BUYER'S GUIDE

Choosing a next-generation sequencing system

A step-by-step guide

Introduction

Next-generation sequencing (NGS) is powering modern biological discoveries. NGS offers exceptional accuracy with higher throughput, analytical sensitivity, and comprehensive genomic coverage compared to classic nucleic acid analysis methods like Sanger sequencing or quantitative PCR.[1-5] Sequencing genomes, exomes, transcriptomes, or proteomes can provide an unbiased view of cells and biological systems for deeper exploration and actionable insights. With hypothesis-free experimental design, NGS is enabling researchers to unearth new knowledge on cancer, microbiology, genetic disease, reproductive health, agriculture, and more.

Researchers are shifting to NGS to expand the scale and discovery power of their genomics studies. NGS equipment—once reserved for the largest and busiest research centers—is now accessible to laboratories of all sizes. Due to this availability, many laboratories are choosing to bring NGS inhouse, rather than send samples to a sequencing service provider. Acquiring their own NGS system can give labs tighter control over their NGS projects and samples for lab efficiency, time savings, and genomic data privacy considerations. Owning an NGS system is an investment in high-quality, reproducible data that can lead to valuable biological insights.

You have an important purchasing decision to make, and the market today offers plenty of choices. NGS systems range from benchtop sequencing systems ideal for smaller scale studies to ultraflexible, high-output instruments that can read hundreds to thousands of samples in a single run. This guide will walk you through the decision process by helping you evaluate your research goals and laboratory needs. Consider these key questions before purchasing your NGS system:

- What methods and applications will I use most?
- What will be the volume of sample numbers processed and the cadence of sample throughput?
- How do I ensure quality in data, instrumentation, and workflow?
- How will I analyze, store, and share NGS data?
- What are the ongoing costs beyond the initial purchase?
- What level of product support will I receive?

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01 Define your methods and applications

What types of questions are you trying to answer with NGS? Which experiments and applications will support these goals? Think broadly here; consider current needs and how needs may evolve in the future. Purchasing NGS instrumentation is an investment, and you want your chosen instrument to provide versatility and scalability to accommodate your lab for years to come. Choose a system demonstrated to work with a large ecosystem of methods if you want access to the widest range of applications.

Workflows for NGS methods generally involve preparing DNA libraries, then sequencing, followed by data analysis. The versatility of NGS allows for measuring multiple biological modalities, such as RNA, methylation, or proteomics, all with a sequencing read-out. Various methods for preparing libraries for sequencing, paired with strategies for turning sequencing data into relevant information, enable a growing catalog of NGS applications.

For each method or application, think about your needs for these three areas: read length, sequencing depth, and throughput per run. Survey the literature to guide experimental design based on the standard sequencing requirements per sample (Table 1).

Read length

Define the read length appropriate for the method or application. For "counting" applications, such as gene expression profiling with RNA sequencing (RNA-Seq), shorter reads of 2 × 75 bp are sufficient. While longer read lengths (eg, 2 × 300 bp) are more valuable for applications like *de novo* sequencing, whole-genome sequencing, and immune repertoire profiling. Some "long-read" sequencing technologies enable reads in the thousands of bases. However standard "short-read" sequencing is the predominant NGS method and most widely used.

Learn about NGS methods and applications

- Large whole-genome sequencing
- Small whole-genome sequencing
- Exome and large-panel sequencing
- Targeted gene sequencing
- Targeted gene expression profiling
- Transcriptome sequencing
- miRNA and small RNA analysis
- Single-cell profiling
- Spatial analysis
- Chromatin analysis
- Methylation sequencing
- 16S metagenomic sequencing
- Metagenomic profiling
- Cell-free DNA sequencing and liquid biopsy analysis
- Multiomics

Many applications benefit from paired-end sequencing, which allows users to sequence from both ends of a DNA fragment. Sequences aligned as read pairs make it easier to detect genomic rearrangements, repetitive sequence elements, gene fusions, and novel transcripts.[6] Find out whether the system you are considering supports paired-end sequencing.

Sequencing depth

Sequencing depth, or depth of coverage, refers to the average number of sequenced bases that align to known reference bases. For example, a whole genome sequenced at 30× coverage means that, on average, each base in the genome was sequenced 30 times. Sequencing depth can also be measured as the number of reads needed per sample. At higher levels of coverage, variant calls can be made with a higher degree of confidence. Defining target coverage provides statistical power and must account for factors like coverage uniformity to reduce coverage bias and errors.

