Gestion de l’information de séquençage numérique

Guy Cochrane

EMBL-EBI

CCTAACCAAGTACTACTTAAAATCAGTAAACTGCCACTCCCTCCCCACTACCCTATATGA
AAATAATTAAAAAAGCTCCTACGCTTCTTTTTTTGACCGCCCTCTACCTCTTTTCACAAA
GGCTATCAACTCGAATTTTCTATTGCTTTATTTTATCAAAAAAAACATAAAAATTTTTGT
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TGAACATAACATGCAATTGTTTTATTTGGACCAAATAGAATGAGTAAATTATATGAATGA
TTAT

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Une séquence isolée

```
CCTAACCAAGTACTACTTAAAATCAGTAAACTGCCACTCCCTCCCCACTACCCTATATGA
AAATAATTAAAAAAGCTCCTACGCTTCTTTTTTTGACCGCCCTCTACCTCTTTTCACAAA
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TCTATGGACACGACTCGAAGATTCATTCATCAATTGGATCGAACGGGTACCTGGCGGCTG
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TGAACATAACATGCAATTGTTTTATTTGGACCAAATAGAATGAGTAAATTATATGAATGA
TTAT
```

.. ne peut être interprétée

.. a peu de chances d’être révélatrice

https://www.ebi.ac.uk/ena/browser/view/HM544578.1

EMBL-EBI

Une collection de séquences

260 millions de séquences

.. peut être interprétée

.. suscite la découverte

EMBL-EBI

Collaboration des bases de données internationales de séquences nucléotidiques (International Nucleotide Sequence Database Collaboration, INSDC)

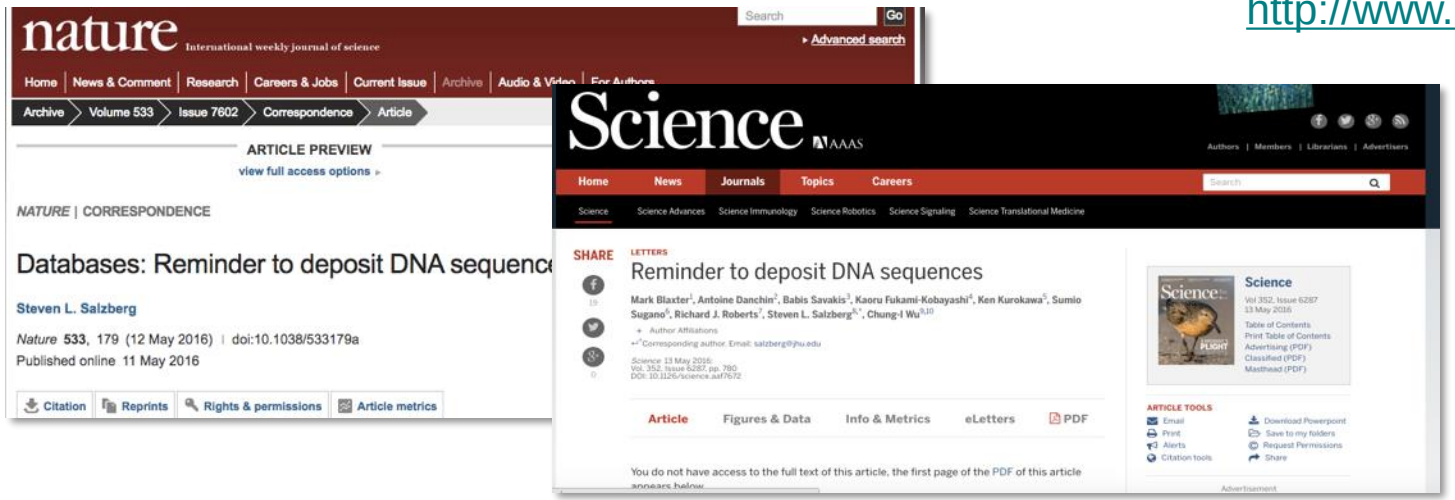
Valeurs

- un accès ouvert à tous
- complet et planétaire
- couvrant les branches des sciences de la vie
- une base de données d'archives
- un forum public pour le processus scientifique

