Novogene

Providing leading genomic services & solutions

Leading Edge Genomic Services & Solutions for FHS

Date：25 Feb 2021

Elaina Lo

澳門大學
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UNIVERSITY OF MACAU

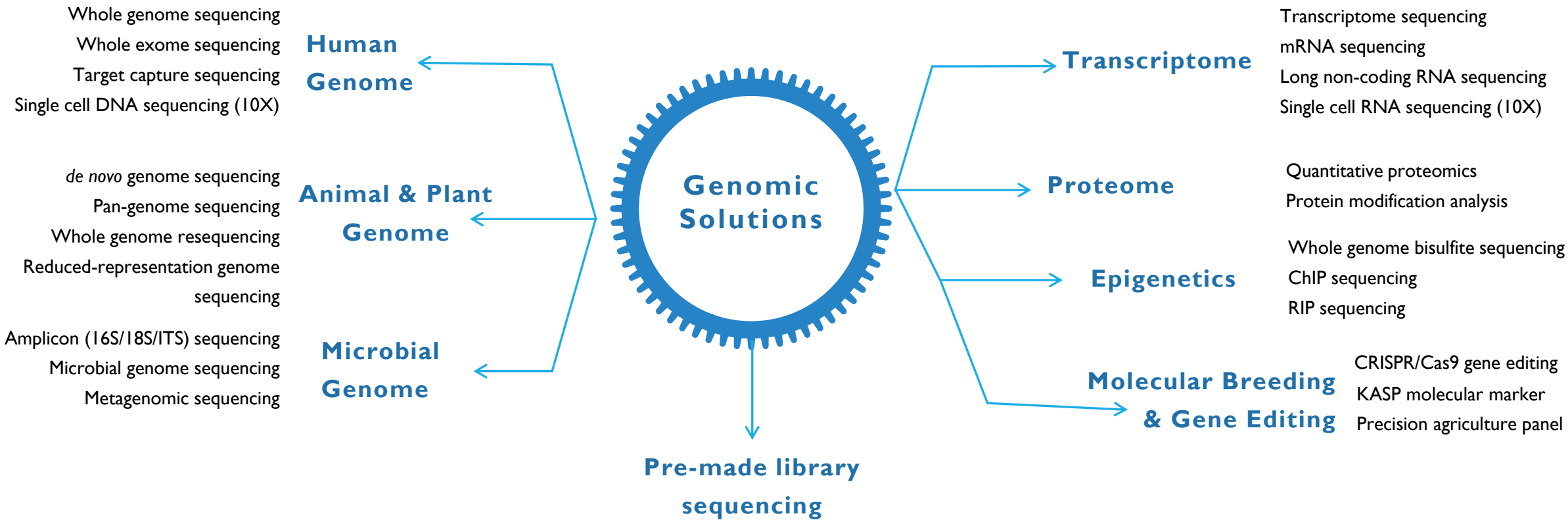
健康科學學院
Faculdade de Ciências da Saúde
Faculty of Health Sciences

Content

Content

Comprehensive Research Services and Solutions

Using our array of state-of-the-art **Illumina, Pacbio and Nanopore** systems, Novogene provides comprehensive genomics, transcriptomics and proteomics research solutions.



Novogene 诺禾致源

First intelligent multi-product NGS delivery platform

Falcon system

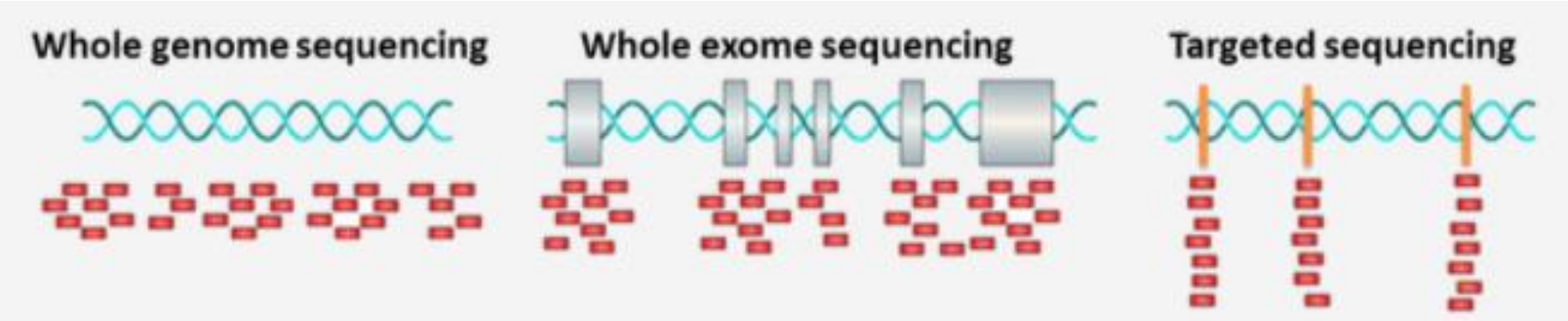
Launched in 2020

Fully automated production line → reduce manual errors and basis

Safe and accurate one-stop ground-breaking solution

Novogene 诺禾致源

DNA Sequencing



Whole genome sequencing	Whole exome sequencing	Targeted sequencing
Sequencing Region: Whole Genome	Sequencing Region: Whole Exome	Sequencing Region: Specific Region (customized)
Sequencing Depth: >30X-50X	Sequencing Depth: >100X-200X	Sequencing Depth: >500X
Identify all variants: SNPs, INDELs, SV, CNV	Identify most variants: SNPs, INDELs in coding region, CNV for paired cancer samples	Identify most variants: SNPs, INDELs in specific region

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Comparison of WGS, WES & TRS

Sequencing strategy	Applications	Advantages	Disadvantages
WGS	• Genetic disease study • Cancer research • Human population evolution • DNA biomarkers • Pharmacogenomics • Evolutionary and demographic history • Biodiversity	• **No need of prior knowledge** of disease • Focus on **non-coding area** mutations • unbiased • Detection of all variant types including CNV & SV • Joint Analysis of Multi-Groups	• High cost • Data interpretation is difficult • Low frequency mutations cannot be detected
WES	• Genetic disease study • Cancer research • Human population evolution • Focus only on **coding areas** for mutation	• **Low cost** • Good detection sensitivity	• Can't detect non-coding region and viral • SV detection is incomplete and inaccurate
TRS	• Cancer research • Human population studies • Linkage analysis for inherited diseases • Discovery of biomarkers and therapeutic targets • Candidate genes screened for pre-sequencing in large sample sizes • High-depth **validation** of mutation information	• Low cost • Can detect low frequency mutations • Offers more targeted detection of mutations	• Needs knowledge of gene of interest • Probes need to be customized and arrive with **long delivery times**

Whole Genome Sequencing (WGS)

Service Parameter

Platform	Illumina
Read length	2×150 bp
Turnaround time	About 15 working days
Standard analysis	additional 10 working days
Advanced analysis	upon request

Project Workflow

Novogene 诺禾致源

Whole Genome Sequencing (WGS)

