



Л е к ц и я № 1

Биоинформатика (введение в дисциплину)

**Разработал профессор П. М. Васильев
Кафедра фармакологии и биоинформатики**

**Для студентов, обучающихся по направлению 06.03.01 «Биология»
профили Биохимия, Генетика
при изучении дисциплины «Биоинформатика»**

П л а н л е к ц и и

- Введение.
- Предмет, цели и задачи биоинформатики.
- Поиск научной информации.
- База данных PubMed.
- Биоинформатика последовательностей.
- Поиск последовательностей.
- База знаний по белкам UniProtKB.
- Банк данных по нуклеотидным последовательностям GenBank.



Николай Павлович Кравков
русский фармаколог, академик
Императорской Военно-медицинской
академии (1914), член-корреспондент
Российской академии наук (1920)
Основоположник советской фармакологии

Василий Васильевич Закусов
фармаколог, академик АМН СССР
Основатель и первый директор
Института фармакологии и химиотерапии
(1952 – 1979)





Артур Викторович Вальдман
фармаколог, академик АМН СССР
Директор НИИ фармакологии (1979 – 1990)



Геннадий Васильевич Ковалев
фармаколог, доктор медицинских наук,
профессор
Заведующий кафедрой фармакологии
ВГМИ (1970 – 1990)



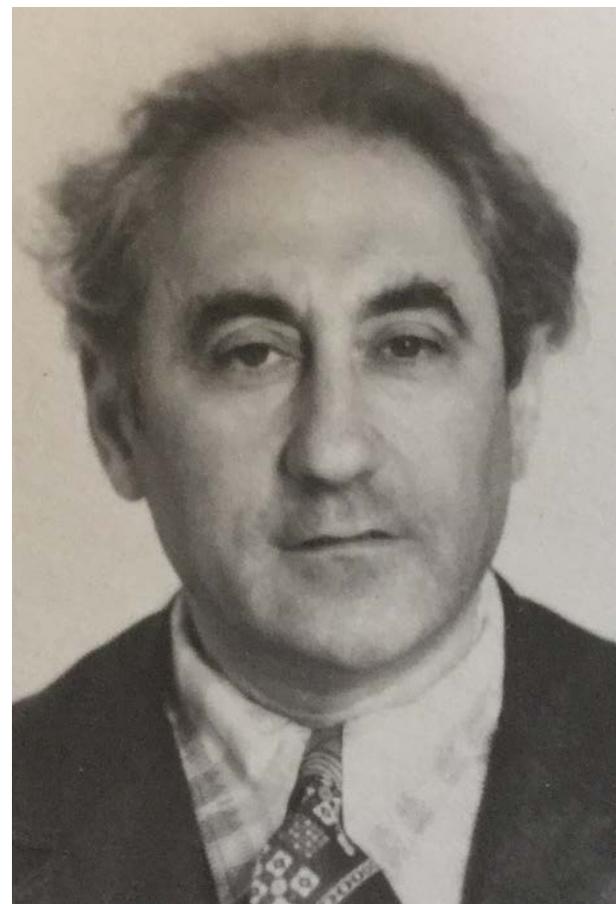
Владимир Иванович Петров
фармаколог, академик РАН
Президент ВолгГМУ, директор НЦИЛС,
заведующий кафедрой клинической
фармакологии и интенсивной терапии



Александр Алексеевич Спасов
фармаколог, академик РАН
Советник ректора по науке ВолгГМУ,
заведующий кафедрой фармакологии и
биоинформатики



Лев Арамович Пирузян
биофизик, академик РАН
Основатель и директор НИИ по
биологическим испытаниям химических
соединений (1972 – 1984)



Ландау Михаил Александрович
химик, доктор химических наук,
профессор
Заведующий лабораторией структурной
химии НИИ по БИХС (1972 – 1984)

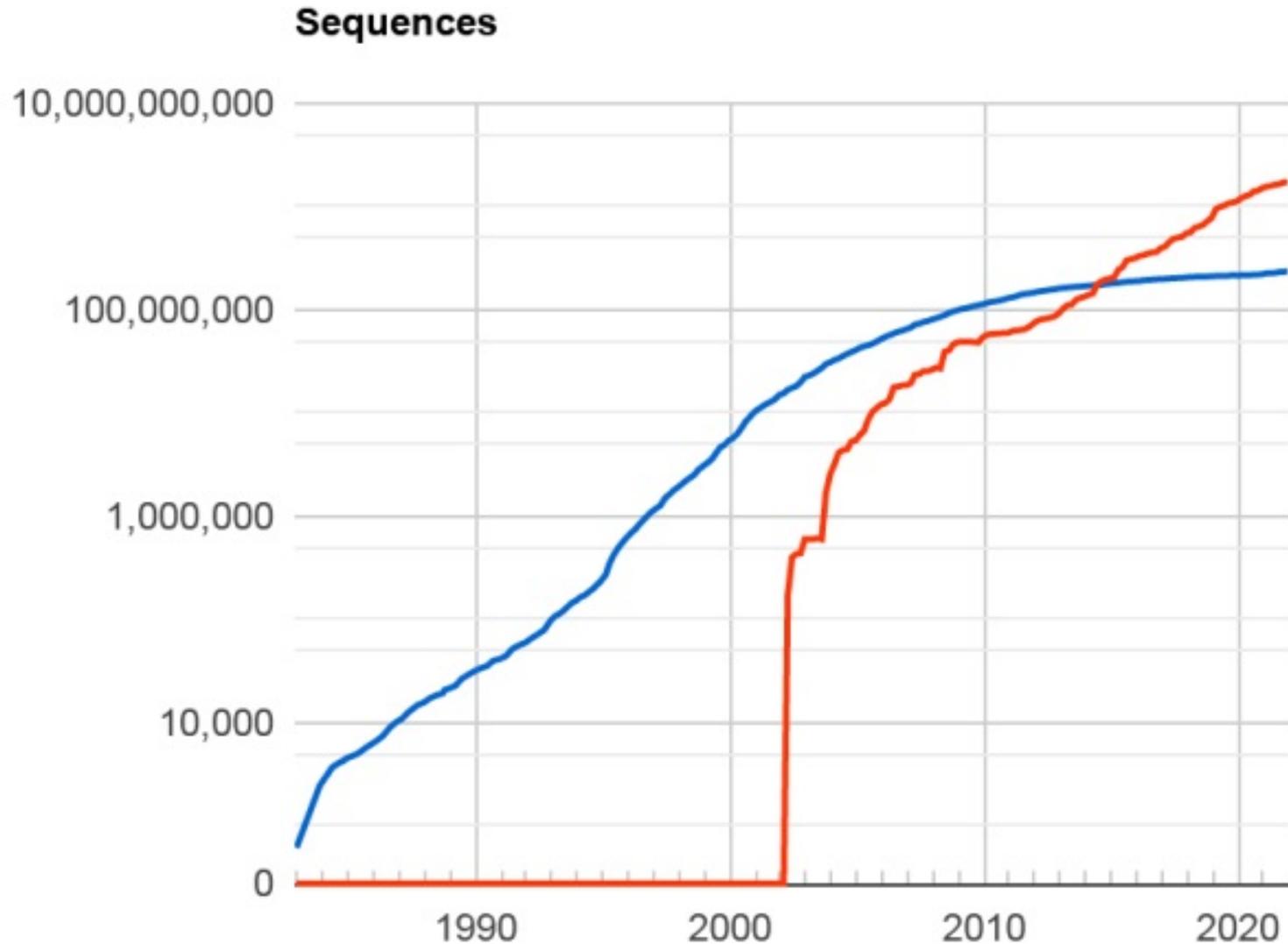
Что такое биоинформатика

Область науки, в которой решаются биологические задачи с помощью вычислительных методов математики и информационных технологий

Этапы развития биоинформатики

Год	Технология	Биоинформатика
1965	Секвенирование tRNA	База данных PIR
1970	Обратная транскрипция	Алгоритм выравнивания NW
1972	Клонирование	
1977	Секвенирование	База данных PDB
1980		База данных NAR
1981		Алгоритм выравнивания SW
1982	Секвенирование ДНК фага лямбда	
1983	PCR	Алгоритм поиска по БД WL
1985	Секвенирование ДНК вирусов	FASTA-поиск по базе данных
1987		GeneBank
1989	Программа "Геном человека"	Swiss-Prot, NCBI
1992	Первая хромосома дрожжей	BLOSSUM
1993	Автоматическое секвенирование	
1995	Первый геном бактерии	База данных SCOP
1996	Первый геном архейный	
1997		PSI-BLAST
1998	Геном червя	
2001	Геном человека	

Рост объема информации по нуклеотидным последовательностям



Разделы биоинформатики (по объектам исследования)

Биоинформатика последовательностей

Структурная биоинформатика

Компьютерная геномика

Разделы биоинформатики (по способам исследования)

**Применение известных методов анализа
для получения новых биологических знаний**

**Разработка новых методов анализа
биологических данных**

Разработка новых баз данных

Задачи биоинформатики

- Разработка алгоритмов для анализа биологических данных большого объема
 - Алгоритм поиска генов в геноме
- Анализ и интерпретация различных типов биологических данных таких, как нуклеотидные и аминокислотные последовательности, домены белков, структура белков и т.д.
 - Изучение структуры активного центра белка
- Разработка программного обеспечения для управления и быстрого доступа к биологическим данным
 - Создание банка данных аминокислотных последовательностей