Organisation

- créée au début des années 1980
- en train de réaliser un investissement important
- structure et gouvernance
- un modèle de collaboration scientifique

http://www.insdc.org/

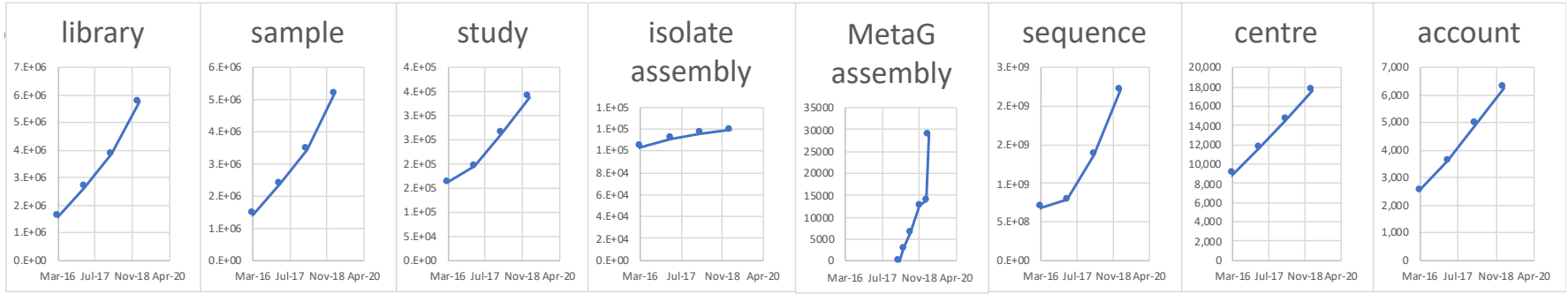


Instruments

- un échange de données régulier
- un régime d'adhésion
- normes de données
- accord de soumission obligatoire
- services et logiciels (au niveau des nœuds)

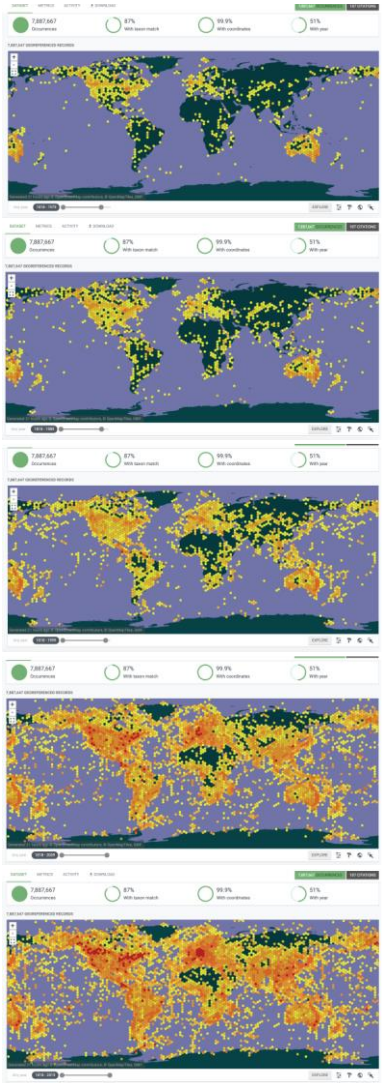
Portée

- **Débit :** 1 nouveau jeu de données toutes les 6 minutes
- **Données** : $2x10^9$ séquences et $1x10^{16}$ paires de base de données de lecture parmi $2x10^6$ taxons
- **Usage** : 2 000 fournisseurs ; 10 000 consommateurs par mois ; 10 millions de visites par mois, dont plusieurs fois dans le monde
- **Soutien** : 46 tickets par jour et une formation en personne dispensée à plus de 350 utilisateurs par an
- **Infrastructure :** comprend des centaines de pétaoctets de stockage, ~50 millions de USD/an



- **Données sur la biodiversité, par exemple**
  - 7,2 millions d'enregistrements pointent vers des musées d'histoire naturelle, des collections culturelles et des biobanques
  - 7,9 millions de séquences avec coordonnées géographiques
  - 2,1 millions de séquences ont une annotation de nom de lieu
  - 1 million d'ensembles de données brutes de séquençage ont des coordonnées

