-IVD marking, giving Illumina a broader regulatory footprint. CAP, CLIA, and ISO 15189 accreditation are platform-agnostic but require platform-specific validation of accuracy, precision, sensitivity, specificity, and reportable range, preferably using NIST/Genome in a Bottle reference materials (HG001–HG007) or equivalent standards.

***Analytical concordance versus clinical validation*.** Concordance with Illumina using GIAB reference materials should not be interpreted as evidence that Illumina-validated clinical assays can be transferred directly to DNBSEQ. Under the EU IVDR, FDA frameworks, and NMPA requirements, clinical performance is tied to the specific combination of instrument, reagents, software, specimen, and assay; changing the sequencing platform therefore requires appropriate re-evaluation or validation [67], [68], [69]. For US laboratory-developed tests, CLIA likewise requires laboratories to establish their own performance specifications before clinical reporting. ACMG and AMP/CAP standards further require end-to-end validation covering

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specimen type, extraction, library preparation, sequencing, bioinformatics, and reporting, including accuracy, reproducibility, variant-specific performance, and limits of detection, with revalidation after major platform or workflow changes [64], [65], [66]. Thus, cross-platform concordance studies should be viewed as evidence of technical transferability and feasibility, supporting investment in DNBSEQ-specific validation, rather than as substitutes for clinical validation. Similarly, CE marking or NMPA registration of a DNBSEQ instrument confirms the regulatory status of the platform itself and does not establish clinical validity for every WGS assay performed on it.

***Ethical, legal and social issues*.** Population WGS raises well-documented issues: informed consent for incidental findings and for adult-onset conditions in children; data sovereignty, particularly relevant to cross-border use of DNBSEQ data generated in China given HGRAC export rules; equitable access; potential insurance discrimination; and genomic data privacy. The BabySeq and GUARDIAN newborn genomic screening programs provide the most rigorous prospective ELSI data, showing no measurable increase in parental anxiety from genomic newborn screening and a high acceptance rate [52], [53]. For DNBSEQ implementation in China, ChinaMAP and similar cohorts have generated population-specific reference data important for variant interpretation in East Asian patients, which is a meaningful advance in equitable access compared with European-oriented population resources [41].

3.8. Future directions

Three areas appear likely to determine the direction of MGI clinical WGS through the end of the decade. Long-read integration: the MGI CycloneSEQ long-read platform paired with the DNBSEQ-T7+ enables hybrid short-plus-long-read clinical assays detecting SVs, repeat expansions and methylation together with small variants from a single submission, with such workflows beginning to emerge for undiagnosed rare disease and refractory oncology [56]. Multi-omics on a single instrument: the DNBSEQ-T7+ explicitly supports DNA, methylation (WGMS), single-cell and Stereo-seq spatial transcriptomics workflows on parallel flow cells, enabling integrated molecular profiling of tumours and tissues [14]. AI/ML-supported bioinformatics: deep-learning variant callers trained with platform-specific error profiles. The Sentieon DNAscope MGI model, DeepVariant retraining, MegaBOLT-DV reduce false positives, and as foundation models for genomic variant calling mature, inter-platform accuracy gaps are likely to narrow further. AI-assisted phenotype-driven variant prioritization (Exomiser, LIRICAL, AI-MARRVEL) is platform-agnostic and applies equally.

Limitation

This review relies on publicly available peer-reviewed publications, vendor specifications, and a number of clearly flagged preprints. Some quantitative platform specifications (Q30/Q40 thresholds, daily throughput, and cost per genome) derive from vendor reports; they represent nameplate maxima and reagent-only prices (Section 3.2) and will differ in real clinical environments. The head-to-head Illumina vs MGI white paper published in 2024, along with the subsequent MGI counter-benchmark, reflect ongoing commercial competition, did not apply a common analysis pipeline across platforms (Section 3.3), and cannot be considered definitive without independent replication. Several preprints cited in this review (the Ultima Genomics UG 100 clinical evaluation, KaryoSeq, the ChinaMAP virome paper) have not passed peer review, and their findings should be treated with commensurate caution. Regulatory status also changes frequently; verification of the current listings from the FDA, NMPA, MHRA, EU-IVDR, and equivalent bodies is needed before clinical implementation. Finally, the literature search was restricted to English-language sources; Chinese-language clinical reports on DNBSEQ platforms, which are likely to be numerous given the installed base, were not systematically searched, and the DNBSEQ-G99 evidence summarized in Section 3.2 is at present entirely manufacturer-generated.

Conclusion

MGI's DNBSEQ-T7 and DNBSEQ-G400 that backed by DNB and cPAS chemistry, the MegaBOLT bioinformatics accelerator, and a continually expanding regulatory footprint (NMPA Class III, CE-IVD) are credible short-read WGS instruments for clinical use across all three coverage-depth tiers examined here. The strongest evidence base exists for three applications:

1) low-pass cfDNA NIPT, where BGISEQ-500/DNBSEQ-G400 workflows have been validated across a cumulative cohort exceeding 200,000 pregnancies with >99% sensitivity for T21;

2) low-pass PGT for aneuploidy, monogenic disorders and structural rearrangements;

3) standard 30× rapid trio WGS for critically ill infants, with a diagnostic yield of 36.6–47.7% and turnaround of 3–7 days.

For deep WGS applications in oncology, tumour heterogeneity and ctDNA MRD DNBSEQ's analytical equivalence with Illumina is supported by concordance studies, although most prospective clinical validation to date still uses Illumina platforms. The benchtop DNBSEQ-G99 extends the family to hospital laboratories for low-pass, panel-based and metagenomic work, but it is not a standard-depth WGS instrument and its clinical evidence base is currently vendor-generated.

Residual limitations were a slightly higher error rate in difficult regions, a still-thin FDA regulatory footprint, and a more limited bioinformatics ecosystem in Western regions. This requires careful local validation against orthogonal truth sets and ongoing comparison with other platforms. This is particularly important amid the emergence of the Illumina NovaSeq X and Ultima Genomics UG 100, which are reshaping the high-throughput sequencing landscape. For clinical laboratories that prioritize cost per genome and high-throughput scalability, particularly for NIPT, PGT and population programs, the DNBSEQ-T7/G400 offers highly attractive value. For laboratories that prioritize regulatory-pathway simplicity in the United States and the largest body of independent clinical validation, Illumina remains the primary choice. Going forward, the approach will most likely be hybrid: DNBSEQ short-read for routine 30× WGS at scale, selectively complemented with long-read sequencing for difficult variants and MRD-style ultra-deep assays for oncology surveillance.

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Конфликт интересов

Не указан.

Conflict of Interest

None declared.

Рецензия

Все статьи проходят рецензирование. Но рецензент или автор статьи предпочли не публиковать рецензию к этой статье в открытом доступе. Рецензия может быть предоставлена компетентным органам по запросу.

Review

All articles are peer-reviewed. But the reviewer or the author of the article chose not to publish a review of this article in the public domain. The review can be provided to the competent authorities upon request.

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