



IV РОССИЙСКИЙ КОНГРЕСС
ЛАБОРАТОРНОЙ МЕДИЦИНЫ

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Пушино*

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 by numbers



Posted on September 3, 2018 at 9:00 am

Genomes Sequenced = 81,179

8 1 1 7 9



Posted on October 1, 2018 at 9:00 am

Genomes Sequenced = 87,231

8 7 2 3 1

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



1 000 000 геномов (PMI)
(2016 - 2018)



THE PRECISION MEDICINE INITIATIVE



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

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



China PMI

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



China National GeneBank (CNGB)

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://webornl.gov/sci/techresources/Human_Genome/index.shtml
deCODE Genetics	Iceland	Variants, anthropology, inherited diseases	160 000	1996—present	http://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?	http://sardinia.lipni.nih.gov/index.html
10K Genomes Project (autism)	United States, China	Autism	10 000	2001—present	http://www.autismpeaks.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	http://www.ncbi.nlm.nih.gov/variation/news/ncbi_releasing_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	http://www.autismpeaks.org/science/initiatives/autism-genome-project
The Cancer Genome Atlas (TCGA)	United States	Cancer	> 11 000	2005—2017	http://cancergenome.nih.gov/
The Genographic Project	International	Anthropology	275 000	2005—present	http://genographic.nationalgeographic.com/
Alzheimer's Genome Project	United States	Alzheimer's Disease	1510	2005—present	http://curialab.org/projects/alzheimers%2080%99-genome-project%2064%9A2
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.epgpg.org/
Gelinger MyCode	United States	Precision medicine	134 379	2007—present	http://www.gelinger.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	2594	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.cligenepe.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/

Table 1. (Continued)

Project	Country	Scope	Population size	Period	URL
UK10K	United Kingdom	Rare variants	10 000	2010—2013	http://www.uk10k.org/
Southern African Human Genome Project	South Africa	Variants	—	2011—2014	http://ahgpa.ac.za/
Paediatric Cancer Genome Project	United States	Cancer	658	2011—present	http://www.rtdg.duke.edu/research/pediatric-cancer-genome-project.html
The Iranian Genome Project	Iran	Variants, precision medicine	77	2011—present	http://irangenome.com/
Defining Developmental Disorders (DDO)	United Kingdom	Developmental disorders	33 000	2011—present	http://www.ddo.king.ac.uk/
International Rare Diseases Research Consortium	International	Rare diseases	—	2011—present	http://www.irdc.org/
Finding Of Rare Disease Genes In Canada (FORGE CANADA)	Canada	Rare diseases	—	2011—present	http://www.genomics.ca/research-programs/projects/finding-of-rare-disease-genes-in-canada-forge-canada/
Belgian Medical Genomics Initiative	Belgium	Variants, precision medicine	—	2012—2017	http://www.belgium.be/health/medgen/program?l=en&CID=17962/43
100K Genomes Project	United Kingdom	Rare diseases, cancer	70 000	2012—present	http://www.genomicsuk.ac.uk/the-100000-genome-project/
Alzheimer's Disease Sequencing Project	United States	Alzheimer's Disease	11 491	2012—present	http://www.alzdisi.org/ledq/identif/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	http://www.projectmin.com/
The Cognomics Project	Netherlands	Genetics and brain function	10 000	2013—present	http://www.cognomics.nl/
Utah Genom Project	United States	Variants, precision medicine	—	2013—present	http://healthsciences.utah.edu/utah-genome-project/
MESNG	United States	Autism	> 10 000	2014—present	http://www.mesng.org/
The Precision Medicine Initiative (PMI)	United States	Variants, precision medicine	1 000 000	2015—present	http://obamawhitehouse.archives.gov/precision-medicine/
Genome Russia Project	Russia	Variants, precision medicine, anthropology	2 980	2015—present	http://genomarusia.spb.ru/
Human Genome Project—Africa	United States	Artificial synthesis of genome	0	2016—2026	http://ciencia.sciencemag.org/content/early/2016/06/01/science.aaf00203v.pdf
AfricaZambia 2 million Genomes	United States, United Kingdom, Finland	Variants, precision medicine	2 000 000	2016—present	http://www.africanzambia2million.org/the-code-of-life-to-develop-new-medicines.html
EpIQ5 Project	International	Epilepsy	25	2016—present	http://epiq5.org/
Projecto Genoma Navarra (NAG20E)	Spain	Rare diseases and cancer	—	2016—present	http://www.navarrabiomed.es/en/precision-tation/navarrabiomed
The Danish Reference Genome Project	Denmark	Variants, precision medicine, anthropology	150	2017	http://www.genomedanmark.dk/english/about/reference-genome/
The Norwegian 1000 Genomes Project	Norway	Variants, precision medicine	1 000	2017	http://norg1000genomes.kibin.no/1000genomes/

