



M1 SM THORAX ED TG: Recherche in silico.



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Team IIA

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Nantes, France

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Search

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Homology

Literature

Proteins

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Taxonomy

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DNA & RNA

Domains & Structures

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Genetics & Medicine

Genomes & Maps

Homology

POPULAR

PubMed

Bookshelf

PubMed Central

BLAST

Nucleotide

Genome

SNP

Cancer

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[NCBI Home](#)

Resource List (A-Z)

- [All Resources](#)
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- [Data & Software](#)
- [DNA & RNA](#)
- [Domains & Structures](#)
- [Genes & Expression](#)
- [Genetics & Medicine](#)
- [Genomes & Maps](#)
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- [Literature](#)
- [Proteins](#)
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SRA High-throughput sequence reads	PubChem Repository of chemical information, molecular pathways, and tools for bioactivity screening	dbVar Genome structural variation studies			
Taxonomy Taxonomic classification and nomenclature	BioAssays Bioactivity screening studies	GTR Genetic testing registry			
	Compounds Chemical information with structures, information and links	MedGen Medical genetics literature and links			
	Pathways Molecular pathways with links to genes, proteins and chemicals	OMIM Online mendelian inheritance in man			
	Substances Deposited substance and chemical information				

Le NCBI abrite une série de bases de données pertinentes pour la biotechnologie et la biomédecine et constitue une ressource importante pour les outils et services de bioinformatique.

OMIM : Le projet Héritage mendélien chez l'humain (en anglais : Mendelian Inheritance in Man) est une base de données originellement compilée par Victor A. McKusick et qui dresse un catalogue de toutes les maladies connues qui relèvent de l'un ou l'autre composant génétique et — si possible — les relie aux gènes adéquats au sein du génome humain. Cette base de données est disponible sous forme d'un livre appelé Mendelian Inheritance in Man (MIM), qui en est à sa 13e édition.

La version en ligne est appelée Online Mendelian Inheritance in Man, OMIM, et peut être consultée à partir de la base de données Entrez1 de la National Library of Medicine2.

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OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is omim.org.

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Items: 1 to 20 of 26

<< First < Prev Page 1 of 2 Next > Last >>

 [#253700 - MUSCULAR DYSTROPHY, LIMB-GIRDLE, AUTOSOMAL RECESSIVE 5; LGMDR5](#)

1. Cytogenetic locations: 13q12.12

OMIM: 253700

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2. Cytogenetic locations: 17q21.33

OMIM: 608099

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3. Cytogenetic locations: Xp21.2-p21.1

OMIM: 310200

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4. Cytogenetic locations: 21q22.3, 1p36, 21q22.3

OMIM: 158810

[Gene summaries](#) [Genetic tests](#) [Medical literature](#) [#310400 - MYOPATHY, CENTRONUCLEAR, X-LINKED; CNMX](#)

5. Cytogenetic locations: Xq28

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[Gene summaries](#) [Genetic tests](#) [Medical literature](#) [#254130 - MIYOSHI MUSCULAR DYSTROPHY 1; MMD1](#)

6. Cytogenetic locations: 2p13.2

OMIM: 254130

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7. Cytogenetic locations: Xq28

OMIM: 310440

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8. Cytogenetic locations: Xq24

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[myopathy \(666\)](#)

OMIM

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OMIM

[dystrophin \(135\)](#)

OMIM

[See more...](#)

#310200

[Table of Contents](#)

310200

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Title[Phenotype-Gene Relationships](#)[Clinical Synopsis](#)**Text**[Description](#)[Clinical Features](#)[Other Features](#)[Inheritance](#)[Cytogenetics](#)[Mapping](#)[Molecular Genetics](#)[Diagnosis](#)[Clinical Management](#)[Population Genetics](#)[Animal Model](#)**See Also**[References](#)[Contributors](#)[Creation Date](#)[Edit History](#)

MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD

Alternative titles; symbols

DUCHENNE MUSCULAR DYSTROPHY

MUSCULAR DYSTROPHY, PSEUDOHYPERTROPHIC PROGRESSIVE, DUCHENNE TYPE

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
Xp21.2-p21.1	Duchenne muscular dystrophy	310200	XLR	3	DMD	300377

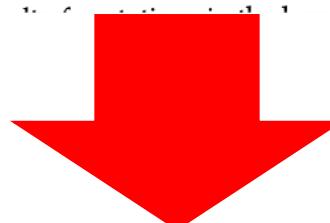
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▼ TEXT

A number sign (#) is used with this entry because Duchenne muscular dystrophy is caused by mutation in the gene encoding dystrophin (DMD; 300377).

▼ Description

Dystrophin-associated muscular dystrophies range from the severe Duchenne muscular dystrophy (DMD) to the milder Becker muscular dystrophy (BMD; 300376). Mapping and molecular genetic



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[▶ Clinical Trials](#)[EuroGentest](#)[▶ Gene Reviews](#)[Genetic Alliance](#)[Genetics Home Reference](#)[GTR](#)[GARD](#)[OrphaNet](#)[POSSUM](#)[▶ Animal Models](#)[▶ Cell Lines](#)

[Title](#)[Phenotype-Gene Relationships](#)[Clinical Synopsis](#)[Text](#)[Description](#)[Clinical Features](#)[Other Features](#)[Inheritance](#)[Cytogenetics](#)[Mapping](#)[Molecular Genetics](#)[Diagnosis](#)[Clinical Management](#)[Population Genetics](#)[Animal Model](#)[See Also](#)[References](#)[Contributors](#)[Creation Date](#)[Edit History](#)

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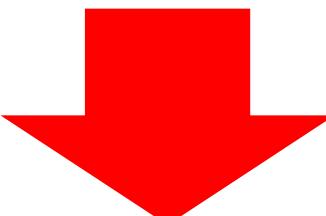
Dystrophin-associated muscular dystrophies range from the severe Duchenne muscular dystrophy (DMD) to the milder Becker muscular dystrophy (BMD; [300376](#)). Mapping and molecular genetic studies indicate that both are the result of mutations in the huge gene that encodes dystrophin, also symbolized DMD. Approximately two-thirds of the mutations in both forms are deletions of one or many exons in the dystrophin gene. Although there is no clear correlation found between the extent of the deletion and the severity of the disorder, DMD deletions usually result in frameshift. [Boland et al. \(1996\)](#) studied a retrospective cohort of 33 male patients born between 1953 and 1983. The mean age at DMD diagnosis was 4.6 years; wheelchair dependency had a median age of 10 years; cardiac muscle failure developed in 15% of patients with a median age of 21.5 years; smooth muscle dysfunction in the digestive or urinary tract occurred in 21% and 6% of the patients, respectively, at a median age of 15 years. In this cohort, death occurred at a median age of 17 years. The authors commented that the diagnosis of DMD is being made at an earlier age but survival has not changed.



▼ Clinical Features

Skeletal Muscle

The most distinctive feature of Duchenne muscular dystrophy is a progressive proximal muscular dystrophy with characteristic pseudohypertrophy of the calves. The bulbar (extraocular) muscles are spared but the myocardium is affected. There is massive elevation of creatine kinase levels in the blood, myopathic changes by electromyography, and myofiber degeneration with fibrosis and fatty infiltration on muscle biopsy. The onset of Duchenne muscular dystrophy usually occurs before age 3 years, and the victim is chairridden by age 12 and dead by age 20. The onset of Becker muscular dystrophy is often in the 20s and 30s and survival to a relatively advanced age is frequent.



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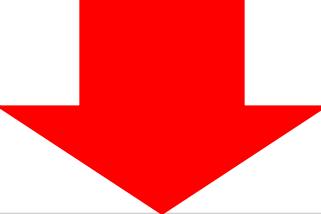
[GARD](#)

[OrphaNet](#)

[POSSUM](#)

► Animal Models

► Cell Lines



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Options ▾

#310200

Table of Contents

Title

Phenotype-Gene Relationships

Clinical Synopsis

Text

Description

Clinical Features

Other Features

Inheritance

Cytogenetics

Mapping

Molecular Genetics

Diagnosis

Clinical Management

Population Genetics

Animal Model

See Also

References

Contributors

Creation Date

Edit History

▼ REFERENCES

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▼ Clinical Resources

Clinical Trials

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► Cell Lines

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[NCBI Home](#)[Resource List \(A-Z\)](#)[All Resources](#)[Chemicals & Bioassays](#)[Data & Software](#)[DNA & RNA](#)[Domains & Structures](#)[Genes & Expression](#)[Genetics & Medicine](#)[Genomes & Maps](#)[Homology](#)[Literature](#)[Proteins](#)[Sequence Analysis](#)[Taxonomy](#)[Training & Tutorials](#)[Variation](#)

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[All Resources](#)

[Chemicals & Bioassays](#)

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[DNA & RNA](#)

[Domains & Structures](#)

[Genes & Expression](#)

[Genetics & Medicine](#)

[Genomes & Maps](#)

[Homology](#)

[Literature](#)

[Proteins](#)

[Sequence Analysis](#)

[Taxonomy](#)

[Training & Tutorials](#)

[Variation](#)

Genomic

[GEO DataSets](#)

[GEO Profiles](#)

GTR

[HomoloGene](#)

[Identical Protein Groups](#)

[MedGen](#)

[MeSH](#)

[NCBI Web Site](#)

[NLM Catalog](#)

[Nucleotide](#)

[OMIM](#)

[PMC](#)

[PopSet](#)

[Protein](#)

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[Gene Expression Omnibus](#)

[Genome Data Viewer](#)

[Human Genome](#)

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- Preparing to Respond to the Next Pandemic: Impact of Key WHO IPC COVID-19 Response Products.

Cite Nzegwu F, Hamilton Hurwitz H, Willet V, Clery E, Ibeto M, Wild J, Baller A.

Public Health Chall. 2025 Sep 30;4(4):e70133. doi: 10.1002/phu2.70133. eCollection 2025 Dec.

PMID: 41036491 [Free PMC article.](#)

An outcomes/impact evaluation was undertaken (July 2023–April 2024) to evaluate the work of the World Health Organization (WHO) infection prevention and control (IPC) coronavirus disease (COVID-19) response team during the COVID-19 pandemic and to iden ...

- Recurrent Dural Arteriovenous Fistula following Cerebral Venous Sinus Thrombosis Related to COVID-19 Vaccination: A Case Report.

Cite Iwabuchi N, Yoshida M, Kumai M, Nishijima Y, Niizuma K, Endo H.

NMC Case Rep J. 2025 Sep 9;12:377-382. doi: 10.2176/jns-nmc.2024-0332. eCollection 2025.

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Cerebral venous sinus thrombosis (CVST) is rare but serious adverse complication of coronavirus disease 2019 (COVID-19) vaccination. CVST can sometimes lead to development of dural arteriovenous fistula (DAVF), but DAVF secondary to CVST following COVID-19 ...

- Isolation of Apigenin from Sungkai (*Peronema canescens*) Leaves and Its Immunomodulatory Effects: An In Vivo Study on Granzyme B, Interferon- γ , and Perforin Expression with Supporting In Silico Analysis.

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Occurrence and transmission potential of asymptomatic and presymptomatic SARS-CoV-2 infections: A living systematic review and meta-analysis

Diana Buitrago-Garcia ^{1 2}, Dianne Egli-Gany ¹, Michel J Counotte ¹, Stefanie Hossmann ¹, Hira Imeri ¹, Aziz Mert Ipekci ¹, Georgia Salanti ¹, Nicola Low ¹

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Abstract

Background: There is disagreement about the level of asymptomatic severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) infection. We conducted a living systematic review and meta-analysis to address three questions: (1) Amongst people who become infected with SARS-CoV-2, what proportion does not experience symptoms at all during their infection? (2) Amongst people with SARS-CoV-2 infection who are asymptomatic when diagnosed, what proportion will

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RESEARCH ARTICLE

Occurrence and transmission potential of asymptomatic and presymptomatic SARS-CoV-2 infections: A living systematic review and meta-analysis

Diana Buitrago-Garcia^{1,2*}, Dianne Egli-Gany^{1,2}, Michel J. Counotte^{1,2}, Stefanie Hossmann³, Hira Imeri⁴, Aziz Meri Ipekci⁵, Georgia Salanti¹, Nicola Low^{1,2}

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Data Availability Statement: The file listing all included studies and files used for all analyses are available from the Harvard Dataverse database.

Abstract

Background

There is disagreement about the level of asymptomatic severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) infection. We conducted a living systematic review and meta-analysis to address three questions: (1) Amongst people who become infected with SARS-CoV-2, what proportion does not experience symptoms at all during their infection? (2)

Amongst people with SARS-CoV-2 infection who are asymptomatic when diagnosed, what proportion will develop symptoms later? (3) What proportion of SARS-CoV-2 transmission is accounted for by people who are either asymptomatic throughout infection or presymptomatic?

Methods and findings

We searched PubMed, Embase, bioRxiv, and medRxiv using a database of SARS-CoV-2 literature that is updated daily, on 25 March 2020, 20 April 2020, and 10 June 2020. Studies of people with SARS-CoV-2 diagnosed by reverse transcriptase PCR (RT-PCR) that documented follow-up and symptom status at the beginning and end of follow-up or modelling studies were included. One reviewer extracted data and a second verified the extraction, with disagreement resolved by discussion or a third reviewer. Risk of bias in empirical studies was assessed with an adapted checklist for case series, and the relevance and credibility of modelling studies were assessed using a published checklist. We included a total of 94 studies. The overall estimate of the proportion of people who become infected with SARS-CoV-2 and remain asymptomatic throughout infection was 20% (95% confidence interval [CI] 17–25) with a prediction interval of 3%–67% in 79 studies that addressed this review question. There was some evidence that biases in the selection of participants influence the estimate. In seven studies of defined populations screened for SARS-CoV-2 and then

Table 1. Characteristics of studies reporting on proportion of asymptomatic SARS-CoV-2 infections.

Author	Country, location	Total SARS-CoV-2, n	Asymptomatic SARS-CoV-2, n	Sex of asymptomatic people	Age of asymptomatic people, years, median	Follow-up method ^a
Contact investigation, single						
Tong, ZD [44]	China, Zhejiang	5	3	2 F, 3 M	28 IQR 12–41	1, 3
Huang, R [74]	China, Suzhou	2	1	1 F, 0 M	54	3
Jiang, XL [26]	China, Shandong	8	3	3 F, 0 M	35 IQR 0–53	3
Jiang, X [75]	China, Chongqing	3	1	1 F, 0 M	8	2
Liao, J [22]	China, Chongqing	12	3	NR	NR	1, 2
Hu, Z [21]	China, Nanjing	4	1	0 F, 1 M	64	2, 3
Luo, SH [23]	China, Anhui	4	1	1 F, 0 M	50	1, 2, 3
Chan, JF [18]	China, Guangdong	5	1	0 F, 1 M	10	1
Ye, F [42]	China, Sichuan	5	1	0 F, 1 M	28	1, 2
Bai, Y [17]	China, Anyang	6	1	1 F, 0 M	20	1
Luo, Y [85]	China, Wuhan	6	5	NR	37 IQR 7–62	1
Zhang, J [50]	China, Wuhan and Beijing	5	2	1 F, 1 M	NR	2
Zhang, B [110]	China, Guangdong	7	2	0 F, 2 M	13.5 IQR 13–14	3
Huang, L [23]	China, Gansu	7	2	2 F, 0 M	44 IQR 38.5–49.5	2
Qian, G [26]	China, Zhejiang	8	2	1 F, 1 M	30.5 IQR 1–60	1, 2
Gao, Y [20]	China, Wuxi	15	6	3 F, 3 M	50 IQR 48–51	1, 2
Contact investigation, aggregated						
Hijnen, D [72]	Germany	11	1	0 F, 1 M	49	1
Brandsma, S [62]	Germany	36	2	NR	NR	2
Zhang, WZ [111]	China, Guiyang	12	4	NR	NR	1, 2, 3
Cheng, HY [56]	Taiwan	22	4	NR	NR	1
Wang, Z [47]	China, Wuhan	47	4	NR	NR	1
Wu, J [105]	China, Zhuhai	83	8	NR	NR	1, 2
Inou, T [16]	China, Guangzhou	129	8	NR	NR	1, 2, 3
Bi, Q [60]	China, Shenzhen	87	17	NR	NR	2, 3
Yang, R [108]	China, Wuhan	78	33	22 F, 11 M	37 IQR 26–45	3
Outbreak investigation						
Danis, K [32]	France	13	1	NR	NR	1, 2
Böhmer, MM [61]	Germany	16	1	NR	NR	1
Roxby, AC [94]	USA	6	3	NR	NR	1
Yang, N [48]	China, Xiaoshan	10	2	1 F, 1 M	NR	1, 2
Schwei策ck, V [35]	Germany	12	2	NR	NR	2
Arons, MM [58]	USA	47	3	NR	NR	2
Park, SY [90]	South Korea	97	4	NR	NR	2
Doria, AV [68]	USA	19	6	0 F, 6 M	75 IQR 72–75	3
Tian, S [43]	China, Shandong	24	7	NR	NR	3
Solbach, W [97]	Germany	97	10	NR	NR	2
Graham, N [71]	United Kingdom	126	46	NR	NR	2

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- S7 Fig. Assessment of credibility of mathematical modelling studies. (PDF)
- S1 Table. Types of study included in successive versions of the living systematic review, as of 10 June 2020. (DOCX)
- S2 Table. Location of studies contributing data to review questions 1 and 2. (DOCX)

Author Contributions

Conceptualization: Diana Buitrago-Garcia, Dianne Egli-Gany, Nicola Low.

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Investigation: Aziz Mert Ipekci.

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Supervision: Nicola Low.

Validation: Diana Buitrago-Garcia, Dianne Egli-Gany, Michel J. Counotte, Stefanie Hossmann, Hira Imeri, Aziz Mert Ipekci, Nicola Low.

Writing – original draft: Diana Buitrago-Garcia, Nicola Low.

Writing – review & editing: Diana Buitrago-Garcia, Dianne Egli-Gany, Michel J. Counotte, Stefanie Hossmann, Hira Imeri, Aziz Mert Ipekci, Georgia Salanti, Nicola Low.

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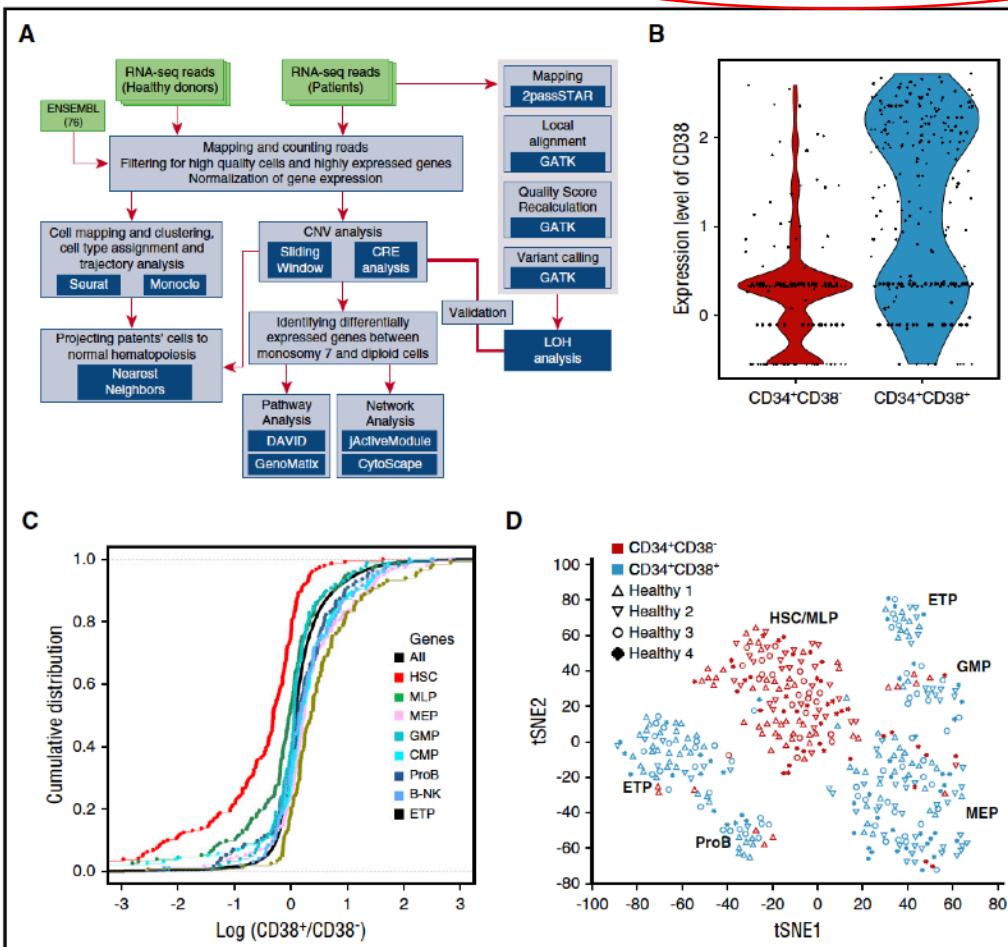


Figure 1. Hematopoietic heterogeneity in healthy donors quantified by scRNA-seq. CD34⁺CD38⁻ and CD34⁺CD38⁺ cells from 4 healthy donors (healthy 1-4) were sorted by surface-membrane markers and subjected to analyses. (A) The schematic pipeline consisting of 3 major analytic components: differentiation analysis with cells from healthy donors, identification and characterization of monosomy 7 cells with gene expression, and validation of monosomy identification with loss of heterozygosity (LOH). CNV, copy-number variation; CRE, chromosome relative expression; GATK, Genome Analysis Toolkit. (B) CD38 expression levels in CD34⁺CD38⁻ and CD34⁺CD38⁺ cells. Each dot represents a single cell. y-axis, batch-corrected gene expression levels. (C) Cumulative distribution of fold changes of expression of hematopoietic cell type signature genes between CD34⁺CD38⁻ and CD34⁺CD38⁺ cells. Each dot represents a gene. B-NK, B cell–natural killer cell precursor; CMP, common myeloid progenitor; ETP, earliest thymic progenitor; GMP, granulocyte–monocyte progenitor; MEP, megakaryocytic–erythroid progenitor; MLP, multilineage progenitor; ProB, pro-B cell. y-axis, cumulative distribution; x-axis, log (marker gene expression levels in CD34⁺CD38⁻ cells/marker gene expression levels in CD34⁺CD38⁺ cells). (D) t-distributed stochastic neighbor embedding (tSNE) plot of single-cell gene expression data. Single cells from 4 healthy donors (healthy 1-4) are represented by different symbols. Highly variable genes (1024) across all healthy donors were used in tSNE analysis.