Биоинформатика последовательностей

- Выравнивание и определение сходства двух последовательностей
- Построение множественных выравниваний
- Распознавание генов
- Предсказание вторичной структуры белков и РНК
- Предсказание сайтов связывания белков

Структурная биоинформатика

- Предсказание функциональных участков белковой молекулы
- Сравнительный анализ структур родственных белков
- Классификация белков на основе их 3D-структуры
- Анализ структур комплексов двух или нескольких молекул белка
- Анализ комплексов молекул белка с другими молекулами
- Предсказание воздействия молекул химических веществ (например, потенциальных лекарств) на молекулы белков
- Предсказание структуры белка по структуре белка с похожей последовательностью

Компьютерная геномика

- Предсказание генов в последовательностях
- Предварительная аннотация генов по сходству белковых последовательностей
- Поиск «пропущенных» генов
- Сравнительный анализ геномов
- Исследование регуляции работы генов (экспрессия, коэкспрессия)
- Полногеномный анализ ассоциаций

ПОИСК И БАЗЫ ДАННЫХ

PubMed – поиск биомедицинской информации

<https://www.ncbi.nlm.nih.gov/pubmed/>

GenBank – нуклеотидные последовательности

<https://www.ncbi.nlm.nih.gov/genbank/>

UniProt – информация о белках

<https://www.uniprot.org/>

PDB – 3D-структуры белков

<https://www.rcsb.org/>

DrugBank – лекарства, действующие на биомишени

<https://www.drugbank.ca/>

Объем баз данных 2022

PubMed – 34 млн. статей

GenBank – 1 трл. 492,8 млрд. оснований
239,9 млн. последовательностей

UniProt – 227,3 млн. белков

PDB – 194,8 тыс. 3D-структур биомолекул

DrugBank – >500 тыс. лекарств и продуктов
29,1 тыс. белков-мишеней

Типы баз данных

- **Архивные базы данных**

Эти базы данных не проверяются

GeneBank, EMBL – первичные последовательности

PDB – пространственные структуры белков

- **Курируемые базы данных**

Информацию отбирают эксперты

Swiss-Prot – аминокислотные последовательности
белков

KEGG – функции генов и сигнальные пути

Типы баз данных

- **Производные базы данных**

 - Результат обработки данных*

 - SCOP – структурная классификация белков

 - PFAM – семейства белков

 - GO – классификация генов (Gene Ontology)

 - ProDom – белковые домены

- **Интегрированные базы данных**

 - Результат объединения всех данных*

 - NCBI Entrez – нуклеотидные и аминокислотные последовательности и структуры

 - Esosuc – гены, белки, метаболизм E. coli

Поиск литературы

<https://www.ncbi.nlm.nih.gov/pubmed/>

PubMed comprises more than **34 million** citations for biomedical literature from MEDLINE, life science journals, and online books.

Citations may include links to full-text content from PubMed Central and publisher web sites.

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PubMed records with recent increases in activity

[Structural basis for channel conduction in the pump-like channelrhodopsin ChRmine.](#)

Latest Literature

New articles from highly accessed journals

[Am J Surg Pathol \(2\)](#)

[Circ Res \(1\)](#)

Простой поиск в PubMed

drug discovery — 141 566 статей

The screenshot displays the PubMed search interface. At the top, the NIH logo and 'National Library of Medicine' are visible. The search bar contains 'drug discovery' and shows '141,566 results'. The results list includes four entries, each with a title, author, citation, and a share button. A red circle highlights the '141,566 results' text.

NIH National Library of Medicine
National Center for Biotechnology Information

PubMed.gov

drug discovery

Advanced Create alert Create RSS Search User Guide

Save Email Send to Sorted by: Best match Display options

MY NCBI FILTERS

141,566 results

RESULTS BY YEAR

1936 2022

TEXT AVAILABILITY

Abstract

Free full text

Full text

ARTICLE ATTRIBUTE

Associated data

ARTICLE TYPE

Books and Documents

Clinical Trial

Meta-Analysis

Randomized Controlled Trial

Review

Systematic Review

1 **Bioinformatics in translational drug discovery.**
Wooler SK, Benstead-Hume G, Chen X, Ali Y, Pearl FMG.
Cite Biosci Rep. 2017 Jul 7;37(4):BSR20160180. doi: 10.1042/BSR20160180. Print 2017 Aug 31.
PMID: 28487472 **Free PMC article.** Review.

Share Bioinformatics approaches are becoming ever more essential in translational **drug discovery** both in academia and within the pharmaceutical industry. Computational exploitation of the increasing volumes of data generated during all phases of **drug discovery** ...

2 **Antibiotic drug discovery.**
Wohleben W, Mast Y, Stegmann E, Ziemert N.
Cite Microb Biotechnol. 2016 Sep;9(5):541-8. doi: 10.1111/1751-7915.12388. Epub 2016 Jul 29.
PMID: 27470984 **Free PMC article.** Review.

Share This has been the consequence of several new and revolutionizing **drug discovery** and development techniques, which is initiating a 'New Age of Antibiotic **Discovery**'. In this review we concentrate on the most significant **discovery** approaches during the ...

3 **Trends in Modern Drug Discovery.**
Eder J, Herrling PL.
Cite Handb Exp Pharmacol. 2016;232:3-22. doi: 10.1007/164_2015_20.
PMID: 26330257 Review.

Share With the advent of modern molecular biology methods and based on knowledge of the human genome, **drug discovery** has now largely changed into a hypothesis-driven target-based approach, a development which was paralleled by significant environmental changes in the phar ...

4 **Cryo-EM in drug discovery: achievements, limitations and prospects.**
Renaud JP, Chari A, Ciferri C, Liu WT, Rémygy HW, Stark H, Wiesmann C.
Cite Nat Rev Drug Discov. 2018 Jul;17(7):471-492. doi: 10.1038/nrd.2018.77. Epub 2018 Jun 8.
PMID: 29880918 Review.

Точный поиск в PubMed

“drug discovery” — 87 012 статей

The screenshot displays the PubMed search interface. At the top, the NIH logo and 'National Library of Medicine' are visible. The search bar contains the query 'drug discovery' and shows '87,012 results' circled in red. The results are sorted by 'Best match' and displayed on page 1 of 8,702. The first three results are listed below, each with a checkbox, title, author, citation, and share options.

MY NCBI FILTERS

RESULTS BY YEAR

1964 2022

TEXT AVAILABILITY

- Abstract
- Free full text
- Full text

ARTICLE ATTRIBUTE

- Associated data

ARTICLE TYPE

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial
- Review
- Systematic Review

87,012 results

Sorted by: Best match Display options

Page 1 of 8,702

Molecular Docking: Shifting Paradigms in Drug Discovery.
1 Pinzi L, Rastelli G.
Cite Int J Mol Sci. 2019 Sep 4;20(18):4331. doi: 10.3390/ijms20184331.
PMID: 31487867 **Free PMC article.** Review.
Share Molecular docking is an established in silico structure-based method widely used in **drug discovery**. Docking enables the identification of novel compounds of therapeutic interest, predicting ligand-target interactions at a molecular level, or delineating structure-ac ...

Bioinformatics in translational drug discovery.
2 Wooller SK, Benstead-Hume G, Chen X, Ali Y, Pearl FMG.
Cite Biosci Rep. 2017 Jul 7;37(4):BSR20160180. doi: 10.1042/BSR20160180. Print 2017 Aug 31.
PMID: 28487472 **Free PMC article.** Review.
Share Bioinformatics approaches are becoming ever more essential in translational **drug discovery** both in academia and within the pharmaceutical industry. Computational exploitation of the increasing volumes of data generated during all phases of **drug discovery** ...

Advancing Drug Discovery via Artificial Intelligence.
3 Chan HCS, Shan H, Dahoun T, Vogel H, Yuan S.
Cite Trends Pharmacol Sci. 2019 Aug;40(8):592-604. doi: 10.1016/j.tips.2019.06.004. Epub 2019 Jul 15.
PMID: 31320117 Review.
Share **Drug discovery** and development are among the most important translational science activities that contribute to human health and wellbeing. ...How to decrease the costs and speed up new **drug discovery** has become a challenging and urgent question in ind ...

Bioinformatics Approaches for Anti-cancer Drug Discovery.
4 Li K, Du Y, Li L, Wei DQ.
Cite Curr Drug Targets. 2020;21(1):3-17. doi: 10.2174/1389450120666190923162203.
PMID: 31549592 Review.