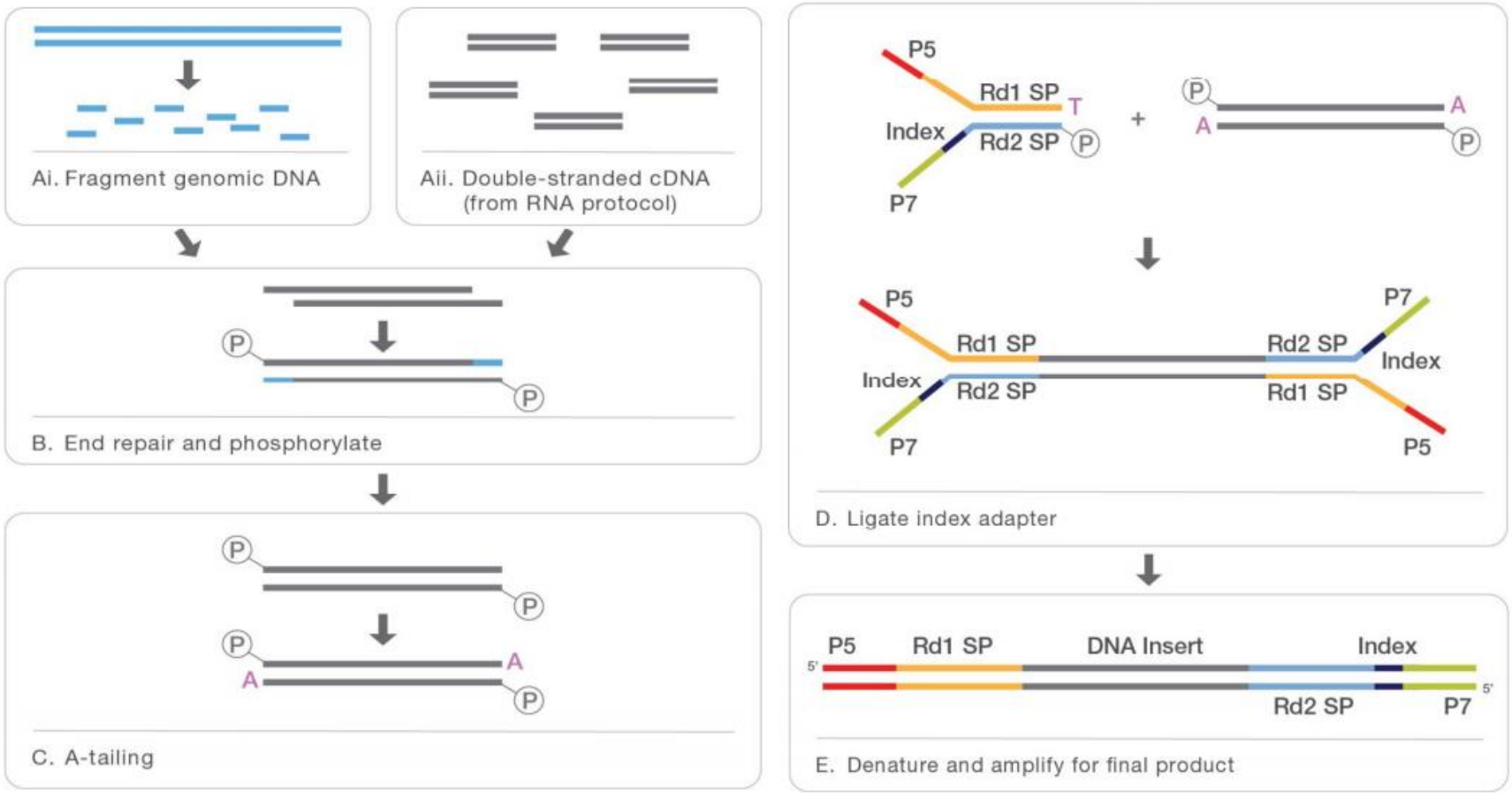
Sample Requirements

Sample Type	Amount (Qubit®)		Volume	Concentration	Purity (NanoDrop™/Agarose Gel)
	Strongly Recommended	Required			
Genomic DNA	≥0.4μg	≥ 0.2μg	≥ 20μL	≥ 10ng/μL	OD260/280 = 1.8-2.0, no degradation, no contamination
Genomic DNA (PCR-free)	≥ 3μg	≥ 1.5μg	≥ 15μL	≥ 10ng/μL	
FFPE	≥ 1.6μg	≥ 0.8μg	≥ 15μL	≥ 10ng/μL	Fragment≥ 1500 bp

Novogene 诺禾致源

Whole Genome Sequencing (WGS)

Library construction



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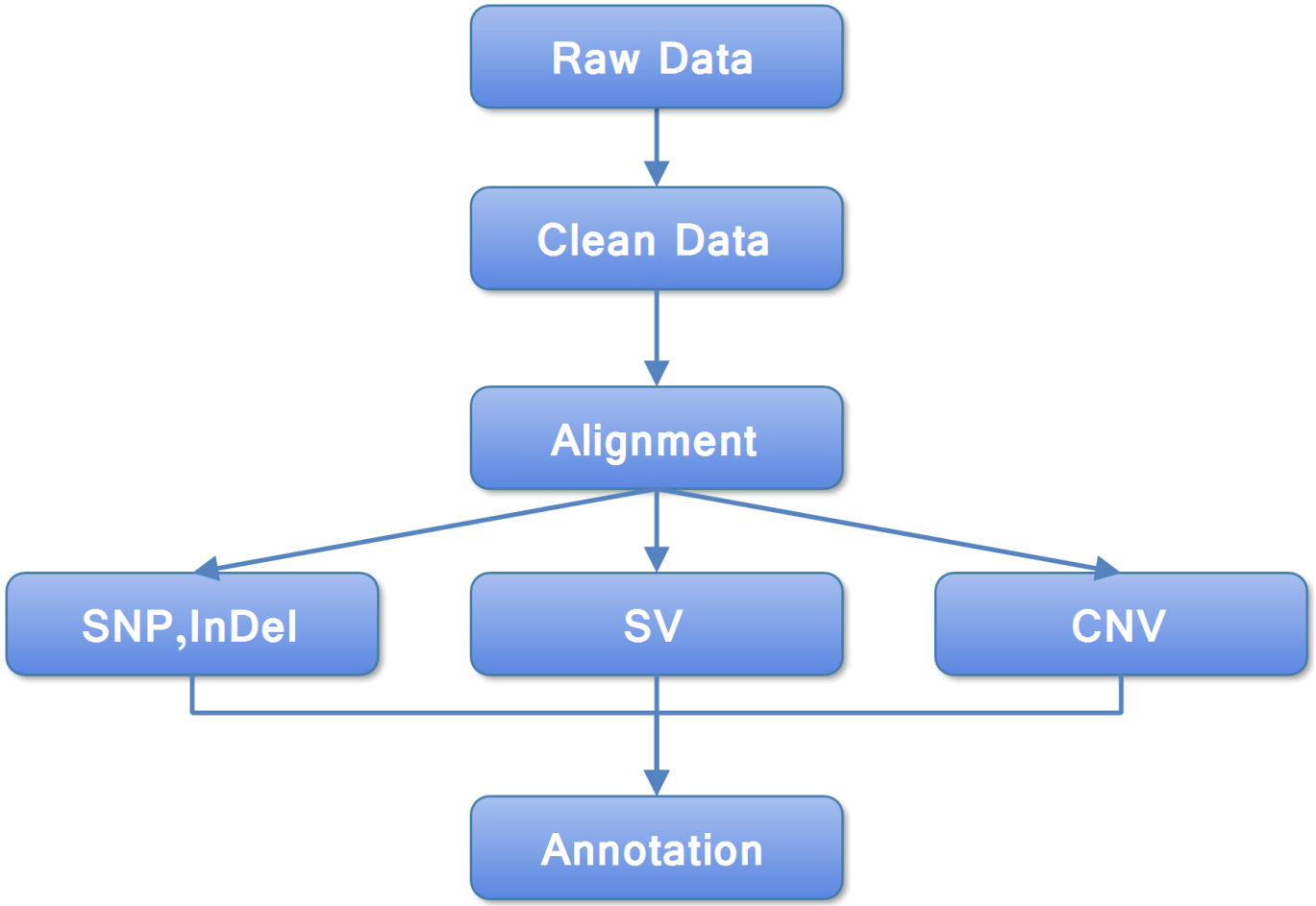
Whole Genome Sequencing (WGS)

Standard Bioinformatics



Novogene 诺禾致源

Whole Genome Sequencing (WGS)

Advanced analysis contents

Single gene disorder	Cancer	Complex disease
1. Filter screened mutations in dbSNP and 1000 Genome database; 2. Screen the variation of exomic or splicing site; 3. Amino acid conservation prediction by SIFT and Polyphen; 4. Gene function and pathway analysis; 5. Encode/miRbase annotation of non-coding region.	1. Oncogene/ Antioncogene/Susceptibility gene screening; 2. Cancer tissue purity and ploidy analysis; 3. Mutation characteristics analysis; 4. Tumor driver gene analysis; 5. Hypermutation analysis; 6. Pathway enrichment analysis; 7. Encode/miRbase annotation of non-coding region.	1. Filter screened mutations in dbSNP and 1000 Genome database; 2. Screen the variation of exomic or splicing site; 3. Amino acid conservation prediction by SIFT and Polyphen; 4. Gene function and pathway analysis; 5. Encode/miRbase annotation of non-coding region; 6. Novo mutation screening 7. Analysis of gene significance; 8. Protein-protein interaction analysis

Novogene 诺禾致源

Whole Exome Sequencing (WES)

Service Parameter

Platform	Illumina
Exome Capture	Agilent SureSelect Kit V6
Read length	2×150 bp
Turnaround time	about 18 working Days
Standard anaylsis	additional 4 working days
Advanced analysis	upon request

Project Workflow

Novogene 诺禾致源

Whole Exome Sequencing (WES)

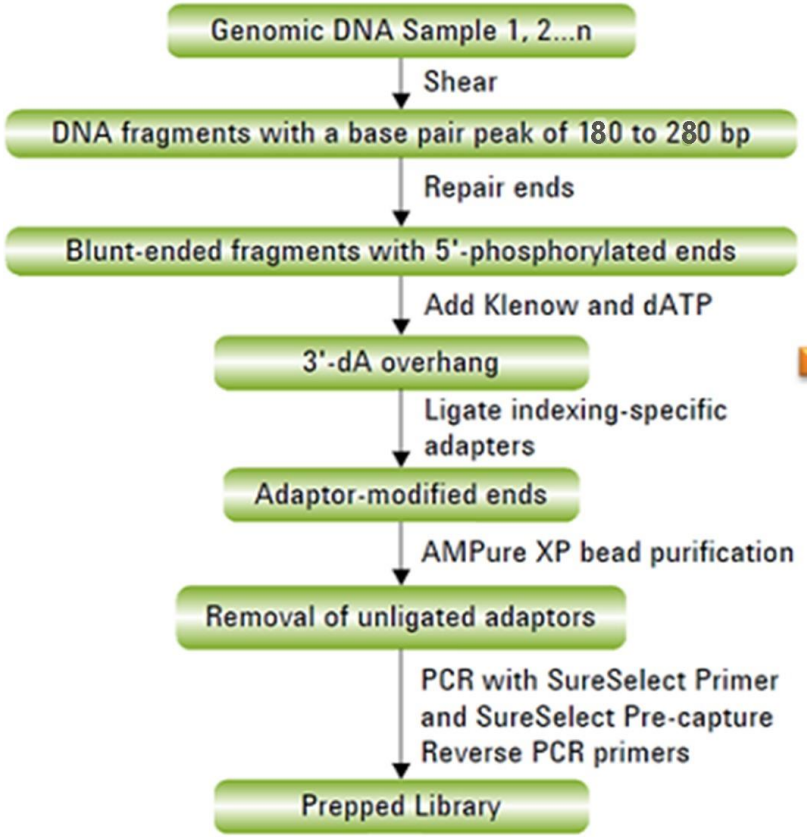
Sample Requirements (Agilent V6)