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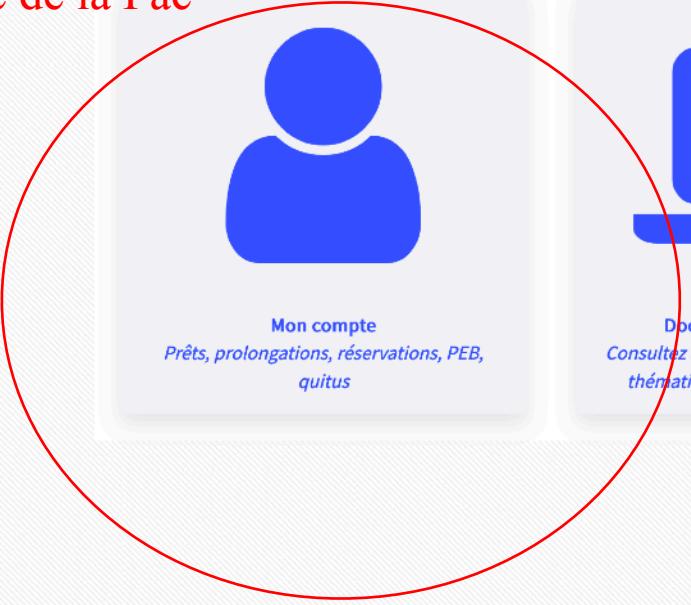
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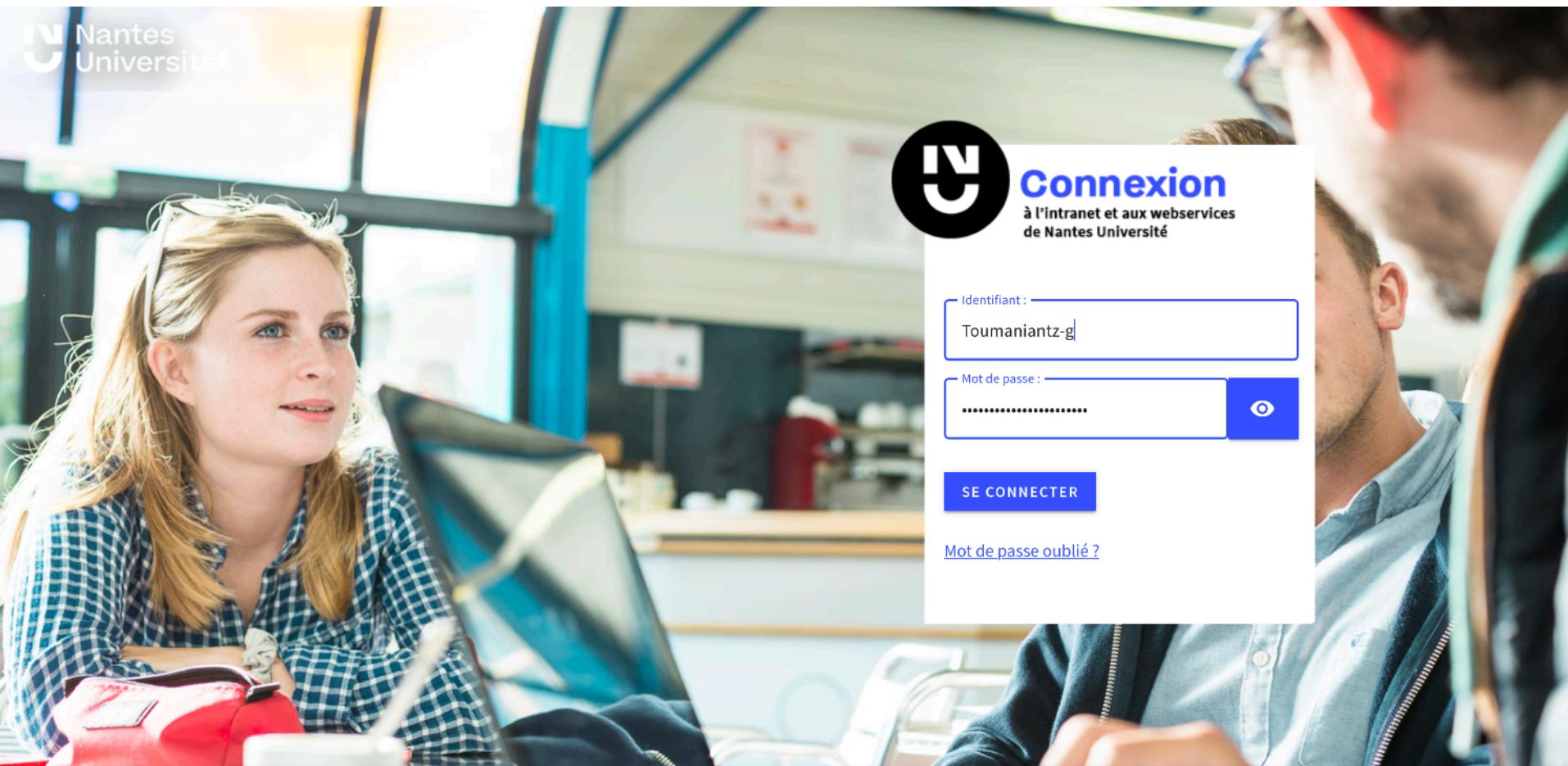
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Bibliothèque

BU Santé

12



BU Sciences

11



Bib. DCS (Droit et Changement Social)

3



Bib. Géothèque A. Vigarié

2



BU Droit

1



BU Lettres

1



Résultat(s) 1 - 20 résultats de 41 pour la requête 'nature', Temps de recherche: 0,05s

Trier

Pertinence

Tout cocher | avec la sélection:



1



Nature

Macmillan Journals 1869-

La bibliothèque possède :

BU Sciences : Vol. 202 (1964) - Vol. 203 (1964) ; Vol. 206 (1965) - Vol. 257 (1975) ; Vol. 259 (1976) - Vol. 312 (1984) ; Vol. 322 (1986) - Vol. 565 (2019)

Egalement en ligne : En ligne [Via Nature](#)

Sommaires en ligne

Revue



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2



Nature

Nature

depuis 1869 jusqu'à 2012

L'accès à cette ressource est contrôlé.

Accès en ligne

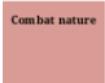
Revue En ligne

Titres liés



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3



Combat nature

Société d'édition pour la nature et l'environnement 1974-2004

La bibliothèque possède :



Mode d'emploi en ligne.
Attention pas Pub Med... Vous savez ce que vous cherchez.

2. Les analyses nucléotidiques

Question à étudier:

2. Les analyses nucléotidiques

Question à étudier:

Suite à un protocole de puce à ADN cet ARNm a été décrit comme étant différentiel dans des préparations de cœur issues d'un modèle d'insuffisance cardiaque par ligature de coronaire chez le rat. Il est en effet sous-exprimé. Déterminez la nature de ce messager.

tgcgtgctgcctccagccagcgaggcgtcagcgccgtgtcgcagcagtggaccgcggatggccta
ctcctggcgctatcgtagtggcaacgtgttgtatcggtggccatgccaagacccc
gcggctgcagacgtcaccaacaccttcatcatgtccctggccagcgccgatctggcatggactgt
gtgggccttcggggccaccattgtggtgtggccgtggagtagcggctcctttgtgagctgt
gacttcggtagacgtgtatgtgtacggccagcatcgagaccctgtgtcatgcctggaccgttc
tcgcacatcagctgccttcgtaccagagcctgtgacgcgcgcgcgcgcggccctgtgtgcac
agtgtggccatctccgcgtgggtgccttcgtccatcctcatgcactggtgccggccgagagcgac
gaagcgccgcgtacaacgacccaaagtgtcgatgtccatggccttcgtgtacccgggtgtccgcgaggccc
tgcgtccgttccttcgtggccctgtgcatcatggccttcgtgtacccgggtgtccgcgaggccc
agaaacaggtgaagaagatcgacagctgcgagcgccgttcctcacggccccggccgcgcctcg
ccgcgcgcctcgcc

2. Les analyses nucléotidiques

A service of the U.S. National Library of Medicine
and the National Institutes of Health
www.ncbi.nlm.nih.gov

All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books

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Limits Preview/Index History Clipboard Details

About Entrez Text Version Entrez PubMed Overview Help | FAQ Tutorials New/Noteworthy  E-Utilities

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Search terms may be [topics](#), [authors](#) or [journals](#).

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Does NIH fund your work?

Then your manuscript must be made available in PubMed Central

How?

If you publish in one of [these journals](#), they will take care of the whole process.

If you publish [anywhere else](#), deposit the manuscript in PubMed Central via [one of the options described at publicaccess.nih.gov](#).

Note: Other funding organizations, including [HHMI](#), [Wellcome Trust](#) and the [MRC](#) also require papers to be made freely available through PMC.

PubMed is a service of the [U.S. National Library of Medicine](#) that includes over 18 million citations from MEDLINE and other life science journals for biomedical articles back to the 1950s. PubMed includes links to full text articles and other related resources.

2. Les analyses nucléotidiques

The Entrez Nucleotide database is a collection of sequences from several sources, including GenBank, RefSeq, and PDB. The number of bases in these databases continues to grow at an exponential rate.

Human Genome

Explore [human genome resources](#) or browse the human genome sequence using the [Map Viewer](#).

Building the human genome

The Human Genome Reference DNA Sequence was completed in April 2003. The current version is listed as a build number on the [Genome View](#) page and includes an accompanying set of [statistics](#) and [release notes](#).

Homo sapiens (human) genome view [BLAST](#) search the human genome

Build 38.2 statistics [Switch to previous build](#)

Chromosome	Hits
2	10
3	30
17	109
22	2
Y	1
not placed	13

The chromosomal locations of several genes believed to be associated with the human BRCA1 gene implicated in breast cancer, highlighted using the Map Viewer query "BRCA1" (build 36).

2. Les analyses nucléotidiques

U.S. National Library of Medicine > NCBI National Center for Biotechnology Information Sign in to NCBI

BLAST® Home Recent Results Saved Strategies Help

Basic Local Alignment Search Tool

BLAST finds regions of similarity between biological sequences. The program compares nucleotide or protein sequences to sequence databases and calculates the statistical significance.

[Learn more](#)

NEWS

Using BLAST Well, How to Maximize Your Search Efforts: Webinar on October 3, 2018

In this webinar, the NCBI BLAST team lead will show you how to be more effective with BLAST.

Thu, 27 Sep 2018 11:00:00 EST [More BLAST news...](#)

Web BLAST

Nucleotide BLAST nucleotide ▶ nucleotide

blastx translated nucleotide ▶ protein

tblastn protein ▶ translated nucleotide

Protein BLAST

protein ▶ protein

BLAST Genomes

Enter organism common name, scientific name, or tax id
Use up and down arrows to choose an item from the autocomplete.

Human Mouse Rat Microbes **Search**

2. Les analyses nucléotidiques

BLAST Basic Local Alignment Search Tool

Home Recent Results Saved Strategies Help

NCBI/ BLAST/ blastn suite

blastn blastp blastx tblastn tbblastx

Enter Query Sequence

Enter accession number, gi, or FASTA sequence Clear

Query subrange From To

Or, upload file Parcourir...

Job Title Enter a descriptive title for your BLAST search

Blast 2 sequences

Choose Search Set

Database Human genomic + transcript Mouse genomic + transcript Others (nr etc.): Nucleotide collection (nr/nt)

Organism Optional Enter organism name or id—completions will be suggested
Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown.

Entrez Query Optional Enter an Entrez query to limit search

Program Selection

Optimize for Highly similar sequences (megablast) More dissimilar sequences (discontiguous megablast) Somewhat similar sequences (blastn)
Choose a BLAST algorithm

l'institut du thorax

2. Les analyses nucléotidiques

Sequence input area (text area with scroll bar) and search parameters:

From _____ To _____

Or, upload file

Job Title Enter a descriptive title for your BLAST search

Blast 2 sequences

Choose Search Set

Database: Human genomic + transcript Mouse genomic + transcript Others (nr etc.): **Nucleotide collection (nr/nt)**

Organism **Optional**: Enter organism name or id—completions will be suggested

Entrez Query **Optional**: Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown.

Program Selection

Optimize for: Highly similar sequences (megablast) More dissimilar sequences (discontiguous megablast) Somewhat similar sequences (blastn)
Choose a BLAST algorithm

BLAST

Search database nr using Megablast (Optimize for highly similar sequences)
 Show results in a new window

2. Les analyses nucléotidiques

Basic Local Alignment Search Tool

My NCBI [Sign In] [Register]

Home Recent Results Saved Strategies Help

NCBI/ BLAST/ blastn suite/ Formatting Results - HRDEGBVV01R

Edit and Resubmit Save Search Strategies > Formatting options >Download

Nucleotide Sequence (716 letters)

Query ID Icl|27220
Description None
Molecule type nucleic acid
Query Length 716

Database Name nr
Description All GenBank+EMBL+DDBJ+PDB sequences (but no EST, STS, GSS,environmental samples or phase 0, 1 or 2 HTGS sequences)
Program BLASTN 2.2.18+ >Citation

Other reports: >Search Summary [Taxonomy reports] [Distance tree of results]

▼ Graphic Summary

Distribution of 45 Blast Hits on the Query Sequence ⓘ

Mouse-over to show define and scores, click to show alignments

Color key for alignment scores

Score Range	Color
<40	Black
40-50	Blue
50-80	Green
80-200	Purple
>=200	Red

Query 0 100 200 300 400 500 600 700

2. Les analyses nucléotidiques



Descriptions

Legend for links to other resources: **U** UniGene **E** GEO **G** Gene **S** Structure **M** Map Viewer

Sequences producing significant alignments: (Click headers to sort columns)							
Accession	Description	Max score	Total score	Query coverage	E value	Max ident	Links
NM_012701.1	Rattus norvegicus adrenergic, beta-1-, receptor (Adrb1), mRNA	1317	1317	100%	0.0	99%	UEG
J05562.1	R.norvegicus beta-1-adrenergic receptor gene, complete cds	1317	1317	100%	0.0	99%	G
D00634.1	Rattus norvegicus gene for beta-1 adrenergic receptor, complete cds	1293	1293	100%	0.0	99%	EG
BC147435.1	Mus musculus adrenergic receptor, beta 1, mRNA (cDNA clone MGC:182135 IMAC	1157	1157	100%	0.0	95%	UG
NM_007419.2	Mus musculus adrenergic receptor, beta 1 (Adrb1), mRNA	1157	1157	100%	0.0	95%	UEG
AC158549.8	Mus musculus chromosome 19, clone RP24-216K4, complete sequence	1157	1157	100%	0.0	95%	
L10084.1	Mus musculus beta-1 adrenergic receptor gene, complete cds	1157	1157	100%	0.0	95%	EG
X75540.1	M.mullata beta 1 adrenergic receptor gene	1057	1057	100%	0.0	93%	
AK018378.1	Mus musculus 16 days embryo lung cDNA, RIKEN full-length enriched library, clon	1037	1037	91%	0.0	95%	UEG
AF169006.1	Homo sapiens beta-1-adrenergic receptor (ADRB1) gene, complete cds	1029	1029	100%	0.0	92%	G
NM_001009375.1	Felis catus adrenergic, beta-1-, receptor (ADRB1), mRNA >gb AF192344.1 AF192	1029	1029	100%	0.0	92%	G
EU332832.1	Homo sapiens adrenergic, beta-1-, receptor (ADRB1) gene, complete cds	1024	1024	100%	0.0	92%	
XM_521608.2	PREDICTED: Pan troglodytes beta-1-adrenergic receptor (ADRB1), mRNA	1024	1024	100%	0.0	92%	G
NM_000684.2	Homo sapiens adrenergic, beta-1-, receptor (ADRB1), mRNA	1024	1024	100%	0.0	92%	UEG
AY567837.1	Homo sapiens adrenergic, beta-1-, receptor (ADRB1) gene, complete cds	1024	1024	100%	0.0	92%	
AF169007.1	Homo sapiens beta-1-adrenergic receptor (ADRB1) gene, complete cds	1024	1024	100%	0.0	92%	G
AL355543.13	Human DNA sequence from clone RP11-86E10 on chromosome 10 Contains the A	1024	1024	100%	0.0	92%	
J03019.1	Human beta-1-adrenergic receptor mRNA, complete cds	1024	1024	100%	0.0	92%	UEG
AC005886.2	b240g16, complete sequence	1018	1018	100%	0.0	92%	
AB334518.1	Sus scrofa ADRB1 gene for beta-1 adrenergic receptor, complete cds, breed: Land	1013	1013	100%	0.0	92%	G
AB334517.1	Sus scrofa ADRB1 gene for beta-1 adrenergic receptor, complete cds, breed: Jinh	1009	1009	100%	0.0	92%	G
NM_001123074.1	Sus scrofa adrenergic, beta-1-, receptor (ADRB1), mRNA	1003	1003	100%	0.0	92%	UG
AF042454.1	Sus scrofa beta-1 adrenergic receptor gene, complete cds	1003	1003	100%	0.0	92%	G
NM_001008713.1	Canis lupus familiaris adrenergic, beta-1-, receptor (ADRB1), mRNA	979	979	100%	0.0	91%	UG
U73207.1	Canis familiaris beta1 adrenergic receptor (dogbeta1) gene, complete cds	979	979	100%	0.0	91%	EG
EU332753.1	Cavia porcellus beta-1 adrenergic receptor (ADRB1) gene, complete cds	935	935	99%	0.0	90%	
DQ538524.1	Bos taurus beta 1 adrenergic receptor gene, complete cds	907	907	100%	0.0	89%	G
	Ovis aries beta-1 adrenergic receptor (ADRB1) gene, complete cds	906	906	100%	0.0	90%	

2. Les analyses nucléotidiques

M14379_1	Turkey beta-adrenergic receptor mRNA, complete cds	501	501	89%	2e-138	81%	U
AF055349_1	Meriones unguiculatus beta-1-adrenergic receptor mRNA, partial cds	364	364	30%	2e-97	96%	
BC169226_1	Homo sapiens cDNA clone IMAGE:9093418, partial cds	261	261	24%	3e-66	93%	
AF041457_1	Cervus dama beta 1 adrenergic receptor mRNA, partial cds	228	228	20%	3e-56	95%	
U51098_1	Cavia porcellus beta3-adrenergic receptor mRNA, partial cds	132	132	52%	3e-27	74%	G

▼ Alignments Select All [Get selected sequences](#) [Distance tree of results](#)

[ref|NM_012701.1] **UEG** Rattus norvegicus adrenergic, beta-1-, receptor (Adrb1), mRNA
Length=1401

GENE ID: 24925 Adrb1 | adrenergic, beta-1-, receptor [Rattus norvegicus]
(Over 10 PubMed links)

Score = 1317 bits (713), Expect = 0.0
Identities = 715/716 (99%), Gaps = 0/716 (0%)
Strand=Plus/Plus

Query	1	TCGCTGCTGCCCTCAGCCAGCGAGGGCTCAGGCCCGCTGTGCAGCAGTGGACCGCGGGT	60
Sbjct	121	TGGCTGCTGCCCTCAGCCAGGGCTCAGGCCCGCTGTGCCAGCAGTGGACCGCGGGT	180
Query	61	ATGGGCCTACTCCCTGGCGCTATCGTGCTCATCGTAGTGGCAACGTGTTGGTGTATC	120
Sbjct	181	ATGGGCCTACTCCCTGGCGCTATCGTGCTCATCGTAGTGGCAACGTGTTGGTGTATC	240
Query	121	GTGGCCATCGCAAGACCCCCCGGCTGCAGACGCTCACCAACCTCTTCATGTCCCTG	180
Sbjct	241	GTGGCCATCGCAAGACCCCCGGCTGCAGACGCTCACCAACCTCTTCATGTCCCTG	300
Query	181	GCCACCGCCGATCTGGTACGGGACTGCTGGTGGTGCTTCCGGGCCACCATTTGGTG	240
Sbjct	301	GCCACCGCCGATCTGGTACGGGACTGCTGGTGGTGCTTCCGGGCCACCATTTGGTG	360
Query	241	TGGGGCCGCTGGAGTACGGCTCCCTCTTGAGCTCTGGACTTCGGTAGACGTGCTA	300
Sbjct	361	TGGGGCCGCTGGAGTACGGCTCCCTCTTGAGCTCTGGACTTCGGTAGACGTGCTA	420
Query	301	TGTGTGACGGCCAGCATCGAGACCCCTGTGTGTCATGCCCTGGACGGCTCCCTGATC	360
Sbjct	421	TGTGTGACGGCCAGCATCGAGACCCCTGTGTGTCATGCCCTGGACGGCTACCTCGATC	480
Query	361	ACGCTGCCCTTCGCTACCAAGACCTGCTGACCGCGCCGGAGGCCGGCCCTCGTGTG	420

Position 470/début séquence référencée

2. Les analyses nucléotidiques

NCBI Nucleotide

PubMed Nucleotide Protein

Search Nucleotide for Go Clear

Display GenBank Show 20 Send to Hide: sequence all but gene, CDS and mRNA features

Range: from begin to end Reverse complemented strand Features: STS Refresh

1: NM_012701. Reports Rattus norvegicus...[gi:6978458]

Comment Features Sequence

LOCUS NM_012701 1401 bp mRNA linear ROD 10-OCT-2008

DEFINITION Rattus norvegicus adrenergic, beta-1-, receptor (Adrb1), mRNA.

ACCESSION NM_012701 XM_001063787

VERSION NM_012701.1 GI:6978458

KEYWORDS .

SOURCE Rattus norvegicus (Norway rat)

ORGANISM Rattus norvegicus

Lineage: Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Glires; Rodentia; Sciurognathii; Muroidea; Muridae; Murinae; Rattus.