Throughput per run

Throughput will largely dictate the sequencing system that's best suited for you. Assess your projected sample volume per month and year, and let this number guide your selection process. For example, if you plan on sequencing microbial genomes with a fairly low number of samples per week, consider a smaller benchtop system. If you plan to run large sample numbers for applications such as human whole-genome or whole-transcriptome sequencing, you're probably a good fit for a high-throughput system.

Also think about sample cadence, whether your lab will focus largely on analyzing samples for single sample reporting or whether you will be conducting large-scale studies across populations. Will you need immediate results per sample to guide your next steps, or will you collect samples to run as batches? Understanding this throughput can help determine how frequently you will need to run your sequencing system and how quickly you need to turn around analysis results.

Table 1: Example sequencing requirements for common NGS methods and applications

Applications or method	Typical read length	Data required per sample
Large whole-genome sequencing	2 × 150 bp	~120 Gb for 30× coverage
Small whole-genome sequencing	2 × 150 bp	~130 Mb for 30× coverage
Exome sequencing	2 × 100 bp	~8 Gb for 100× coverage
Targeted gene expression profiling	2 × 75 bp	10M reads
Total RNA-Seq	2 × 75 bp	50M reads
mRNA-Seq	2 × 75 bp	25M reads
miRNA and small RNA analysis	1 × 50 bp	11M reads
Single-cell gene expression	2 × 50 bp	20K reads per cell

02 Understanding instrumentation options

With your applications and methods defined, it's time to begin exploring the market for NGS systems. The best type of sequencing system for your lab is mainly determined by the sample throughput and data requirements of your methods and applications (Table 2). You'll find a full spectrum of options available, depending on your budget, the needs of your laboratory, and the nature of your research (Figure 1).

When comparing system costs, you can look at the cost per base or cost per sample. These values are rough estimates dictated by the amount of DNA that can be sequenced per run. Sequencing systems that fit on a benchtop can have output that is scalable from millions of bases (megabases or Mb) to billions of bases (gigabases or Gb) per run. Many production-scale sequencing systems can output up to trillions of bases (terabases or Tb) per run. Ultimately, you want to consider cost per answer or total cost of ownership, including library prep, skilled labor, data analysis and storage, and support (Figure 2). For example, with higher output applications, economies of scale can help reduce running costs.

Figure 1: The main categories of NGS instrumentation

	Low-throughput benchtop system	Mid-throughput benchtop system	High-throughput production-scale system
Output	14 Mb to 50 Gb	10 Gb to 540 Gb	65 Gb to 16 Tb
Reads per run	1 million to 100 million	75 million to 1.8 billion	65 million to 26 billion
Information	• Ideal for small- to medium-scale studies • Fast workflow for targeted samples • Well suited for pilot studies and library QC before progressing to larger projects on higher throughput equipment • May include common data analysis pipelines on the instrument • Most affordable option in terms of initial purchase price	• Flexible for a wide range of applications • Broad output range while maintaining a benchtop footprint • May include common data analysis pipelines on the instrument • Mid-range price point for both initial purchase price and cost per sample	• Highest throughput available, with outputs of over a hundred human whole genomes per run • Highest operational efficiency to complete projects in least number of runs • May include common data analysis pipelines on the instrument • Largest investment in terms of initial capital outlay; delivers the lowest price per sample for labs that handle large volumes or perform data-intensive applications
Costs	Capital expense $ Cost per base $$$	Capital expense $$ Cost per base $$	Capital expense $$$ Cost per base $

Cost per base and cost per sample are very rough estimates and not always the best indicators of total cost of ownership. Instead, consider cost per answer, including library prep, skilled labor, data analysis and storage, and support.

Table 2: Your applications and methods help determine the best type of sequencing system for your lab

Popular applications and methods	Low-throughput benchtop sequencing system	Mid-throughput benchtop sequencing system	High-throughput production-scale sequencing system
Large whole-genome sequencing		●●	●●●
Small whole-genome sequencing	●●●	●●●	●●
Exome and large panel sequencing		●●●	●●●
Targeted gene sequencing	●●●	●●	●●
Single-cell profiling		●●●	●●●
Transcriptome sequencing		●●●	●●●
Targeted gene expression profiling	●●●	●●	
miRNA and small RNA analysis	●●●	●●●	
Chromatin analysis		●●●	●●●
Methylation sequencing		●●	●●●
16S metagenomic sequencing	●●●	●●	
Metagenomic profiling		●●	●●●
Cell-free sequencing		●●	●●●

●●● Key application ●● Supported application

For labs that primarily perform molecular diagnostic applications in a clinical setting, there are NGS instruments that have gone through the regulatory process and are specifically intended for clinical use. Learn more

03 Ensure the best data quality

Exceptional data quality is at the core of why NGS has become such a huge driver of discovery. However, not all systems can offer the same reassurance that a sequence is read correctly. Ultimately, any accuracy claim must be supported by final assay and application performance and demonstrated to be reproducible.