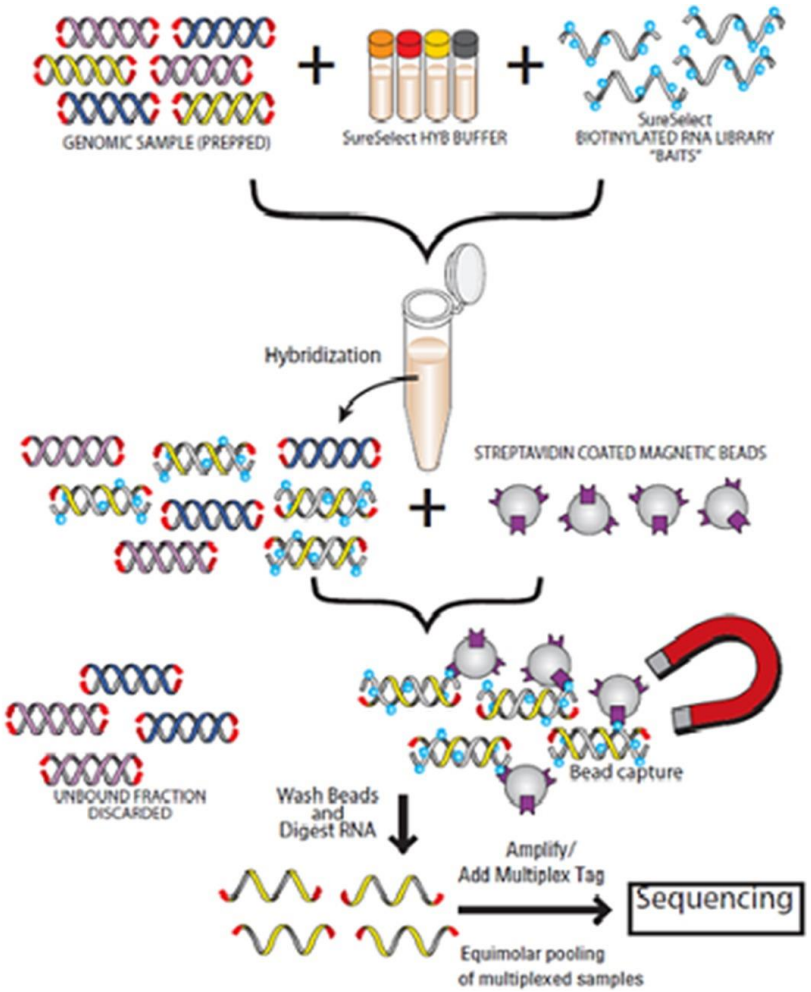
Sample Type	Amount (Qubit®)		Volume	Concentration	Purity (NanoDrop™/Agarose Gel)
	Strongly Recommended	Required			
Genomic DNA	≥0.8μg	≥ 0.4μg	≥ 15μL	≥ 20ng/μL	OD260/280 = 1.8-2.0, no degradation, no contamination
FFPE	≥ 1.6μg	≥ 0.8μg	≥ 15μL	≥ 20ng/μL	Fragment≥ 1000 bp
cfDNA/ctDNA	≥ 80ng	≥ 40ng	≥ 15μL	≥ 0.5ng/μL	Fragment in multiples of 170 bp, no genomic contamination

Novogene 诺禾致源

Whole Exome Sequencing (WES)

Library construction

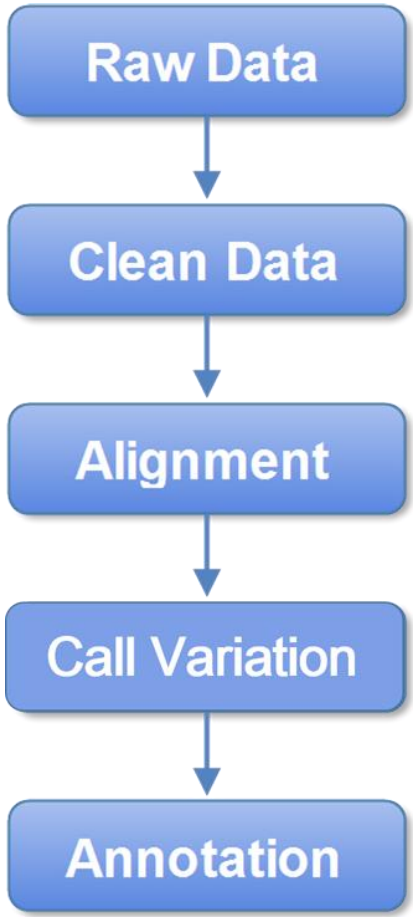




Novogene 诺禾致源

Whole Exome Sequencing (WES)

Standard Bioinformatics



Novogene 诺禾致源

Whole Exome Sequencing (WES)

Advanced analysis contents

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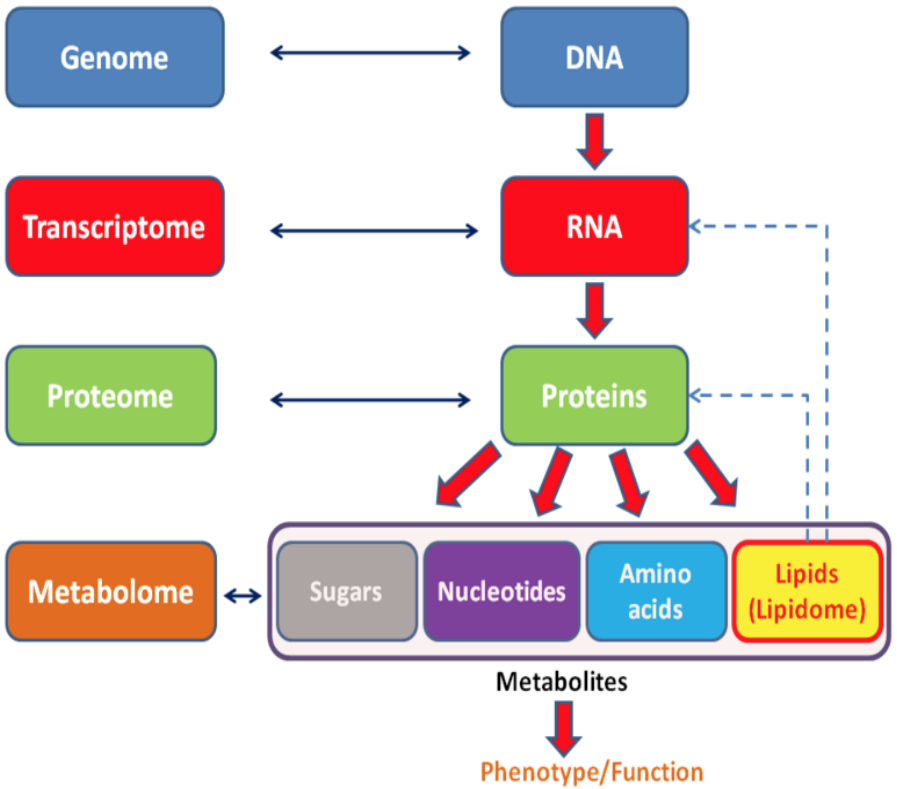
RNA Sequencing



	Classification	Name		Percentage
RNA Family	Coding RNA	Messenger RNA, **mRNA**		**2%-5%**
	Noncoding RNA	Long noncoding RNA, **lncRNA**		**~5%**
		Small RNA	microRNA, **miRNA**	
			Small interference RNA, **siRNA**	
			Piwi-protein interacting RNA, **piRNA**	
		Ribosomal RNA, **rRNA**		**80%-85%**
	Transfer RNA, **tRNA**			**~15%**

The transcriptome is the set of all RNA molecules in one cell or a population of cells. Based on next generation sequencing technique, transcriptome sequencing is capable of fully and quickly acquiring the overall transcripts information in the specific tissues or organs of eukaryotic species under a certain status.

