IV РОССИЙСКИЙ КОНГРЕСС
ЛАБОРАТОРНОЙ МЕДИЦИНЫ

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Пущино

Project «Jim»
(первый индивидуальный геном, 2005...2007)

Dr. James Watson, co-discoverer of the DNA helix and father of the Human Genome Project, became the first human to receive the data encompassing his personal genome sequence at Baylor College of Medicine in Houston in **May 31, 2007**.

James Watson (left) receives a digital copy of his genome sequence from Jonathan Rothberg in May 2007.

The 100 000 Genomes Project (2013 - 2018)

The 100,000 Genomes Project by numbers

Posted on September 3, 2018 at 9:00 am

Genomes Sequenced = 81,179

Posted on October 1, 2018 at 9:00 am

Genomes Sequenced = 87,231

https://www.genomicsengland.co.uk/the-100000-genomes-project-by-numbers/

День ДНК в США
(2018 г.)

Broad Institute sequences its 100,000th whole human genome on National DNA Day

By Broad Communications

Milestone crossed on the 15th anniversary of the completion of the Human Genome Project, as the worldwide estimate for whole human genomes sequenced approaches one million

https://www.broadinstitute.org/journalists/logos-graphics

1 000 000 геномов (PMI)
(2016 - 2018)

https://obamawhitehouse.archives.gov/precision-medicine

1 000 000 китайских геномов
(2018 - 2019 г.г.)

China Province of Jiansu will genome sequence one million people within two years

https://www.nextbigfuture.com/2017/11/china-province-of-jiansu-will-genome-sequence-one-million-people-within-two-years.html

China PMI

(100 000 000 геномов; $9,2; 2017 - 2030)

China National GeneBank (CNGB)