1980

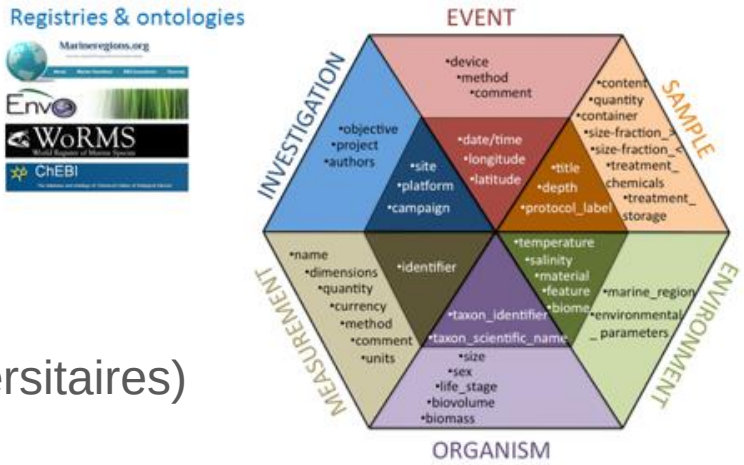


2020

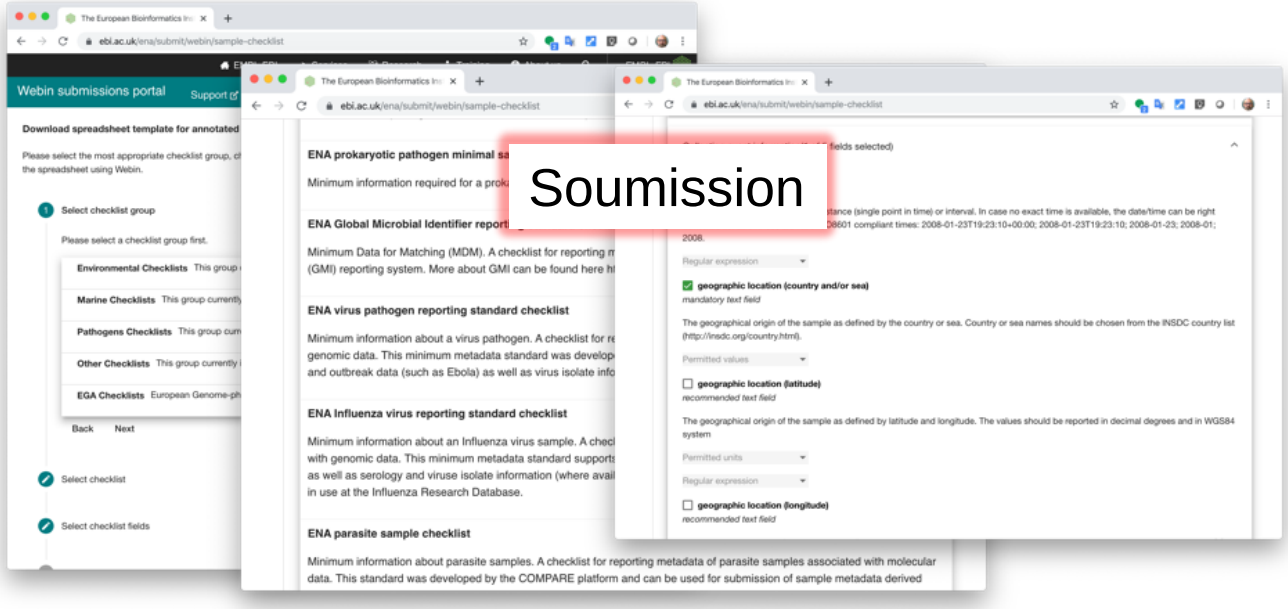
https://doi.org/10.15468/cndomv, avec l'aimable autorisation du Centre EMBL-EBI

Le cycle des données de la Collaboration internationale en bases de données de séquences nucléotidiques (INSDC)

- Fournisseur de données
  - Validation, organisation, ajout de structure et édition
  - Respect des normes et des conventions établies
  - Liens vers des données externes (p. ex., publications universitaires)
- Base de données
  - Édition et intégration dans l'ensemble du corpus
  - Indexation pour la découverte et la réutilisation
  - Des services ouverts, librement accessibles au monde entier