REFERENCE 1 (bases 1 to 1401)

AUTHORS Bidasee,K.R., Zheng,H., Shao,C.H., Parbhoo,S.K., Rozanski,G.J. and Patel,K.P.

TITLE Exercise training initiated after the onset of diabetes preserves myocardial function: effects on expression of beta-adrenoceptors

JOURNAL J. Appl. Physiol. 105 (3), 907-914 (2008)

PUBMED 18583384

REMARK GeneRIF: Myocardial Adrb1 reduction in type 1 diabetes is prevented using exercise training.

REFERENCE 2 (bases 1 to 1401)

AUTHORS Fu,A., Li,X. and Zhao,B.

TITLE Role of betal-adrenoceptor in the basolateral amygdala of rats with anxiety-like behavior

JOURNAL Brain Res. 1211, 85-92 (2008)

PUBMED 18423428

REMARK GeneRIF: results suggested that the betal-AR played an important role in anxiety-like behavior

REFERENCE 3 (bases 1 to 1401)

AUTHORS Abraham,P.A., Xing,G., Zhang,L., Yu,E.Z., Post,R., Gamble,E.H. and Li,H.

TITLE betal- and beta2-adrenoceptor induced synaptic facilitation in rat basolateral amygdala

JOURNAL Brain Res. 1209, 65-73 (2008)

PUBMED 18396264

REMARK GeneRIF: These data suggest that beta-adrenoceptor mediated synaptic facilitation in the amygdala is mediated by both betal and beta2-adrenoceptor activation.

REFERENCE 4 (bases 1 to 1401)

AUTHORS Plante,E., Lachance,D., Champetier,S., Drolet,M.C., Roussel,E., Arsenault,M. and Couet,J.

TITLE Benefits of long-term beta-blockade in experimental chronic aortic regurgitation

JOURNAL Am. J. Physiol. Heart Circ. Physiol. 294 (4), H1888-H1895 (2008)

PUBMED 18296565

REMARK GeneRIF: Long-term beta-blockade in chronic aortic regurgitation improved heart function and restored betal/beta2 adrenergic receptor

Identité

Bibliographie

2. Les analyses nucléotidiques

FEATURES	Location/Qualifiers
source	1..1401 /organism="Rattus norvegicus" /mol_type="mRNA" /db_xref="taxon:10116" /chromosome="1" /map="1q55"
gene	1..1401 /gene="Adrb1" /gene_synonym="BLAR" /gene_synonym="RATBLAR" /note="adrenergic, beta-1-, receptor" /db_xref="GeneID:24925" /db_xref="RATMAP:34960" /db_xref="RGD:2059"
CDS	1..1401 /gene="Adrb1" /gene_synonym="BLAR" /gene_synonym="RATBLAR" /note="beta 1-adrenergic receptor beta 1-AR; adrenergic receptor, beta 1" /codon_start=1 /product="adrenergic, beta-1-, receptor" /protein_id="NP_036833.1" /db_xref="GI:6978459" /db_xref="GeneID:24925" /db_xref="RATMAP:34960" /db_xref="RGD:2059" /translation="MGAGALALGASEPCNLSSAAPLPDGAATAARLLVLASPPASLLPASEGSAPLSQQWTAGMGLLALLVLLIVVGNVLVIVAIAKTPRLTQLTNLFIMSLAADLVMGLLVVPFGATIVVWGRWEYGSFFCELWTSVDLCVTSIETLCVIALDRYLALTLPRYQSLTRARALARLVTVAISALVSFLPILMHWWRAESDEARRCYNDPKCDCVTNRAYIAASSVVSFYPLCIMAFLVRFREAQKQVKKIDSERRFLTGPPRPPSPVPSPPGPPRPAIDLNGRSSKRRPSRLVALREQKALKTLGIIMGVFTLCWLPPFLANTVKAFHRDLVPDRLFVFWLGYANSAFNPIIYCRSPDFRKAFQRLLCARRAACRRRAHGDPRPASGCLARAGPPSPGAPSDDDDDAAGATPPARLLEPWAGCNGGTTVDSDSLDEPGHQGFSSESKV"
STS	274..795 /gene="Adrb1" /gene_synonym="BLAR" /gene_synonym="RATBLAR" /standard_name="Adrb1" /db_xref="UniSTS:256461"
STS	1252..1351 /gene="Adrb1" /gene_synonym="BLAR" /gene_synonym="RATBLAR" /standard_name="Adrb1" /db_xref="UniSTS:141043"
ORIGIN	1 atgggcggcg gggcgtcgcc cttggggccc tccgaaacct gcaacactgtc gtggccggcg 61 ccgtcgcccg acggcggggc caccggggca cgactgtctgg tgctcgccgtc gcttcccgcc 121 tcgtctgtcc ctccagccag cgagggtctca gggccgtgtt cgacggatgtc gaccgggggt 181 atgggcctac tccctgggtt catcggtgtt ctatcgatgt tggccaaactt gtgtgtgtatc 241 gtggccatcc ccaagacggcc ggggttcgtac acgttcacca acctttcatatc catgtccctgt 301 gccaaggcccg atctgggtatc gggactgtgtc gtgtgtccgtt tggggccac catttgtgtgt 361 tggggccgtt gggaggtatgg ctccttttgc tttgtgtgtt ggacttgggtt agacgtgtata 421 ttgtgtgtaccc ccagcatgtca gaccgtgtgtt gttatcgccgg tggacgtgtt ctctggccatc 481 acgtgtgcctt ttctgtatcc gagectgtgtc acgegtgcgtc gagegtggccgtt ctgtgtgtatc 541 acatgtgtggg ccatctccgc gttgtgttgc ttctgtatccca ttctgtatccca ttgtgtgtatc

Phase ouverte de lecture

Protéine

Défaut en position
470/début
séquence
référencée



2. Les analyses nucléotidiques

Question à étudier:

Mutation silencieuse, faux-sens ou non-sens chez mon patient ?

2. Les analyses nucléotidiques

Question à étudier:

Mutation silencieuse, faux-sens ou non-sens chez mon patient ?

Basic BLAST

Choose a BLAST program to run.

[nucleotide blast](#)

Search a **nucleotide** database using a **nucleotide** query
Algorithms: blastn, megablast, discontiguous megablast

[protein blast](#)

Search **protein** database using a **protein** query
Algorithms: blastp, psi-blast, phi-blast

[blastx](#)

Search **protein** database using a **translated nucleotide** query

[tblastn](#)

Search **translated nucleotide** database using a **protein** query

[tblastx](#)

Search **translated nucleotide** database using a **translated nucleotide** query

2. Les analyses nucléotidiques

BLAST Basic Local Alignment Search Tool

NCBI/ BLAST/ tblastx

blastn blastp blastx tblastn

tblastx TBLASTX search translated nucleotide database using a translated nucleotide query

Enter Query Sequence

Enter accession number, gi, or FASTA sequence Clear

Query subrange From To

Or, upload file

Genetic code

Job Title

Enter a descriptive title for your BLAST search

Blast 2 sequences

Choose Search Set

Database Nucleotide collection (nr/nt)

Organism Enter organism name or id--completions will be suggested

Entrez Query Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown.

Optional Enter an Entrez query to limit search

BLAST Search database nr using Tblastx (Search translated nucleotide database using a translated nucleotide query)

Show results in a new window

2. Les analyses nucléotidiques

BLAST Basic Local Alignment Search Tool My NCBI [Sign in] [Register]

NCBI/ BLAST/ tblastx/ Formatting Results - HRFE3ZC7015

Edit and Resubmit Save Search Strategies ►Formatting options ►Download

Nucleotide Sequence (716 letters)

Query ID Icl|22459
Description None
Molecule type nucleic acid
Query Length 716

Database Name nr
Description All GenBank+EMBL+DDBJ+PDB sequences (but no EST, STS, GSS, environmental samples or phase 0, 1 or 2 HTGS sequences)
Program TBLASTX 2.2.18+ ►Citation

Other reports: ►Search Summary [Taxonomy reports]

▼ Graphic Summary

Distribution of 1000 Blast Hits on the Query Sequence ⓘ
Mouse-over to show details and scores, click to show alignments

Color key for alignment scores

Query	<40	40-50	50-80	80-200	>=200		
0	100	200	300	400	500	600	700

►

2. Les analyses nucléotidiques

▼ Alignments Select All [Get selected sequences](#) [Distance tree of results](#)

> [gb|J05561.1|RATB1AR](#) G R.norvegicus beta-1-adrenergic receptor gene, complete cds
Length=1645

GENE ID: 24925 Adrb1 | adrenergic, beta-1-, receptor [Rattus norvegicus]
[\(Over 10 PubMed links\)](#)

Score = 439 bits (952), Expect = 6e-154
Identities = 177/178 (99%), Positives = 177/178 (99%), Gaps = 0/178 (0%)
Frame = -3/-3

Query	534	EIAALGVVVAAARFVALGPPPVEDGQEGHQRGDGPHCAHEGPRSRARQQALVAKGQRDG	355
		EIAALGVVVAAARFVALGPPPVEDGQEGHQRGDGPHCAHEGPRSRARQQALVAKGQRDG	
Sbjct	722	EIAALGVVVAAARFVALGPPPVEDGQEGHQRGDGPHCAHEGPRSRARQQALVAKGQRDG	543
Query	354	EAVVQGDDTQGLDAGRHT*HVYRSPELTEEGAVLPAAAPHNNGPERHHQQSHDQIGAGQQ	175
		E AVQGDDTQGLDAGRHT*HVYRSPELTEEGAVLPAAAPHNNGPERHHQQSHDQIGAGQQ	
Sbjct	542	EAVVQGDDTQGLDAGRHT*HVYRSPELTEEGAVLPAAAPHNNGPERHHQQSHDQIGAGQQ	363
Query	174	HDEEVGERLQPRGLGDGHDHQHVAHYDEQHDERQE*AHTRGPLLRQRR*ALAGWRQQR	1
		HDEEVGERLQPRGLGDGHDHQHVAHYDEQHDERQE*AHTRGPLLRQRR*ALAGWRQQR	
Sbjct	362	HDEEVGERLQPRGLGDGHDHQHVAHYDEQHDERQE*AHTRGPLLRQRR*ALAGWRQQR	189

2. Les analyses nucléotidiques

Legend for links to other resources:  UniGene  GEO  Gene  Structure  Map Viewer

Position sur le génome ???

2. Les analyses nucléotidiques

Legend for links to other resources: [U UniGene](#) [E GEO](#) [G Gene](#) [S Structure](#) [M Map Viewer](#)

Position sur le génome ???

NCBI Entrez Gene

All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books

Search Gene for 6978458[NUID] Go Clear Save Search

Limits Preview/Index History Clipboard Details

Display Full Report Show 20 Sort by Relevance Send to

All: 1 Current Only: 1 Genes Genomes: 1 SNP GeneView: 0

1: Adrb1 adrenergic, beta-1-, receptor [Rattus norvegicus]

GeneID: 24925 updated 15-Oct-2008

Summary

Official Symbol Adrb1 provided by RGD

Official Full Name adrenergic, beta-1-, receptor provided by RGD

Primary source RGD:2059

See related Ensembl:ENSRNOG00000017002; RATMAP:34960

Gene type protein coding

RefSeq status PROVISIONAL

Organism [Rattus norvegicus](#)

Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Glires; Rodentia; Sciurognathi; Muroidea; Muridae; Murinae; Rattus

Also known as B1AR; RATB1AR; Adrb1

Summary binds beta-adrenergic receptor agonists isoproterenol, norepinephrine, and epinephrine; mediates adenylylcyclase induction; involved in regulation of calcium current [RGD]

Genomic regions, transcripts, and products

Go to reference sequence details Try our new Sequence Viewer

NC_005100.2

The diagram shows a horizontal line representing the genomic region NC_005100.2. Two arrows at the ends indicate the direction of transcription. The left arrow points to a blue box labeled 'NM_012791.1' and '5'-. The right arrow points to a red box labeled 'NP_036833.1' and '3+'. Below the line, a legend indicates that a red box represents the 'coding region' and a blue box represents the 'untranslated region'.

Links

- Conserved Domains
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- PubChem Compound
- PubChem Substance
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- Taxonomy
- UniSTS
- Ensembl
- Evidence Viewer
- KEGG
- ModelMaker
- RATMAP
- RGD
- UniGene
- LinkOut

Entrez Gene Info

2. Les analyses nucléotidiques

Legend for links to other resources:  UniGene  GEO  Gene  Structure  Map Viewer

Position sur le génome ???

Genomic regions, transcripts, and products



Genomic Sequence NC_000010 chromosome 10 reference GRCh37.p10 Primary Assembly

Go to [reference sequence details](#)

[Go to nucleotide](#) [Graphics](#) [FASTA](#) [GenBank](#)



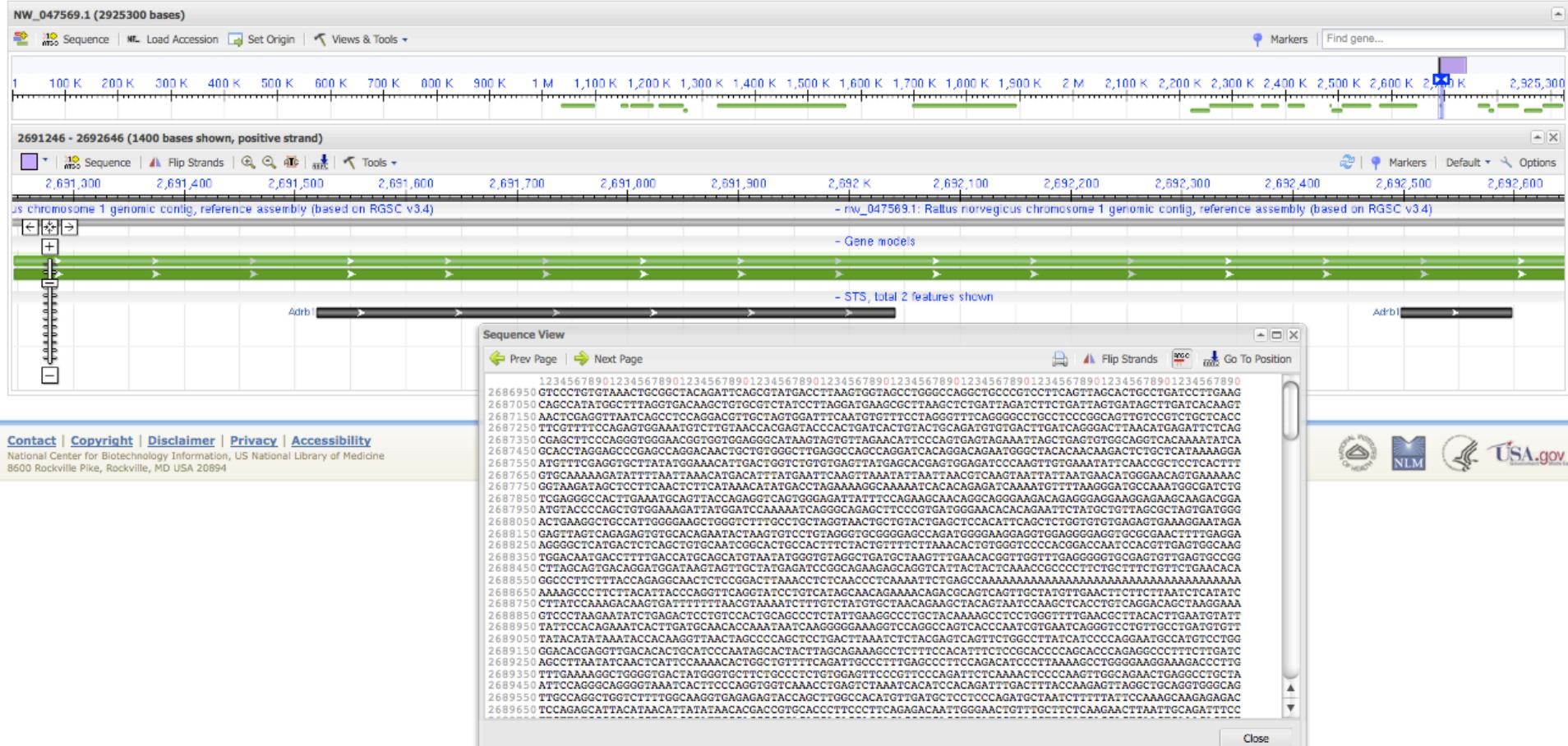
Exons, introns, SNP, etc...

2. Les analyses nucléotidiques

Rattus norvegicus chromosome 1 genomic contig, reference assembly (based on RGSC v3.4)
gi|34864585|refNW_047569.1|Rn1_WGA2091_4

Position sur le génome ???

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2. Les analyses nucléotidiques

Legend for links to other resources:  UniGene  GEO  Gene  Structure  Map Viewer

NCBI Map Viewer

PubMed Entrez BLAST OMIM

Search Find Find in This View

Rattus norvegicus (rat) RGSC v3.4

Chromosome: [1] [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) X Y MT

Query: 24925[[gene_id](#)] [[clear](#)]

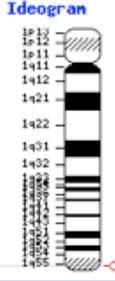
Master Map: Genes On Sequence Summary of Maps

Region Displayed: 263,025,480-263,027,230 bp

Hs	UniG	Model	Rn	UniG	ensGenes	RefSeq	RNA	Genes_seq	Symbol	Links	Cyto	Description
	Hs.591914	Hs.21631		Rn.87064				NM_012781	Adrb1			

You are here:

Ideogram



default master

La carte chromosomique...

Adrb1 + [svprdllevmmhmrgdsts](#) 1q55 best RefSeq adrenergic receptor, beta 1

l'ins du th

3. Les pathologies héréditaires

3. Les pathologies héréditaires

NCBI

All Databases PubMed Nucleotide Pro

Search OMIM for beta drenergic receptor Go Clear

Limits Preview/Index History Clipboard Details

About Entrez
Entrez Nucleotide
Help | FAQ
Entrez Tools
Check sequence revision history
LinkOut
My NCBI (Cubby)
Related resources
BLAST
Reference sequence project
Search for Genes
Submit to GenBank
Search for full length cDNAs

The Entrez Nucleotide database is a collection of sequences from several sources, including GenBank, RefSeq, and PDB. The number of bases in these databases continues to grow at an exponential rate.

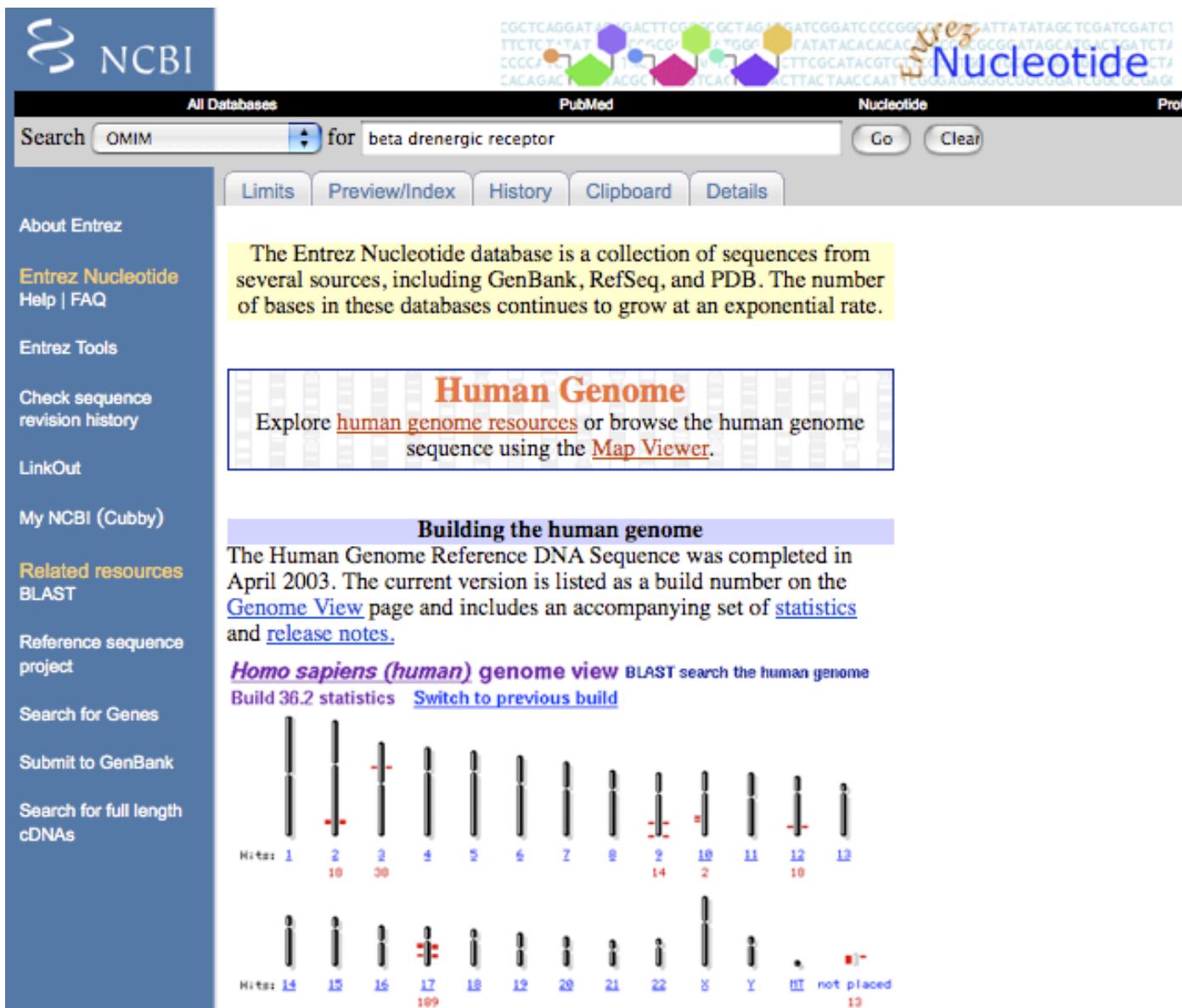
Human Genome
Explore [human genome resources](#) or browse the human genome sequence using the [Map Viewer](#).

Building the human genome
The Human Genome Reference DNA Sequence was completed in April 2003. The current version is listed as a build number on the [Genome View](#) page and includes an accompanying set of [statistics](#) and [release notes](#).