https://www.presstv.com/Detail/2016/09/22/485893/China-national-gene-bank

Геномные проекты

Table 1. Completed and ongoing human genome project (HGP)-like projects

Project	Country	Scope	Population size	Period	URL
Human Genome Project—Read	International	Genome	4	1990—2004	http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml
DeCODE Genetics	Iceland	Variants, anthropology, inherited diseases	160 000	1996—present	https://www.decode.is/
Cancer Genome Project	United Kingdom	Cancer	—	2000—present	http://www.sanger.ac.uk/science/groups/cancer-genome-project
SardiNIA Project	International	Variants, disease and age-related	6700	2001—2015?	https://sardinia.irp.nia.nih.gov/index.html
10K Genomes Project (autism)	United States, China	Autism	10 000	2001—present	https://www.autismspeaks.org/site-wide/autism-10k-genome-project
Estonian Genome Project	Estonia	Precision medicine	—	2001—present?	http://www.genomics.ee/genome/index.html
International HapMap Project	International	Variants, precision medicine	1184	2002—2016	https://www.ncbi.nlm.nih.gov/variation/news/NCBI_retiring_HapMap/
African Genome Initiative	International	Variants, precision medicine, anthropology	—	2003—2015	http://www.africagenome.co.za/
Autism Genome Project (AGP)	International	Autism	> 1200	2004—present	https://www.autismspeaks.org/science/initiatives/autism-genome-project
The Cancer Genome Atlas (TCGA)	United States	Cancer	>11 000	2005—2017	https://cancergenome.nih.gov/
The Genographic Project	International	Anthropology	275 000	2005—present	https://genographic.nationalgeographic.com/
Alzheimer's Genome Project	United States	Alzheimer's Disease	1510	2005—present	http://curealz.org/projects/alzheimer%E2%80%99s-genome-project%E2%84%A2
Personal Genomes Project	International	Precision medicine	100 000	2005—present	http://www.personalgenomes.org/
Neanderthal genome project	Germany, United States	Anthropology	5	2006—2013	http://www.eva.mpg.de/neandertal/index.html
Human Variome Project (HVP)	International	Variants, precision medicine	—	2006—present	http://www.humanvariomeproject.org/
Epilepsy Phenome/Genome Project (EPGP)	International	Epilepsy	4000	2006—present?	http://www.epgp.org/
Geisinger MyCode	United States	Precision medicine	134 379	2007—present	http://www.geisinger.org/for-researchers/partnering-with-patients/pages/mycode-health-initiative.html
Human Heredity & Health in Africa (H3Africa)	International	Genetic variability in infectious diseases, genetic diseases	>10 000	2007—present?	http://h3africa.org/
1000 Genomes Project	International	Variants	2504	2008—2015	http://www.internationalgenome.org/
Human Microbiome Project (HMP)	United States	Metagenomics	300	2008—present	http://hmpdacc.org/
Chronic Lymphocytic Leukemia Genome Project	Spain	Cancer	500	2009—2014	http://www.cllgenome.es/
Roadmap Epigenomics Project	United States	Methylation patterns	111	2009—2015?	http://www.roadmapepigenomics.org/
GONL: Genome Of The Netherlands	Netherlands	Variants	769	2009—present	http://www.nlgenome.nl/
UK10K	United Kingdom	Rare variants	10 000	2010—2013	https://www.uk10k.org/
Southern African Human Genome Project	South Africa	Variants	—	2011—2014	http://sahgp.sanbi.ac.za/
Pediatric Cancer Genome Project	United States	Cancer	658	2011—present	https://www.stjude.org/research/pediatric-cancer-genome-project.html
The Iranian Genome Project	Iran	Variants, precision medicine	77	2011—present	https://irangenes.com/
Deciphering Developmental Disorders (DDD)	United Kingdom	Developmental disorders	33 000	2011—present	https://www.ddduk.org/
International Rare Diseases Research Consortium	International	Rare diseases	—	2011—present	http://www.irdirc.org/
Finding Of Rare Disease Genes In Canada (FORGE CANADA)	Canada	Rare diseases	—	2011—present	http://www.genomebc.ca/research-programs/projects/health/finding-of-rare-disease-genes-in-canada-forge-canada/
Belgian Medical Genomics Initiative	Belgium	Variants, precision medicine	—	2012—2017	https://www.belspo.be/belspo/fedra/proj.asp?l=en&COD=P7%2F43
100K Genomes Project	United Kingdom	Rare diseases, cancer	70 000	2012—present	https://www.genomicsengland.co.uk/the-100000-genomes-project/
Alzheimer's Disease Sequencing Project	United States	Alzheimer's Disease	11 491	2012—present	https://www.niagads.org/adsp/content/home
Human Longevity Inc.	United States	Variants	10 545	2013—present	http://www.humanlongevity.com/
Project MinE	International	Amyotrophic Lateral Sclerosis (ALS)	15 000	2013?—present	https://www.projectmine.com/
The Cognomics Project	Netherlands	Genetics and brain function	10 000	2013?—present	http://www.cognomics.nl/
Utah Genome Project	United States	Variants, precision medicine	—	2013?—present	http://healthsciences.utah.edu/utah-genome-project/
MSSNG	United States	Autism	>10 000	2014—present	https://www.mss.ng/
The Precision Medicine Initiative (PMI)	United States	Variants, precision medicine	1 000 000	2015—present	https://obamawhitehouse.archives.gov/precision-medicine
Genome Russia Project	Russia	Variants, precision medicine, anthropology	2980	2015—present	http://genomerussia.spbu.ru/
Human Genome Project—Write	United States	Artificial synthesis of genome	0	2016—2026	http://science.sciencemag.org/content/early/2016/06/01/science.aaf6850/tab-pdf
AstraZeneca 2 million Genomes	United States, United Kingdom, Finland	Variants, precision medicine	2 000 000	2016—present	https://www.astrazeneca.com/Harnessing-the-code-of-life-to-develop-new-treatments.html
Epi25 Project	International	Epilepsy	25	2016—present	http://epi25.org/
Proyecto Genoma Navarra (NAGEN)	Spain	Rare diseases and cancer	—	2016—present	http://www.navarrabiomed.es/en/presentation/navarrabiomed
The Danish Reference Genome Project	Denmark	Variants, precision medicine, anthropology	150	2017	http://www.genomedenmark.dk/english/about/referencegenome/
The Norwegian 1000 Genomes Project	Norway	Variants, precision medicine	1000	2017	http://kreftgenomikk.no/en/1000genomes/
Swedish Genomes Program	Sweden	Variants, precision medicine	1000	2017	https://www.scilifelab.se/research/national-projects/swedish-genomes-program/
Singapore Genome Variation Project	Singapore	Variants	292	Finished	http://www.statgen.nus.edu.sg/~SGVP/default.html
Singapore Sequencing Malay	Singapore	Variants	100	Finished?	http://www.statgen.nus.edu.sg/~SSMP/
Melanoma Genome Project	Australia	Melanoma	500	Ongoing	https://www.melanoma.org.au/research/current-projects/melanoma-genome-project/
GCAT Genomes for life	Spain	Cancer, diabetes, cardiovascular disease respiratory disease and others	50 000	Ongoing	http://www.gcatbiobank.org/qui-som/en_index/
4D Genome Project	Spain	3D genome structure	—	Ongoing	http://www.crg.eu/en/content/research/4dgenome-erc-synergy-project
RD-connect	International	Rare diseases databases platform	—	Ongoing	http://rd-connect.eu/
Metastatic Breast Cancer Project	United States	Metastatic breast cancer	—	Ongoing	https://www.mbcproject.org/
Human Cell Atlas Project	International	Health and disease	—	Ongoing	https://www.humancellatlas.org/
Centers for Mendelian Genomics (CMG)	United States	Mendelian diseases	>20 000	Ongoing	http://mendelian.org/
International Cancer Genome Consortium (ICGC)	International	Many cancers	>16 000	Ongoing	http://icgc.org/
Qatar Genome Programme	Qatar	Genome	27 000	Ongoing	http://www.qatargenome.org.qa/
GUARDIAN	India	Rare diseases	—	Ongoing	http://guardian.meragenome.com/home
Initiative on Rare and Undiagnosed Diseases (IRUD)	Japan	Rare diseases	—	Ongoing	http://www.amed.go.jp/en/program/IRUD/
GenomeAsia100K	South Korea, India	Asian reference genomes, variation	100 000	Ongoing?	http://www.genomeasia100k.com/
Saudi Human Genome Program	Saudi Arabia	Variation and genetic disease	100 000	Ongoing?	http://shgp.kacst.edu.sa/site/
African Genome Variation Program	United Kingdom, Uganda	Variants	1000	Ongoing	http://www.sanger.ac.uk/science/collaboration/african-genome-variation-project
Epi4K	International	Epilepsy	4	Ongoing	http://www.epi4k.org/
China Precision Medicine Initiative	China	Variants, precision medicine	1 000 000	—	–
African Ancestry Project	United States	Ancestry and health	—	—	https://blog.23andme.com/23andme-research/23andmes-african-ancestry-project/
France Population-scale Seq	France	Genome	—	—	http://www.fiercebiotech.com/it/france-plans-745m-investment-to-build-235-000-genome-a-year-sequencing-operation
Korean Reference Genome Project	South Korea	Variants, precision medicine	—	—	http://koreagenome.kobic.re.kr/