Поисковые запросы

AND, & – логическое “И”

OR – логическое “ИЛИ”

diabetes – 856 238 статей

alzheimer – 193 397 статей

antioxidant – 657 255 статей

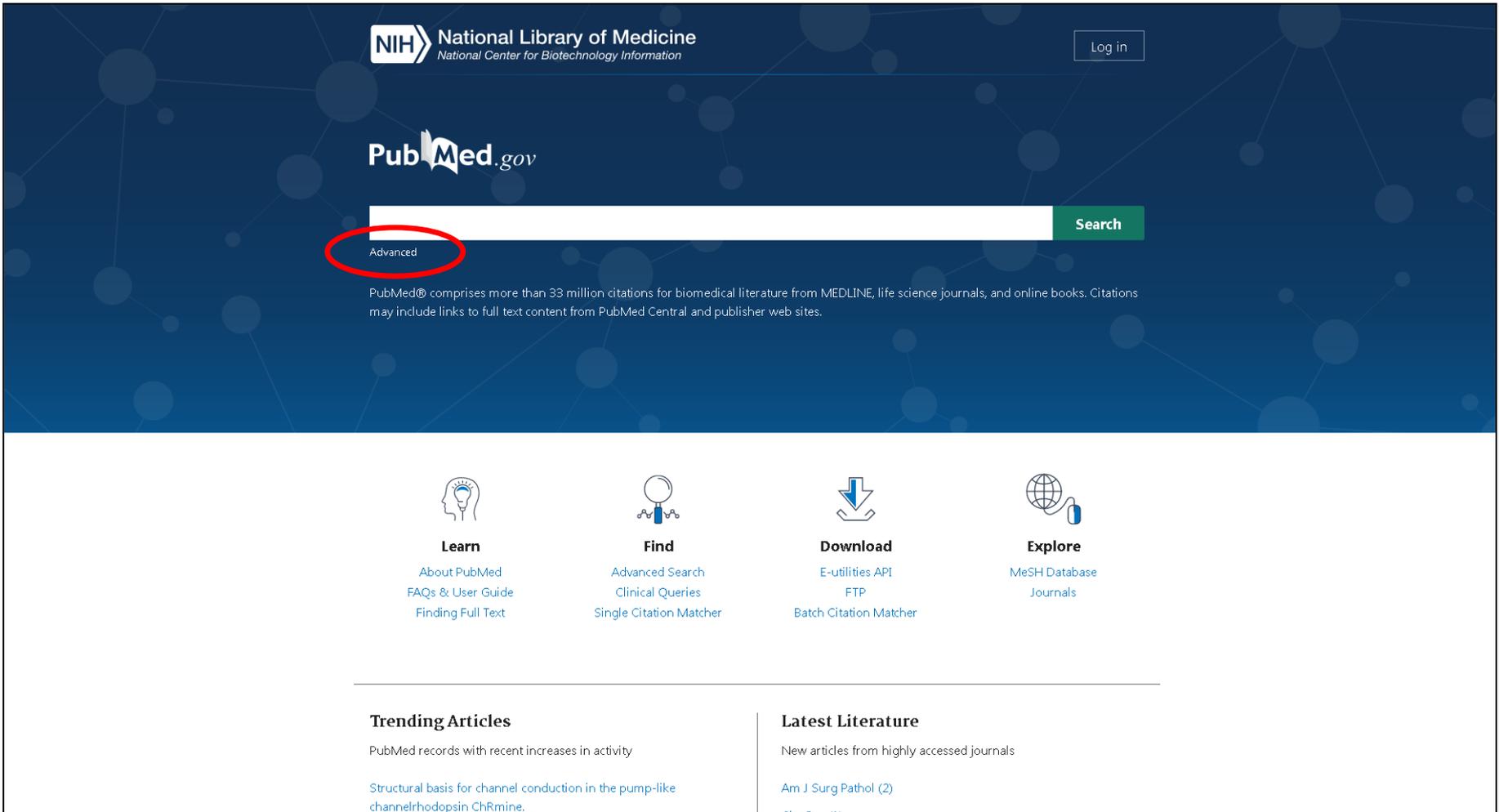
diabetes OR alzheimer – 1 041 368 статей

diabetes AND alzheimer – 8 267 статей

diabetes alzheimer antioxidant – 763 статьи

Расширенный поиск в PubMed

Advanced



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National Center for Biotechnology Information

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Advanced

Search

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Latest Literature
New articles from highly accessed journals
[Am J Surg Pathol \(2\)](#)
[Circ Res \(1\)](#)

Расширенный поиск в PubMed

Title/Abstract

The screenshot displays the PubMed Advanced Search Builder interface. At the top, the NIH logo and 'National Library of Medicine' are visible. The main area is titled 'PubMed Advanced Search Builder'. A search term 'Title/Abstract' is entered in the query box, highlighted with a red circle. The search operator 'AND' is selected, highlighted with a blue circle. The resulting query is shown in the 'Query box' and highlighted with a green circle: `((diabetes[Title/Abstract]) AND (alzheimer[Title/Abstract])) AND (antioxidant[Title/Abstract])`. The 'Search' button is highlighted with a pink circle. Below the search area, the 'History and Search Details' section shows a table of previous searches.

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National Center for Biotechnology Information

Log in

PubMed Advanced Search Builder

PubMed.gov
User Guide

Add terms to the query box

Title/Abstract Enter a search term

AND Show Index

Query box

((diabetes[Title/Abstract]) AND (alzheimer[Title/Abstract])) AND (antioxidant[Title/Abstract])

Search

History and Search Details

Download Delete

Search	Actions	Details	Query	Results	Time
#11	...	>	Search: diabetes alzheimer antioxidant	763	06:10:14
#10	...	>	Search: diabetes AND alzheimer	8,267	06:09:21
#9	...	>	Search: diabetes OR alzheimer	1,041,368	06:08:51
#8	...	>	Search: antioxidant	657,255	06:08:33
#7	...	>	Search: alzheimer	193,397	06:08:13
#4	...	>	Search: diabetes	856,238	06:07:52
#6	...	>	Search: "drug discovery"	87,012	06:05:40
#5	...	>	Search: drug discovery	141,566	06:03:17
#3	...	>	Search: diabetes Filters: Review, in the last 10 years	66,527	06:02:46
#2	...	>	Search: "drug discovery" Filters: Review, in the last 10 years	17,117	05:57:40

Расширенный поиск в PubMed

376 статей

NIH National Library of Medicine
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PubMed.gov

Search: ((diabetes[Title/Abstract]) AND (alzheimer[Title/Abstract])) AND (antioxidant[Ti

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MY NCBI FILTERS **376 results** Page 1 of 38

RESULTS BY YEAR



TEXT AVAILABILITY

- Abstract
- Free full text
- Full text

ARTICLE ATTRIBUTE

- Associated data

ARTICLE TYPE

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial
- Review
- Systematic Review

1 Mediterranean diet: The role of long-chain omega-3 fatty acids in fish; polyphenols in fruits, vegetables, cereals, coffee, tea, cacao and wine; probiotics and vitamins in prevention of stroke, age-related cognitive decline, and **Alzheimer disease**.
Share Román GC, Jackson RE, Gadhia R, Román AN, Reis J. Rev Neurol (Paris). 2019 Dec;175(10):724-741. doi: 10.1016/j.neurol.2019.08.005. Epub 2019 Sep 11. PMID: 31521398 Review. The mechanisms of action of the dietary components of the Mediterranean diet are reviewed in prevention of cardiovascular disease, stroke, age-associated cognitive decline and **Alzheimer disease**. A companion article provides a comprehensive review of extra-virgin olive oil. ...

2 Benefits of curcumin in brain disorders.
Share Bhat A, Mahalakshmi AM, Ray B, Tuladhar S, Hediyaal TA, Manthiannem E, Padamati J, Chandra R, Chidambaram SB, Sakharkar MK. Biofactors. 2019 Sep;45(5):666-689. doi: 10.1002/biof.1533. Epub 2019 Jun 11. PMID: 31185140 Review. Curcumin has got its global recognition because of its strong **antioxidant**, anti-inflammatory, anti-cancer, and antimicrobial activities. Additionally, it is used in **diabetes** and arthritis as well as in hepatic, renal, and cardiovascular diseases. ...

3 Cinnamon and Chronic Diseases.
Share Hariri M, Ghiasvand R. Adv Exp Med Biol. 2016;929:1-24. doi: 10.1007/978-3-319-41342-6_1. PMID: 27771918 Review. Cinnamon contains derivatives, such as cinnamaldehyde, cinnamic acid, cinnamate, and numerous other components such as polyphenols and **antioxidant**, anti-inflammatory, antidiabetic, antimicrobial, anticancer effects. ...Recently, many trials have explored the beneficial eff ...

Полные тексты статей

164 статьи

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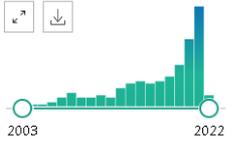
ie/Abstract]) AND (alzheimer[Title/Abstract])) AND (antioxidant[Title/Abstract]) X Search

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MY NCBI FILTERS 164 results

RESULTS BY YEAR



2003 2022

TEXT AVAILABILITY

Abstract

Free full text

Full text

ARTICLE ATTRIBUTE

Associated data

ARTICLE TYPE

Books and Documents

Clinical Trial

Meta-Analysis

Randomized Controlled Trial

Review

Systematic Review

Filters applied: Free full text. Clear all

1 Diabetes and Alzheimer's Disease: Might Mitochondrial Dysfunction Help Deciphering the Common Path?

Cite Potenza MA, Sgarra L, Desantis V, Nacci C, Montagnani M. Antioxidants (Basel). 2021 Aug 6;10(8):1257. doi: 10.3390/antiox10081257. PMID: 34439505 Free PMC article. Review.

Share A growing number of clinical and epidemiological studies support the hypothesis of a tight correlation between type 2 diabetes mellitus (T2DM) and the development risk of Alzheimer's disease (AD). Indeed, the proposed definition of Alzheimer's disease as type ...

2 Beneficial Effects of Walnuts on Cognition and Brain Health.

Cite Chauhan A, Chauhan V. Nutrients. 2020 Feb 20;12(2):550. doi: 10.3390/nu12020550. PMID: 32093220 Free PMC article. Review.

Share Our recent study in AD-tg mice has shown that a walnut-enriched diet significantly improves antioxidant defense and decreases free radicals' levels, lipid peroxidation, and protein oxidation when compared to a control diet without walnuts. These findings suggest that a die ...