Homo sapiens (human) genome view BLAST search the human genome
Build 36.2 statistics [Switch to previous build](#)

Hits: 1 2 3 4 5 6 7 8 9 10 11 12 13

Hits: 14 15 16 17 18 19 20 21 22 23 Y M not placed 13



3. Les pathologies héréditaires

NCBI

OMIM
Online Mendelian Inheritance in Man

Johns Hopkins University

All Databases PubMed Nucleotide Protein Genome

Search OMIM for beta drenergetic receptor Go Clear Save Search

Limits Preview/Index History Clipboard Details

Entrez

The following term was not found: drenergetic.
See [Details](#).

Did you mean: [beta adrenergic receptor](#) (99 items)

Display Titles Show 20 Send to

All: 1464 OMIM UniSTS: 190 OMIM dbSNP: 227

Items 1 - 20 of 1464

1: [+141900](#)
HEMOGLOBIN--BETA LOCUS; HBB
BETA-THALASSEMIAS, INCLUDED
Gene map locus [11p15.5](#)

2: [+109690](#)
BETA-2-ADRENERGIC RECEPTOR; ADRB2
BETA-2-ADRENORECEPTOR AGONIST, REDUCED RESPONSE TO, INCLUDED
Gene map locus [5q32-q34](#)

3: [*608886](#)
PEROXISOME PROLIFERATOR-ACTIVATED RECEPTOR-GAMMA, COACTIVATOR 1, BETA; PPARGC1B
Gene map locus [5q33](#)

4: [*104760](#)
AMYLOID BETA A4 PRECURSOR PROTEIN; APP
Gene map locus [21q21](#)

L'onglet OMIM est un outils de synthèse

3. Les pathologies héréditaires

NCBI

OMIM
Online Mendelian Inheritance in Man

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All Databases PubMed Nucleotide Protein Genome Structure PMC OMIM

Search OMIM for Go Clear

Limits Preview/Index History Clipboard Details

Display Detailed Show 20 Send to

*109691

BETA-3-ADRENERGIC RECEPTOR; ADRB3

Gene map locus [8p12-p11.2](#)

TEXT

CLONING

[Emorine et al. \(1989\)](#) isolated a third beta-adrenergic receptor, beta-3-adrenergic receptor (ADRB3). (See ADRB1 ([109630](#)) and ADRB2 ([109690](#)).) Exposure of eukaryotic cells transfected with this gene to adrenaline or noradrenaline promoted the accumulation of adenosine 3-prime,5-prime-monophosphate. The potency of beta-AR agonists and inhibitors was described. ☺

[Van Spronsen et al. \(1993\)](#) demonstrated that the transcription start sites of the mouse and human ADRB3 mRNA are located in a region comprised between 150 and 200 nucleotides 5-prime from the ATG translation start codon. Motifs potentially implicated in heterologous regulation of ADRB3 expression by glucocorticoids and by beta-adrenergic agonists were identified upstream from these cap sites. ☺

GENE STRUCTURE

[Van Spronsen et al. \(1993\)](#) described the exon/intron structure of the mouse and human ADRB3 genes. Their results suggested that utilization of alternate promoters and/or 3-prime untranslated regions may allow tissue-specific regulation of the expression of ADRB3.

MAPPING

[Wilkie et al. \(1993\)](#) presented a list of G protein-coupled receptor genes (their Table 3), indicating that the ADRB3 gene had been mapped to 8p12-p11.2 and the homologous gene to mouse chromosome 8.

MOLECULAR GENETICS

The beta-3-adrenergic receptor, located mainly in adipose tissue, is involved in the regulation of lipolysis and thermogenesis. The potential relevance of this receptor to obesity (see [501665](#)) in humans led [Clement et al. \(1995\)](#) to screen obese patients for the mutation in the ADRB3 gene that results in replacement of tryptophan by arginine at position 64 (W64R; [109691_0001](#)). They studied DNA extracted from leukocytes of 94 normal subjects and 185 unrelated patients with morbid obesity, as defined by a body-mass index (BMI; the weight in kilograms divided by the square of the height in meters) greater than 40. The mutation was detected by analysis of RFLPs with the restriction enzyme BstNI, which discriminates between the normal and mutant sequences. The frequency of the W64R variant was similar in the morbidly obese patients and the normal subjects: 0.08 and 0.10, respectively. However, patients with morbid obesity who were heterozygous for the allele had an increased capacity to gain weight: the mean weight in the 14 heterozygous patients was 140 kg, as compared with 126 kg in the 171 patients without the mutation ($P = 0.03$). There were no homozygotes in this sample. The cumulative 25-year change in weight (from the age of 20 years) was 67 kg in W64R heterozygotes, as compared with 51 kg in those without the mutation. The maximum weight differential (the maximal lifetime weight minus the weight at 20 years of age) in the heterozygotes was 74 kg, as compared with 59 kg in the patients without the mutation ($P = 0.02$). [Clement et al. \(1995\)](#) interpreted the findings as indicating that the ADRB3 gene mutation W64R increases the capacity to gain weight. ☺

ANIMAL MODEL

To determine whether the sympathetic nervous system is the efferent arm of diet-induced thermogenesis, [Bachman et al. \(2002\)](#) created mice that lacked the beta-adrenergic receptors ADRB1, ADRB2, and ADRB3. Beta-less mice on a chow

3. Les pathologies héréditaires

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ANIMAL MODEL

To determine whether the sympathetic nervous system is the efferent arm of diet-induced thermogenesis, [Bachman et al. \(2002\)](#) created mice that lacked the beta-adrenergic receptors ADRB1, ADRB2, and ADRB3. Beta-less mice on a chow diet had a reduced metabolic rate and were slightly obese. On a high-fat diet, beta-less mice, in contrast to wildtype mice, developed massive obesity that was due entirely to a failure of diet-induced thermogenesis. [Bachman et al. \(2002\)](#) concluded that the beta-adrenergic receptors are necessary for diet-induced thermogenesis and that this efferent pathway plays a critical role in the body's defense against diet-induced obesity. [?]

ALLELIC VARIANTS (selected examples)

.0001 OBESITY, SUSCEPTIBILITY TO [ADRB3, TRP64ARG]

Using a candidate gene approach to study the genetics of obesity ([601665](#)), [Clement et al. \(1995\)](#) found evidence suggesting that the trp64-to-arg (W64R) variant of the ADRB3 gene increases the capacity to gain weight. [Gagnon et al. \(1996\)](#) failed to find an association between W64R and obesity in studies in 2 cohorts: the Quebec Family Study (QFS) and the Swedish Obese Subjects (SOS). [?]

[Walston et al. \(1995\)](#) found that Pima Indians homozygous for the W64R ADRB3 mutation had an earlier onset of noninsulin-dependent diabetes mellitus (NIDDM; [125853](#)) and tended to have a lower resting metabolic rate. The authors suggested that the mutation may accelerate the onset of NIDDM by altering the balance of energy metabolism in visceral adipose tissue. [?]

[Elbein et al. \(1996\)](#) tested the hypothesis that the beta-3-adrenergic receptor locus affects diabetes susceptibility, obesity as measured by body mass index (BMI), and components of the insulin ([176730](#)) resistance syndrome, by examining ADRB3 allele sharing in families ascertained for 2 or more sibs with NIDDM. They found no evidence for linkage to NIDDM as a dichotomous trait and no evidence for linkage to BMI, waist/hip ratio, insulin levels, or glucose levels as quantitative traits or to reported age of onset among NIDDM individuals. The W64R mutation present in 11% of the population also did not show linkage or association. They concluded that the beta-3-adrenergic receptor locus does not play an important role in NIDDM susceptibility or in the insulin resistance syndrome among members of families with a strong predisposition to NIDDM. [?]

[Kim-Motoyama et al. \(1997\)](#) examined the frequency of the W64R variant in 278 Japanese men in relation to visceral obesity assessed by computerized tomography. They found that the mutation was more frequent in subjects with higher BMI. In subjects with a moderate degree of obesity, the mutation (homozygotes and heterozygotes) was associated with visceral obesity (higher ratio of visceral to subcutaneous fat area). Furthermore, the W64R variant was more frequent in subjects with lower serum triglyceride levels, and homozygotes, but not heterozygotes, exhibited lower triglyceride levels. [Kim-Motoyama et al. \(1997\)](#) suggested that the mutation may describe a subset of subjects characterized by decreased lipolysis in visceral adipose tissue. [?]

To examine the effect of W64R on body weight during adult life, the ADRB3 genotypes of 186 unselected Japanese men, most of whom had records of body weight measured yearly from 25 to 53 years of age, were determined by [Nagase et al. \(1997\)](#). Of these subjects, 26 were diagnosed as having noninsulin-dependent diabetes mellitus (NIDDM) and 41 as having impaired glucose tolerance. The results suggested that ADRB3 is not a major contributing factor to obesity or NIDDM in Japanese men. [?]

[Buechner et al. \(1998\)](#) examined the prevalence of the 2 ADRB3 alleles in Germany and looked for associations between the ADRB3 genotype and obesity and NIDDM. The frequencies of the different genotypes in the examined cohort were as follows: trp64/trp64, 88.3%; trp64/arg64, 10.8%; and arg64/arg64, 0.8%. The authors found no significant differences between the different genotypes when comparing age, BMI, weight, total and high density lipoprotein, cholesterol, fasting insulin, HbA1c, and blood pressure. They concluded that the NIDDM phenotype did not differ significantly between the different genotype groups in terms of age of diabetes onset or HbA1c. [?]

Using hyperinsulinemic/euglycemic clamp methodology, [Garcia-Rubi et al. \(1998\)](#) measured insulin sensitivity in 13 obese women heterozygous for the W64R ADRB3 variant and in 14 women homozygous for the normal gene. Exogenous glucose infusion during the clamp was significantly lower ($P = 0.03$) in W64R heterozygotes (241 ± 135 mg/min) compared with normal homozygotes (379 ± 172 mg/min). They concluded that obese postmenopausal women who are heterozygous for the W64R variant have greater insulin resistance than women homozygous for the normal gene matched for age, body composition, and physical activity. [?]

[Mitchell et al. \(1998\)](#) detected an effect of the W64R variant on obesity in a Mexican-American population. They had previously identified a major quantitative trait locus (QTL) influencing the serum concentrations of leptin on 2p in a Mexican-American population in south Texas ([Comuzzie et al., 1997](#)). They studied 45 sib pairs who were concordant (identical by descent) for this locus on chromosome 2, which had been shown previously to be tightly linked to obesity in this population. The W64R variant, detected by PCR-RFLP analysis, was present in 1 sib within each of the 45 sib pairs. Presence of the variant was associated with a significantly higher values in body mass index, fat mass, and waist circumference. The paired-sib design enhanced their ability to detect the effects of this variant by allowing them to account for variation attributable to another obesity susceptibility locus and to background genes. [?]



MIM *109691

Cloning

Gene Structure

Mapping

Molecular Genetics

Animal Model

Allergic Variants

View List

References

Contributors

Creation Date

Edit History

Gene map

Entrez Gene

Nomenclature

RefSeq

GenBank

Protein

UniGene

LinkOut

HGVS

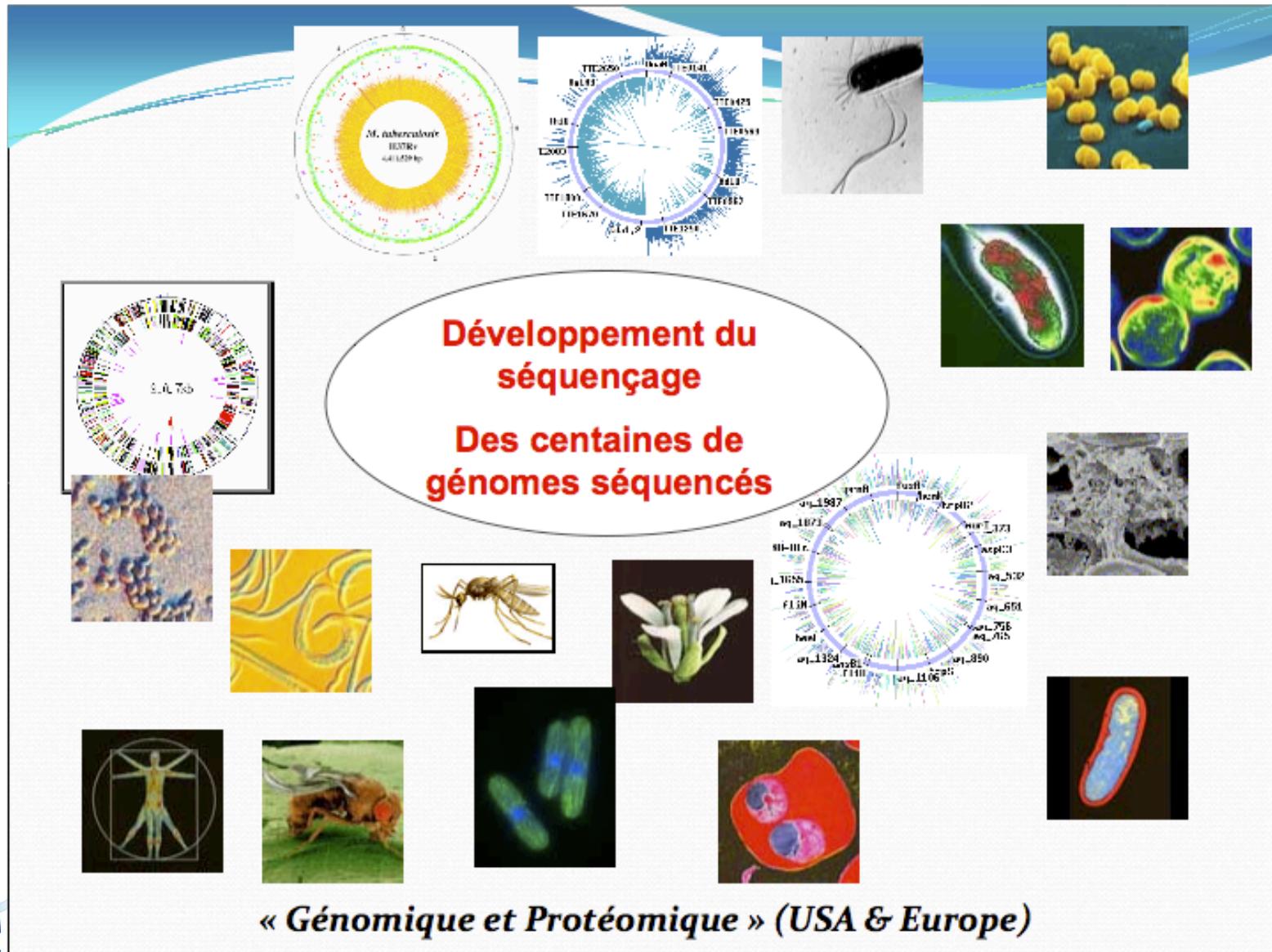
HGMD

GAD

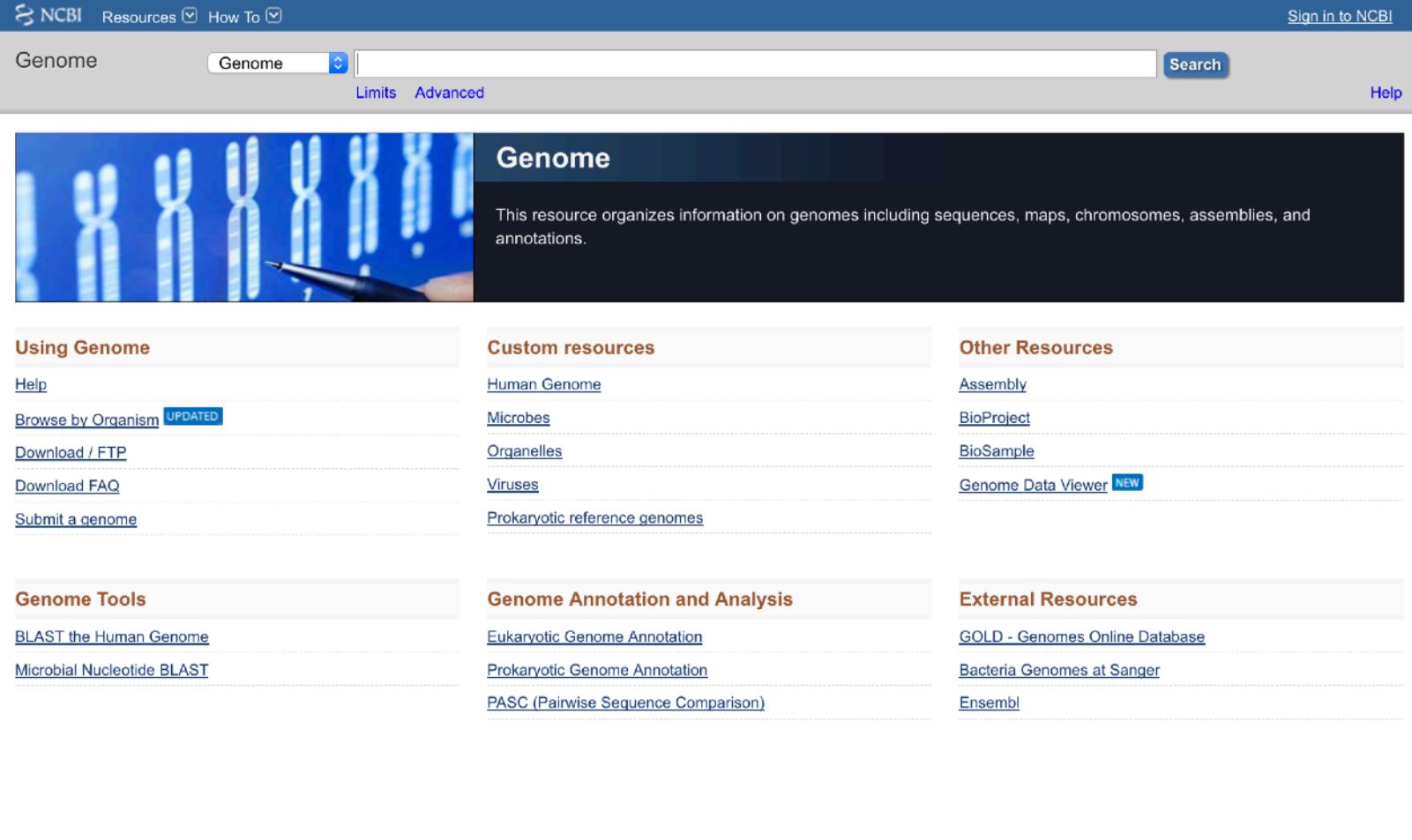
MGI

4. Les analyses génomiques

4. Les analyses génomiques



4. Les analyses génomiques



The screenshot shows the NCBI Genome homepage. At the top, there's a navigation bar with links for "Resources" and "How To". On the right, there's a "Sign in to NCBI" link. Below the navigation, the word "Genome" is repeated in a dropdown menu and a search bar. Underneath the search bar are "Limits" and "Advanced" buttons. A "Help" link is also present. The main content area features a large image of chromosomes with a pen pointing at one. The title "Genome" is displayed above a text block that reads: "This resource organizes information on genomes including sequences, maps, chromosomes, assemblies, and annotations." Below this, there are several sections: "Using Genome" (with links to Help, Browse by Organism, Download / FTP, Download FAQ, and Submit a genome); "Custom resources" (with links to Human Genome, Microbes, Organelles, Viruses, and Prokaryotic reference genomes); "Other Resources" (with links to Assembly, BioProject, BioSample, and Genome Data Viewer); "Genome Tools" (with links to BLAST the Human Genome and Microbial Nucleotide BLAST); "Genome Annotation and Analysis" (with links to Eukaryotic Genome Annotation, Prokaryotic Genome Annotation, and PASC (Pairwise Sequence Comparison)); and "External Resources" (with links to GOLD - Genomes Online Database, Bacteria Genomes at Sanger, and Ensembl).

4. Les analyses génomiques

The screenshot shows the NCBI Genome homepage. At the top, there is a navigation bar with links for "Resources" and "How To". Below the navigation bar, there is a search bar with the word "Genome" selected in the dropdown menu. The main content area features a large image of chromosomes and a pen pointing at one. The title "Genome" is displayed, followed by a description: "This resource organizes information on genomes including sequences, maps, chromosomes, assemblies, and annotations." On the left side, there is a sidebar titled "Using Genome" with links for "Help", "Browse by Organism" (which is marked as "UPDATED"), "Download / FTP", "Download FAQ", and "Submit a genome". In the center, there is a section titled "Custom resources" which includes links for "Human Genome", "Microbes", "Organelles", "Viruses", and "Prokaryotic reference genomes". A red circle highlights the "Human Genome" link. On the right side, there are sections titled "Other Resources" and "External Resources", each containing several links. The "Other Resources" section includes "Assembly", "BioProject", "BioSample", and "Genome Data Viewer" (marked as "NEW"). The "External Resources" section includes "GOLD - Genomes Online Database", "Bacteria Genomes at Sanger", and "Ensembl".