Human genomics projects and precision medicine / F. Carrasco-Ramiro, R. Peiró-Pastor, B. Aguado
Gene Therapy (2017) 24, 551–561

“Нацпроекты”

- Saudi human genome project (SGP) ~ 33 млн. геномов

- Qatar genome project (QGP) ~ 0,3 млн. геномов

- Iceland’s Genome Project ~ 0,3 млн. геномов

- Korea's Personal Genome Project ~ 51 млн. геномов

- Dubai Genomics ~ 3 млн. геномов

Ещё «немного» китайских геномов

(100 + 200 + 500 тысяч)

GENOME DECODE PROGRAM (GDP) was launched at the 12th International Conference on Genomics (ICG-12) in October 2017. The program is composed of three phases: to advance the sequencing cost reduction, to achieve the“hundred dollars genome”, and to achieve Whole Genome Resequencing for every individual. With BGI, decoding the genome has never been easier.

http://gdp.bgi.com/page/index/en/

Китайские секвенаторы

MGISEQ-200

MGISEQ-2000

Performance Parameters

Chip types	FC	**Average Effective Signal Point**	300M	**Average Output**	15-60Gb/run
Chip lanes number	1 lane	**Read Length** *	SE50 SE100 PE50 PE100	Run time**	≤ 48 hours

FCS Chip					
Lane numbers	2 lanes	**Read Length** *	SE50 SE100 PE50 PE100	Run time **	≤ 36 hours
Average Effective Signal Point	Single-chip: 375M	**Average Output**	Single-chip: 18.75-75Gb/run		
	Double-chip: 750M		Double-chip: 37.5-150Gb/run		
FCL Chip					
Lane numbers	4 lanes	**Read Length** *	SE50 SE100 PE50 PE100	Sequencing time**	≤ 48 hours
Average Effective Signal Point	Single-chip: 1500M	**Average Output**	Single-chip: 75-300Gb/run		
	Double-chip: 3000M		Double-chip: 150-600Gb/run		