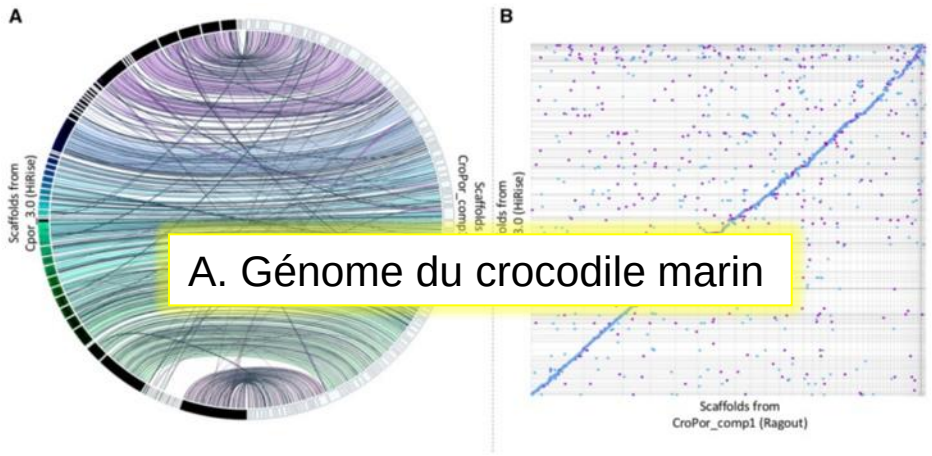
Ten Hoopen P., Pesant S., Kottmann R., Kopf A., Bicak M., Claus S., Deneudt K., Borremans C., Thijsse P., Dekeyzer S., Schaap D., **Bowler C.**, Glöckner F.O., **Cochrane G.** Data standards for Marine Microbial Biodiversity, Bioinformatics and Biotechnology (M2B3) *Standards in Genomic Sciences* 10:20 doi:10.1186/s40793-015-0001-5 (2015). http://www.standardsingenomics.com/content/10/1/20



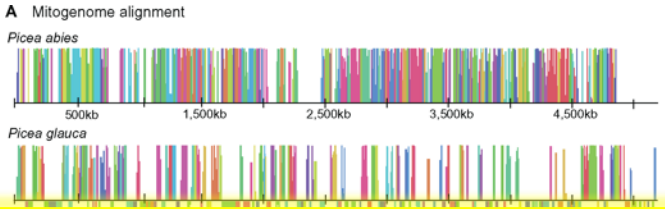
EMBL-EBI

Utilisation directe

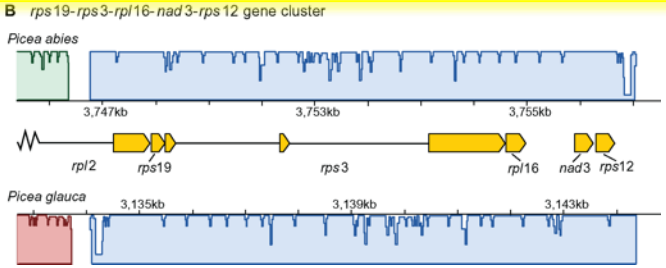
- 1 600 publications citant les accessions de séquences par mois en 2020