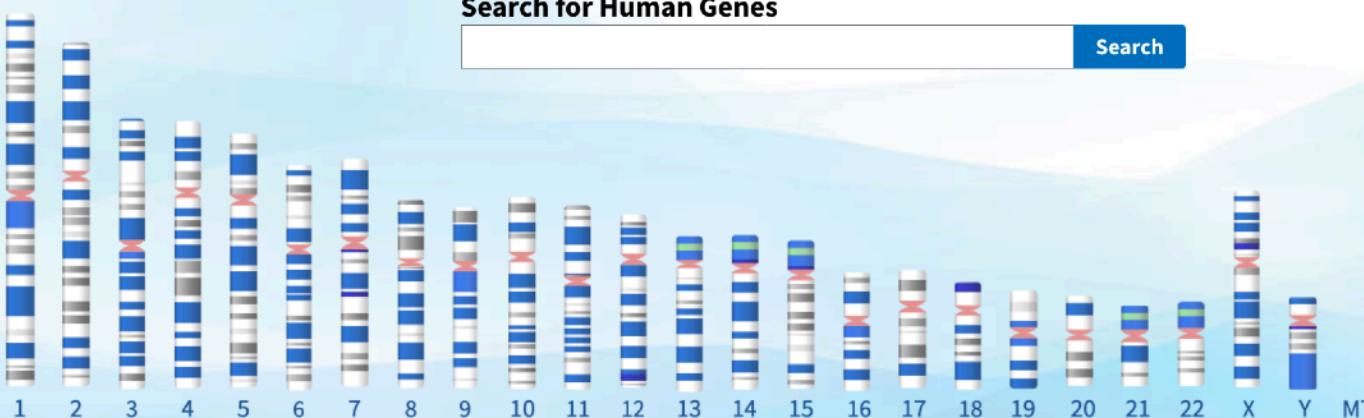
4. Les analyses génomiques

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Select a chromosome to access the [Genome Data Viewer](#)

▼

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	GRCh38	GRCh37
Reference Genome Sequence	Fasta	Fasta
RefSeq Reference Genome Annotation	gff3	gff3

 l'institut du thorax

4. Les analyses génomiques

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RefSeq Reference Genome Annotation	gff3	gff3

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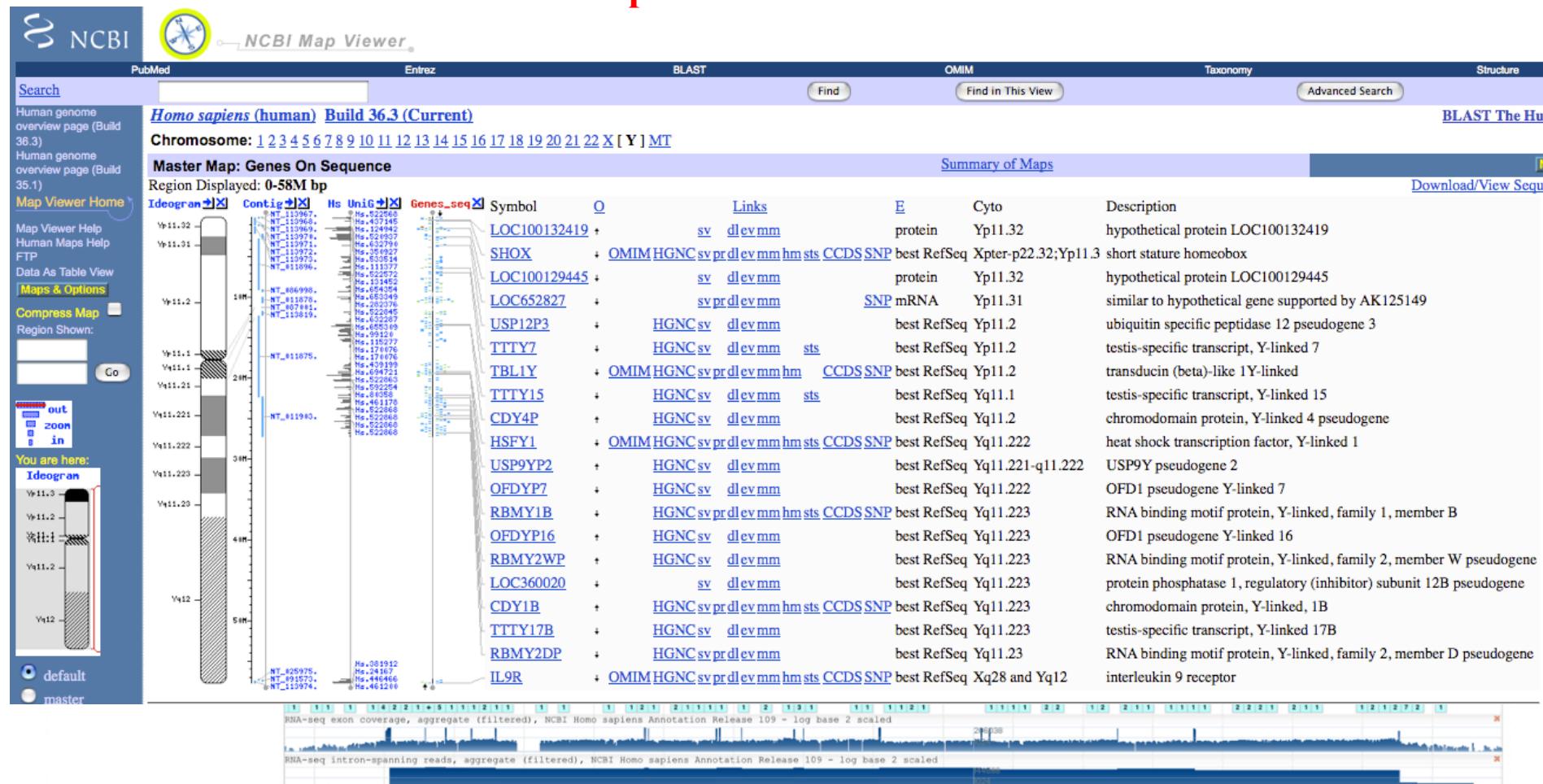
4. Les analyses génomiques



J'ai accès à tout ce qui est annoté sur le chromosome 21 ici...

4. Les analyses génomiques

Ici un Map Viewer sur le chromosome Y.



4. Les analyses génomiques



Genome Home ▶ Genome Resource Guides

NCBI
**Genome
Resource
Guides**

NCBI Genome Resource guides provide links, scoped searches, and alerts for a variety of genome-oriented data sources.

NCBI Genome Resource Guides

Access to genome resource guides for selected organisms.

Mammals

Organism	Reference Assembly	Current NCBI Build	Map Viewer Release date	Resource Links
human	Build 36.3	36.3	24 Mar 2008	G B M P
mouse	Build 37.1	37.1	5 Jul 2007	G B M P
rat	RGSC v3.4	4.1	6 Jul 2006	G B M P
cow	Btau_4.0	4.1	5 Aug 2008	G B M P
dog	Build 2.1	2.1	8 Sep 2005	G B M P

Show (+)

Birds

Organism	Reference Assembly	Current NCBI Build	Map Viewer Release date	Resource Links
chicken	Build 2.1	2.1	29 Nov 2006	G B M P
zebra finch	na	na	na	G B M P

Amphibians

Organism	Reference Assembly	Current NCBI Build	Map Viewer Release date	Resource Links
frog	na	na	na	G B M P

Echinoderms

Organism	Reference Assembly	Current NCBI Build	Map Viewer Release date	Resource Links
sea urchin	Build 2.1	2.1	18 Oct 2006	G B M P

Fish

Organism	Reference Assembly	Current NCBI Build	Map Viewer Release date	Resource Links
zebrafish	Zv7	3.1	12 Jul 2008	G B M P
fugu	Truv4.0	na	na	P
pufferfish	Tniv7	na	na	P
stickleback	Broad v1.0	na	na	P

Les autres espèces...

5. Les analyses protéiques

5. Les analyses protéiques

U.S. National Library of Medicine > NCBI National Center for Biotechnology Information Sign in to NCBI

BLAST® Home Recent Results Saved Strategies Help

Basic Local Alignment Search Tool

BLAST finds regions of similarity between biological sequences. The program compares nucleotide or protein sequences to sequence databases and calculates the statistical significance.

[Learn more](#)

N E W S

Using BLAST Well, How to Maximize Your Search Efforts: Webinar on October 3, 2018
In this webinar, the NCBI BLAST team lead will show you how to be more effective with BLAST.
Thu, 27 Sep 2018 11:00:00 EST [More BLAST news...](#)

Web BLAST

Nucleotide BLAST nucleotide ▶ nucleotide

blastx translated nucleotide ▶ protein

tblastn protein ▶ translated nucleotide

Protein BLAST protein ▶ protein

A red circle highlights the Protein BLAST button.

BLAST Genomes

Enter organism common name, scientific name, or tax id
Use up and down arrows to choose an item from the autocomplete.

Human Mouse Rat Microbes **Search**

Idem sur les protéines...

5. Les analyses protéiques

BLAST Basic Local Alignment Search Tool

Home Recent Results Saved Strategies Help

► NCBI/ BLAST/ blastp suite

blastn blastp blastx tblastn tblastx

Enter Query Sequence

Enter accession number, gi, or FASTA sequence

Query subrange

Or, upload file

Job Title
Enter a descriptive title for your BLAST search

Blast 2 sequences

Choose Search Set

Database Non-redundant protein sequences (nr)

Organism Optional
Enter organism name or id--completions will be suggested

Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown.

Entrez Query Optional
Enter an Entrez query to limit search

Program Selection

Algorithm blastp (protein-protein BLAST)
 PSI-BLAST (Position-Specific Iterated BLAST)
 PHI-BLAST (Pattern Hit Initiated BLAST)
Choose a BLAST algorithm

BLAST Search database nr using Blastp (protein-protein BLAST)

5. Les analyses protéiques

Basic Local Alignment Search Tool

My NCBI ?
[Sign In] [Register]

NCBI/ BLAST/ blastp suite/ Formatting Results - HRGK4VXW013

Edit and Resubmit Save Search Strategies ►Formatting options ►Download

Protein Sequence (138 letters)

Query ID Icl|7334
Description None
Molecule type amino acid
Query Length 138

Database Name nr
Description All non-redundant GenBank CDS translations+PDB+SwissProt+PIR+PRF excluding environmental samples from WGS projects
Program BLASTP 2.2.18+ ►Citation

Other reports: ►Search Summary [Taxonomy reports] [Distance tree of results]

▼ Graphic Summary

▼ Show Conserved Domains

Putative conserved domains have been detected, click on the image below for detailed results.

Query seq. Superfamilies 1 20 40 60 80 100 120 138

7tm_1 superfamily

Distribution of 100 Blast Hits on the Query Sequence ⓘ

Mouse-over to show defline and scores, click to show alignments

Color key for alignment scores

Query	<40	40-50	50-80	80-200	>=200	
0	20	40	60	80	100	120

5. Les analyses protéiques

▼ Alignments Select All [Get selected sequences](#) [Distance tree of results](#)

> ref|NP_037240.1| **UG** adrenergic, beta-3-, receptor [Rattus norvegicus]
gb|AAA74470.1| **G** beta-adrenergic receptor
Length=400

[GENE ID: 25645 Adrb3](#) | adrenergic, beta-3-, receptor [Rattus norvegicus]
[\(Over 10 PubMed links\)](#)

Score = 276 bits (706), Expect = 4e-73, Method: Compositional matrix adjust.
Identities = 138/138 (100%), Positives = 138/138 (100%), Gaps = 0/138 (0%)

Query 1	AQECHSNPRCCSFASNMPYALLSSSVSFYLPLLVMFLFYARVFVVAKRQRRFVRRELGRF	60
Sbjct 183	AQECHSNPRCCSFASNMPYALLSSSVSFYLPLLVMFLFYARVFVVAKRQRRFVRRELGRF	242
Query 61	PPEESPRSPSRSPSPATVGPTTASDGVPSCGRRPARLLPLGEHRALRTLGLIMGIFSLCW	120
Sbjct 243	PPEESPRSPSRSPSPATVGPTTASDGVPSCGRRPARLLPLGEHRALRTLGLIMGIFSLCW	302
Query 121	LPFFLANVLRALVGPSLV	138
Sbjct 303	LPFFLANVLRALVGPSLV	320

6. L'intelligence artificielle

6. L'intelligence artificielle

Des algorithmes au service de la médecine

6. L'intelligence artificielle

- L'intelligence artificielle (IA) est un domaine de recherche en pleine expansion et promis à un grand avenir.
- Ses applications, qui concernent toutes les activités humaines, permettent notamment d'améliorer la qualité des soins.
- L'IA est en effet au cœur de la médecine du futur avec **les opérations assistées, le suivi des patients à distance, les prothèses intelligentes, ou encore les traitements personnalisés** grâce au recouplement de données (*big data*)...

6. L'intelligence artificielle

- Les chercheurs développent des approches et techniques multiples, du traitement des langues et de la construction d'ontologies, à la fouille de données et à l'apprentissage automatique...
- Comment fonctionnent ces systèmes pour savoir ce qu'ils font et surtout ce qu'ils ne font pas?



L'IA s'appuie sur **2 approches** :

- **symbolique**
fondée sur la logique
- **numérique**
fondée sur les données

Les enjeux de la recherche :

- des **données** mieux structurées et protégées
- des **systèmes** plus performants et transparents



6. L'intelligence artificielle

- Le but de l'IA c'est de développer toutes les technologies disponibles pour **concevoir des machines capables d'aider les humains dans leurs tâches.**



Les domaines d'application de l'IA en médecine

6. L'intelligence artificielle

Les différentes approches

- 1) Dans les années 1980, cette **approche, dite symbolique**, a permis le développement d'outils capables de reproduire les mécanismes cognitifs d'un expert. C'est pourquoi on les a baptisés « **systèmes experts** ».

Cette approche s'appuie sur l'ensemble des connaissances médicales dans un domaine donné et une formalisation des raisonnements des spécialistes qui lient ces connaissances entre elles pour aboutir à un diagnostic.

- Les systèmes actuels, qualifiés **d'aide à la décision**, de **gestion des connaissances** ou **d'e-santé**, sont plus sophistiqués. La mécanique algorithmique est globalement la même, mais les langages de description sont plus efficaces et les machines plus puissantes.

Ils ne cherchent plus à remplacer le médecin, mais à l'épauler dans un raisonnement fondé sur les connaissances médicales de sa spécialité.

6. L'intelligence artificielle

Les différentes approches

- 2) Contrairement à l'approche symbolique, **l'approche dite *numérique*** raisonne sur les données. Le système cherche des régularités dans les données disponibles pour extraire des connaissances, sans modèle préétabli.

Cette méthode, née avec le connexionnisme et les réseaux de neurones artificiels dans les années 1980, se développe aujourd'hui grâce à l'augmentation de puissance des ordinateurs et à l'accumulation des gigantesques quantités de données, le fameux big data.

La plupart des systèmes actuels procèdent par apprentissage automatique, une méthode fondée sur la représentation mathématique et informatique de neurones biologiques.

6. L'intelligence artificielle

Les différentes approches

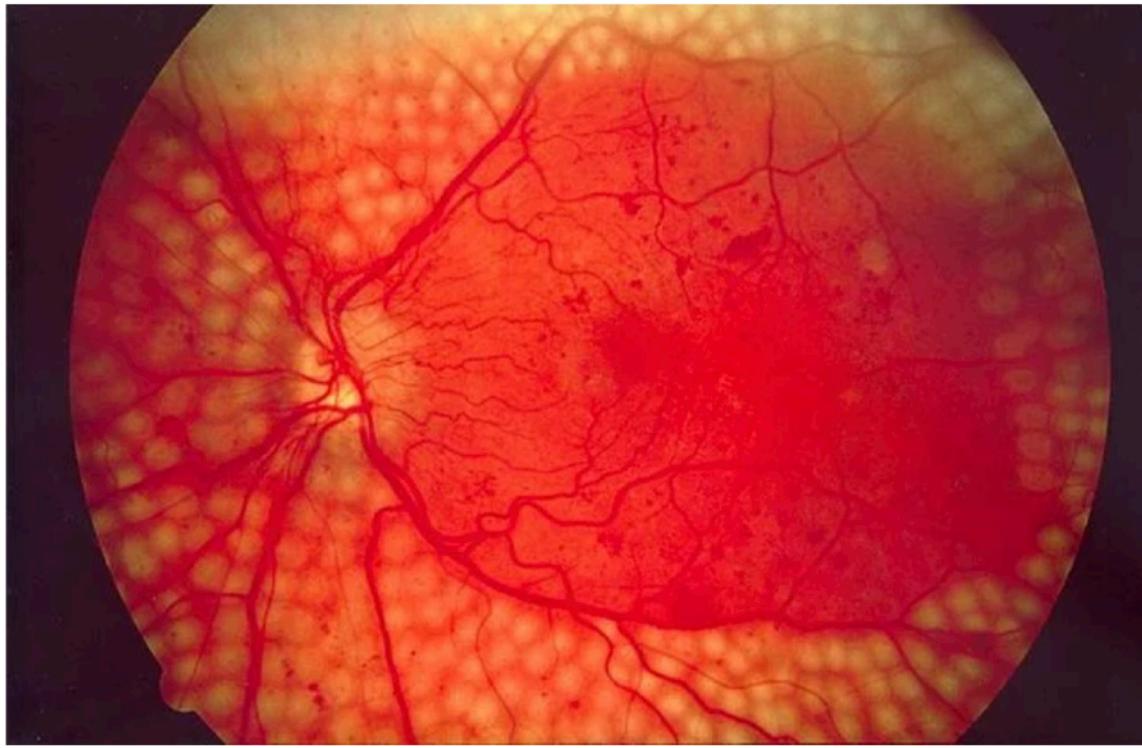
- Les algorithmes d'apprentissage profond (deep learning) par exemple, dont l'usage explose depuis une dizaine d'années, font une analogie lointaine avec le fonctionnement cérébral en simulant un réseau de neurones organisés en différentes couches, échangeant les uns avec les autres.

=> *La force de cette approche est que l'algorithme apprend la tâche qui lui a été assignée par « essais et erreurs », avant de se débrouiller tout seul.*

- Des applications de *deep learning* existent en traitement d'images, par exemple pour repérer de possibles mélanomes sur les photos de peau, ou bien pour dépister des rétinopathies diabétiques sur des images de rétines.



Fond d'œil. Ce fond d'œil montre la répartition des points de coagulation proposée par un logiciel de pilotage du laser pour traiter une rétinopathie diabétique. L'IA permet de dépister automatiquement les patients qui devraient pouvoir subir une telle intervention. © National Eye Institute, NIH.



Fond d'œil. Ce fond d'œil montre la répartition des points de coagulation proposée par un logiciel de pilotage du laser pour traiter une rétinopathie diabétique. L'IA permet de dépister automatiquement les patients qui devraient pouvoir subir une telle intervention. © National Eye Institute, NIH.

Leur mise au point nécessite de grands échantillons d'apprentissage : 50 000 images dans le cas des mélanomes, et 128 000 dans celui des rétinopathies, ont été nécessaires pour entraîner l'algorithme à identifier les signes de pathologies. Pour chacune de ces images on lui indique si elle présente ou non des signes pathologiques. A la fin de l'apprentissage, l'algorithme arrive à reconnaître avec une excellente performance de nouvelles images présentant une anomalie.

Review

Artificial Intelligence in the Diagnosis and Treatment of Brain Gliomas

Kyriacos Evangelou ^{1,*}, Ioannis Kotsantis ², Aristotelis Kalyvas ^{1,3}, Anastasios Kyriazoglou ², Panagiota Economopoulou ², Georgios Velonakis ⁴, Maria Gavra ⁵, Amanda Psyri ², Efstathios J. Boviatsis ¹ and Lampis C. Stavrinou ¹

Abstract

Brain gliomas are highly infiltrative and heterogenous tumors, whose early and accurate detection as well as therapeutic management are challenging. Artificial intelligence (AI) has the potential to redefine the landscape in neuro-oncology and can enhance glioma detection, imaging segmentation, and non-invasive molecular characterization better than conventional diagnostic modalities through deep learning-driven radiomics and radio-genomics. AI algorithms have been shown to predict genotypic and phenotypic glioma traits with remarkable accuracy and facilitate patient-tailored therapeutic decision-making. Such algorithms can be incorporated into surgical planning to optimize resection extent while preserving eloquent cortical structures through preoperative imaging fusion and intraoperative augmented reality-assisted navigation. Beyond resection, AI may assist in radiotherapy dose distribution optimization, thus ensuring maximal tumor control while minimizing surrounding tissue collateral damage. AI-guided molecular profiling and treatment response prediction models can facilitate individualized chemotherapy regimen tailoring, especially for glioblastomas with MGMT promoter methylation. Applications in immunotherapy are emerging, and research is focusing on AI to identify tumor microenvironment signatures predictive of immune checkpoint inhibition responsiveness. AI-integrated prognostic models incorporating radiomic, histopathologic, and clinical variables can additionally improve survival stratification and recurrence risk prediction remarkably, to refine follow-up strategies in high-risk patients. However, data heterogeneity, algorithmic transparency concerns, and regulatory challenges hamstring AI implementation in neuro-oncology despite its transformative potential. It is therefore imperative for clinical translation to develop interpretable AI frameworks, integrate multimodal datasets, and robustly validate externally. Future research should prioritize the creation of generalizable AI models, combine larger and more diverse datasets, and integrate multimodal imaging and molecular data to overcome these obstacles and revolutionize AI-assisted patient-specific glioma management.