Sequencing chemistry and underlying technology

The major NGS suppliers rely on different underlying technologies for each sequencing platform. Sequencing by synthesis, semiconductor technology, single-molecule sequencing, and nanopore technology are a few of the terms you'll come across when researching the different platforms. As you explore your options, learn about how each system's technology works, which can build confidence in your investment choice.

Look into the breadth of adoption and number of publications to determine whether the chemistry and technology are trustworthy and have potential longevity. Explore user groups and see what experiences people report having and how that compares with competing instruments.

Think about quality holistically, from Q-scores to variant calling

Research how instrument design and performance affect overall quality. For example, some NGS technologies have encountered problems correctly counting the number of bases in even short regions of a repetitive sequence and identifying relatively common variants. The consequences of poor quality from incorrect base calling are serious: Not only can errors give you the wrong answer to your clinical and research questions (false positive), but they can also cause you to miss the right answer (false negative). False positives and false negatives can lead to inaccurate or faulty results that reduce the utility of testing.

For this reason, when comparing systems, you should start by paying attention to quality, often reported by NGS systems as a quality score or Q-score. Q-scores measure the probability that a base is called incorrectly, reflected as a log value. Thus, a higher quality score indicates a smaller probability of error. This chart will help you understand how Q-scores are calculated and what they mean for your research (Table 3).

Table 3: Relationship between quality and base call accuracy

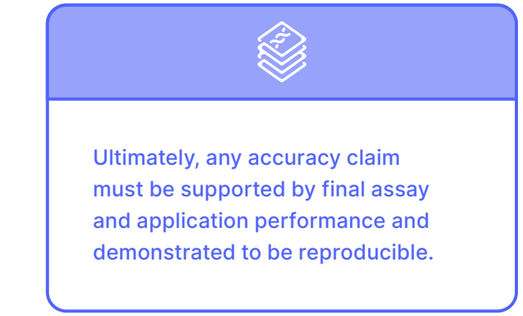
Quality score	Probability of incorrect base call	Inferred base call accuracy
10 (Q10)	1 in 10	90%
20 (Q20)	1 in 100	99%
30 (Q30)	1 in 1000	99.9%
40 (Q40)	1 in 10,000	99.99%

Different platforms and technologies calculate Q-scores differently or may focus on consensus read accuracy vs raw read accuracy. In many NGS workflows, sample manipulations like extraction and library preparation add upstream noise and errors that will impose a limit on total accuracy for a given assay. This makes very high accuracy (above Q30) challenging to distinguish for most applications. For this reason, Q30 has become a standard metric across platforms.

Additonal factors to consider are coverage uniformity and bias. For example, because of random sampling during extraction and library preparation, not all regions of the genome are read at equal levels during whole-genome sequencing (WGS). Thus, to make sure that all regions are sequenced at sufficient depth (eg, > 10×), WGS is often performed at an average depth of ≥ 30×. This added depth also helps to improve confidence to differentiate a true variant from an error.

***Tip:** In certain "needle-in-a-haystack" applications, such as identification of ultrarare genetic variants, unique molecular identifiers (UMIs) can be introduced during library preparation to identify and remove errors and increase accuracy beyond 99.9% (Q30).*

Finally, consider the algorithms used to identify genetic variants from the sequence data. Data analysis software can make a significant difference in accuracy of results. When comparing systems, evaluate quality holistically based on assay performance using similar benchmark samples or data sets. For example, WGS accuracy is commonly measured with the PrecisionFDA Truth Challenge, an FDA-organized community-wide assessment of variant calling accuracy that uses well-established benchmark samples with known variants.[7]



Ultimately, any accuracy claim must be supported by final assay and application performance and demonstrated to be reproducible.