3 Therapeutic applications of pomegranate (Punica granatum L.): a review.

Cite Jurenka JS. Altern Med Rev. 2008 Jun;13(2):128-44. PMID: 18590349 Free article. Review.

Share The synergistic action of the pomegranate constituents appears to be superior to that of single constituents. In the past decade, numerous studies on the antioxidant, anticarcinogenic, and anti-inflammatory properties of pomegranate constituents have been published, focusi ...

New Insights into the Biological and Pharmaceutical Properties of Royal Jelly.

Полные тексты обзоров

64 статьи

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le/Abstract) AND (alzheimer[Title/Abstract]) AND (antioxidant[Title/Abstract]) X Search

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MY NCBI FILTERS 64 results << < Page 1 of 7 > >>

RESULTS BY YEAR



TEXT AVAILABILITY

- Abstract
- Free full text
- Full text

ARTICLE ATTRIBUTE

- Associated data

ARTICLE TYPE

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial
- Review
- Systematic Review

Filters applied: Free full text, Review. Clear all

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New Insights into the Biological and Pharmaceutical Properties of Royal Jelly.

Обзоры за 5 последних лет

42 статьи

The screenshot shows a PubMed search results page for the query: `Title/Abstract] AND (alzheimer[Title/Abstract]) AND (antioxidant[Title/Abstract])`. The search is filtered for "Free full text" and "Review" articles published in the last 5 years. The results are sorted by "Best match" and displayed on page 1 of 5. The first three results are highlighted with blue circles, and the total number of results (42) is circled in red.

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PubMed.gov

Search: `Title/Abstract] AND (alzheimer[Title/Abstract]) AND (antioxidant[Title/Abstract])`

Advanced Create alert Create RSS User Guide

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MY NCBI FILTERS **42 results** Page 1 of 5

RESULTS BY YEAR

Filters applied: Free full text, Review, in the last 5 years. Clear all

TEXT AVAILABILITY

- Abstract
- Free full text
- Full text

ARTICLE ATTRIBUTE

- Associated data

ARTICLE TYPE

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial
- Review
- Systematic Review

PUBLICATION DATE

- 1 year
- 5 years
- 10 years

1 Diabetes and Alzheimer's Disease: Might Mitochondrial Dysfunction Help Deciphering the Common Path?
Cite Potenza MA, Sgarra L, Desantis V, Nacci C, Montagnani M. Antioxidants (Basel). 2021 Aug 6;10(8):1257. doi: 10.3390/antiox10081257. PMID: 34439505 Free PMC article. Review.
A growing number of clinical and epidemiological studies support the hypothesis of a tight correlation between type 2 diabetes mellitus (T2DM) and the development risk of Alzheimer's disease (AD). Indeed, the proposed definition of Alzheimer's disease as type ...

2 Beneficial Effects of Walnuts on Cognition and Brain Health.
Cite Chauhan A, Chauhan V. Nutrients. 2020 Feb 20;12(2):350. doi: 10.3390/nu12020350. PMID: 32093220 Free PMC article. Review.
Our recent study in AD-tg mice has shown that a walnut-enriched diet significantly improves antioxidant defense and decreases free radicals' levels, lipid peroxidation, and protein oxidation when compared to a control diet without walnuts. These findings suggest that a die ...

3 New Insights into the Biological and Pharmaceutical Properties of Royal Jelly.
Cite Ahmad S, Campos MG, Fratini F, Altaye SZ, Li J. Int J Mol Sci. 2020 Jan 8;21(2):382. doi: 10.3390/ijms21020382. PMID: 31936187 Free PMC article. Review.
It is also the most studied bee product, aimed at unravelling its bioactivities, such as antimicrobial, antioxidant, anti-aging, immunomodulatory, and general tonic action against laboratory animals, microbial organisms, farm animals, and clinical trials. It is commonly us ...

4 A Review: The Bioactivities and Pharmacological Applications of Polygonatum sibiricum polysaccharides.
Cite Cui X, Wang S, Cao H, Guo H, Li Y, Xu F, Zheng M, Xi X, Han C. Molecules. 2018 May 14;23(5):1170. doi: 10.3390/molecules23051170. PMID: 29757991 Free PMC article. Review.
This article summarizes Polygonatum sibiricum polysaccharides (PSP) have many pharmacological

Реферат обзора

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Title/Abstract)) AND (alzheimer[Title/Abstract])) AND (antioxidant[Title/Abstract]))

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User Guide

Search results

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Send to

Display options

Review > Antioxidants (Basel). 2021 Aug 6;10(8):1257. doi: 10.3390/antiox10081257.

Diabetes and Alzheimer's Disease: Might Mitochondrial Dysfunction Help Deciphering the Common Path?

Maria Assunta Potenza ¹, Luca Sgarra ¹, Vanessa Desantis ¹, Carmela Nacci ¹, Monica Montagnani ¹

Affiliations + expand

PMID: 34439505 PMID: PMC8389322 DOI: 10.3390/antiox10081257

Free PMC article

Abstract

A growing number of clinical and epidemiological studies support the hypothesis of a tight correlation between type 2 diabetes mellitus (T2DM) and the development risk of Alzheimer's disease (AD). Indeed, the proposed definition of Alzheimer's disease as type 3 diabetes (T3D) underlines the key role played by deranged insulin signaling to accumulation of aggregated amyloid beta (A β) peptides in the senile plaques of the brain. Metabolic disturbances such as hyperglycemia, peripheral hyperinsulinemia, dysregulated lipid metabolism, and chronic inflammation associated with T2DM are responsible for an inefficient transport of insulin to the brain, producing a neuronal insulin resistance that triggers an enhanced production and deposition of A β and concomitantly contributes to impairment in the microtubule-associated protein Tau, leading to neural degeneration and cognitive decline. Furthermore, the reduced antioxidant capacity observed in T2DM patients, together with the impairment of cerebral glucose metabolism and the decreased performance of mitochondrial activity, suggests the existence of a relationship between oxidative damage, mitochondrial impairment, and cognitive dysfunction that could further reinforce the common pathophysiology of T2DM and AD. In this review we discuss the molecular mechanisms by which insulin-signaling dysregulation in T2DM can contribute to the pathogenesis and progression of AD, deepening the analysis of complex mechanisms involved in reactive oxygen species (ROS) production under oxidative stress and their possible influence in AD and T2DM. In addition, the role of current therapies as tools for prevention or treatment of damage induced by oxidative stress in T2DM and AD will be debated.

Keywords: Alzheimer's disease (AD); mitochondrial dysfunction; type 2 diabetes (T2DM).

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2 of 42 >

Поиск аминокислотных последовательностей

UniProt

<https://www.uniprot.org/>

227,3 млн. белков

Формат FASTA

W. R. Pearson, D. J. Lipman – 1988

Строка-заголовок (the definition line)

> [уникальный ID] | [описание]

Последовательность белка (или ДНК) в однобуквенном коде

```
>sp|P35557|HXK4_HUMAN Hexokinase-4 OS=Homo sapiens
MLDDRARMEAAKKEKUEQILAEFQLQEEDLKKUMRRMQKEMDRGLRLETH
EEASUKMLPTYURSTPEGSEUGDFLSLDLGGTNFRUMLUKUGE GEEGQWS
UKTKHQMYSIPEDAMTGTAEMLFDYISECISDFLDKHQMKHKKLPLGFTF
SFPURHEDIDKGILLNWTKGFKASGAEGNNUUGLLRDAIKRRGDFEMDUU
AMUNDTUATMISCYEDHQCEUGMIUGTG CNAC YMEEMQNVELUEGDEGR
MCUNTEWGAFGDSGELDEFLL EYDRLUDESSANPGQQLYEKLIGGKYMGE
LURLULLRLUDENLLFHGEASEQLRTRGAFETRFUSQUESDTGDRKQIYN
ILSTLGLRPSTTDCDIURRACESUSTRAAHMCSAGLAGUINRMRESRSED
UMRITUGUDGSUYKLHPSFKERFHASURRLTPSCEITFIESEEGSGRGAA
LUSAUACKKACMLGQ
```

Коды нуклеотидов

a	adenine
c	cytosine
g	guanine
t	thymine
u	uracil
r	a or g
y	c or t
s	g or c
w	a or t

k	g or t
m	a or c
b	c or g or t
d	a or g or t
h	a or c or t
v	a or c or g
n	any base
. or -	gap

Коды аминокислот

A	Ala	Alanine
B	Asx	Aspartic acid or Asparagine
C	Cys	Cysteine
D	Asp	Aspartic Acid
E	Glu	Glutamic Acid
F	Phe	Phenylalanine
G	Gly	Glycine
H	His	Histidine
I	Ile	Isoleucine
K	Lys	Lysine
L	Leu	Leucine
M	Met	Methionine
N	Asn	Asparagine
P	Pro	Proline

Q	Gln	Glutamine
R	Arg	Arginine
S	Ser	Serine
T	Thr	Threonine
V	Val	Valine
W	Trp	Tryptophan
X	Xaa	Any amino acid
Y	Tyr	Tyrosine
Z	Glx	Glutamine or Glutamic acid
U	Sec	SelenoCysteine
O	Pyl	Pyrrolysine
*		translation stop
-		gap of indeterminate length

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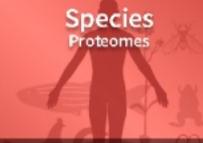
Accessing UniProt programmatically? Have a look at the [new API documentation](#).
If you still need it, the legacy version of the website is available until the 2022_04 release.