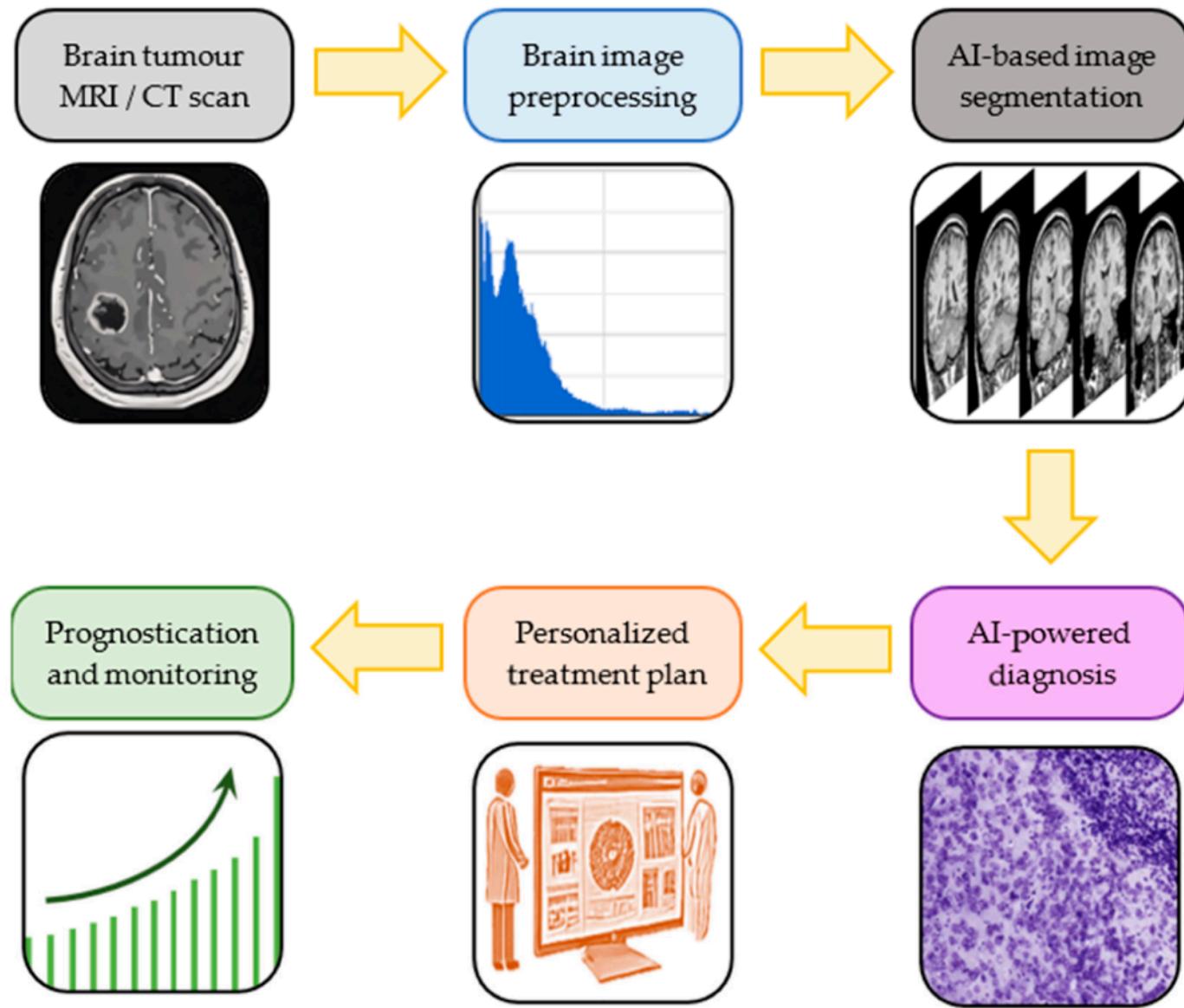
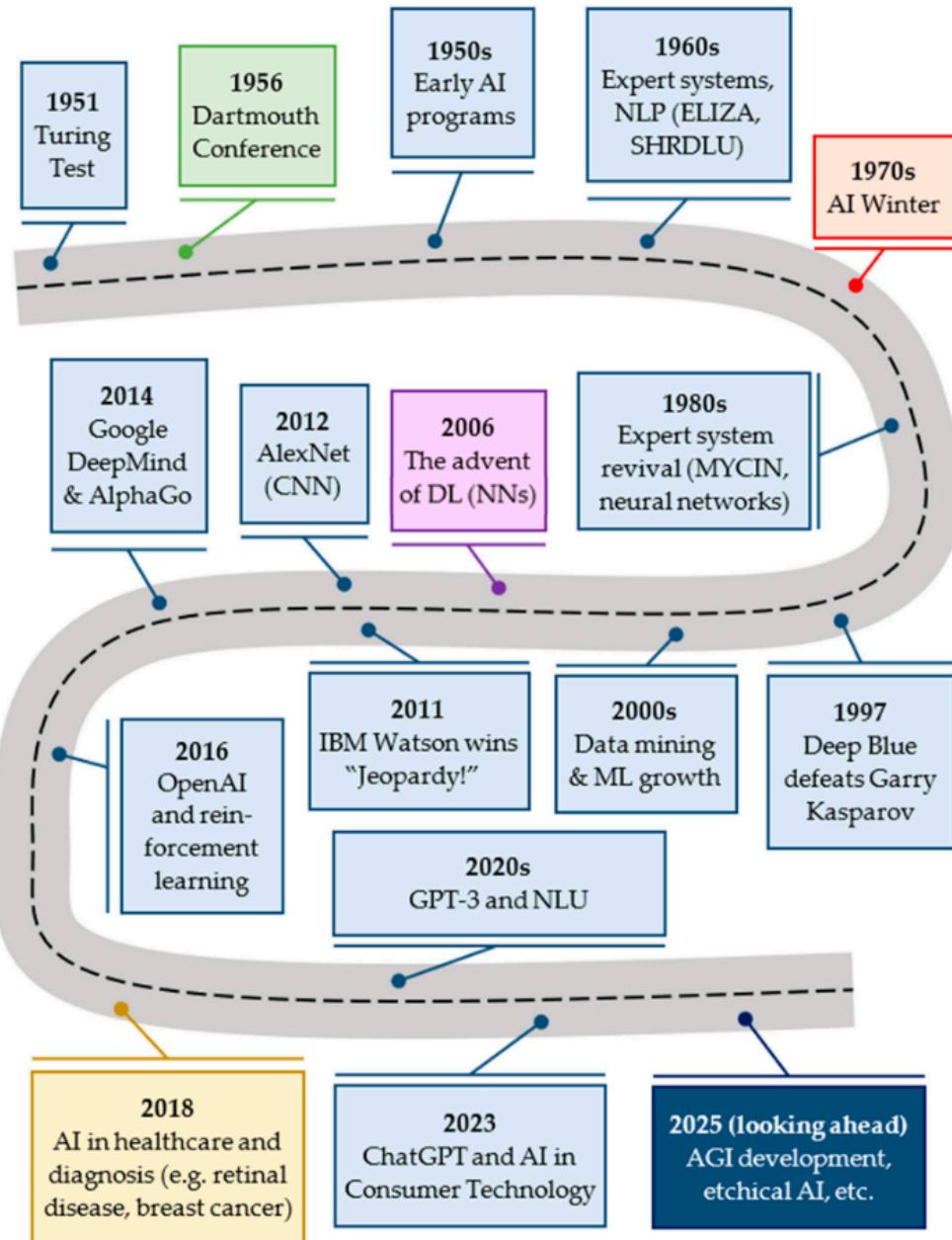


Figure 1. Artificial intelligence (AI) workflow sequential steps in brain glioma diagnosis and treatment. Multimodal imaging-derived brain tumour images are preprocessed and segmented with the help of AI; this facilitates accurate diagnosis and the development of personalized treatment plans, which improve patient prognostication and monitoring.

Figure 3. Timeline of key milestones in AI development from its inception in the 1950s to the groundbreaking advancements of the 2020s [33]. The Turing Test was proposed by Alan Turing to determine if a machine could exhibit human-like intelligence, something which later became a foundational concept in AI. Deep Blue of the International Business Machines Corporation (IBM) was a chess-playing computer that defeated world champion Garry Kasparov, marking a significant achievement for AI in strategy and game-playing. Watson is a computer system that defeated human champions in the quiz show “Jeopardy!”, showcasing advancements in natural language processing (NLP) and AI’s ability to understand and respond to complex questions. AlexNet was a convolutional neural network (CNN) that won the ImageNet competition by a large margin, revolutionizing the field of computer vision. In the present environment, efforts to develop Artificial General Intelligence (AGI) continue to progress, with a focus on creating AI that can understand, learn, and apply knowledge in a human-like manner across diverse tasks. Ethical considerations, including AI governance and fairness, will play an increasingly significant role as AI becomes more integrated into daily life. AI = artificial intelligence; DL = deep learning; GPT-3 = generative pre-trained transformer 3; ML = machine learning; NLU = natural language understanding.



Most important artificial intelligence (AI) algorithms in neuro-oncology

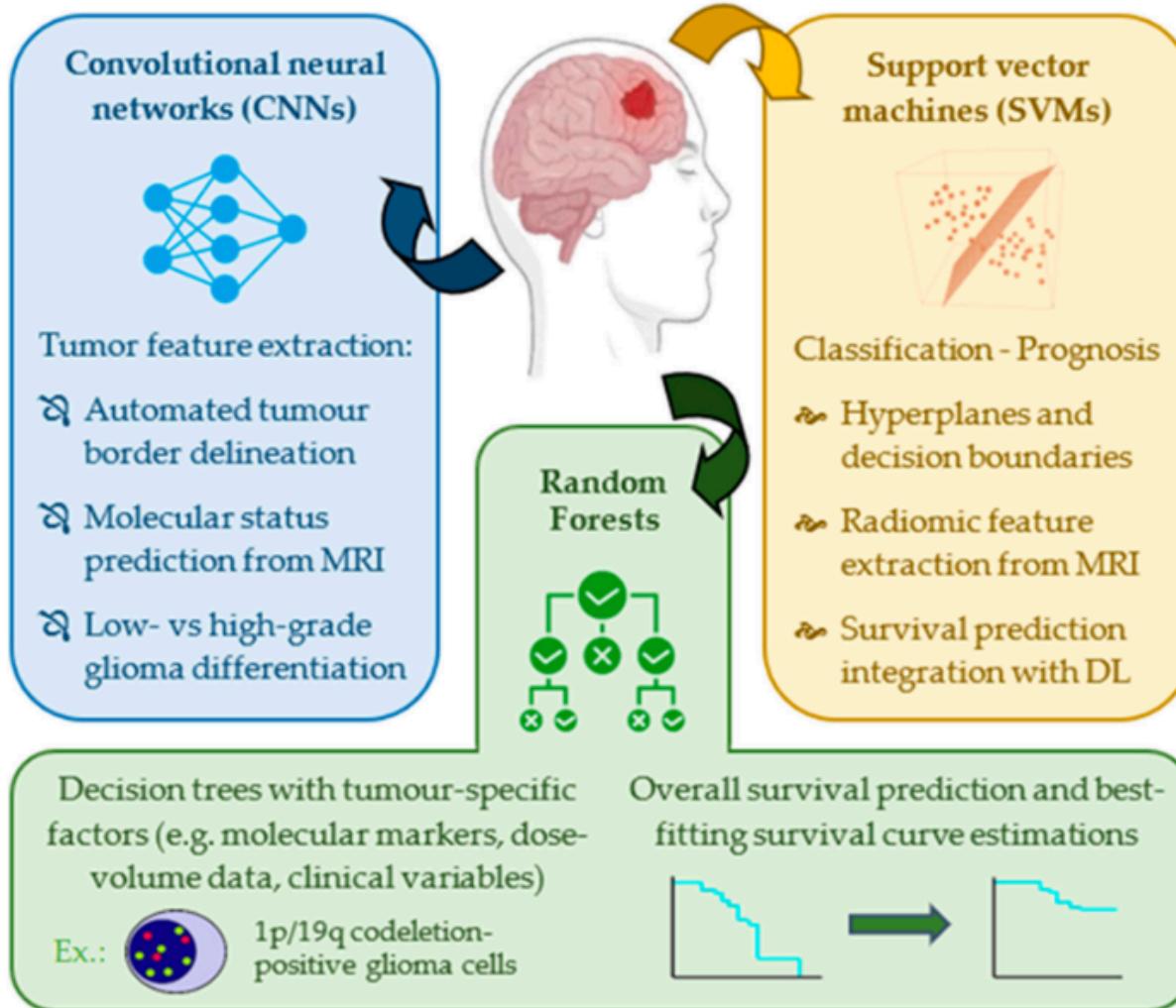


Figure 4. Artificial intelligence (AI) algorithms in neuro-oncology for glioma diagnosis, prognosis assessment, and treatment facilitation. 1p = short arm of chromosome 1; 19q = long arm of chromosome 19; DL = deep learning; MRI = magnetic resonance imaging.

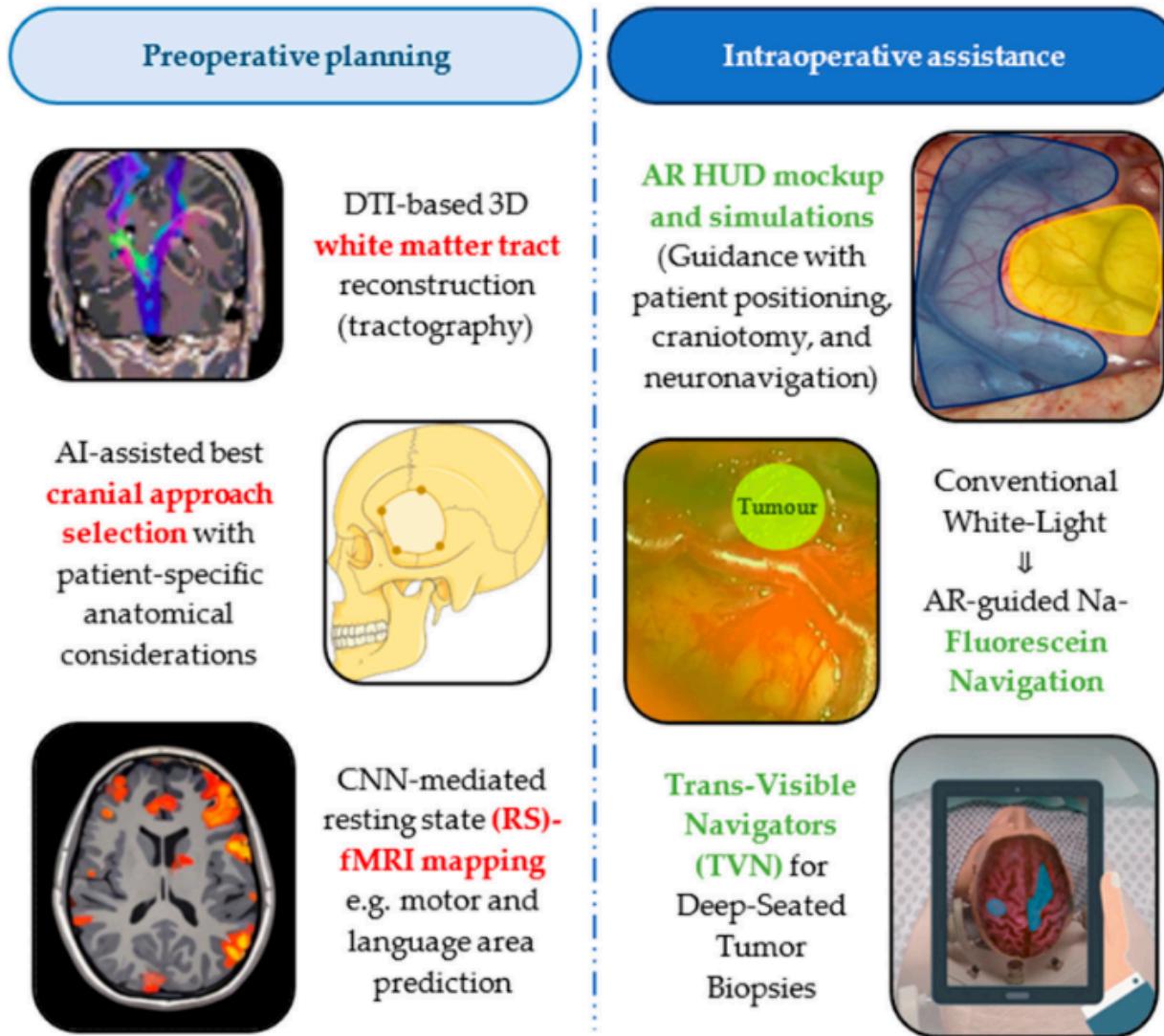


Figure 5. Applications of artificial intelligence (AI) derivatives in postoperative planning and intraoperative assistance for glioma surgery. AR = Augmented reality; CNN = Convolutional neural network; DTI = Diffusion tensor imaging; fMRI = functional magnetic resonance imaging; HUD = heads-up display.

7. Exemple publication utilisant la bioinfo

7. Exemple publication utilisant la bioinfo

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<https://doi.org/10.1186/s13023-023-02885-1>

Orphanet Journal of Rare Diseases

RESEARCH

Open Access



The metabolomic plasma profile of patients with Duchenne muscular dystrophy: providing new evidence for its pathogenesis

Huayan Xu^{1†}, Xiaotang Cai^{2†}, Ke Xu¹, Qihong Wu¹ and Bei Xu^{3,4*} 

7. Exemple publication utilisant la bioinfo

Huayan Xu^{1†}, Xiaotang Cai^{2†}, Ke Xu¹, Qihong Wu¹ and Bei Xu^{3,4*} 

Abstract

Background Duchenne muscular dystrophy (DMD) is a fatal genetic muscle-wasting disease that affects 1 in 5000 male births with no current cure. Despite great progress has been made in the research of DMD, its underlying pathological mechanism based on the metabolomics is still worthy of further study. Therefore, it is necessary to gain a deeper understanding of the mechanisms or pathogenesis underlying DMD, which may reveal potential therapeutic targets and/or biomarkers.

Results Plasma samples from 42 patients with DMD from a natural history study and 40 age-matched healthy volunteers were subjected to a liquid chromatography-mass spectrometry-based non-targeted metabolomics approach. Acquired metabolic data were evaluated by principal component analysis, partial least squares-discriminant analysis, and metabolic pathway analysis to explore distinctive metabolic patterns in patients with DMD. Differentially expressed metabolites were identified using publicly available and integrated databases. By comparing the DMD and healthy control groups, 25 differential metabolites were detected, including amino acids, unsaturated fatty acids, carnitine, lipids, and metabolites related to the gut microbiota. Correspondingly, linoleic acid metabolism, D-glutamine and D-glutamate metabolism, glycerophospholipid metabolism, and alanine, aspartate, and glutamate metabolism were significantly altered in patients with DMD, compared with those of healthy volunteers.

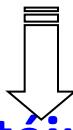
Conclusions Our study demonstrated the abnormal metabolism of amino acids, energy, and lipids in patients with DMD, consistent with pathological features, such as recurrent muscle necrosis and regeneration, interstitial fibrosis, and fat replacement. Additionally, we found that metabolites of intestinal flora were disordered in DMD patients, providing support for treatment of intestinal microbia disturbance in DMD diseases. Our study provides a new research strategy for understanding the pathogenesis of DMD.

Keywords Duchenne muscular dystrophy, Metabonomics, Mass spectrometry, Plasma

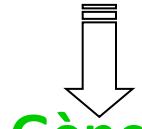
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Physiologie « classique »

Fonction



Protéines

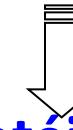


Gènes

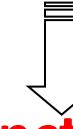
Vision restreinte

Physiologie « inverse »

Gènes



Protéines



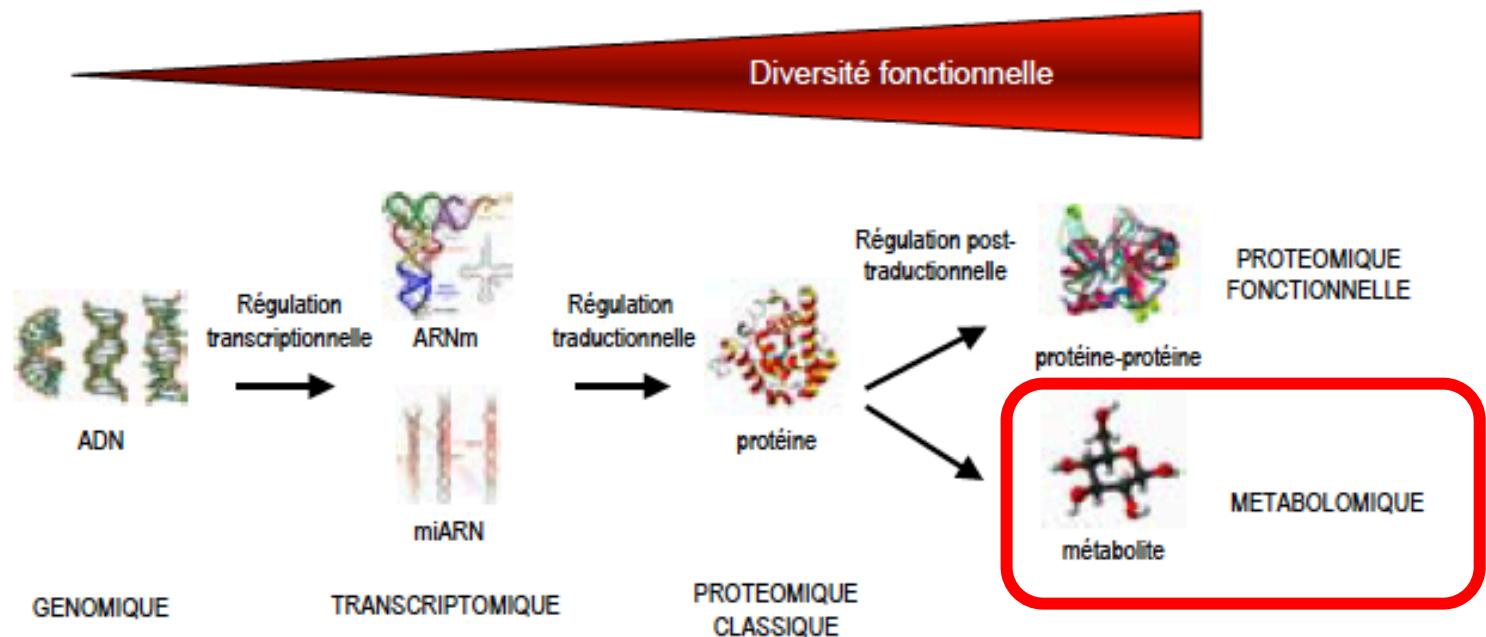
Fonction

Vision globale

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Intérêt de la recherche « OMICS »