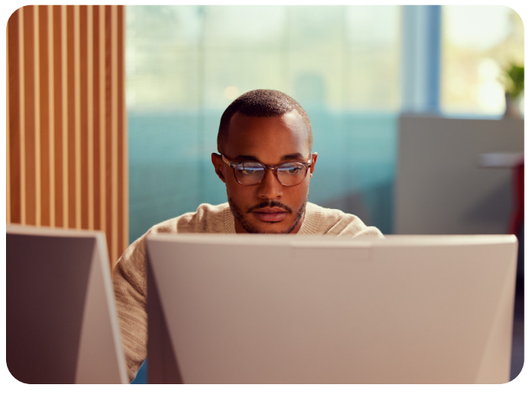
04 Plan how you will connect data to insights

The informatics phase of sequencing studies can often be a workflow bottleneck. As you move from raw data to meaningful results, you want the process to be as simple and streamlined as possible. Explore what types of analysis tools and protocols are available to users of a given platform. Consider the data analysis–focused questions in Figure 2 to help you plan an integrated NGS workflow for the greatest accuracy and ease of use.

Choosing a well-established NGS platform with standardized data formats can make it easier to share and compare data. For example, find out what file output types are compatible with your analysis solution.

Informatics pipelines

To manage the volume of data from NGS studies, you need bioinformatics tools that are accurate, efficient, and easy to use. Think about how you will use your NGS data and make sure that your bioinformatics pipelines are scalable and customizable to match your lab needs.



- Will your analytical workflow stop at a single sample report or will you run analysis across a large population for population-wide discoveries?
- Will you be integrating other types of data like phenotypic or electronic health records with your genomic data?

A wide range of data analysis algorithms for various applications are available to purchase from software vendors or as open-source tools from academic institutions. When creating your bioinformatics workflow, it's important to plan for maintaining current software. Some open-source tools, while free, might not be frequently updated or have the latest security features in place. Commercially purchased software often has a team of developers and security engineers maintaining analytical accuracy and security. A commercial software solution also offers a higher level of support for implementation and ongoing maintenance.

Consider who will be managing the informatics workflow. Labs that choose to build an informatics infrastructure inhouse may need to employ a team of bioinformaticians, IT professionals, data security experts, and data scientists. In addition to analyzing and interpreting results, this team needs to maintain, support, and continually monitor analysis pipelines for data security and compliance.

Think about your current and future security and accuracy needs. For users with workflows handling large amounts of data, building an informatics environment with the durability and security of a commercially available one can take several years and require highly specialized team members, incurring significant expense. Ready-to-use data pipelines can simplify the lift required to manage and develop future-proofed variant analysis processes.

Figure 2: Questions to consider for your holistic sequencing needs

Installation	Laboratory	Data analysis	Data storage	Support
What is the installation process? How should you prepare your lab space? Will you need additional equipment?	What is the ongoing cost for consumables? How much hands-on labor is needed? How is automation supported? How much cold storage space will you need? How will you manage waste?	Will you build your own analysis pipelines or purchase software? How will you integrate data analysis and interpretation with sequencing? How is data transferred from the instrument? How will you track and manage runs and samples?	Will you store and access your data onsite or in the cloud? How will you ensure data security and compliance?	What training is available? What warranty, reliability, and proactive support are offered? Is there a broad user community?

Data integration

You want your NGS system to foster the easiest workflow possible, and integration is key. Considering the scope of many sequencing experiments, you should seek out opportunities to save time and ensure accuracy. Figure out how you will transfer data from your instrument to your downstream analysis pipelines. Features such as preconfigured workflows, automated sequencing steps, push-button operation, and onboard analysis can make it easier to get started and maximize use of your NGS system.

Consider how each supplier handles data integration and find out whether it offers comprehensive workflow solutions for the applications that matter most to you. When gauging quality in this area, look for customer reviews and community forums. Ask colleagues for advice and speak in depth with representatives about how easy it will be for you to extract answers to your research questions in an integrated, meaningful way.

The volume of data from NGS studies requires bioinformatics tools that are accurate, efficient, and easy to use.

05 Think about budget holistically, from sample to results

Your budget can help you narrow your decision on the sequencing system to purchase; however, assessing the holistic cost can be more complex. There are many important factors that contribute to the total cost of ownership. Beyond the initial capital expenditure of the instrument itself, consider the cost of installation and ongoing operations, the skilled hands-on time required, the quality of the data (ensuring the process won't have to be repeated at an additional cost), and the cost of data analysis, storage, and any associated analysis of aggregate data (Figure 2).

Budgeting is where it's critical to consider your research needs, as well as the labor required to prepare samples for sequencing and analyze your results. Scope out the most common NGS workflows. Examine your budget per sample and make sure that everything is included in that cost, DNA extraction, sequencing reagents, informatics, and long-term data storage and access requirements.

