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IV РОССИЙСКИЙ КОНГРЕСС  
ЛАБОРАТОРНОЙ МЕДИЦИНЫ

# Секвенирование - 2018

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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	<a href="http://webornl.gov/sci/techresources/Human_Genome/index.shtml">http://webornl.gov/sci/techresources/Human_Genome/index.shtml</a>
DeCODE Genetics	Iceland	Variants, anthropology, inherited diseases	160 000	1996—present	<a href="http://www.decode.is/">http://www.decode.is/</a>
Cancer Genome Project	United Kingdom	Cancer	—	2000—present	<a href="http://www.sanger.ac.uk/science/groups/cancer-genome-project">http://www.sanger.ac.uk/science/groups/cancer-genome-project</a>
Sardinia Project	International	Variants, disease and age-related	6700	2001—2015?	<a href="http://sardinia.lipni.nih.gov/index.html">http://sardinia.lipni.nih.gov/index.html</a>
10K Genomes Project (autism)	United States, China	Autism	10 000	2001—present	<a href="http://www.autismpeaks.org/site-wide/autism-10k-genome-project">http://www.autismpeaks.org/site-wide/autism-10k-genome-project</a>
Estonian Genome Project	Estonia	Precision medicine	—	2001—present?	<a href="http://www.genomics.ee/genome/index.html">http://www.genomics.ee/genome/index.html</a>
International HapMap Project	International	Variants, precision medicine	1184	2002—2016	<a href="http://www.ncbi.nlm.nih.gov/variation/news/ncbi_releasing_HapMap/">http://www.ncbi.nlm.nih.gov/variation/news/ncbi_releasing_HapMap/</a>
African Genome Initiative	International	Variants, precision medicine, anthropology	—	2003—2015	<a href="http://www.africagenome.co.za/">http://www.africagenome.co.za/</a>
Autism Genome Project (AGP)	International	Autism	> 1200	2004—present	<a href="http://www.autismpeaks.org/science/initiatives/autism-genome-project">http://www.autismpeaks.org/science/initiatives/autism-genome-project</a>
The Cancer Genome Atlas (TCGA)	United States	Cancer	> 11 000	2005—2017	<a href="http://cancergenome.nih.gov/">http://cancergenome.nih.gov/</a>
The Geographic Project	International	Anthropology	275 000	2005—present	<a href="http://genographic.nationalgeographic.com/">http://genographic.nationalgeographic.com/</a>
Alzheimer's Genome Project	United States	Alzheimer's Disease	1510	2005—present	<a href="http://curialz.org/projects/alzheimers%2080%99-genome-project%2064%9A2">http://curialz.org/projects/alzheimers%2080%99-genome-project%2064%9A2</a>
Personal Genomes Project	International	Precision medicine	100 000	2005—present	<a href="http://www.personalgenomes.org/">http://www.personalgenomes.org/</a>
Neanderthal genome project	Germany, United States	Anthropology	5	2006—2013	<a href="http://www.eva.mpg.de/neanderthal/index.html">http://www.eva.mpg.de/neanderthal/index.html</a>
Human Variome Project (HVP)	International	Variants, precision medicine	—	2006—present	<a href="http://www.humanvariomeproject.org/">http://www.humanvariomeproject.org/</a>
Epilepsy Phenome/Genome Project (EPGP)	International	Epilepsy	4000	2006—present?	<a href="http://www.epgpg.org/">http://www.epgpg.org/</a>
Gelinger MyCode	United States	Precision medicine	134 379	2007—present	<a href="http://www.gelinger.org/for-researchers/partnering-with-patients/pages/mycode-health-initiative.html">http://www.gelinger.org/for-researchers/partnering-with-patients/pages/mycode-health-initiative.html</a>
Human Heredity & Health in Africa (H3Africa)	International	Genetic variability in infectious diseases, genetic diseases	> 10 000	2007—present?	<a href="http://h3africa.org/">http://h3africa.org/</a>
1000 Genomes Project	International	Variants	2594	2008—2015	<a href="http://www.internationalgenome.org/">http://www.internationalgenome.org/</a>
Human Microbiome Project (HMP)	United States	Metagenomics	300	2008—present	<a href="http://hmpdpc.org/">http://hmpdpc.org/</a>
Chronic Lymphocytic Leukemia Genome Project	Spain	Cancer	500	2009—2014	<a href="http://www.cligenepe.es/">http://www.cligenepe.es/</a>
Roadmap Epigenomics Project	United States	Methylation patterns	111	2009—2015?	<a href="http://www.roadmapepigenomics.org/">http://www.roadmapepigenomics.org/</a>
GONL: Genome Of The Netherlands	Netherlands	Variants	769	2009—present	<a href="http://www.nlgenome.nl/">http://www.nlgenome.nl/</a>