Table 1. (Continued)

Project	Country	Scope	Population size	Period	URL
Swedish Genomes Program	Sweden	Variants, precision medicine	1000	2017	https://www.scilifelab.se/research/national-projects/swedish-genome-program/
Singapore Genome Variation Project	Singapore	Variants	292	Finished	https://www.sitgen.nus.edu.sg/~SGVP/default.htm
Singapore Sequencing Malay	Singapore	Variants	100	Finished?	http://www.sitgen.nus.edu.sg/~SSMP/
Melanoma Genome Project	Australia	Melanoma	500	Ongoing	https://www.melanoma.org.au/research/australian-genetic-melanoma-genome-project/
GCAT Genomes for Life	Spain	Cancer, diabetes, cardiovascular disease and others	50 000	Ongoing	http://www.gcatbiobanque.org/qui-est-nous/index/
4D Genome Project	Spain	3D genome structure	—	Ongoing	http://www.4dgenome.com/content/research/4dgenome-enc-synseq-y-project
RD-connect	International	Rare diseases databases platform	—	Ongoing	http://rd-connect.eu/
Metastatic Breast Cancer Project	United States	Metastatic breast cancer	—	Ongoing	http://www.mbcproject.org/
Human Cell Atlas Project	International	Health and disease	—	Ongoing	http://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.svgenomics.com/home
Initiative on Rare and Undiagnosed Diseases (IRUD)	Japan	Rare diseases	—	Ongoing	http://www.irdc.jp/en/program/IRUD/
Genome Asia100K	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://h3g.scienceministry.gov.sa/
African Genome Variation Program	United Kingdom, Uganda	Variants	1000	Ongoing?	http://www.sanger.ac.uk/science/collaboration/african-genome-variation-project
EpIK	International	Epilepsy	4	Ongoing?	http://www.epik.org/
China Precision Medicine Initiative	China	Variants, precision medicine	1 000 000	—	—
African Ancestry Project	United States	Ancestry and health	—	—	https://biog.23andme.com/23andme-research/23andme-african-ancestry-project/
French Population-scale Sequencing	France	Genome	—	—	https://www.francobioinformatics.fr/france-plans-2025-en-biomedecine-04-25-2020-genome-sequence-1-seqan-claq-operat-on
Korean Reference Genome Project	South Korea	Variants, precision medicine	—	—	http://koreagenomebio.kribic.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.



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

iSeq 100



NovaSeq 6000



System Specifications

 1.2 Gb MAX OUTPUT	 4 million READS PER RUN	 2 x 150 bp MAX READ LENGTH
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 OUTPUT	167 – 6000 Gb
 READ NUMBER	1.6 – 20B
 RUN TIME	Fastest (40 Hr. for 2T Run)
 Flow Cells	4 Types

Секвенаторы 2018 года



1 Tbp



NovaSeq 6000



PromethION



MGISEQ-2000



GridION X5



MGISEQ-200



MinION

10 Gbp



iSeq 100



MinION Dx

100 Gbp

Британские секвенаторы



SmidgION



MinION



Flongle



GridION



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



GridIONx5



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)



Длина чтения:

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

Количество ридов:

до 20×10^9 до 3×10^9

Точность чтения:

99,9% (>80% ридов)

Основная мишень:

SNP

(single nucleotide polymorphism)

Нанопоровая
(мономолекулярная)



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)

Happy New Sequencing!

Благодарю за внимание