The following questions are some of the most important considerations for hidden costs.

Installation: Prepararing an end-to-end NGS workflow

How should you prepare your lab space?

NGS systems range from small enough to fit on a benchtop to the size of a large major appliance. Laboratory space can be a limiting factor, so consider the instrument footprint, weight, power, ventilation, and connectivity requirements. Determine whether the NGS system will need specialty water lines, drainage, or other facility upgrades and include that in your cost calculations.

Will you need additional equipment?

Consider what additional equipment you may need for your workflow, including quality control (QC) steps. Find out if the NGS system requires ancillary equipment, such as a separate instrument for cluster generation, which can add significant expense. Make sure your lab is equipped for your planned applications. For example, enrichment experiments may require incubation equipment for hybridization steps.

Factors that contribute to the total cost of ownership include library prep, skilled labor, and data analysis and storage.

Laboratory: Ongoing operating costs

What reagents will you need on an ongoing basis?

Consumables are one of the biggest drivers of operational expenses, as they're required for both library preparation and sequencing. To prepare libraries for sequencing, the library prep workflow uses enzymes to add platform-specific oligonucleotide adapters to the DNA fragments. Library prep costs will vary depending on the study size, application, and vendor. Many third-party vendors sell library prep kits, but not all kits will work on every machine. That's why it's helpful to select a sequencing technology that is compatible with a wide variety of kits to support multiple methods. This ensures a larger range of options and, oftentimes, more competitive prices.

For sequencing reagents, look for a broad portfolio of consumable formats for different read lengths and throughputs, ensuring you have the appropriate kit configurations to match your projects as you scale.

Also consider the shelf life and storage space needed for consumables. How much room do kits take up in your lab refrigerator or freezer? How will you need to handle waste? Waste and unused, expired reagents can be hidden sources of expense.

***Tip:** Sample multiplexing allows you to pack more samples into each sequencing run, lowering the cost per base. For labs running high sample numbers, the higher expense of a high-throughput sequencing system is offset over time by the lower operating costs associated with each sample.*

What hands-on time is required for the workflow?

Consider efficiency and workflow simplicity as you compare platform options. Look specifically at how long it will take to generate the libraries and the hands-on time required by lab technicians. The more time a lab tech must spend to carry out a given experiment, the less time that person has available for other important projects. It becomes an issue of both money and time. Plan for the types of experiments you do and how many samples they usually involve.

Pay attention to the number of repetitive tasks and the amount of user intervention needed per experiment. Automation equipment for library prep can streamline NGS workflows and reduce hands-on time and user error. Ask the equipment vendor how their systems integrate with automation options. Also consider the ease of use of the NGS instrument. Are automation of sequencing steps (such as denature and dilution of libraries, cluster generation, washing, analysis) and remote monitoring built in to reduce the number of manual touchpoints?

***Tip:** Laboratory information management systems (LIMS) can help you automate workflows, connect instruments, and track runs, samples, and associated data. Ask if your NGS provider has scalable, accessible tools for run monitoring and data management.*

Data storage and security

How and where will you store the data?

The quantity of data that NGS produces can seem overwhelming at first. In 2007, a single sequencing run could produce a maximum of around 1 Gb of data. Today that can exceed 16 Tb of data in a single sequencing run—more than a 10,000× increase. This data volume necessitates sophisticated data storage solutions. Labs have a choice to store their data in the cloud or servers onsite.

Regulations in some countries require that data be stored onsite. This provision can come with added administrative burdens, such as maintaining the physical infrastructure to store and secure data. Also, in these instances, users may encounter challenges in sharing data with others outside of their network. The physical storage capacity may need to be continually scaled as larger volumes of sequencing data are generated.

As the volume of data grows, many genomics labs have moved to the cloud to manage and store their data. Cloud-based storage platforms do not require onsite compute and storage infrastructure. Sequencing data are loaded from the instrument directly into the cloud. Users can analyze data in the cloud, aggregate and mine their data and publicly available data bases, and selectively share data as needed. Cloud-based systems easily scale to meet growing sequencing needs.

In addition to comparing the costs of your various data storage options, you may want to explore solutions for compressing and archiving data. The cost required in space and maintenance for storing large volumes of data (on-premises or in the cloud) is growing at an exponential rate as we sequence more. Considering informatics workflows that can easily compress and archive data can save on storage costs.