**Table 1.** (Continued)

Project	Country	Scope	Population size	Period	URL
UK10K	United Kingdom	Rare variants	10 000	2010—2013	<a href="http://www.uk10k.org/">http://www.uk10k.org/</a>
Southern African Human Genome Project	South Africa	Variants	—	2011—2014	<a href="http://ahgpa.ac.za/">http://ahgpa.ac.za/</a>
Paediatric Cancer Genome Project	United States	Cancer	658	2011—present	<a href="http://www.rtdg.duke.edu/research/pediatric-cancer-genome-project.html">http://www.rtdg.duke.edu/research/pediatric-cancer-genome-project.html</a>
The Iranian Genome Project	Iran	Variants, precision medicine	77	2011—present	<a href="http://irangenome.com/">http://irangenome.com/</a>
Defining Developmental Disorders (DOD)	United Kingdom	Developmental disorders	33 000	2011—present	<a href="http://www.doduk.org/">http://www.doduk.org/</a>
International Rare Diseases Research Consortium	International	Rare diseases	—	2011—present	<a href="http://www.irdrc.org/">http://www.irdrc.org/</a>
Finding Of Rare Disease Genes In Canada (FORGE CANADA)	Canada	Rare diseases	—	2011—present	<a href="http://www.genomics.ca/research-programs/projects/finding-of-rare-disease-genes-in-canada-forge-canada/">http://www.genomics.ca/research-programs/projects/finding-of-rare-disease-genes-in-canada-forge-canada/</a>
Belgian Medical Genomics Initiative	Belgium	Variants, precision medicine	—	2012—2017	<a href="http://www.belga.be/belstop/medgen/program?l=en&amp;CID=17962/43">http://www.belga.be/belstop/medgen/program?l=en&amp;CID=17962/43</a>
100K Genomes Project	United Kingdom	Rare diseases, cancer	70 000	2012—present	<a href="http://www.genomicsuk.ac.uk/the-100000-genome-project/">http://www.genomicsuk.ac.uk/the-100000-genome-project/</a>
Alzheimer's Disease Sequencing Project	United States	Alzheimer's Disease	11 491	2012—present	<a href="http://www.alzdisi.org/ledq/identif/home">http://www.alzdisi.org/ledq/identif/home</a>
Human Longevity Inc.	United States	Variants	10 545	2013—present	<a href="http://www.humanlongevity.com/">http://www.humanlongevity.com/</a>
Project MinE	International	Amyotrophic Lateral Sclerosis (ALS)	15 000	2013—present	<a href="http://www.projectmin.com/">http://www.projectmin.com/</a>
The Cognomics Project	Netherlands	Genetics and brain function	10 000	2013—present	<a href="http://www.cognomics.nl/">http://www.cognomics.nl/</a>
Utah Genom Project	United States	Variants, precision medicine	—	2013—present	<a href="http://healthsciences.utah.edu/utah-genome-project/">http://healthsciences.utah.edu/utah-genome-project/</a>
MESNG	United States	Autism	> 10 000	2014—present	<a href="http://www.mesng.org/">http://www.mesng.org/</a>
The Precision Medicine Initiative (PMI)	United States	Variants, precision medicine	1 000 000	2015—present	<a href="http://obamawhitehouse.archives.gov/precision-medicine/">http://obamawhitehouse.archives.gov/precision-medicine/</a>
Genome Russia Project	Russia	Variants, precision medicine, anthropology	2 980	2015—present	<a href="http://genomarusia.spb.ru/">http://genomarusia.spb.ru/</a>
Human Genome Project—Africa	United States	Artificial synthesis of genome	0	2016—2026	<a href="http://ciencia.sciencemag.org/content/early/2016/06/01/science.aaf00203v.pdf">http://ciencia.sciencemag.org/content/early/2016/06/01/science.aaf00203v.pdf</a>
AfricaZambia 2 million Genomes	United States, United Kingdom, Finland	Variants, precision medicine	2 000 000	2016—present	<a href="http://www.africanzambia2million.org/the-code-of-life-to-develop-new-medicines.html">http://www.africanzambia2million.org/the-code-of-life-to-develop-new-medicines.html</a>
EpIQ5 Project	International	Epilepsy	25	2016—present	<a href="http://epiq5.org/">http://epiq5.org/</a>
Projecto Genoma Navarra (NAG20E)	Spain	Rare diseases and cancer	—	2016—present	<a href="http://www.navarrabiomed.es/en/prevencion/navarrabiomed">http://www.navarrabiomed.es/en/prevencion/navarrabiomed</a>
The Danish Reference Genome Project	Denmark	Variants, precision medicine, anthropology	150	2017	<a href="http://www.genomedanmark.dk/english/about/reference-genome/">http://www.genomedanmark.dk/english/about/reference-genome/</a>
The Norwegian 1000 Genomes Project	Norway	Variants, precision medicine	1 000	2017	<a href="http://norg1000genomes.kibex.no/1000genomes/">http://norg1000genomes.kibex.no/1000genomes/</a>