Novogene 诺禾致源

Comparison of mRNA Seq, small RNA Seq & lncRNA

Sequencing strategy	Applications	Remarks
mRNA Seq	• Pathological mechanism • Tumor-subtypes classification • Molecular markers • Human evolution • Drug target • Clinical diagnostics • Biology Development	• Captures both **known and novel** features included gene expression structure and quantification • Identify biomarkers across the broadest range of transcripts • Can detect alternative Splicing
Small RNA Seq (size < 200nt) e.g., miRNA, piRNA	• Expression quantification of small RNA transcripts • Function verification, such as gene knockout, over-expression of miRNA genes • Advanced Analysis: miRNA target gene verification, piRNA identification and expression quantification	• Understand how post-transcriptional regulation contributes to phenotype • Identify novel biomarkers • Only available for eukaryotic species with reference genome
lncRNA Seq (size ≥ 200nt) e.g., lincRNA, NAT	• Expression Quantification of lncRNA transcripts • Gene or RNA subcellular Localization and Expression • Function Verification, such as gene knockout, over-expression of lncRNA genes • Protein Interaction	• Included mRNA information

Experimental Planning of RNA Seq

1. Number and type of replicates
   - 3 biological replicates being the **minimum** for any inferential analysis
   - More replicate is better, especially for clinical samples
   - Try to avoid pooling of individuals/experiments

2. Avoiding confounding
   - Ensure samples in each condition are all the same sex, age, litter, and batch, if possible.

3. Addressing batch effects
   - Avoid batch or split replicates across batches

4. Using same sample for sequencing and validation

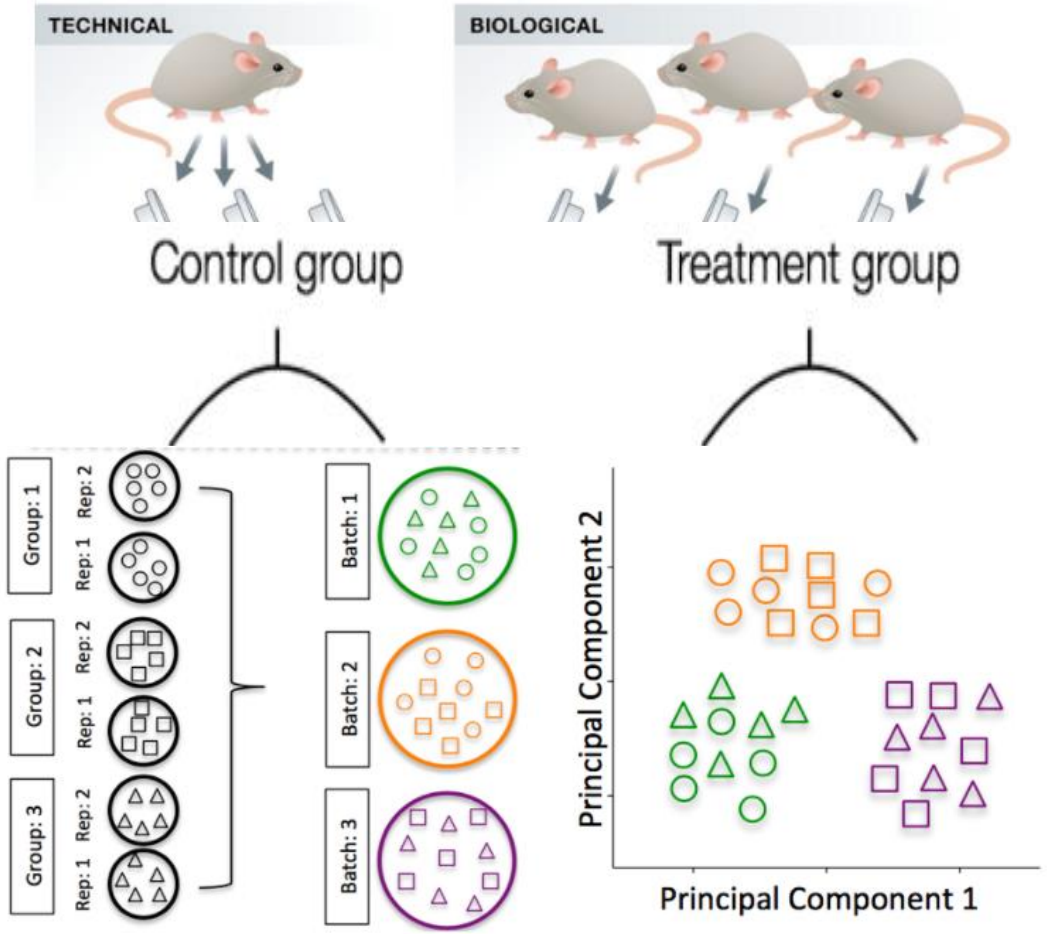


Image credit: Klaus B., EMBO J (2015) 34: 2727-2730

mRNA Sequencing

Service Parameter

Platform	Illumina
Read length	2×150 bp
Turnaround time	15-18 working days
Standard anaylsis	additional 5-15 working days
Advanced analysis	upon request

Project Workflow

Novogene 诺禾致源

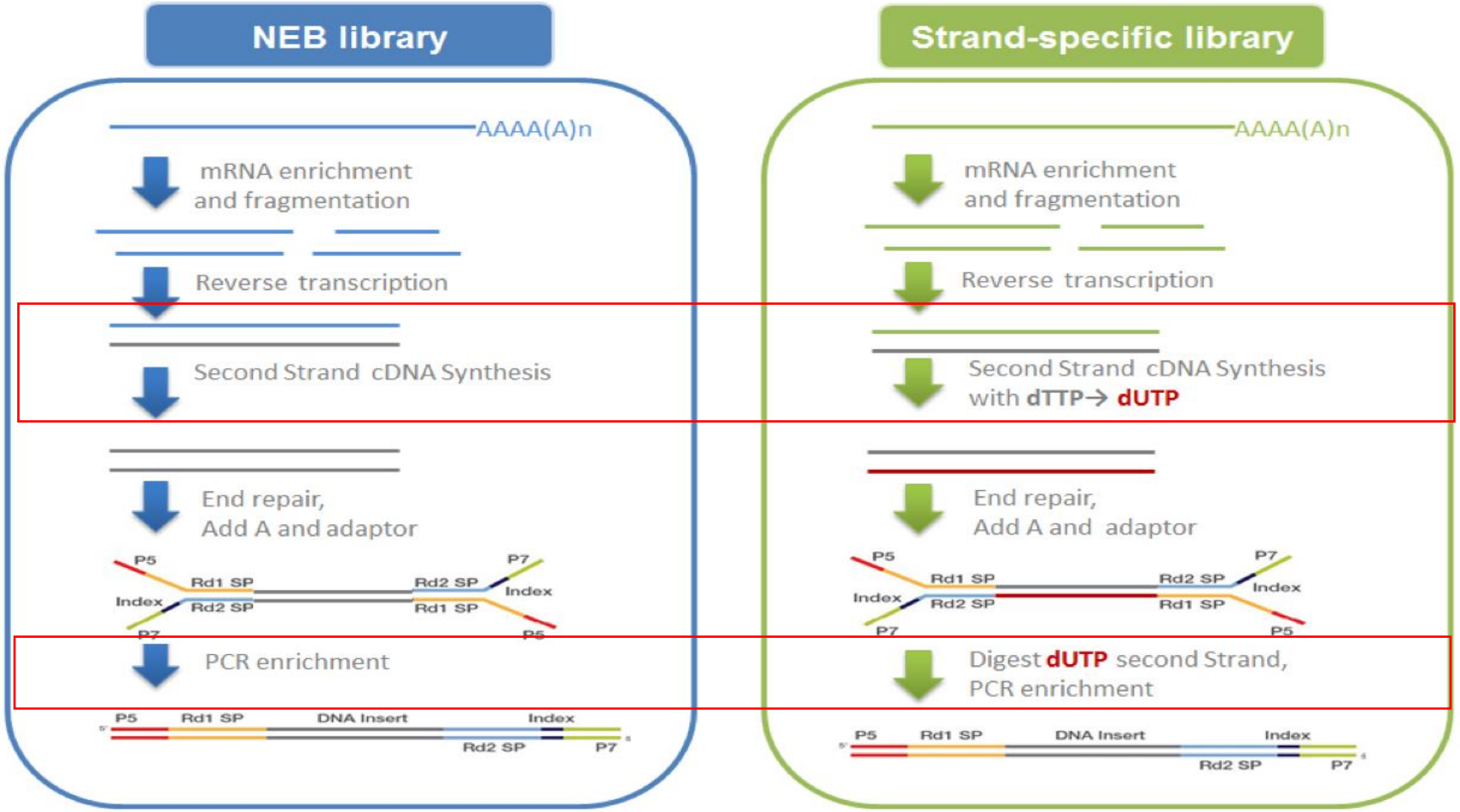
mRNA Sequencing

Sample Requirements

Library Type	Sample Type	Amount (Qubit®)		RIN	Volume	Concentration	Purity (NanoDrop™/Agarose Gel)
		Strongly Recommended	Required				
Eukaryotic (PolyA enrichment)	Total RNA (Eukaryotic)	≥0.8μg	≥ 0.4μg	≥ 6.8 (animals)/ 6.3 (plants & fungi)	≥ 20μL	≥ 20ng/μL	OD260/280 ≥ 2.0, no degradation, no contamination
Eukaryotic (Directional)	Total RNA (Eukaryotic)	≥1.6μg	≥ 0.8μg				
Prokaryotic (Directional, rRNA removal)	Total RNA (Prokaryotic)	≥6μg	≥ 3μg	≥ 6	≥ 20μL	≥ 50ng/μL	