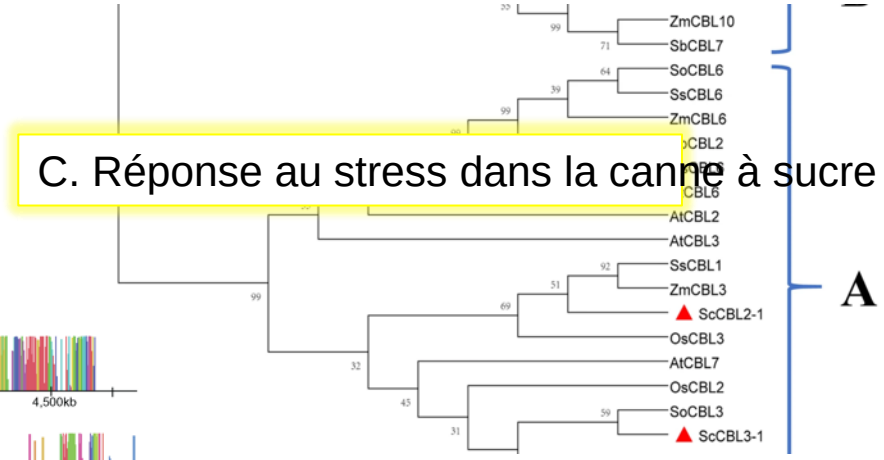


A. Génome du crocodile marin

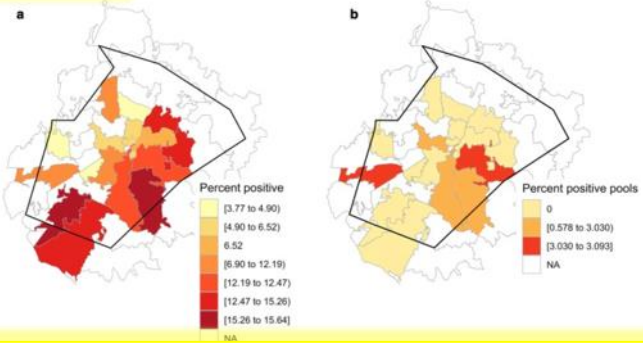


B. Biologie évolutive de l'épicéa de Norvège

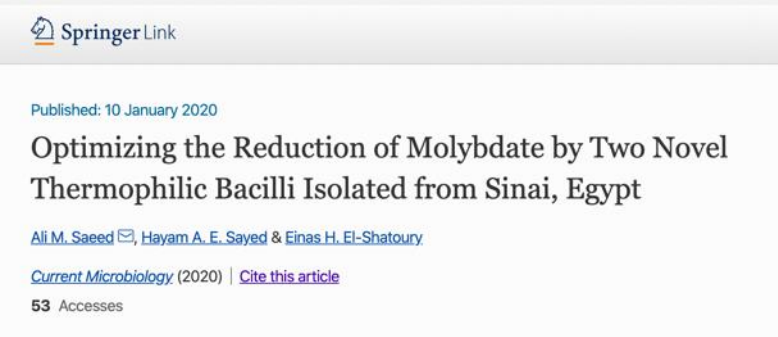




C. Réponse au stress dans la canne à sucre



E. Ver du cœur transmis par les moustiques chez le chien


Springer Link

Published: 10 January 2020

Optimizing the Reduction of Molybdate by Two Novel Thermophilic Bacilli Isolated from Sinai, Egypt

Ali M. Saeed, Hayam A. E. Sayed & Einas H. El-Shatoury

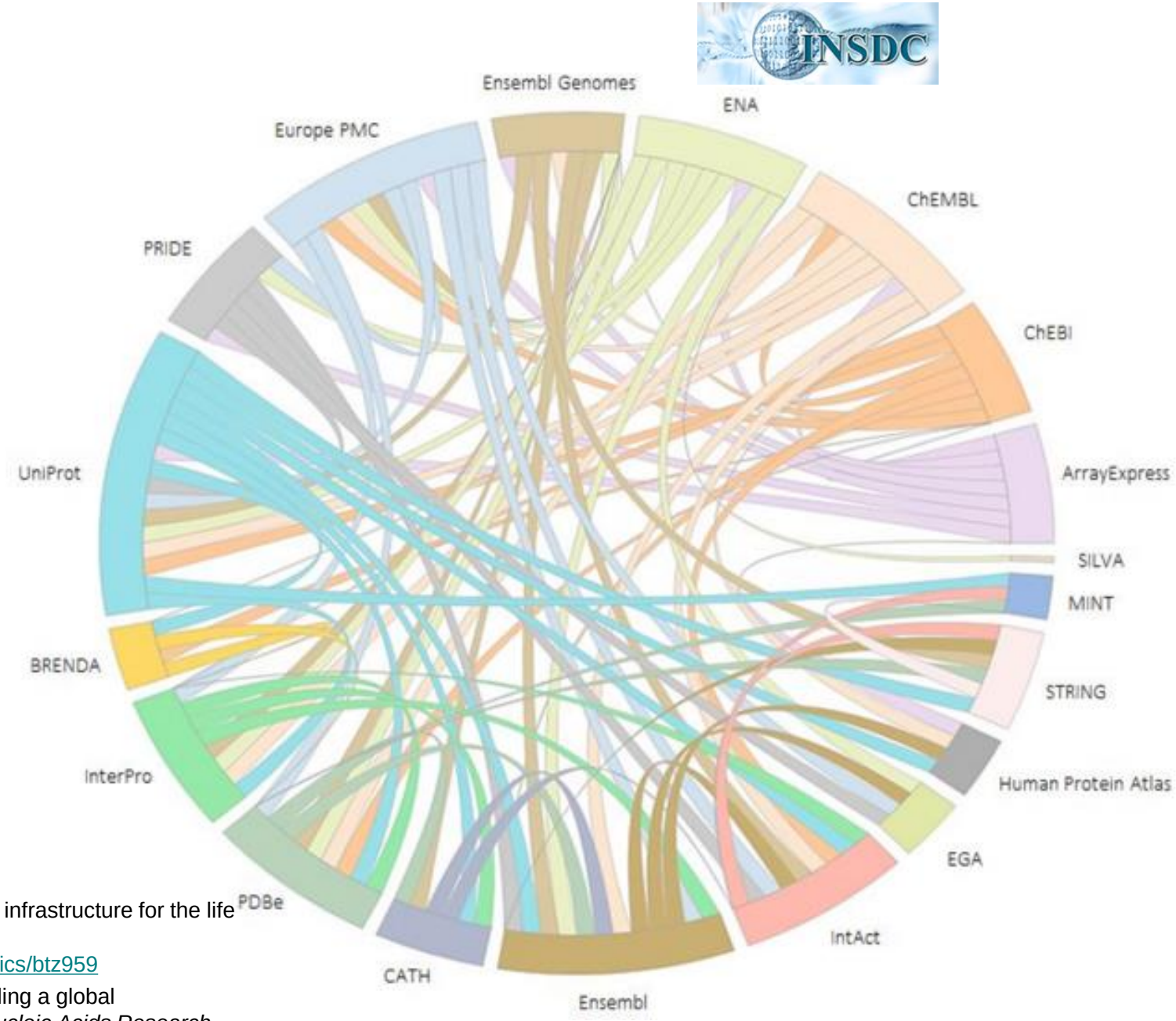
Current Microbiology (2020) | Cite this article