**Table 1.** (Continued)

Project	Country	Scope	Population size	Period	URL
Swedish Genomes Program	Sweden	Variants, precision medicine	1000	2017	<a href="https://www.scilifelab.se/research/national-projects/swedish-genome-program/">https://www.scilifelab.se/research/national-projects/swedish-genome-program/</a>
Singapore Genome Variation Project	Singapore	Variants	292	Finished	<a href="https://www.sitgen.nus.edu.sg/~SGVP/default.htm">https://www.sitgen.nus.edu.sg/~SGVP/default.htm</a>
Singapore Sequencing Malay	Singapore	Variants	100	Finished?	<a href="https://www.sitgen.nus.edu.sg/~SSMP/">https://www.sitgen.nus.edu.sg/~SSMP/</a>
Melanoma Genome Project	Australia	Melanoma	500	Ongoing	<a href="https://www.melanoma.org.au/research/australian-genetic-melanoma-genome-project/">https://www.melanoma.org.au/research/australian-genetic-melanoma-genome-project/</a>
GCAT Genomes for Life	Spain	Cancer, diabetes, cardiovascular disease and others	50 000	Ongoing	<a href="http://www.gcatbiobanque.org/qui-est-nous/index/">http://www.gcatbiobanque.org/qui-est-nous/index/</a>
4D Genome Project	Spain	3D genome structure	—	Ongoing	<a href="http://www.crg.eu/en/content/research/4d-genome-3d-structure-project">http://www.crg.eu/en/content/research/4d-genome-3d-structure-project</a>
RD-connect	International	Rare diseases databases platform	—	Ongoing	<a href="http://rd-connect.eu/">http://rd-connect.eu/</a>
Metastatic Breast Cancer Project	United States	Metastatic breast cancer	—	Ongoing	<a href="http://www.mbcproject.org/">http://www.mbcproject.org/</a>
Human Cell Atlas Project	International	Health and disease	—	Ongoing	<a href="http://www.humancellatlas.org/">http://www.humancellatlas.org/</a>
Centers for Mendelian Genomics (CMG)	United States	Mendelian diseases	> 20 000	Ongoing	<a href="http://mendelian.org/">http://mendelian.org/</a>
International Cancer Genome Consortium (ICGC)	International	Many cancers	> 16 000	Ongoing	<a href="http://icgc.org/">http://icgc.org/</a>
Qatar Genome Programme	Qatar	Genome	27 000	Ongoing	<a href="http://www.qatar-genome.org.qa/">http://www.qatar-genome.org.qa/</a>
GUARDIAN	India	Rare diseases	—	Ongoing	<a href="http://guardian.sanger.ac.uk/home">http://guardian.sanger.ac.uk/home</a>
Initiative on Rare and Undiagnosed Diseases (IRUD)	Japan	Rare diseases	—	Ongoing	<a href="http://www.irdc.jp/en/program/IRUD/">http://www.irdc.jp/en/program/IRUD/</a>
Genome Asia100K	South Korea, India	Asian reference genomes, variation	100 000	Ongoing?	<a href="http://www.genomesasia100k.com/">http://www.genomesasia100k.com/</a>
Saudi Human Genome Program	Saudi Arabia	Variation and genetic disease	100 000	Ongoing?	<a href="http://h3g.scienceministry.gov.sa/">http://h3g.scienceministry.gov.sa/</a>
African Genome Variation Program	United Kingdom, Uganda	Variants	1000	Ongoing?	<a href="http://www.sanger.ac.uk/science/collaboration/african-genome-variation-project">http://www.sanger.ac.uk/science/collaboration/african-genome-variation-project</a>
EpIK	International	Epilepsy	4	Ongoing?	<a href="http://www.epik.org/">http://www.epik.org/</a>
China Precision Medicine Initiative	China	Variants, precision medicine	1 000 000	—	—
African Ancestry Project	United States	Ancestry and health	—	—	<a href="https://biog.23andme.com/23andme-research/23andme-african-ancestry-project/">https://biog.23andme.com/23andme-research/23andme-african-ancestry-project/</a>
Iran-ai Population-scale Sequencing	France	Genome	—	—	<a href="https://www.francobioinformatics.fr/france-aians-74555-en-brevet-03-03-16-225-000-g-enom-sequen-clin-q-orientee">https://www.francobioinformatics.fr/france-aians-74555-en-brevet-03-03-16-225-000-g-enom-sequen-clin-q-orientee</a>
Korean Reference Genome Project	South Korea	Variants, precision medicine	—	—	<a href="http://koreagen.genome.kribic.ac.kr/">http://koreagen.genome.kribic.ac.kr/</a>

*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 <sup>1</sup>
Instrument price	\$19.9K	\$49.5K	\$99K	\$275K	\$900K	\$6M <sup>2</sup> /\$10M <sup>2</sup>	\$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



illumina®



NovaSeq 6000

1 Tbp



MGI  
华大智造



MGISEQ-2000



MGISEQ-200



Oxford  
NANOPORE  
Technologies®



PromethION



GridION X5



MinION



MinION Dx



iSeq 100

10 Gbp

100 Gbp

1 Tbp

# 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  $20 \times 10^9$       до  $3 \times 10^9$

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*