Proteins
UniProt Knowledgebase



Reviewed (Swiss-Prot) 568,002
Unreviewed (TrEMBL) 226,771,948

Species
Proteomes



Protein sets for species with sequenced genomes from across the tree of life

Protein Clusters
UniRef



Clusters of protein sequences at 100%, 90% & 50% identity

Sequence Archive
UniParc



Non-redundant archive of publicly available protein sequences seen across different databases

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5-HT_{2a} рецептор человека

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB 5-HT2a human Advanced | List Search

Status
Reviewed (Swiss-Prot) (9) UniProtKB 9 results

Popular organisms BLAST Align Map IDs Download Add View: Cards Table Customize columns Share

	Entry Name	Protein Names	Gene Names	Organism	Length
Human (5)	<input type="checkbox"/> P28223	5HT2A_HUMAN	5-hydroxytryptamine receptor 2A[...]	Homo sapiens (Human)	471 AA
Mouse (1)	<input type="checkbox"/> P50129	5HT2A_PIG	5-hydroxytryptamine receptor 2A[...]	Sus scrofa (Pig)	470 AA
Taxonomy	<input type="checkbox"/> P50128	5HT2A_MACMU	5-hydroxytryptamine receptor 2A[...]	Macaca mulatta (Rhesus macaque)	471 AA
Filter by taxonomy	<input type="checkbox"/> Q5R4Q6	5HT2A_PONPY	5-hydroxytryptamine receptor 2A[...]	Pongo pygmaeus (Bornean orangutan)	471 AA
Proteins with	<input type="checkbox"/> Q14416	GRM2_HUMAN	Metabotropic glutamate receptor 2[...]	Homo sapiens (Human)	872 AA
3D structure (6)	<input type="checkbox"/> P28335	5HT2C_HUMAN	5-hydroxytryptamine receptor 2C[...]	Homo sapiens (Human)	458 AA
Alternative products (isoforms) (4)	<input type="checkbox"/> P14416	DRD2_HUMAN	D(2) dopamine receptor[...]	Homo sapiens (Human)	443 AA
Alternative splicing (4)	<input type="checkbox"/> P41595	5HT2B_HUMAN	5-hydroxytryptamine receptor 2B[...]	Homo sapiens (Human)	481 AA
Beta strand (6)	<input type="checkbox"/> Q811D0	DLG1_MOUSE	Disks large homolog 1[...]	Mus musculus (Mouse)	905 AA
Binary interaction (6)					
More items					
Protein existence					
Protein level (6)					
Transcript level (3)					
Annotation score					
5 (7)					
4 (2)					
Sequence length					
401 - 600 (7)					
>= 801 (2)					

Feedback Help

5-HT_{2a} рецептор человека

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB Advanced | List Search Help

P28223 · 5HT2A_HUMAN

5-hydroxytryptamine receptor 2A · Homo sapiens (Human) · Gene: HTR2A (HTR2) · 471 amino acids · Evidence at protein level · Annotation score: 9/9

Entry Feature viewer Publications External links History

BLAST Align Download Add Add a publication Entry feedback

Function¹

G-protein coupled receptor for 5-hydroxytryptamine (serotonin) (PubMed:1330647, PubMed:18703043, PubMed:19057895). Also functions as a receptor for various drugs and psychoactive substances, including mescaline, psilocybin, 1-(2,5-dimethoxy-4-iodophenyl)-2-aminopropane (DOI) and lysergic acid diethylamide (LSD) (PubMed:28129538). Ligand binding causes a conformation change that triggers signaling via guanine nucleotide-binding proteins (G proteins) and modulates the activity of down-stream effectors (PubMed:28129538). Beta-arrestin family members inhibit signaling via G proteins and mediate activation of alternative signaling pathways (PubMed:28129538). Signaling activates phospholipase C and a phosphatidylinositol-calcium second messenger system that modulates the activity of phosphatidylinositol 3-kinase and promotes the release of Ca²⁺ ions from intracellular stores (PubMed:18703043, PubMed:28129538). Affects neural activity, perception, cognition and mood (PubMed:18297054). Plays a role in the regulation of behavior, including responses to anxiogenic situations and psychoactive substances. Plays a role in intestinal smooth muscle contraction, and may play a role in arterial vasoconstriction [7 Publications](#)

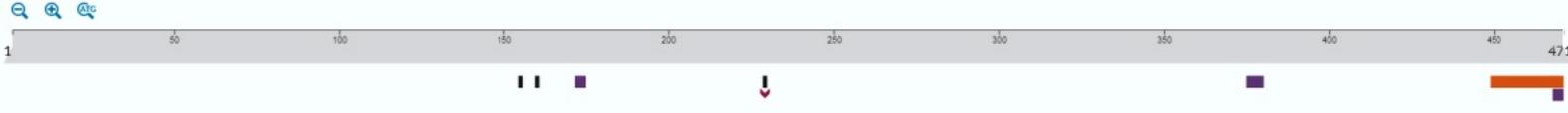
(Microbial infection) Acts as a receptor for human JC polyomavirus/JCPyV. [1 Publication](#)

Miscellaneous

Binds lysergic acid diethylamine (LSD) in the orthosteric pocket (Probable). Bound LSD dissociates extremely slowly, with a residence time of about 221 minutes at 37 degrees Celsius. [1 Publication](#)

Features

Showing features for binding site¹, motif¹, site¹, region¹.



TYPE	ID	POSITION(S)	DESCRIPTION
Binding site		155	ergotamine (UniProtKB ChEBI ¹); agonist By Similarity
Binding site		160	ergotamine (UniProtKB ChEBI ¹); agonist By Similarity

Последовательность 5-HT_{2A} Human

Sequences (2+)ⁱ

Sequence statusⁱ: Complete.

This entry describes **2** isoformsⁱ produced by **alternative splicing**. [Align](#) [Add to basket](#)

This entry has 2 described isoforms and 2 potential isoforms that are computationally mapped. [Show all](#) [Align All](#)

Isoform 1 (identifier: **P28223-1**) [UniProt] [FASTA](#) [Add to basket](#)

This isoform has been chosen as the canonicalⁱ sequence. All positional information in this entry refers to it. This is also the sequence that appears in the downloadable versions of the entry.

[« Hide](#)

Length: 471

Mass (Da): 52,603

Last modified: June 1, 1994 - v2

Checksum:ⁱ EF8AAC0BC5379DA2

BLAST [GO](#)

```
      10      20      30      40      50
MDILCEENTS LSSTTNSLMQ LNDDTRLYSN DFNSGEANTS DAFNWTVDSE
      60      70      80      90     100
NRTNLSCEGC LSPSCLSL LH LQEKNSALL TAVVIILTIA GNILVIMAVS
     110     120     130     140     150
LEKKLQ NATN YFLMSLAIAD MLLGFLVMPV SMLTILYGYR WPLPSKLC AV
     160     170     180     190     200
WIYLDVLFST ASIMHLCAIS LDRYVAIQNP IHHSRFNSRT KAFLKIIAVW
     210     220     230     240     250
TISVGISMPI PVFGLQDDSK VFKEGSCLLA DDNFVLIGSF VSFFIPLTIM
     260     270     280     290     300
VITYFLTIKS LQKEATLCVS DLGTRAKLAS FSFLPQSSLS SEKLFQRSIH
     310     320     330     340     350
REPGSYTGRR TMSISNEQK ACKVLGIVFF L FVVMWCPFF ITNIMAVICK
     360     370     380     390     400
ESCNE DVIGA LLNVFVWIGY LSSAVNPLVY TLFNKTYRSA FSRYIQ CQYK
     410     420     430     440     450
ENKKPLQLIL VNTIPALAYK SSQLQMGQKK NSKQDAKTTD NDCSMVALGK
     460     470
QHSEEASKDN SDGVNEKVSC V
```