mRNA Sequencing

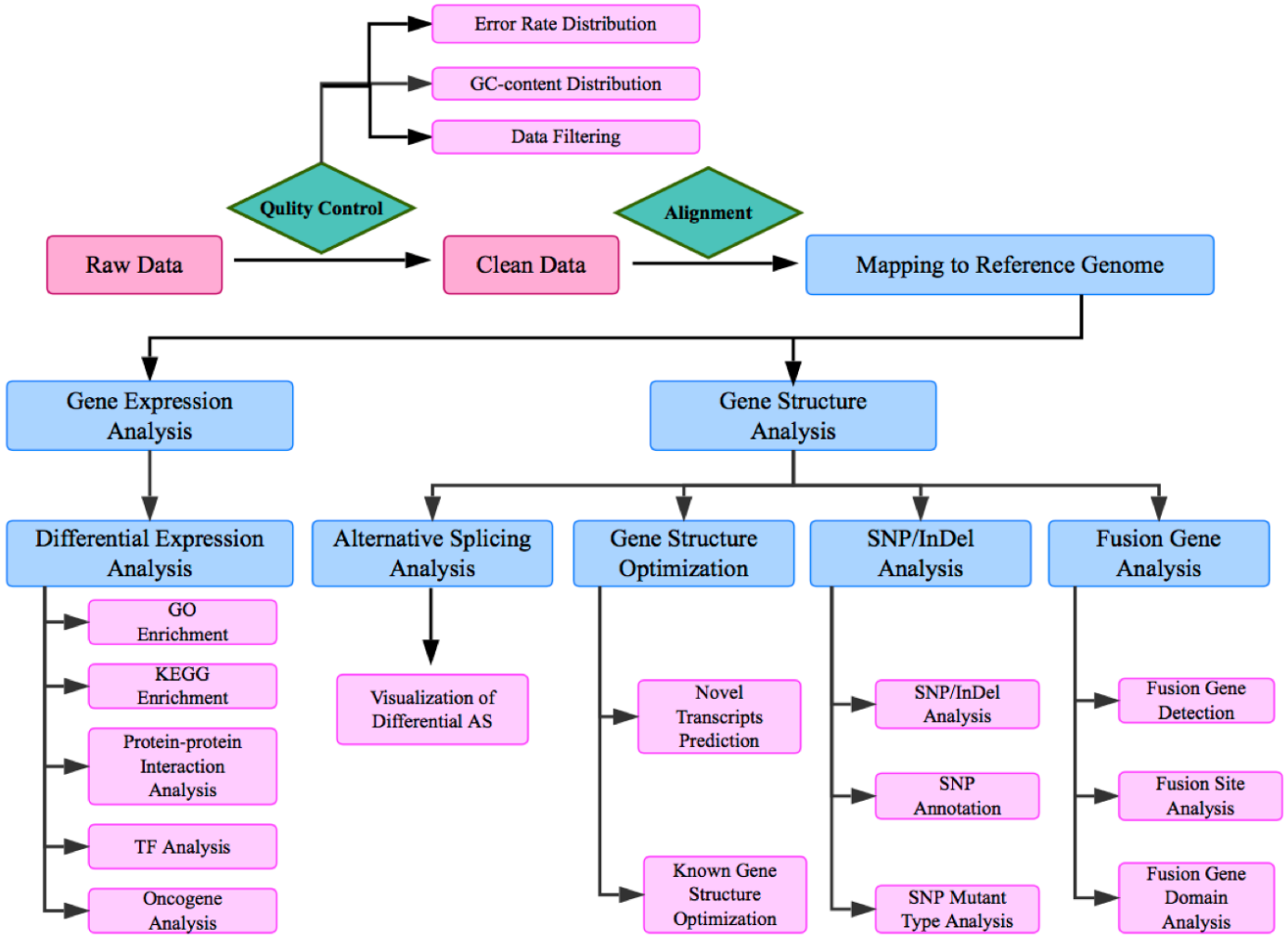
Library construction



The strand specificity of origin for each transcript provides a more accurate information with the un-stranded RNA-seq.

mRNA Sequencing

Standard Bioinformatics



Novogene 诺禾致源

Small RNA Sequencing

Service Parameter

Platform	Illumina
Read length	SE50
Library type	18-40 bp insert sRNA library
Turnaround time	20-22 working days
Standard anaylsis	additional 5-10 working days
Advanced analysis	upon request

Project Workflow

Sample Preparation → Library Preparation → Sequencing → Data Quality Control → Bioinformatics Analysis

Novogene 诺禾致源

Small RNA Sequencing

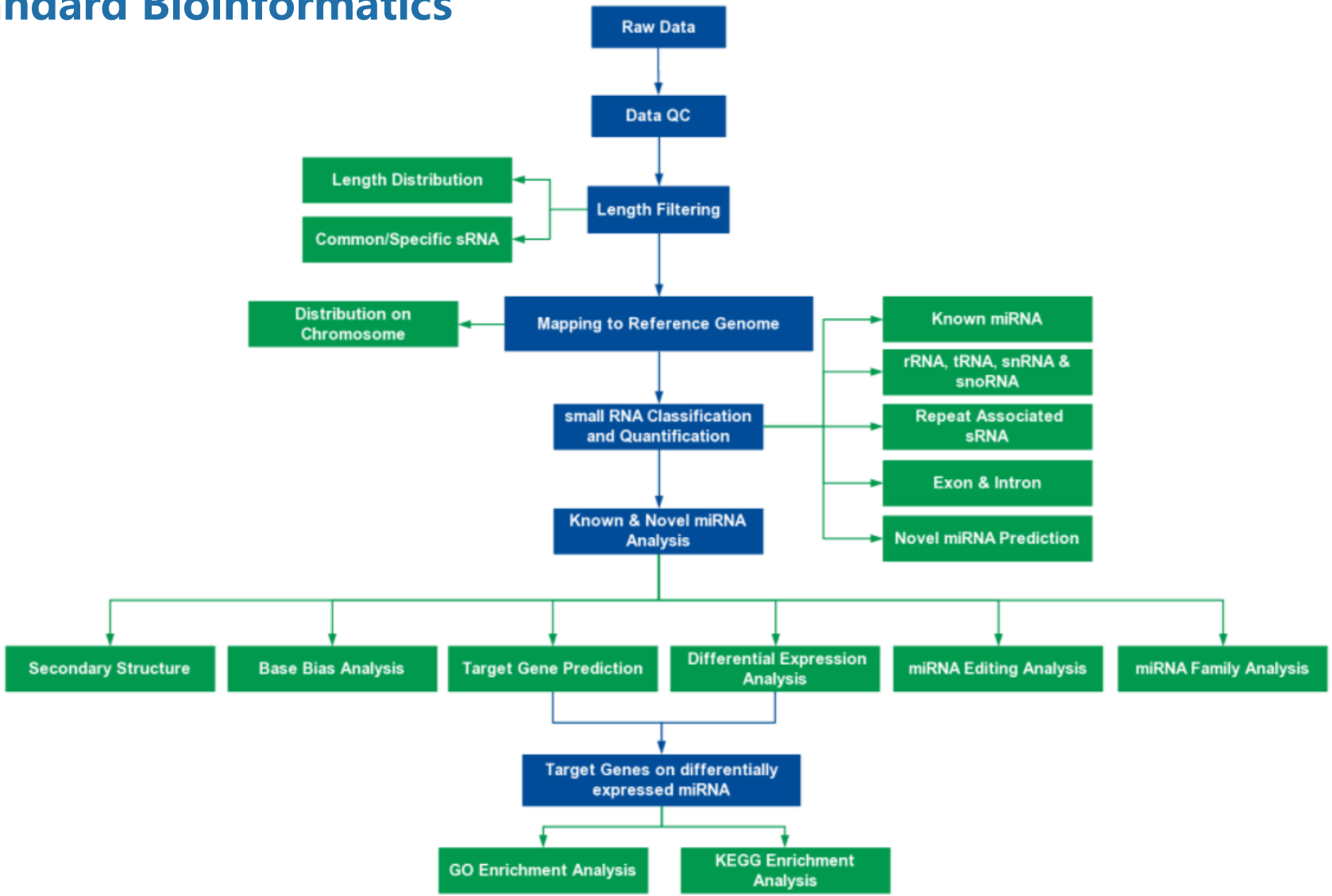
Sample Requirements

Sample Type	Amount (Qubit®)		RIN	Volume	Concentration	Purity (NanoDrop™/Agarose Gel)
	Strongly Recommended	Required				
Total RNA	≥4μg	≥ 2μg	≥ 7.5 (animals)/ 7 (plants & fungi)	≥ 20μL	≥ 50ng/μL	OD260/280 ≥ 2.0, OD260/230 ≥ 2.0, no degradation, no contamination
Total RNA (Exosome)	≥40ng	≥ 20ng	Peak Range: 25-200 nt, FU≥ 10, with no peak >2000nt			

Novogene 诺禾致源

Small RNA Sequencing

Standard Bioinformatics



Novogene 诺禾致源

Whole Genome Bisulfite Sequencing

Service Parameter

Platform	Illumina
Read length	PE 150 bp
Turnaround time	25 working days (< 10 samples)
Standard anaylsis	additional 10 working days

Project Workflow

Sample Preparation → Library Construction → Bisulfite Treatment → Sequencing → Bioinformatics Analysis