Data transfers can also slow workflows. Take note of file sizes generated by the instrument and connection speed. Some systems allow for onboard data analysis and/or automated data uploads to the cloud or local servers.

***Tip:** Some institutions require data to be stored locally vs in the cloud. If you're interested in the integrated computing and storage of a cloud platform, ask the provider if an onsite version is available.*

How will you ensure data security?

Genomic sequence data is especially sensitive information that needs to be protected. Make plans for data quality and security management that meets compliance with government requirements and regulations, such as HIPAA and GDPR.* Look for a vendor that complies with a secure product development lifecycle and has enterprise-level quality management in place. Ask your NGS provider how they can help support data security management at scale.

* HIPAA, United States Health Insurance Portability and Accountability Act; GDPR, European Union General Data Protection Regulation.

06 Choose a reliable NGS partner

When making any new purchase, there can be concerns over whether the vendor will be able to provide the reliability, proven performance, future-proof solution you need. Making a switch to a new technology. can be daunting, but it doesn't have to be. This is where a company's strong track record, established community of users, and responsive service and support will help put you at ease and ensure your success.

Proven performance

Leveraging established, trusted tools and methods adds confidence to scientific results. Investigate published research that relied on the equipment and software used to generate data. Ask your sales representatives directly about the number of peer-reviewed publications and public data sets available and compare from different vendors. Consider each vendor's track record for reliability and robust performance. The more you can learn, the more comfortable you'll feel with your purchasing decision.

Community

Look for an established community of users who specialize in your key methods and applications. Engage with this community to learn about others' experiences using the systems you're considering. Ask if the workflow is easy. Scientists are always inventing new ways of doing things; as such, an active community offers the benefit of deep knowledge, innovative ideas, and new methods. Another benefit is access to built-out methods, supported by hundreds of publications you can follow for your own experiments.

Support

Find out what happens after the purchase. You want to do business with a company that will help you get up and running, not simply drop off an instrument at your door. Look for a well-defined and personalized onboarding process—a company that will help you set up your machine, train your lab personnel, collaborate with your bioinformaticians, and follow up to make sure everything is working as planned. Ask your sales representative whether the company offers in-person and virtual training that will help you keep your knowledge fresh.

Consider vendors that offer proactive instrument monitoring and easy access to onsite support. Remote monitoring can streamline troubleshooting, and even anticipate system issues to make repairs preemptively, all to make sure that your system performs optimally. Look at response times for support calls and repairs and the availability of local, in-language support to maximize instrument uptime.

Learn more

- Introduction to NGS
- A beginner's guide to NGS
- Illumina sequencing platforms
- Library prep selection tool

NGS Buyer's checklist

01 Define your methods and applications

What types of NGS methods will you use most? Consider the needs for these three areas:

- ◯ Read length
- ◯ Sequencing depth
- ◯ Throughput per run

02 Understand the instrumentation options

Buying equipment is a commitment; consider your needs now and for the future.

- ◯ Low-throughput benchtop sequencing system
- ◯ Mid-throughput benchtop sequencing system
- ◯ High-throughput production-scale sequencing system

03 Ensure the best data quality

Ultimately, any accuracy claim must be supported by final assay and application performance.

- ◯ Sequencing chemistry: Compare pros and cons based on the underlying technology of each NGS platform
- ◯ Quality: Think about quality holistically, from Q-scores to variant calling
- ◯ Accuracy: Consider the accuracy of variant calling algorithms and quality of tertiary analysis software

04 Plan how to connect data to insight

There's more to NGS than sequencing. Make plans for data analysis and management.

- ◯ Data analysis: Explore what types of analysis tools and protocols are available for a given platform
- ◯ Informatics: Decide whether you will use commercially available software or build your own pipelines
- ◯ Data integration: Look for integrated solutions that help streamline lab operations

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05 Think about budget holistically

There are many factors to consider beyond the initial capital expenditure.

- ◯ Laboratory space: Determine whether additional equipment or laboratory renovations will be needed
- ◯ Running expenses: Factor in the costs of consumables and library prep kits
- ◯ Hands-on labor: Consider the element of efficiency for lab technicians
- ◯ Data compute and storage: Onsite or in the cloud? Learn about the costs and benefits of each solution

06 Choose a reliable NGS partner

Ensure that there's an established base of users and responsive service and support.

- ◯ Track record: Investigate published research and ask existing users about ease of workflow
- ◯ Community: Look for an established group of users who specialize in your key applications
- ◯ Support: Seek out a well-defined and personalized onboarding process

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