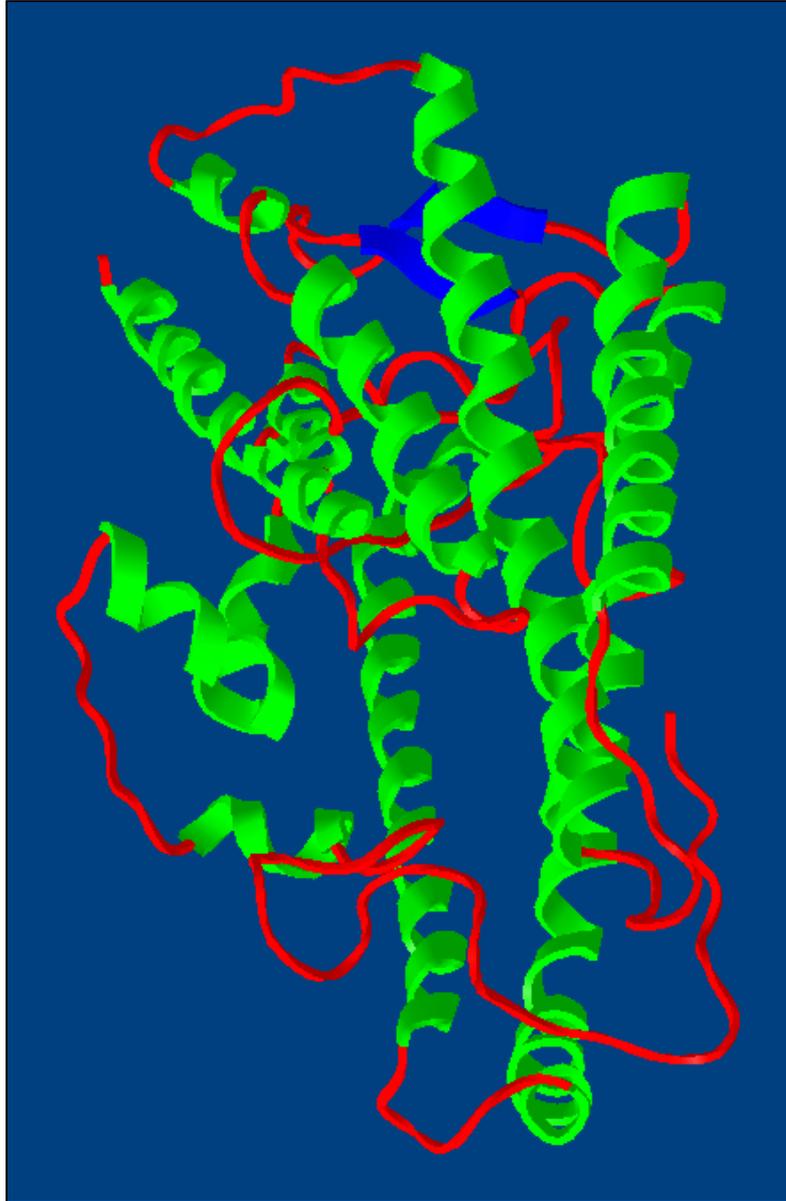
FASTA 5-HT_{2A} Human

```
>sp|P28223|5HT2A_HUMAN 5-hydroxytryptamine receptor 2A OS=Homo sapiens GN=HTR2A PE=1 SV=2
MDILCEENTSLSSSTNSLMQLNDDTRLYSNDNFNSGEANTSDAFNWTVDSENRTNLSCEGC
LSPSCLSLHLQEKNEWSALLTAVVIILTIAGNILVIMAVSLEKKLQNATNYFLMSLAIAD
MLLGFLVMPVSMLTILYGYRWPLPSKLCVWIIYLDVLFSTASIMHLCAISLDRYVAIQNP
IHHSRFNSRTKAFLKIIAVWTISVGISMPIPVFGLQDDSKVFKEGSCLLADDNFVLIGSF
VSFFIPLTIMVITYFLTIKSLQKEATLCVSDLGTRAKLASFSLPQSSLSSEKLFQRSIH
REPGSYTGRRTMQSISNEQKACKVLGIVFFLFVVMWCPFFITNIMAVICKESCNEVDVIGA
LLNVFVMWIGYLS SAVNPLVYTLFNKTYRSAFSRYIQCYKENKKPLQLILVNTIPALAYK
SSQLQMGQKNSKQDAKTTDNC SMVALGKQHSEEASKDNSDGVNEKVSCV
```

PDB формат

```
HEADER      ModPipe Model of UP B2RAC5                      2010-09-2
TITLE       Model of 5-hydroxytryptamine receptor 2A isoform 1 [Homo sa
SOURCE
AUTHOR      URSULA PIEPER, EASHWAR NARAYANAN, ANDREJ SALI
REMARK 220 Original ID: ENSP0000036795
REMARK 220 EXPERIMENTAL DETAILS
REMARK 220 EXPERIMENT TYPE: THEORETICAL MODEL
REMARK 220 METHOD: HOMOLOGY MODELING
REMARK 220 PROGRAM: MODPIPE
REMARK 220 SEQUENCE IDENTITY:                18.00
REMARK 220 GA341 SCORE:                      0.29
REMARK 220 EVALUE:                          0
REMARK 220 MPQS:                             0.671047
REMARK 220 zDOPE SCORE:                     0.78
REMARK 220 TEMPLATE PDB:                   2ziy
REMARK 220 TEMPLATE CHAIN:                 A
REMARK 220 TARGET LENGTH:                 471
REMARK 220 TARGET BEGIN:                 75
REMARK 220 TARGET END:                   439
REMARK 220 TEMPLATE BEGIN:               35
REMARK 220 TEMPLATE END:                 369
REMARK 220 MODPIPE RUN:                   human_2010_I1
REMARK 220 MODPIPE MODEL ID:              5d6b98e9e00432ae919177b2b38d636c
REMARK 220 MODPIPE ALIGN ID:              ca14ce341fce7d514026f126f0842823
REMARK 220 MODPIPE SEQUENCE ID:           3d8573062bfc3a41e50f3ea5cd5f1570MDILUSCU
EXPDTA      THEORETICAL MODEL, MODELLER SUN 2010/07/22 23:00:26
REMARK      6 MODELLER OBJECTIVE FUNCTION: 3242.1162
REMARK      6 MODELLER BEST TEMPLATE % SEQ ID: 18.209
REMARK      6 GENERATED BY MODPIPE UERSION SUN.r1188:1193
ATOM        1  N  ASN  75      53.564  25.782  27.022  1.00  43.29      N
ATOM        2  CA ASN  75      53.307  26.958  26.181  0.50  43.29      C
ATOM        3  CB ASN  75      54.466  27.190  25.199  0.50  43.29      C
ATOM        4  CG ASN  75      55.636  27.743  25.995  0.50  43.29      C
ATOM        5  OD1 ASN 75      56.654  27.076  26.170  1.00  43.29      O
ATOM        6  ND2 ASN 75      55.487  28.999  26.496  1.00  43.29      N
ATOM        7  C  ASN  75      52.053  26.820  25.395  1.00  43.29      C
ATOM        8  O  ASN  75      51.000  26.472  25.925  1.00  43.29      O
ATOM        9  N  TRP  76      52.159  27.075  24.082  1.00125.00    N
ATOM       10  CA TRP  76      51.022  27.089  23.216  1.00125.00    C
```

3D-модель 5-HT_{2a}-рецептора человека



Поиск нуклеотидных последовательностей

GenBank

<https://www.ncbi.nlm.nih.gov/genbank/>

234,6 млн. последовательностей

GenBank

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Nucleotide Nucleotide **(homo sapiens[Organism]) AND (whole genome[Title])** Search

Species **245 766** полных геномов человека

Animals (245,766) Customize ...

Molecule types genomic DNA/RNA (245,766) Customize ...

Source databases INSDC (GenBank) (97,175) Customize ...

Sequence Type Nucleotide (245,766)

Genetic compartments Mitochondrion (22)

Sequence length Custom range...

Release date Custom range...

Revision date Custom range...

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Items: 1 to 20 of 245766

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- [Homo sapiens isolate NA24385 chromosome 1, whole genome shotgun sequence](#)
251,946,536 bp linear DNA
Accession: CM039011.1 GI: 2188936806
[GenBank](#) [FASTA](#) [Graphics](#)
- [Homo sapiens isolate NA24385 chromosome 2, whole genome shotgun sequence](#)
241,746,906 bp linear DNA
Accession: CM039012.1 GI: 2188936183
[GenBank](#) [FASTA](#) [Graphics](#)
- [Homo sapiens isolate NA24385 chromosome 3, whole genome shotgun sequence](#)
199,578,765 bp linear DNA
Accession: CM039013.1 GI: 2188936182
[GenBank](#) [FASTA](#) [Graphics](#)
- [Homo sapiens isolate NA24385 chromosome 1, whole genome shotgun sequence](#)
252,114,745 bp linear DNA
Accession: CM039034.1 GI: 2188936181
[GenBank](#) [FASTA](#) [Graphics](#)
- [Homo sapiens isolate NA24385 chromosome 2, whole genome shotgun sequence](#)
242,054,856 bp linear DNA
Accession: CM039035.1 GI: 2188936176
[GenBank](#) [FASTA](#) [Graphics](#)
- [Homo sapiens isolate NA24385 chromosome 3, whole genome shotgun sequence](#)
199,186,228 bp linear DNA
Accession: CM039036.1 GI: 2188936175
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Database: Select
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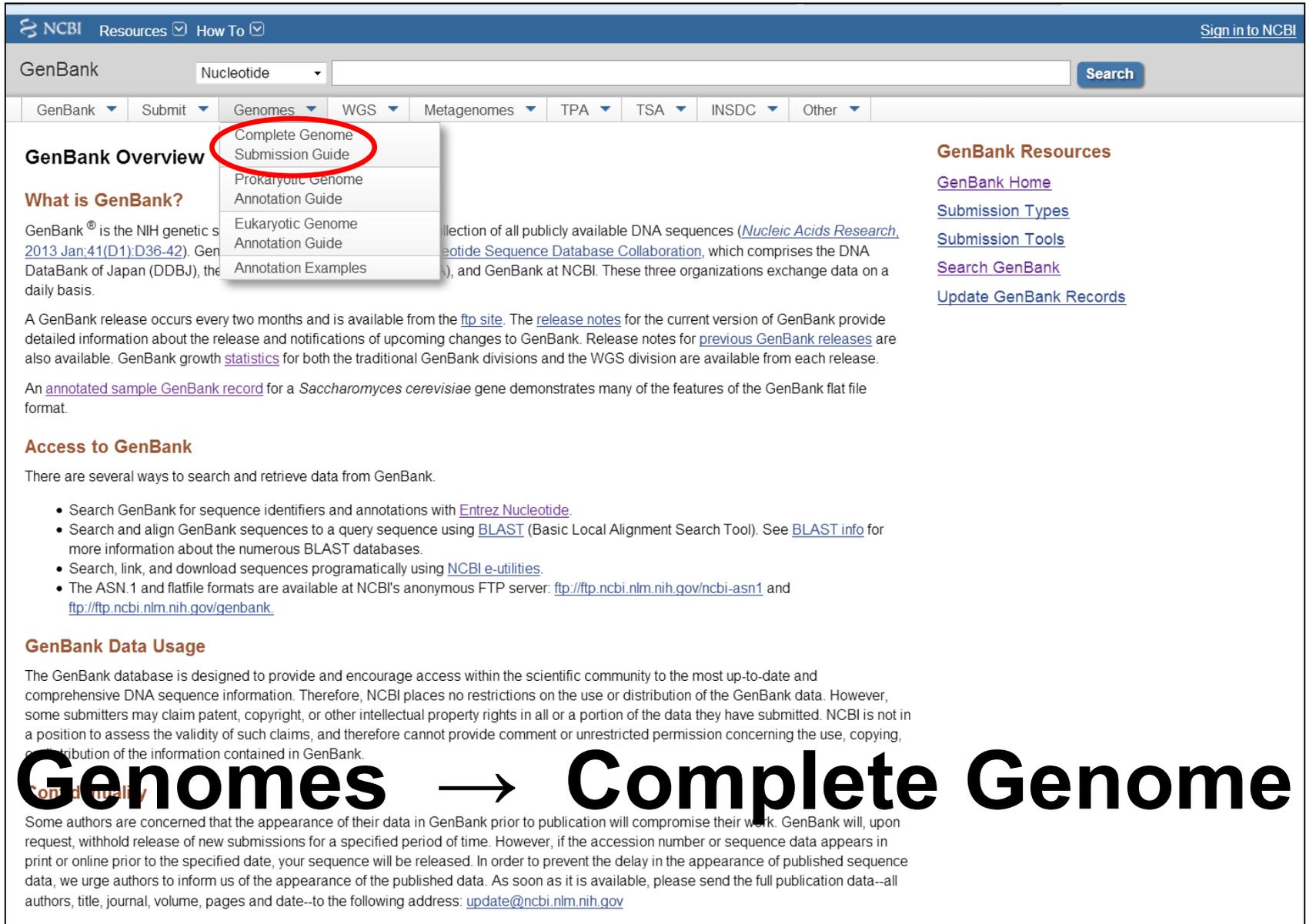
Search details
"Homo sapiens"[Organism] AND whole genome[Title]
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- (homo sapiens[Organism]) AND (whole genome[Title]) (245766) Nucleotide
- GABA Potency at GABAA Receptors Found in Synaptic and Extrasynaptic Zones
- Development of Polarizable Models for Molecular Mechanical Calculations IV: van ...
- AutoDock VinaXB: implementation of XBSF, new empirical halogen bond scoring func...
- MetStabOn—Online Platform for Metabolic Stability Predictions