Novogene 诺禾致源

Whole Genome Bisulfite Sequencing

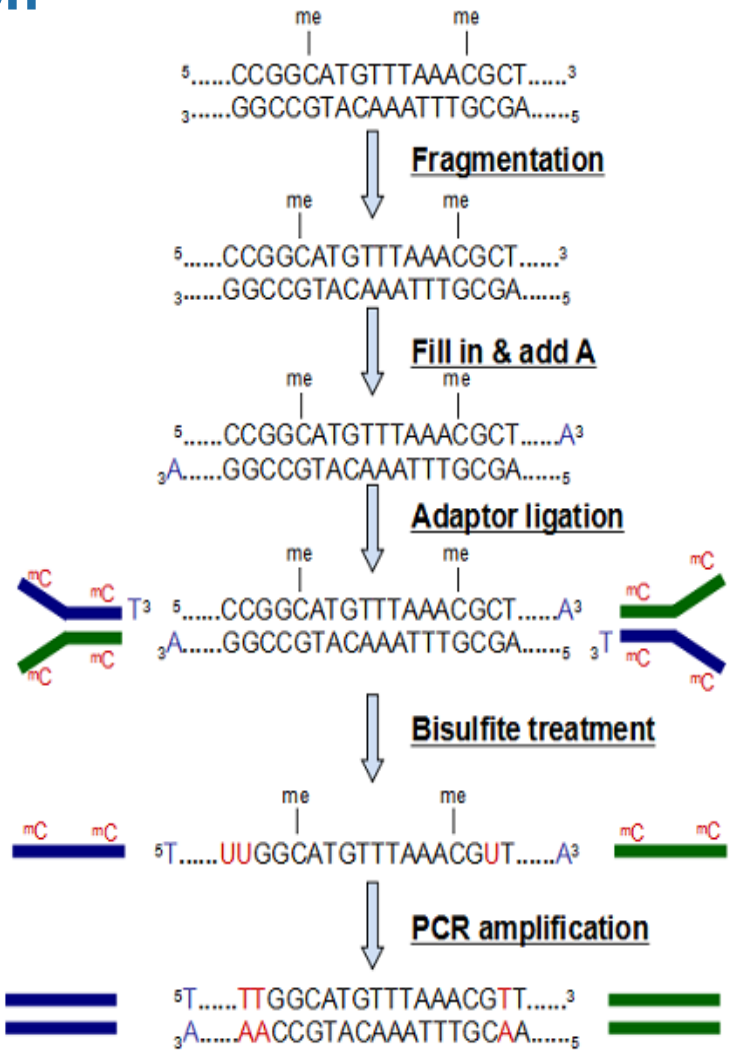
Sample Requirements

Sample Type	Amount (Qubit®)		Volume	Concentration	Purity (NanoDrop™/Agarose Gel)
	Strongly Recommended	Required			
Genomic DNA	≥5μg	≥ 2.5μg	≥ 15μL	≥ 20ng/μL	OD260/230 = 0-3, no degradation, no color, no RNA or protein contamination
Low-input Genomic DNA	≥ 0.4μg	≥ 0.2μg	-	≥ 20ng/μL	

Novogene 诺禾致源

Whole Genome Bisulfite Sequencing

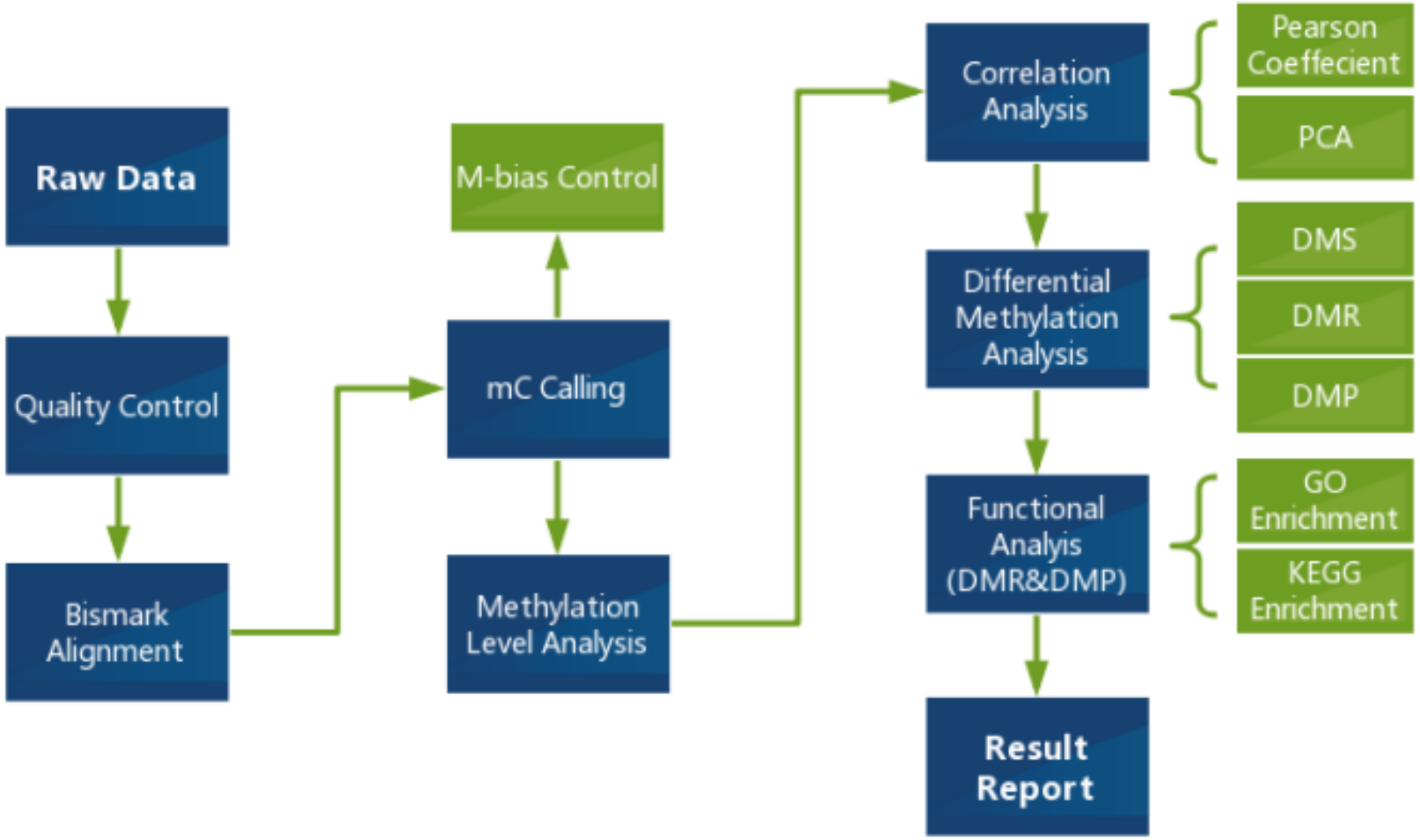
Library construction



Novogene 诺禾致源

Whole Genome Bisulfite Sequencing

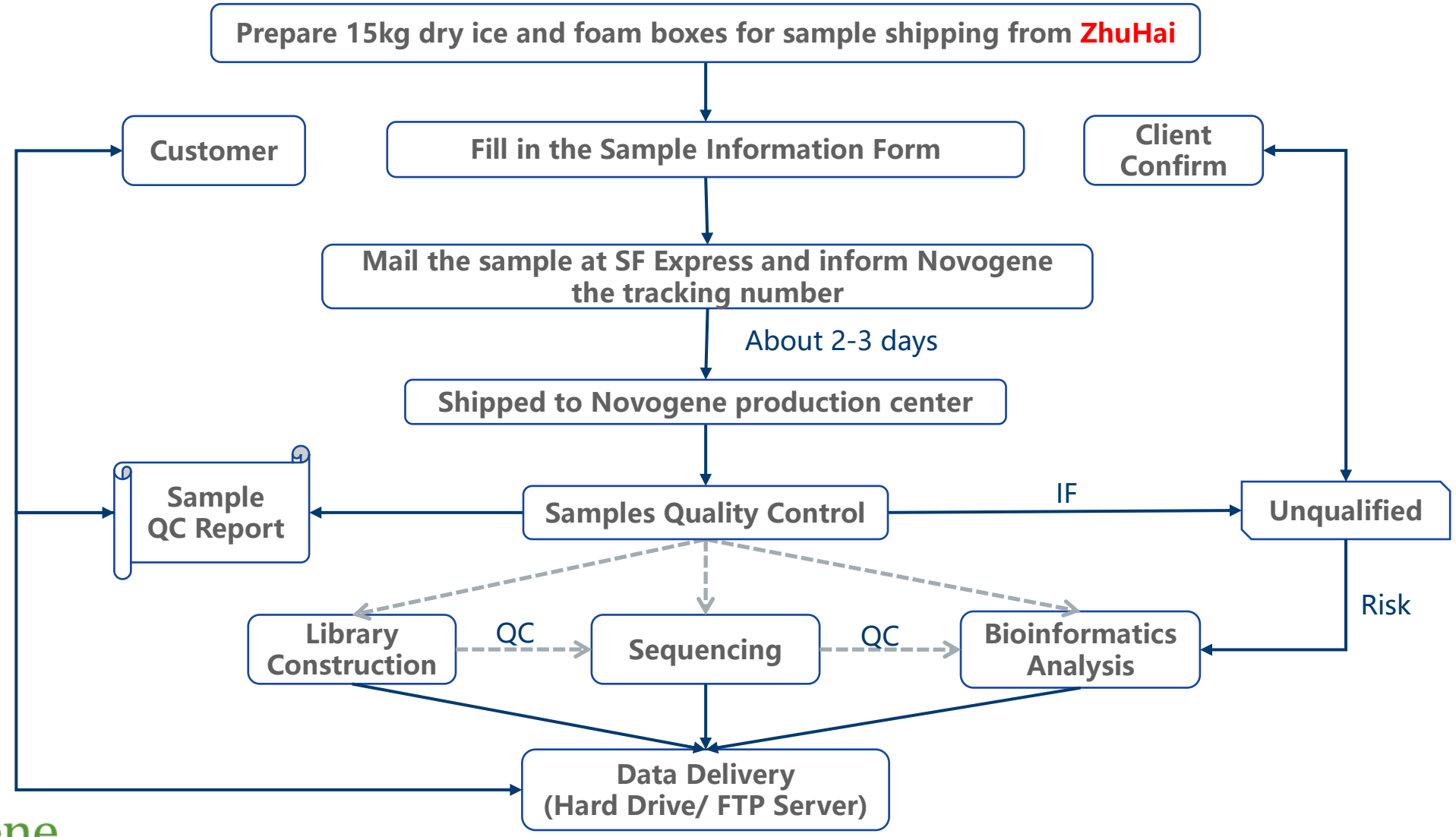
Standard Bioinformatics



Novogene 诺禾致源

Content

Sample Shipping Workflow (Macau)