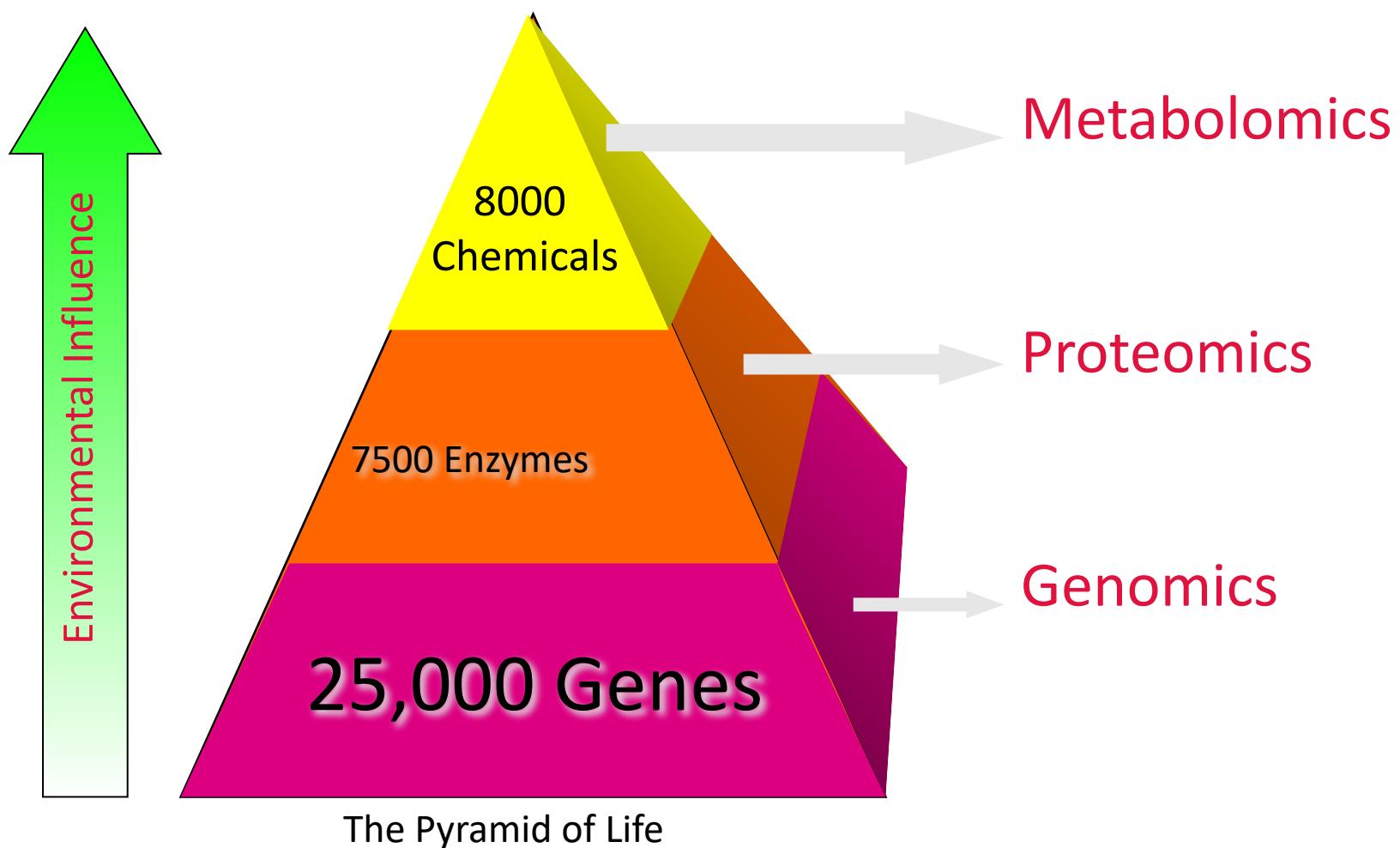
- Approche globale sans aucun *a priori*



- Protéomique

Caractérisation qualitative et quantitative de l'ensemble des protéines présentes dans un échantillon biologique obtenu dans des conditions définies

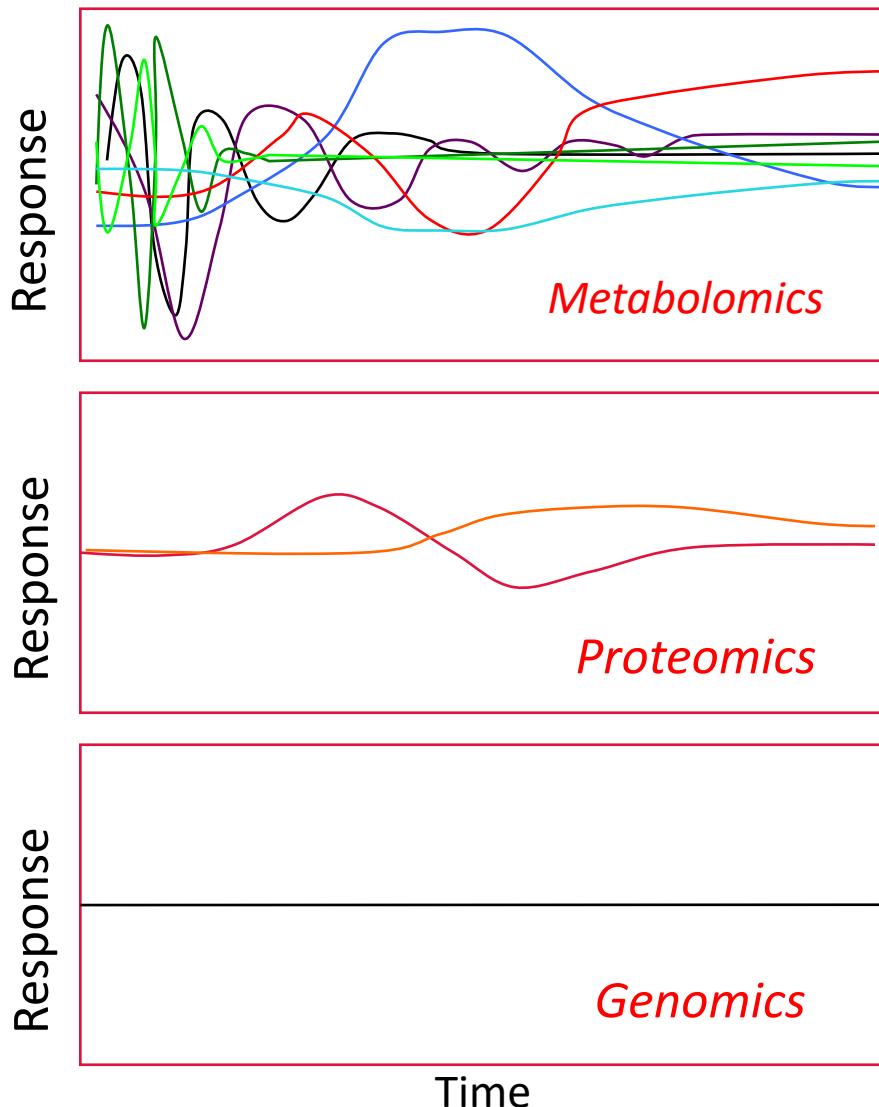
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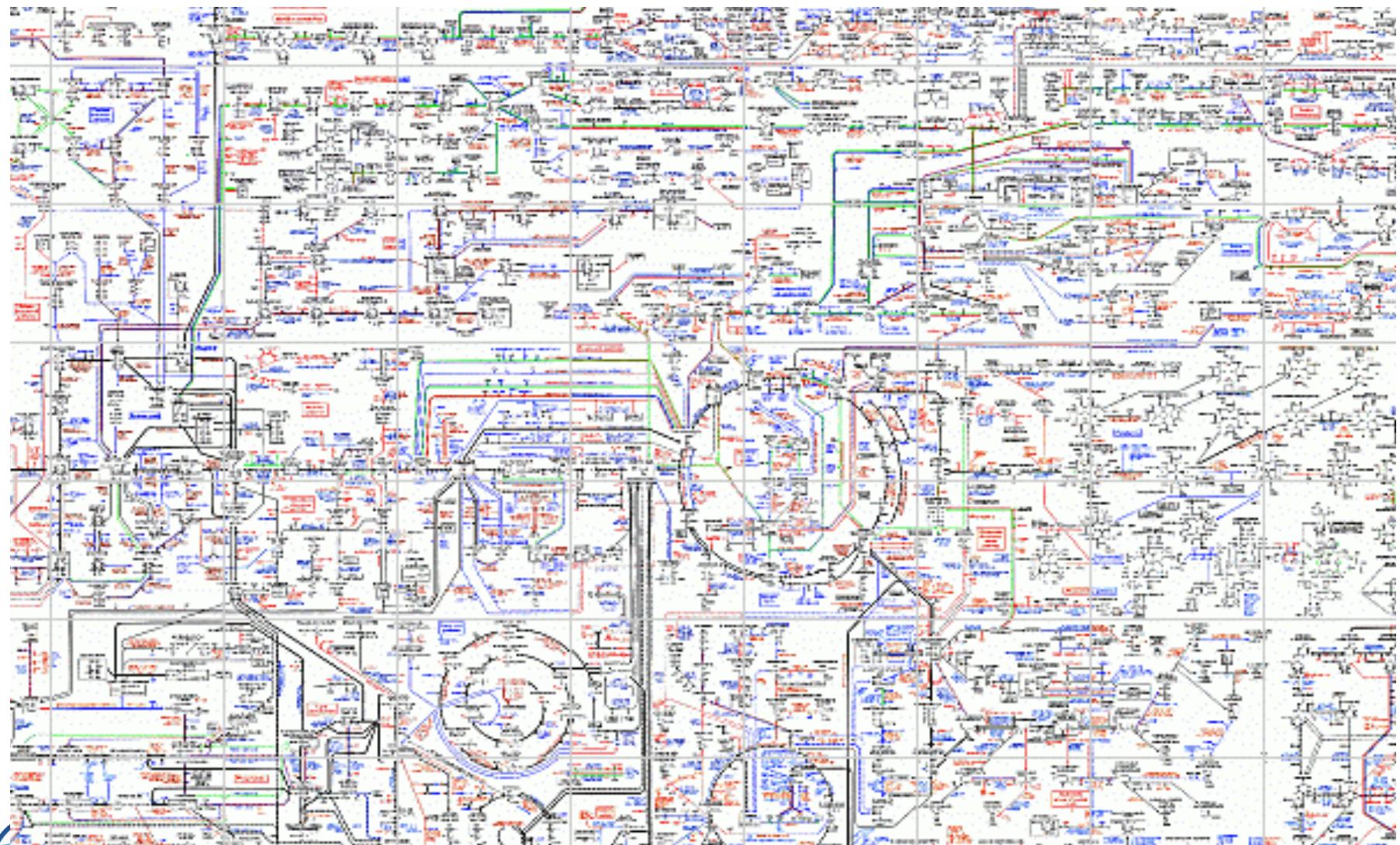
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La métabolomique est plus sensible
dans le temps que d'autres "omiques"



7. Exemple publication utilisant la bioinfo



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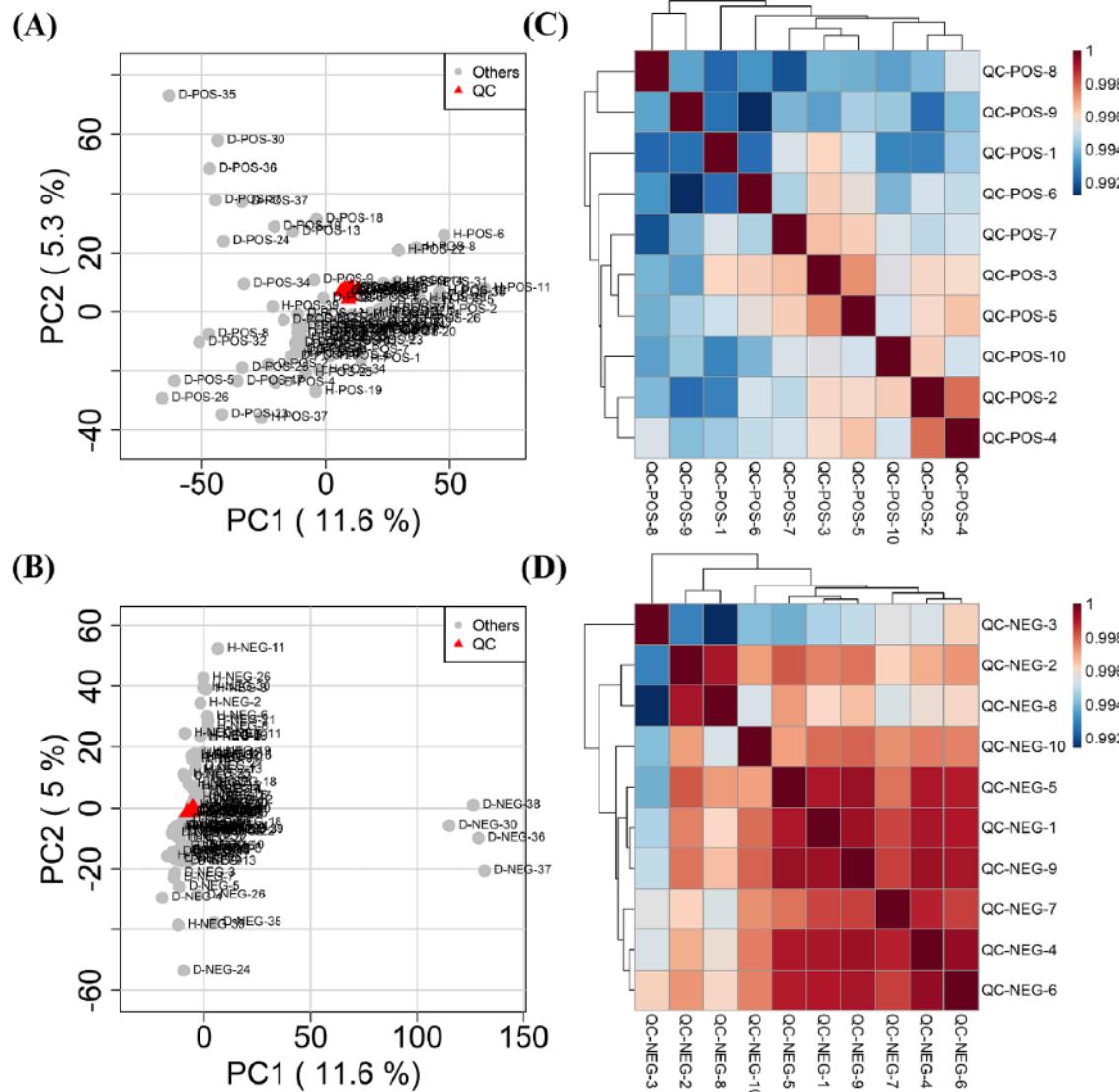


Fig. 1 PCA score plots and correlation analysis of QC samples in ESI+ (**A, C**) and ESI- (**B, D**) scan modes

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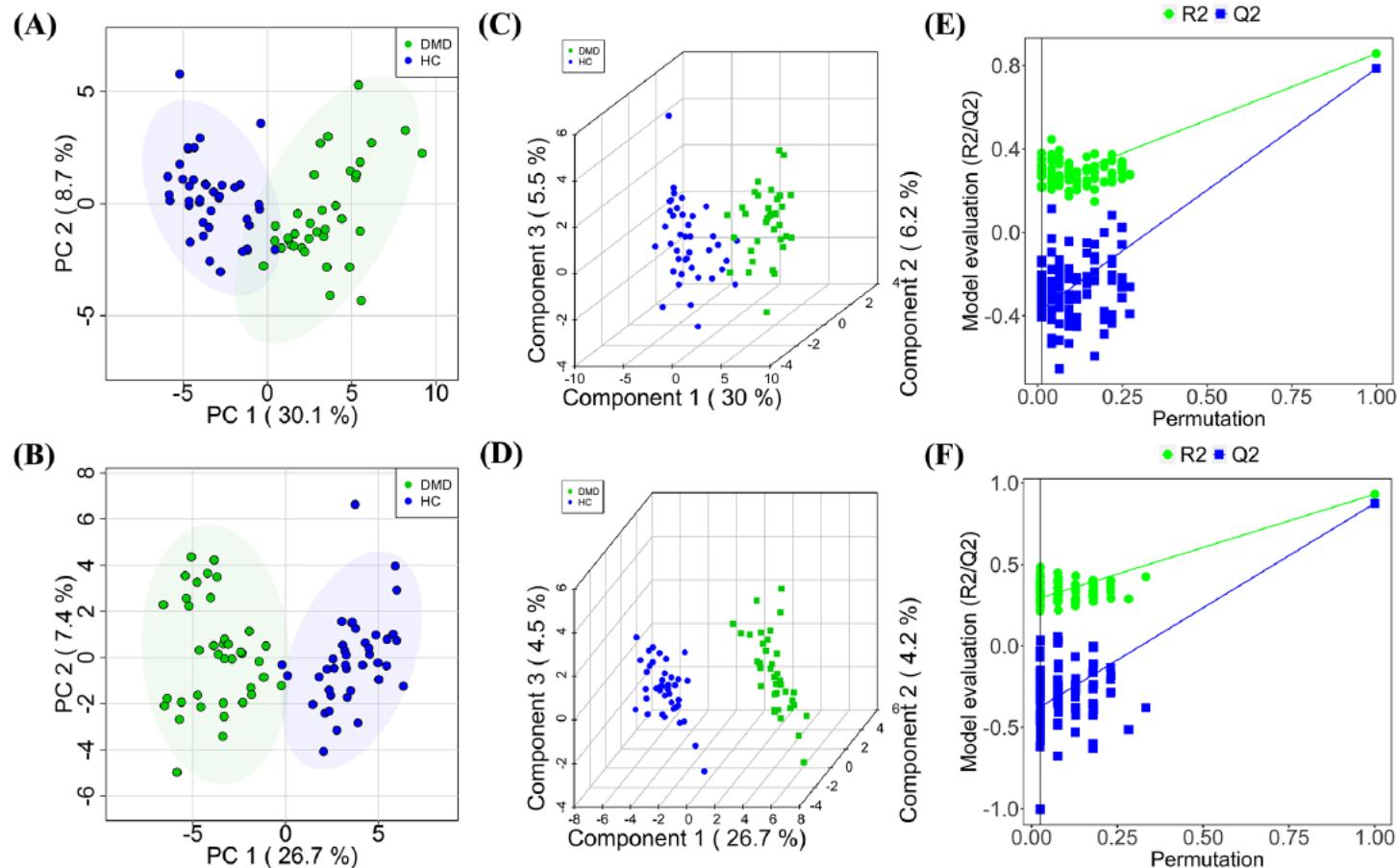


Fig. 2 Plots of PCA (**A, B**) and PLS-DA scores (**C, D**) with permutation testing (**E, F**) for healthy controls and DMD patients comparison in the ESI+ and ESI- scan modes

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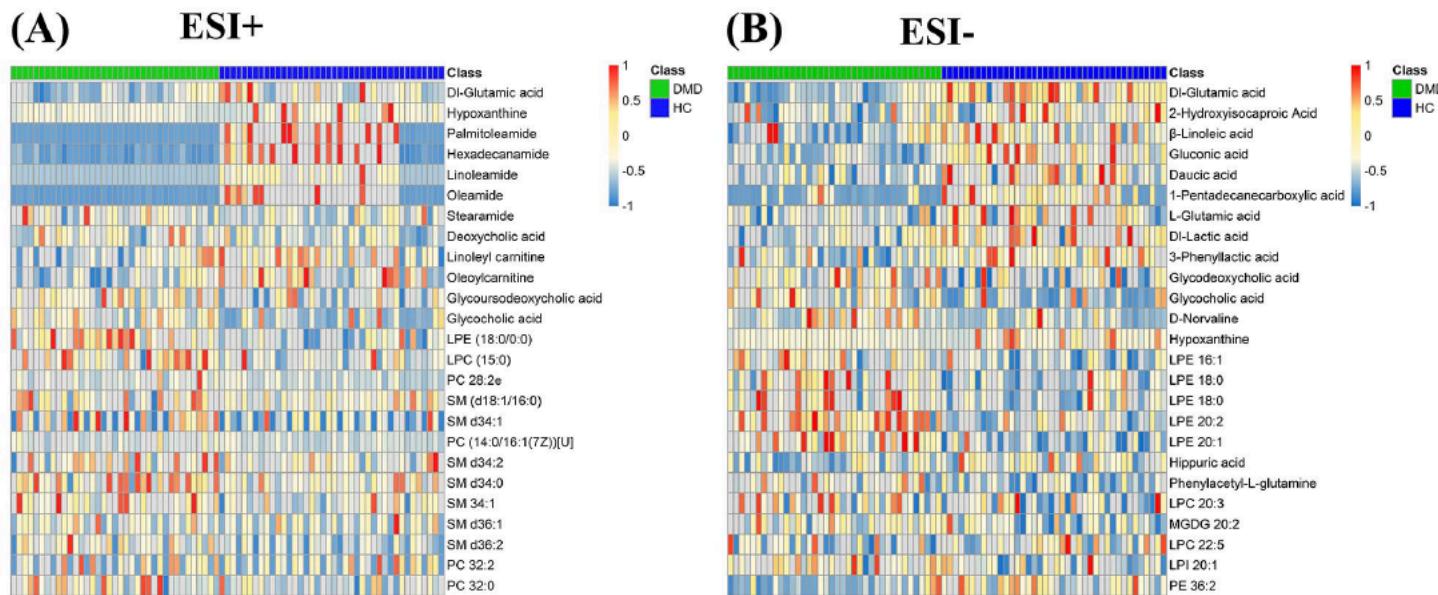


Fig. 3 Differential metabolite heat maps in ESI+ (A) and ESI- (B) scan modes. The columns represent samples, the rows represent metabolites, and the relative content of the metabolites is displayed by color

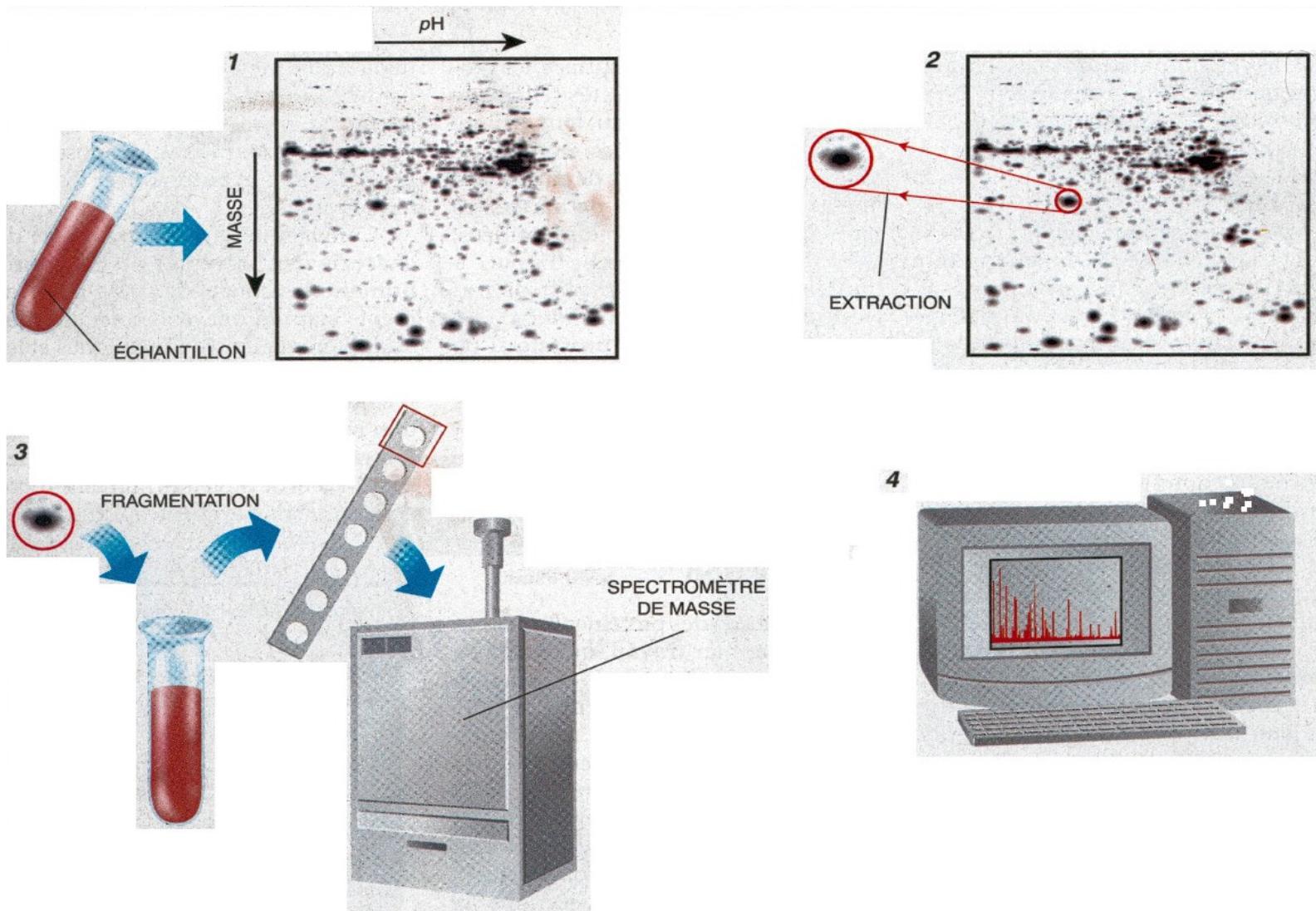
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Metabolite detection

An ultra-performance liquid chromatography (UPLC) system (Agilent1290 Infinity II; Agilent Technologies Inc., CA, USA) connected to a high-resolution tandem mass spectrometer (TripleTOF 5600 Plus; AB SCIEX, Framingham, MA, USA) was used to conduct the metabolomic analysis. Reversed-phase separation was performed on an ACQUITY HSS T3 column (100×2.1 mm, i.d. $1.8 \mu\text{m}$; Waters, Milford, USA). The mobile phase composition was determined using a gradient elution of solvent A (0.1% formic acid in water) and solvent B (0.1% formic acid in acetonitrile), as previously described [19]. The flow rate was constant at 0.30 mL/min , and the column temperature was set at 30°C .