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GenBank



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GenBank Overview

What is GenBank?

GenBank® is the NIH genetic sequence database, an annotated collection of all publicly available DNA sequences ([Nucleic Acids Research](#), 2013 Jan;41(D1):D36-42). GenBank is part of the International Nucleotide Sequence Database Collaboration, which comprises the DNA DataBank of Japan (DDBJ), the European Nucleotide Archive (ENA), and GenBank at NCBI. These three organizations exchange data on a daily basis.

A GenBank release occurs every two months and is available from the [ftp site](#). The [release notes](#) for the current version of GenBank provide detailed information about the release and notifications of upcoming changes to GenBank. Release notes for [previous GenBank releases](#) are also available. GenBank growth [statistics](#) for both the traditional GenBank divisions and the WGS division are available from each release.

An [annotated sample GenBank record](#) for a *Saccharomyces cerevisiae* gene demonstrates many of the features of the GenBank flat file format.

Access to GenBank

There are several ways to search and retrieve data from GenBank.

- Search GenBank for sequence identifiers and annotations with [Entrez Nucleotide](#).
- Search and align GenBank sequences to a query sequence using [BLAST](#) (Basic Local Alignment Search Tool). See [BLAST info](#) for more information about the numerous BLAST databases.
- Search, link, and download sequences programatically using [NCBI e-utils](#).
- The ASN.1 and flatfile formats are available at NCBI's anonymous FTP server: <ftp://ftp.ncbi.nlm.nih.gov/hcbi-asn1> and <ftp://ftp.ncbi.nlm.nih.gov/genbank>.

GenBank Data Usage

The GenBank database is designed to provide and encourage access within the scientific community to the most up-to-date and comprehensive DNA sequence information. Therefore, NCBI places no restrictions on the use or distribution of the GenBank data. However, some submitters may claim patent, copyright, or other intellectual property rights in all or a portion of the data they have submitted. NCBI is not in a position to assess the validity of such claims, and therefore cannot provide comment or unrestricted permission concerning the use, copying, or distribution of the information contained in GenBank.

Genomes → Complete Genome

Some authors are concerned that the appearance of their data in GenBank prior to publication will compromise their work. GenBank will, upon request, withhold release of new submissions for a specified period of time. However, if the accession number or sequence data appears in print or online prior to the specified date, your sequence will be released. In order to prevent the delay in the appearance of published sequence data, we urge authors to inform us of the appearance of the published data. As soon as it is available, please send the full publication data--all authors, title, journal, volume, pages and date--to the following address: update@ncbi.nlm.nih.gov

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Prokaryotic and Eukaryotic Genomes Submission Guide

Both WGS and non-WGS genomes, including gapless complete bacterial chromosomes, can be submitted via the Submission Portal. You will be asked to choose whether the genome being submitted is considered WGS or not. The differences for GenBank purposes are:

non-WGS genome

WGS genome

= Gap (Ns)

non-WGS

- Each chromosome is in a single sequence and there are no extra sequences
- Each sequence in the genome must be assigned to a chromosome or plasmid or organelle
- Plasmids and organelles can still be in multiple pieces.

WGS

- One or more chromosomes are in multiple pieces and/or some sequences are not assembled into chromosomes

In both cases

- There can still be gaps within the sequences; you will supply that information in the submission
- Plasmids and organelles can still be in multiple pieces
- Internal sequences must be arranged in the correct order and orientation
- Sequences concatenated in unknown order are not allowed.

WGS Browser

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- [Annotation Example Files](#)
- [Validation Error Explanations for Genomes](#)
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GenBank — WGS Browser

NCBI Sequence Set Browser

Facet Panel

- Available Facets
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 - Source database
 - Targeted Locus Name
 - DIV
 - Organism**
 - Bioproject
 - Biosample
 - Strain
 - Breed
 - Cultivar
 - Isolate
 - Tissue Type
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 - Isolation Source
 - Dev Stage
- Organism
 - homo sapiens** 1,043
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 - Homo sapiens [1043]
 - human oral metagenome [392]

Description

This site is for browsing WGS (Whole Genome Shotgun) genomes, TSA (Transcriptome Shotgun Assemblies) and TLS (Targeted Locus Study) sets. Sequences are incomplete genomes that have been sequenced by a whole genome shotgun strategy. TSA sequences are transcript sequences that have been computationally assembled from primary RNA sequence data. TLS sequences are large-scale marker gene sequencing studies. Please consult [WGS Submission](#) or [TSA Submission](#) pages for more details.

Project type:

Search – search in all fields. Use wildcard "" to search in the middle of a field's text.

Term

Found 1,435 projects Page 2 3 4 5 6 7 8 ... 29

#	Prefix	Type	DIV	Organism	Bioproject	Biosample	Intraspecific Name	Other Source	Contigs				
									Total Length (Mbases)	#	# Prot	Has Annot	
1	AACC02	WGS	PRI	Homo sapiens	PRJNA10793	SAMN02981216			137.7	171	109	Y	
2	AADB02	WGS	PRI	Homo sapiens	PRJNA1431	SAMN02981219			3,087.3	401,931			14
3	AACC02	WGS	PRI	Homo sapiens	PRJNA10793	SAMN02981216			2,656	169,156			2
4	AADD01	WGS	PRI	Homo sapiens	PRJNA1431	SAMN02981219			2,695.6	211,493			1

Human whole genome

GenBank — Project: AACCC02

 **Sequence Set Browser**  Sign In to NCBI

Project:

AACC00000000.2 Homo sapiens

# of Contigs:	171	<p>On Jul 19, 2004 this sequence version replaced gi:29727032.</p> <p>The Homo sapiens whole genome shotgun (WGS) project has the project accession AACC00000000. This version of the project (02) has the accession number AACC02000000, and consists of sequences AACC02000001-AACC02000171.</p>
# of Proteins:	818	
# of Scaffolds/Chrs:	20	
Total length:	137,712,494 bp	
BioProject:	PRJNA10793	
BioSample:	SAMN02981216	
Keywords:	WGS	
Annotation:	Contigs, Scaffolds	
Organism:	Homo sapiens – show lineage	
Biosource:	/chromosome = 7 /mol_type = genomic genomic	
WGS:	AACC02000001:AACC02000171	
Scaffolds:	CH236947:CH236966	
Reference:	20 scaffolds, 709 proteins, total length is 123,763,301 bases Human chromosome 7: DNA sequence and biology : Science 300 (5620), 767-772 (2003) – show 90 authors	
Submission:	Submitted (27-MAR-2003) Department of Genetics and Genomic Biology, The Hospital for Sick Children, 555 University Avenue, Toronto, Ontario M5G 1X8, Canada – show 88 authors	
Submission:	Submitted (29-JUN-2004) Department of Genetics and Genomic Biology, The Hospital for Sick Children, 555 University Avenue, Toronto, Ontario M5G 1X8, Canada – show 88 authors	

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Поиск нуклеотидных последовательностей

1000 Genomes

IGSR: The International Genome Sample Resource

<https://www.internationalgenome.org/>

**5019 полных геномов людей
разных национальностей**

1000 Genomes

IGSR: The International Genome Sample Resource

Supporting open human variation data

Home About **Data** Help

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The International Genome Sample Resource

The 1000 Genomes Project created a catalogue of common human genetic variation, using openly consented samples from people who declared themselves to be healthy. The reference data resources generated by the project remain heavily used by the biomedical science community.