** Macau to HK – depends on COVID-19 Situation

Sample Shipping Workflow (Macau)

Dry ice provider (Taobao):

1) 源太干冰
2) 火星干冰

Please order 1 day in advance and ship to ZhuHai

Shipping address of Tianjin:

Novogene BuildingB07,Venture Headquarter Base,Wuqing Development Area,Tianjin,China.
天津市武清区创业总部基地B07栋
Contact: Sample Receiving Department
联系人：天津科服收样组
Tel: +86 18522699037

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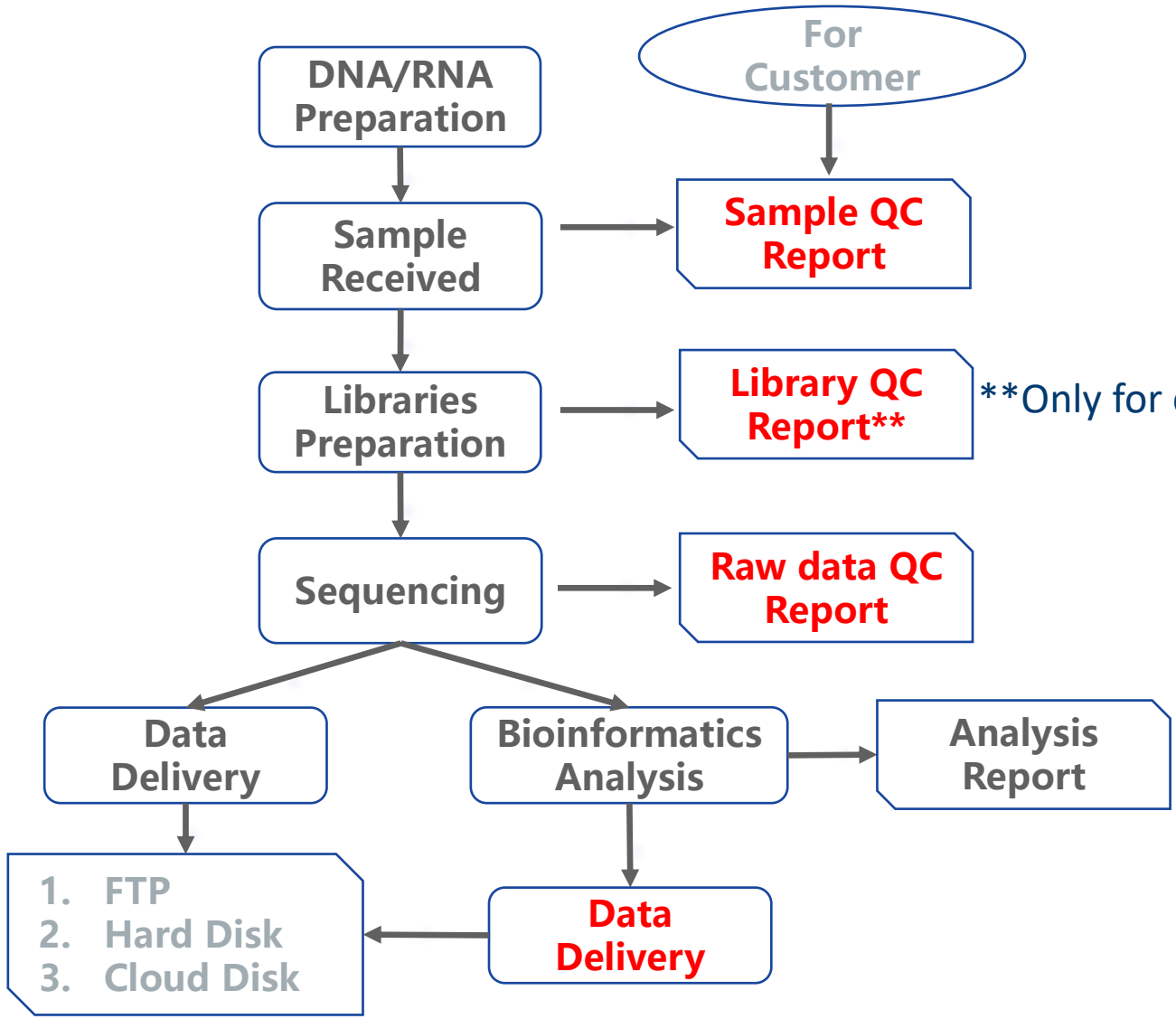
Extensive Quality Control



**Only for customer submit library samples

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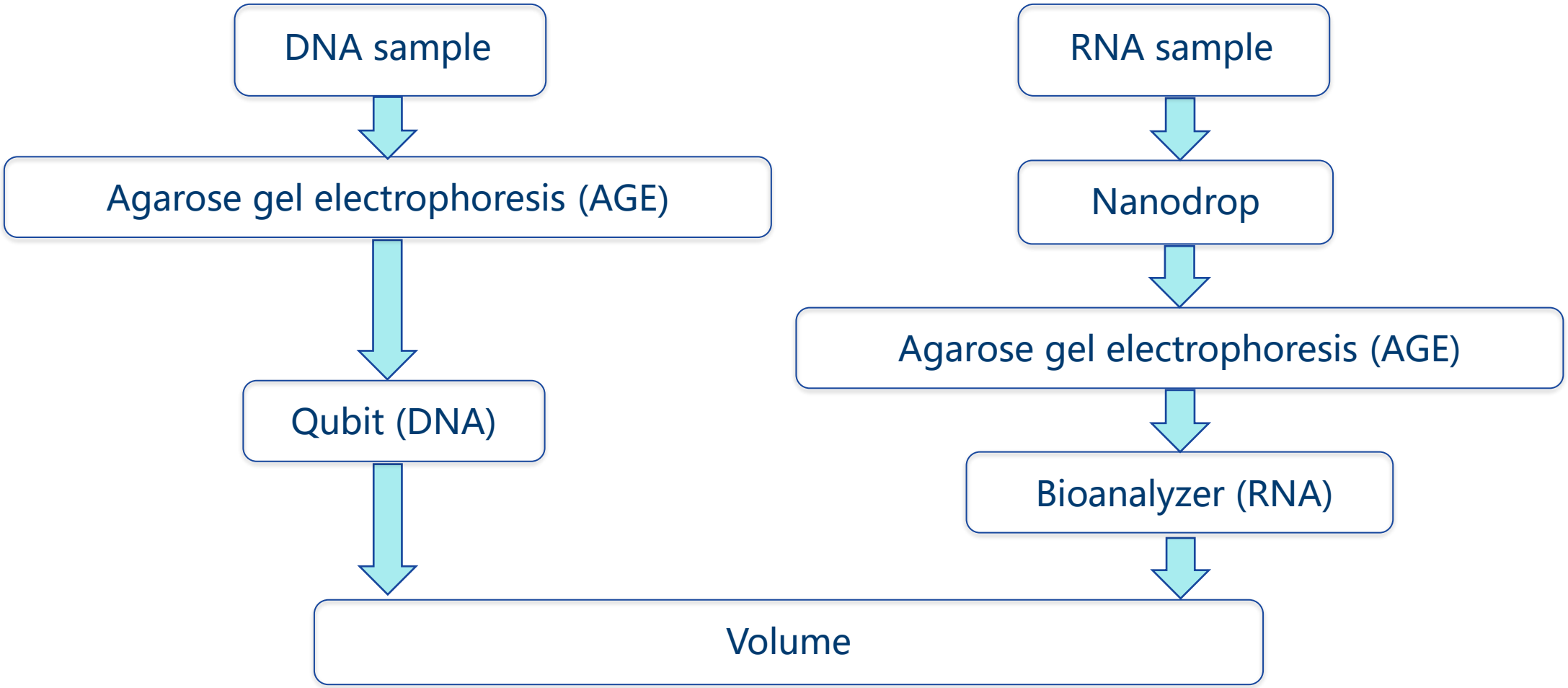
Sample QC



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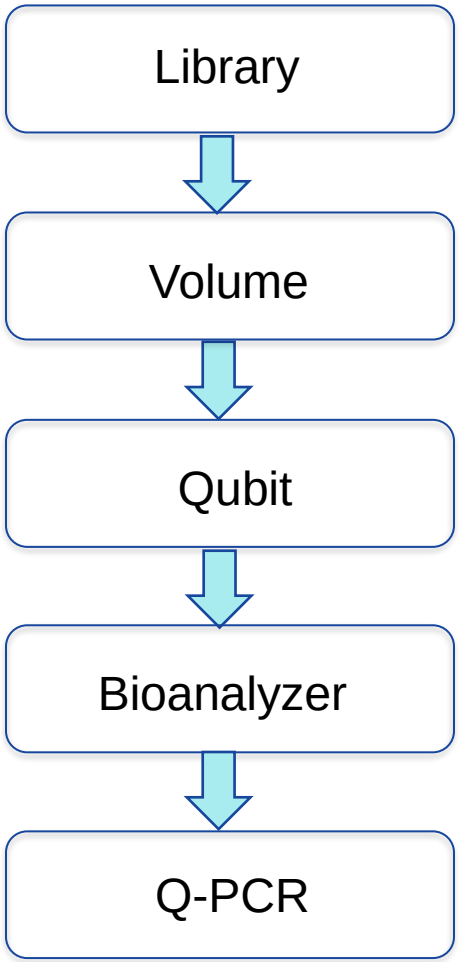
Library QC



We have strict quality control steps, only qualified library can be used for sequencing and analysis. The quality control include volume, concentration and library peak shape.