- La spectrométrie de masse (mass spectrometry ou MS) est une technique physique d'analyse permettant de détecter et d'identifier des molécules d'intérêt par mesure de leur masse mono-isotopique.
- De plus, la spectrométrie de masse permet de caractériser la structure chimique des molécules en les fragmentant.

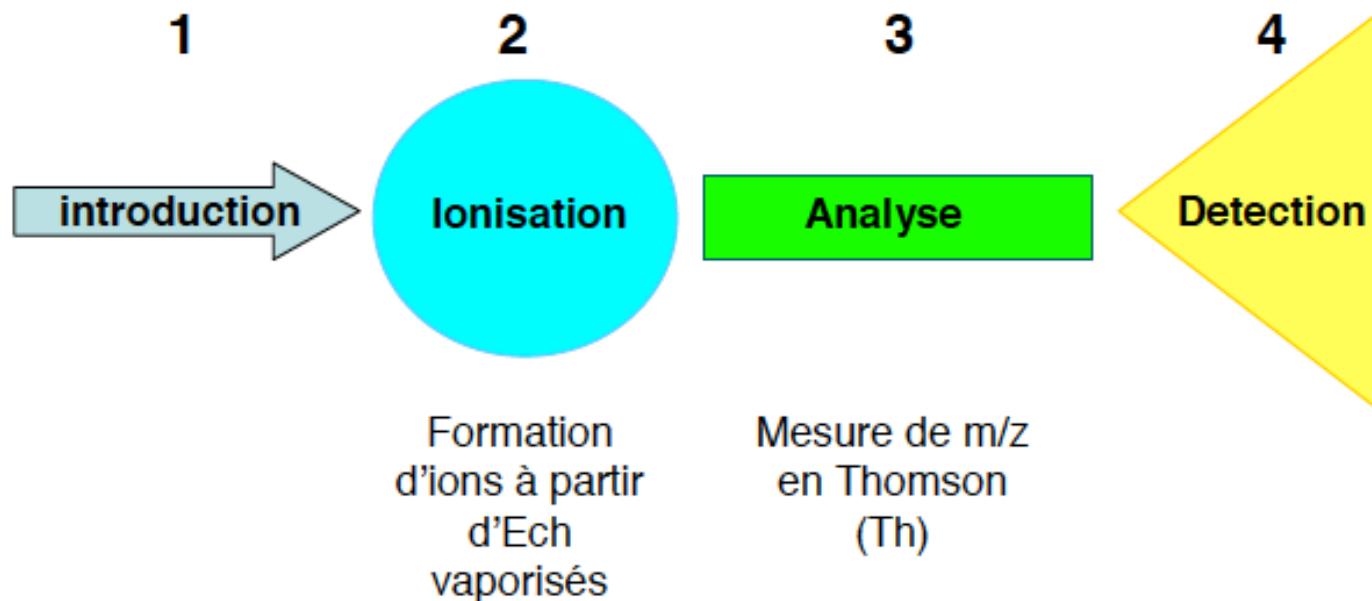
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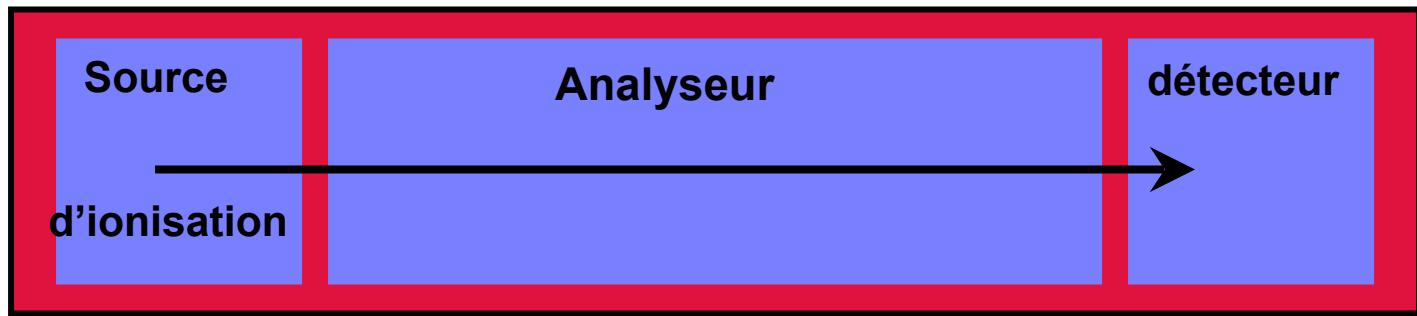
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Le spectromètre de masse se compose donc de quatre parties :

- 1- Le système d'introduction de l'échantillon
- 2- La source d'ionisation: elle consiste à vaporiser les molécules et à les ioniser.
- 3- L'analyseur
- 4- Le détecteur et système de traitement

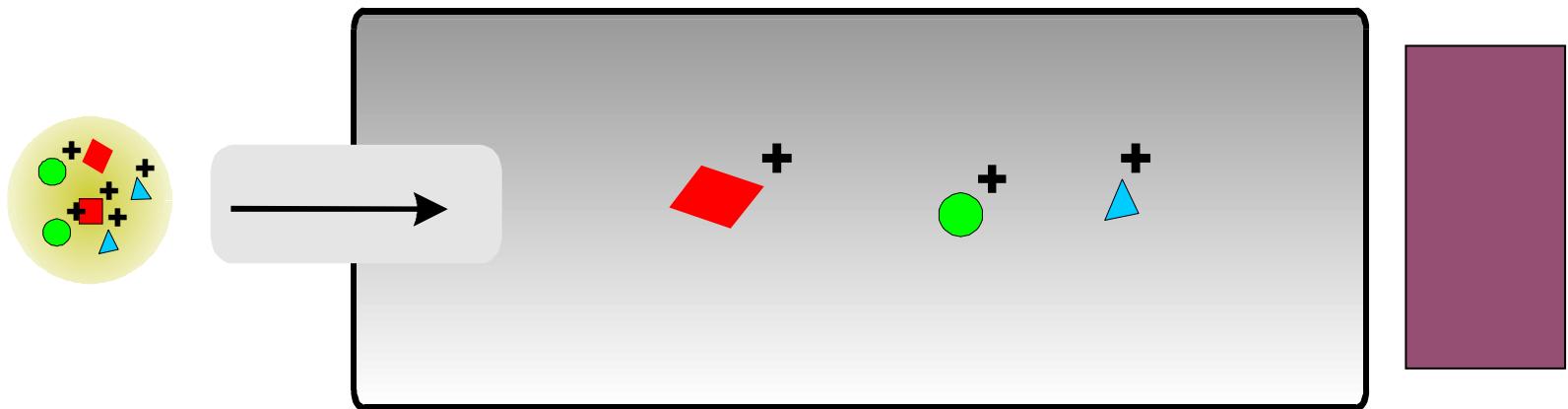


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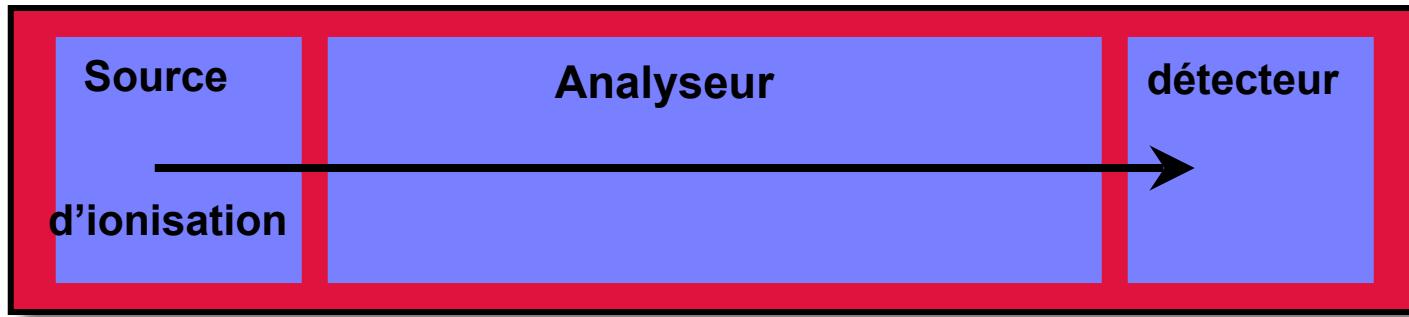


L'analyseur de masse

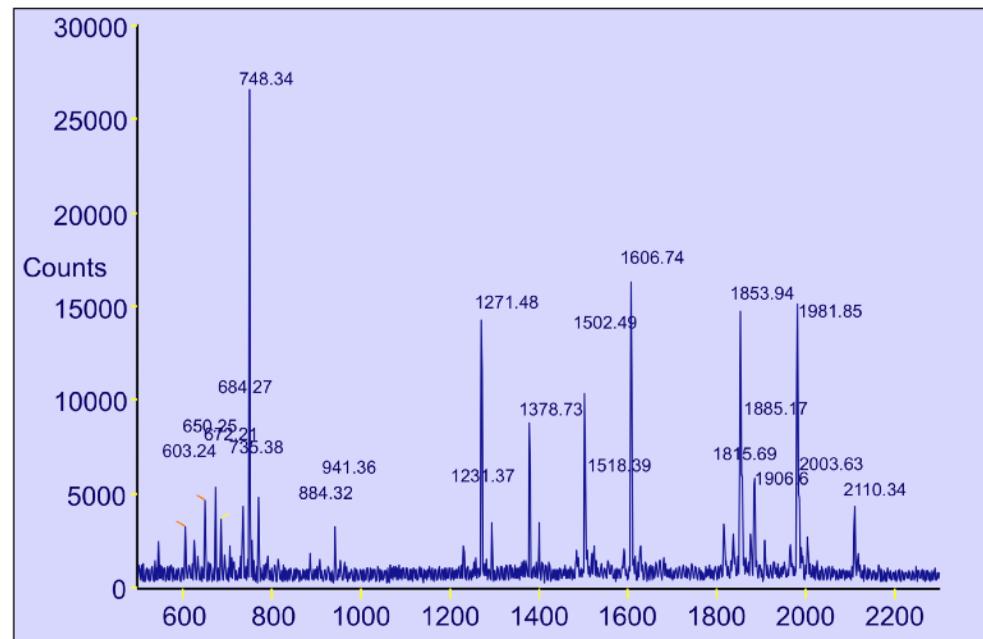
Détecteur



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Détecteur



7. Exemple publication utilisant la bioinfo

Cartographie massique par SM



4148.1061
3221.5113
3392.5756
2993.3522
2468.2415
2325.0207
2144.9770
1948.9585
2005.9800
1906.8817
1883.0497
1721.7765
1618.7577
1732.8006
1508.7889
1288.7259
1134.6742
1008.5360
1008.5295
1065.5509
950.4499
1007.4714
880.4159
832.3658
792.3709

La carte massique obtenue est **l'empreinte massique** de la protéine. Elle est très spécifique.

7. Exemple publication utilisant la bioinfo

Liste de masses expérimentale

4148.1061
3221.5113
3392.5756
2993.3522
2468.2415
2325.0207
2144.9770
1948.9585
2005.9800
1906.8817
1883.0497
1721.7765
1618.7577
1732.8006
1508.7889
1288.7259
1134.6742
1008.5360
1008.5295
1065.5509
950.4499
1007.4714
880.4159
832.3658
792.3709

Interrogation des banques de données

Comparaison avec tous les profils théoriques des séquences de protéines connues présentes dans les banques

Sélection de la protéine la plus probable.



7. Exemple publication utilisant la bioinfo

Table 2 List of statistically significant metabolites in DMD vs. HC comparisons

Metabolites		Scan mode	Rt (s)	m/z	Adducts	DMD vs. HC			
						Log2(FC)	P (T test)	VIP	Trends
Amino acids	Glutamic acid	ESI-	73.431	128.036	M-H ₂ O-H	-1.181	<0.001	1.955	↓
	Glutamine	ESI+	47.218	146.080	M+H	-1.072	<0.001	1.857	↓
	Hippuric acid	ESI-	300.935	178.051	M-H	-0.802	0.021	1.009	↓
	Phenylacetyl-glutamine	ESI-	348.896	527.213	2 M-H	1.143	<0.001	2.008	↑
	Valine	ESI-	420.136	293.176	2 M+Hac-H	1.072	0.005	1.263	↑
Unsaturated fatty acids	β-Linolenic acid	ESI-	419.308	279.232	M-H	-0.766	0.0001	1.582	↓
Carnitine	Linoleyl carnitine	ESI+	435.747	424.342	M+H	-0.784	<0.001	1.536	↓
	Oleoylcarnitine	ESI+	461.927	426.358	M+H	-0.766	<0.001	1.635	↓
Bile acids	Glycocholic acid	ESI-	372.288	464.302	M-H	1.121	0.005	1.122	↑
	Glycodeoxycholic acid	ESI-	406.477	448.307	M-H	0.950	0.0004	1.460	↑
	Glycoursodeoxycholic acid	ESI+	406.026	450.322	M+H	1.031	<0.001	1.543	↑
	Deoxycholic acid	ESI+	462.698	357.279	M+H-2H ₂ O	1.174	0.002	1.323	↑
Lipids	PC 32:0	ESI+	1117.300	756.555	M+Na	0.712	0.006	1.058	↑
	PC 32:2	ESI+	730.026	730.539	M+H	0.872	0.029	1.018	↑
	LPE 18:0	ESI-	552.975	480.310	M-H	0.655	<0.001	2.289	↑
	LPE 20:1	ESI-	590.366	506.326	M-H	0.748	<0.001	1.656	↑
	LPE 20:2	ESI-	526.317	504.311	M-H	0.856	<0.001	2.269	↑
	LPE (18:0/0:0)	ESI+	574.100	482.324	M+H	0.785	<0.001	2.489	↑
	LPI 20:1	ESI-	386.676	625.361	M-H	0.799	<0.001	1.997	↑
	LPE 16:1	ESI-	444.569	450.263	M-H	0.803	<0.001	1.733	↑
	LPC 20:3	ESI-	405.790	590.235	M+FA-H	0.976	<0.001	1.525	↑
	LPC 15:0	ESI+	487.913	504.309	M+Na	0.634	<0.001	1.675	↑
	SM d34:0	ESI+	1118.730	705.591	M+H	0.851	<0.001	1.360	↑
	SM d36:2	ESI+	812.360	729.590	M+H	0.983	0.002	1.203	↑
	SM 34:1	ESI+	729.672	725.557	M+Na	1.034	<0.001	1.383	↑

"↑": Compared with HC group, the differential metabolites were significantly increased in DMD group

"↓": Compared with HC group, the differential metabolites were significantly decreased in DMD group

7. Exemple publication utilisant la bioinfo

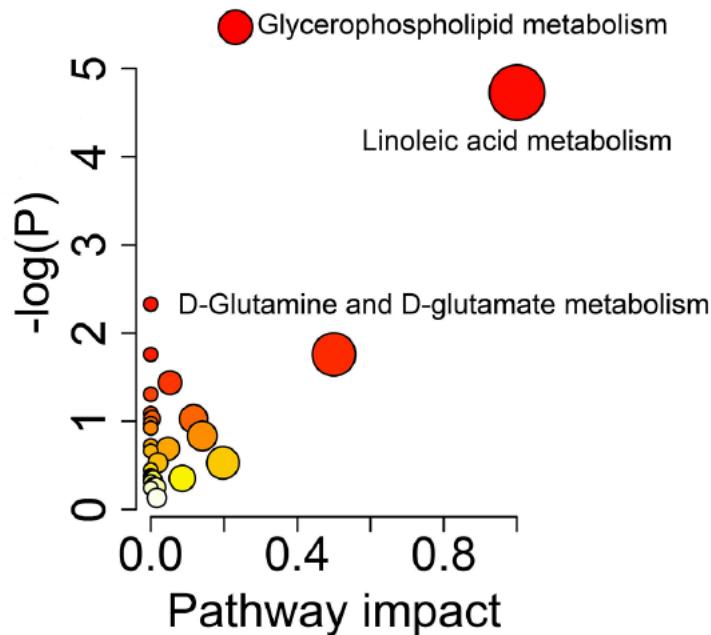


Fig. 5 Bubble diagram of metabolic pathways between DMD and HC groups

Table 3 Significantly altered metabolic pathways between DMD and HC groups

Pathway name	KEGG.id	-log(P)	Impact	Hits
Linoleic acid metabolism	Hsa00591	4.73	1	2
D-Glutamine and D-glutamate metabolism	Has00471	1.76	0.5	1
Glycerophospholipid metabolism	Hsa00564	5.47	0.23	5
Alanine, aspartate and glutamate metabolism	Hsa00250	0.53	0.20	1

7. Exemple publication utilisant la bioinfo

Conclusions

Overall, our study demonstrated the abnormal metabolism of amino acids, energy, and lipids in patients with DMD, consistent with pathological features, such as recurrent muscle necrosis and regeneration, interstitial fibrosis, and fat replacement. In addition, we also identified a number of differential metabolites associated with gut microbiota, which may be related to nutritional disorders and intestinal muscle dysfunction in DMD patients. Although our study provides a new research strategy for the pathogenesis of DMD, there are some limitations. First, the sample size was small, so we hope to conduct a multi-center study with a large sample size in a later stage to reduce sampling error. Second, due to the different types, treatment courses, and doses of corticosteroids used by DMD group in this article, we were unable to completely distinguish the corticosteroid-treated group from the untreated group using PCA. Therefore, we could not obtain differences in disease metabolism at corticosteroid treated or nontreated conditions. Although we cannot separate DMD patients into treated and untreated group, this article can still be considered as the first exploratory study on metabolic changes in clinical patients with DMD (regardless of medication use) in natural research history. In the future, we will conduct a prospective study with larger samples to focus on drug treatments (such as glucocorticoids, calcium channel

blockers and vitamin D) and explore their impacts on the metabolic spectrum of DMD patients. Furthermore, target validation should be applied in an in-depth study to validate our selected metabolic indicators.

List of abbreviations

DMD	Duchenne muscular dystrophy
HC	healthy control
UPLC-MS/MS	ultra-high performance liquid chromatography-tandem mass spectrometry
IRB	Institutional Review Board
QC	quality control
UPLC	ultra-high performance liquid chromatography
IDA	independent data acquisition
KEGG	Kyoto Encyclopedia of Genes and Genomes
HMDB	Human Metabolome Database
PCA	principal component analysis
PLS-DA	partial least-squares discriminant analysis
VIP	variable importance in projection
ANOVA	analysis of variance
LDS	least significant difference
PC	phosphatidylcholines
LPE	lysophosphatidylethanolamines
SM	sphingomyelin
FDR	false discovery rate
GLS	glutaminases
LPC	lysophosphatidylcholine
PA	phosphatidic acid
PS	phosphatidylserine
PAGln	phenylacetylglutamine
FXR	farnesoid X receptor
GPBAR-1	G-protein-coupled bile acid receptor-1
GUDCA	glycoursoodeoxycholic acid
FGF	fibroblast growth factor

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Not applicable.

MERCI à tous!

umr1087.univ-nantes.fr



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Inserm UMR 1087 / CNRS UMR 6291
Nantes, France



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