Секвенаторы компании Illumina

Sequencing System	iSeq™	MiniSeq™	MiSeq®	NextSeq®	HiSeq® 4000	HiSeq® X Five/Ten	NovaSeq® 6000
Output per run	1.2 Gb	7.5 Gb	15 Gb	120 Gb	1.5 Tb	1.8 Tb	1 Tb - 6 Tb[1]
Instrument price	$19.9K	$49.5K	$99K	$275K	$900K	$6M[2]/$10M[2]	$985K

Illumina Investor Presentation
May 3, 2018

Американские секвенаторы

iSeq 100

System Specifications

1.2 Gb	4 million	2 × 150 bp
MAX OUTPUT	READS PER RUN	MAX READ LENGTH

NovaSeq 6000

OUTPUT	167 – 6000 Gb
READ NUMBER	1.6 – 20B
RUN TIME	Fastest (40 Hr. for 2T Run)
Flow Cells	4 Types

Секвенаторы 2018 года

illumina

MGI 华大智造

Oxford NANOPORE Technologies

NovaSeq 6000

PromethION

1 Tbp

MGISEQ-2000

GridION X5

100 Gbp

MGISEQ-200

MinION

10 Gbp

iSeq 100

MinION Dx

Британские секвенаторы

MinION

GridION

SmidgION

Flongle

PromethION

MinION

Вес – 78 г
Мощность – 1 Вт
Размеры – 105x23x33 мм
Количество нанопор – до 512 (2048)
Интерфейс – USB 3.0 (до 5 Gbps)
Стоимость проточной ячейки - $500…$900
Производительность – 10…20 Gbp за 48 часов

GridION x5

Вес – 10 кг
Мощность – 600 Вт
Размеры – 360x200x360 мм
Количество нанопор – до 512x5
Стоимость проточной ячейки - $299…$900
Производительность – 50…100 Gbp за 48 часов

Технологии секвенирования 2018 года

Флуоресцентная (мультимолекулярная)

illumina

华大基因 BGI

Длина чтения:

до 150 п.н. до 100 п.н.
или до 150x2 п.н. или до 100x2 п.н.

Количество ридов:

до $20x10^9$ до $3x10^9$

Точность чтения:

99,9% (>80% ридов)

Основная мишень:

SNP

(single nucleotide polymorphism)

Нанопоровая (мономолекулярная)

Oxford NANOPORE Technologies®

Средняя длина чтения:

10 000 … 100 000 п.н. (до 1 000 000 п.н.)

Количество нанопор:

MinION	PromethION
512 (2048)	3000 (6000)

Точность чтения:

~ 90% (2D/1D2 ~95%)

Основная мишень:

CNV

(copy number variations)

Геномная геополитика
(2018 год)



Обладание технологиями геномного секвенирования - прерогатива сверхдержав.

«Россия была и будет сверхдержавой. Или России не будет.»
(Яков Кедми)

Если Россия хочет оставаться сверхдержавой, то она должна обладать технологиями геномного секвенирования.

Закупка №0195100000218000261

ИНФОРМАЦИЯ ОБ ОБЪЕКТЕ ЗАКУПКИ

Описание объекта закупки

Выполнение опытно-конструкторской работы на тему: «Создание аппаратно-программного комплекса, набора реагентов и расходных материалов для расшифровки последовательности нуклеиновых кислот патогенных микроорганизмов методом массового параллельного секвенирования» (Шифр «Сиквенс-Био»)

КОД ПОЗИЦИИ	НАИМЕНОВАНИЕ ТОВАРА, РАБОТЫ, УСЛУГИ ПО КТРУ	ЕДИНИЦА ИЗМЕРЕНИЯ	КОЛИЧЕСТВО	ЦЕНА ЗА ЕД.ИЗМ.	СТОИМОСТЬ
72.19.50.000	Работы оригинальные научных исследований и экспериментальных разработок в области естественных и технических наук, кроме биотехнологии	Штука	1	346 559 010,00	346 559 010,00
				Итого:	346 559 010,00 (Российский рубль)

http://www.zakupki.gov.ru/epz/order/notice/ok44/view/common-info.html?regNumber=0195100000218000261

Happy New Sequencing!

Благодаря за вниманието