The International Genome Sample Resource (IGSR) maintains and shares the human genetic variation resources built by the 1000 Genomes Project. We also update the resources to the current reference assembly, add new data sets generated from the 1000 Genomes Project samples and add data from projects working with other openly consented samples.



	G: 0.934 (185)	GIG: 0.879 (87)	
GWD	A: 0.066 (15) G: 0.934 (211)	AIA: 0.009 (1) GIG: 0.876 (99)	AIG: 0.115 (13)
LWK	A: 0.111 (22)	AIA: 0.020 (2) GIG: 0.876 (79)	AIG: 0.182 (18)
MSL	A: 0.024 (4) G: 0.976 (166)	AIG: 0.047 (4)	GIG: 0.953 (81)
YRI	A: 0.079 (17) G: 0.921 (199)	AIG: 0.157 (17)	GIG: 0.843 (91)

View variants in genomic context in Ensembl

Latest Announcements

Thursday September 30, 2021

[A variation call set obtained from the analysis of Gambian Genome Variation Project samples on GRCh38](#)

We have recently published a [Data Note](#) describing our analysis of 505 samples from four Gambian populations in the Gambian Genome Variation Project (GGVP) on GRCh38.

For the analysis we have used a multi-caller site discovery approach along with imputation and phasing to produce a phased biallelic single nucleotide variant (SNV) and insertion/deletion (INDEL) call set. Variation had not previously been explored on the GRCh38 human genome assembly for 387 of the samples. Compared to our previous work with the 1000 Genomes Project data on GRCh38 described [here](#), we identified over nine million novel SNVs and over 870 thousand novel INDELS.

The files generated in this analysis can be accessed from our FTP. Including the alignment files used in the variant identification http://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/gambian_genome_variation_project/data/ and the call set itself http://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/gambian_genome_variation_project/release/20200217_biallelic_SNV/

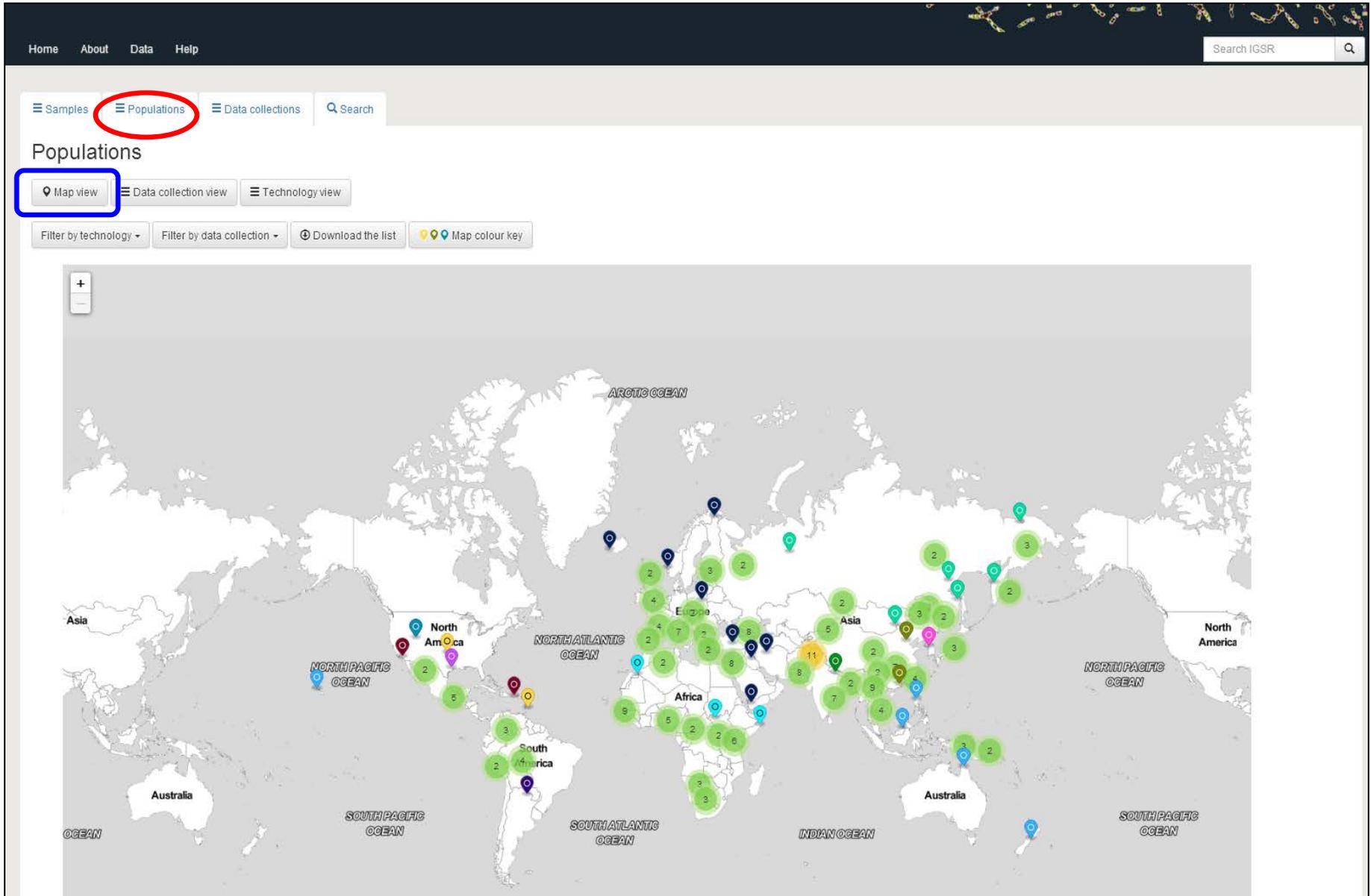
More information on the samples analysed in this work can be found in the [IGSR portal](#).

Frequency distributions and genotypes are available in [Ensembl](#).

[All announcements](#)

Data → **Data Portal**

1000 Genomes — Map



1000 Genomes — Samples

IGSR: The International Genome Sample Resource

Supporting open human variation data



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Samples Populations Data collections Search

Samples

Data collection view Technology view

Filter by population Filter by technology Filter by data collection Download the list

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Sample	Sex	Populations	30x GRCh38	HGSVC2	GRCh38	Phase 3	Phase 1	Structural variation	HGDP	SGDP	GGVP GRCh38	GGVP GRCh37	Genradis	Platinum Pedigree	90 Han Chinese	HGDP Transcriptome
HG00152	Male	British in England and Scotland		•	•	•						•				
HG00157	Male	British in England and Scotland	•		•	•	•					•				
HG00171	Female	Finnish in Finland	•	•	•	•	•					•				
HG00176	Female	Finnish in Finland	•		•	•	•					•				
HG00183	Male	Finnish in Finland	•		•	•	•					•				
HG00188	Male	Finnish in Finland	•		•	•	•					•				
HG00190	Male	Finnish in Finland Finnish in Finland (SGDP)	•		•	•	•		•							
HG00234	Male	British in England and Scotland	•		•	•	•					•				
HG00239	Female	British in England and Scotland	•		•	•	•					•				
HG00246	Male	British in England and Scotland	•		•	•	•					•				

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1000 Genomes — HG00152

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☰ Samples ☰ Populations ☰ Data collections 🔍 Search

Sample HG00152

HG00152 details

Sex: Male
Populations: [British in England and Scotland, European Ancestry](#)
Biosample ID: [SAME124593](#)
Cell line source: [HG00152 at Coriell](#) 🔍

Data collections for HG00152

1000 Genomes on GRCh38

1000 Genomes phase 3 release

1000 Genomes phase 1 release

Geuvadis

[Data reuse policy for 1000 Genomes on GRCh38](#)

60 matching data files

Download the list

Data types

- Variants
- Sequence
- Alignment

Technologies

- Integrated variant call sets
- Exome
- Low coverage WGS

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File	Technology
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr14.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr7.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz.tbi	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr8.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz.tbi	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr19.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz.tbi	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr1.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz.tbi	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr18.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz.tbi	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr12.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz.tbi	Integrated variant call sets
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data_collections/1000_genomes_project/release/20190312_biallelic_SNV_and_INDEL/ALL.chr20.shapeit2_integrated_snvindels_v2a_27022019.GRCh38.phased.vcf.gz	Integrated variant call sets

1000 Genomes — Exome

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Sample HG00152

HG00152 details

Sex: Male
Populations: [British in England and Scotland](#), European Ancestry
Biosample ID: [SAME124593](#)
Cell line source: [HG00152 at Coriell](#)

Data collections for HG00152

1000 Genomes on GRCh38

1000 Genomes phase 3 release

1000 Genomes phase 1 release

Geuvadis

[Data reuse policy for 1000 Genomes on GRCh38](#)

2 matching data files

Download the list

Data types

- Variants
- Sequence
- Alignment

Technologies

- Integrated variant call sets
- Exome
- Low coverage WGS

File

- ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR769/SRR769545/SRR769545_2.fastq.gz
- ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR769/SRR769545/SRR769545_1.fastq.gz

Technology

Exome
Exome

To be continued ...