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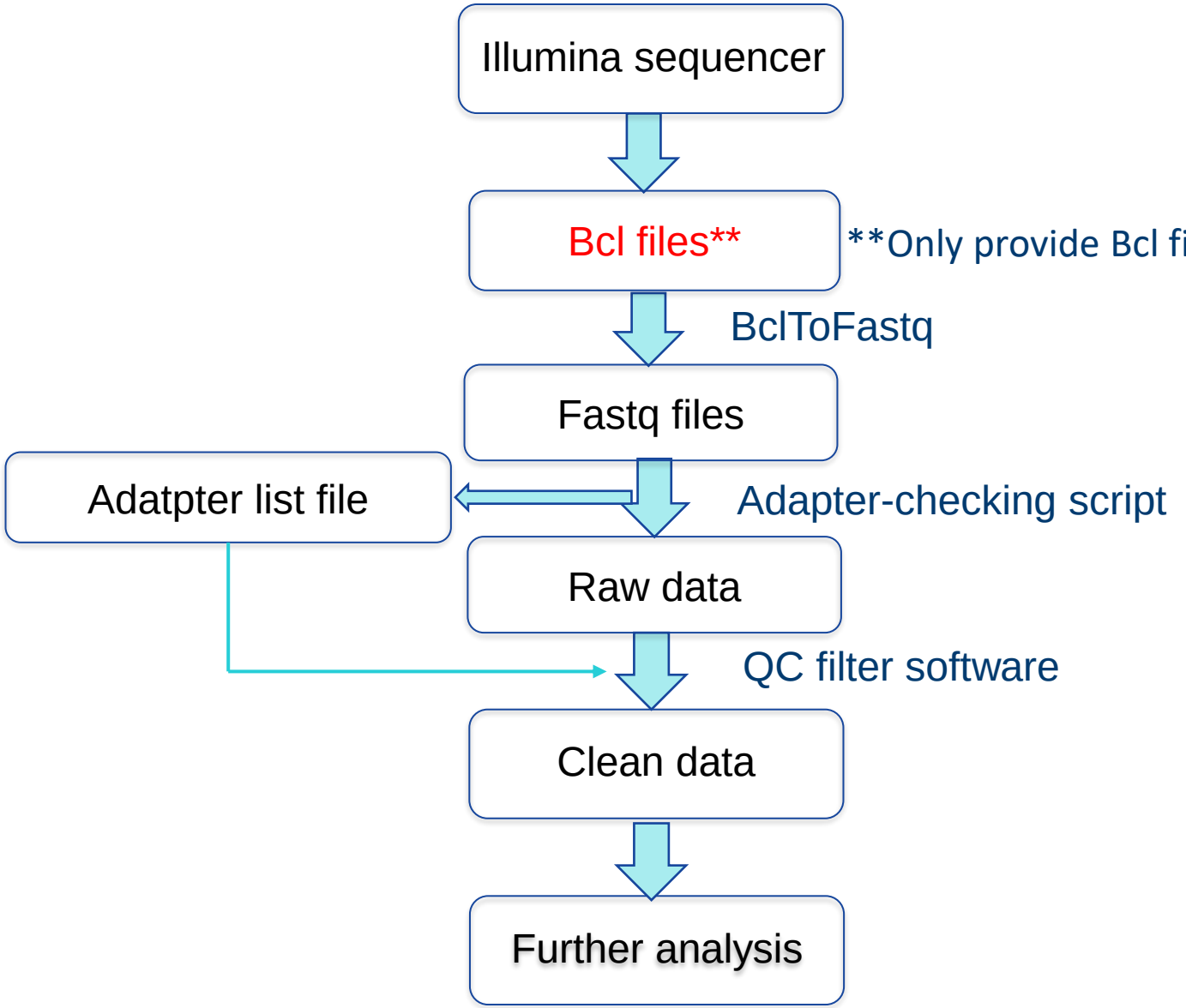
Data Quality



Novogene 诺禾致源

Content

Novogene Mission

Founded in March 2011

With cutting-edge genomic technologies and high-performance computing, Novogene is dedicated to improving life science research and human healthcare.

Executive Team

Dr. Ruiqiang Li, Ph.D.

Founder

Dr. Ruiqiang Li, is widely recognized as one of the world's leading experts in genomics and bioinformatics, owning 17 patents and has authored over 100 publications that have been cited more than 42,000 times. Best known for developing the software SOAP (Short Oligonucleotide Analysis Package) for ultra-fast sequence mapping, variation detection, and de novo genome assembly, Dr. Li has focused the company on being a full-service provider, performing NGS services and providing bioinformatic analysis to fully empower and enable our customers to move forward rapidly and effectively in their scientific endeavors.

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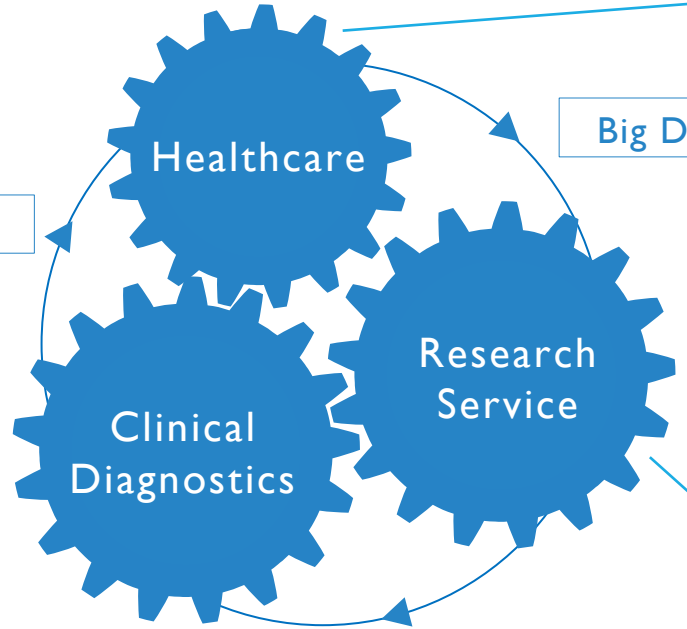
Novogene Strategy

- Carrier Testing
- New-born Screening
- Personal Genomics



Big Data

Clinical Quality

- CAP, CLIA, ISO13485
- NGS based IVD
- Digital PCR based IVD

- Largest Sequencing Facility in the World
- Cutting Edge & Comprehensive Solutions
- Global Research Network

Solid Scientific Foundation and Innovation Capability

NGS | **Super Computing/ Data Centre** | **Bioinformatics** | **Clinical Lab** | **GMP** | **Global Commercial Network**

Novogene 诺禾致源

Novogene Global Presence

2011 Beijing

2014 Tianjin Sequencing Center & Clinical Center

2017 Nanjing Sequencing Center

2013 Hong Kong Branch

2014 U.S. Branch

2016 UC Davis Sequencing Center

2014 U.K. Branch

2018 U.K. Sequencing Center

2016 Singapore Branch

2016 Singapore Sequencing Center

Novogene 诺禾致源

Largest Range of Platforms to Serve Diverse Needs

>280,000 Human WGS/Year

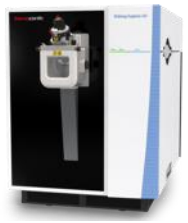
NovaSeq6000

Pacbio Sequel & Sequel II

Nanopore

HiSeq X

HiSeq 2000/2500/4000



Orbitrap Exploris™480

DA8600

Q Exactive™ HF-X

S5XL

NextSeq 500

Miseq

QTRAP® 6500+

Novogene 诺禾致源

First intelligent multi-product NGS delivery platform

Falcon system

Launched in 2020

Fully automated production line → reduce manual errors and basis

Safe and accurate one-stop ground-breaking solution

Novogene 诺禾致源

Super Computing and Data Center

280,000 Human WGS Analysis / Year

1,000 Human WGS Analysis / Day

High Performance Computing

Capability	Capacity
Memory Size	410 TB
Computing power	1727 T flops
Storage	60.2 PB

Why Novogene?

- Quality
- Analysis
- Technology
- Capacity
- Turnaround

Summary of Novogene Competitive Advantages

- ✓ **Corporation** – Global company, multi-geographic laboratories with multinational talents.
- ✓ **Platform** – The largest sequencing center equipped with the latest and comprehensive platforms.
- ✓ **Team** – 1,800 professionals, dedicated and specialised groups.
- ✓ **Quality** – Clinical standards (CAP, CLIA & ISO13485), automatic workflow, LIMS.
- ✓ **Experience** – 28,000+ projects and one million samples in 2017, 26,000+ global customers.
- ✓ **Innovation** – Sustainable development powered by R&D (over £15 Million), comprehensive product portfolio
- ✓ **Research** – 580+ Publications, global collaboration

Novogene 诺禾致源

Thank You !

www.novogene.com

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