53 Accesses

D. Biorémédiation du molybdène

A. Ghosh A et al., Genome Biol Evol. 2020 Jan;12(1) 3635-3646. doi:10.1093/gbe/evz269. PMID : 31821505; © The Author(s) 2020; http://creativecommons.org/licenses/by/4.0/
B. Sullivan AR et al., Genome Biol Evol. 2020 Jan;12(1) 3586-3598. doi:10.1093/gbe/evz263. PMID : 31774499; © The Author(s) 2020; http://creativecommons.org/licenses/by-nc/4.0/
C. Su W et al., Sci Rep. 2020 Jan;10(1) 167. doi:10.1038/s41598-019-57058-7. PMID : 31932662; http://creativecommons.org/licenses/by/4.0/
D. Saeed AM et al., Curr Microbiol. 2020 Jan . doi:10.1007/s00284-020-01874-y. PMID : 31925514.
E. Spence Beaulieu MR et al., Parasit Vectors. 2020 Jan;13(1) 12. doi:10.1186/s13071-019-3874-0. PMID : 31924253; PMCID: PMC6953185. http://creativecommons.org/licenses/by/4.0/; http://creativecommons.org/publicdomain/zero/1.0/

Portée des données

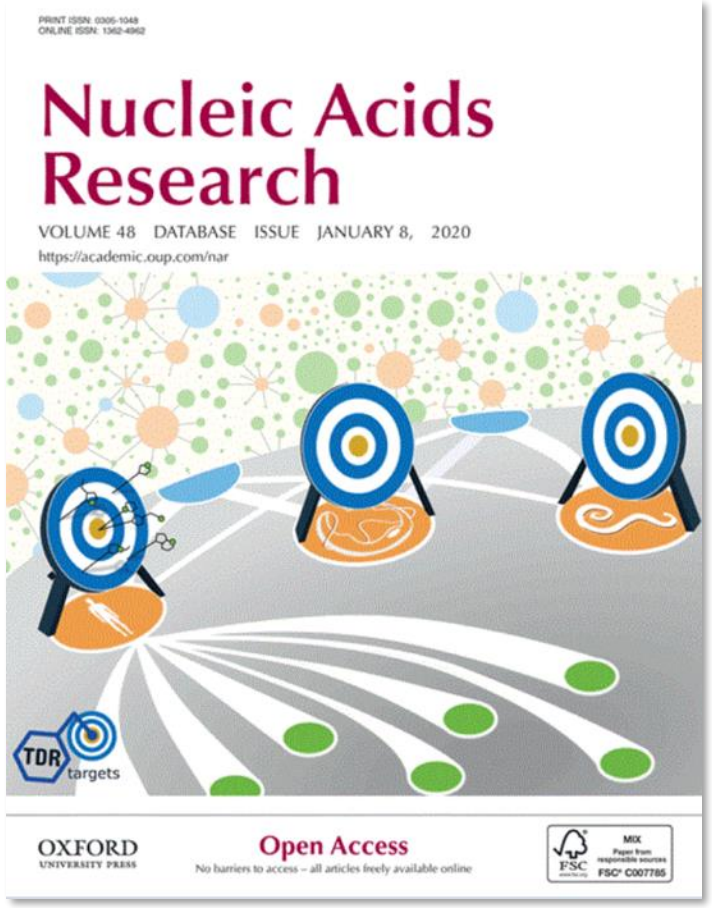


Core Data Resource

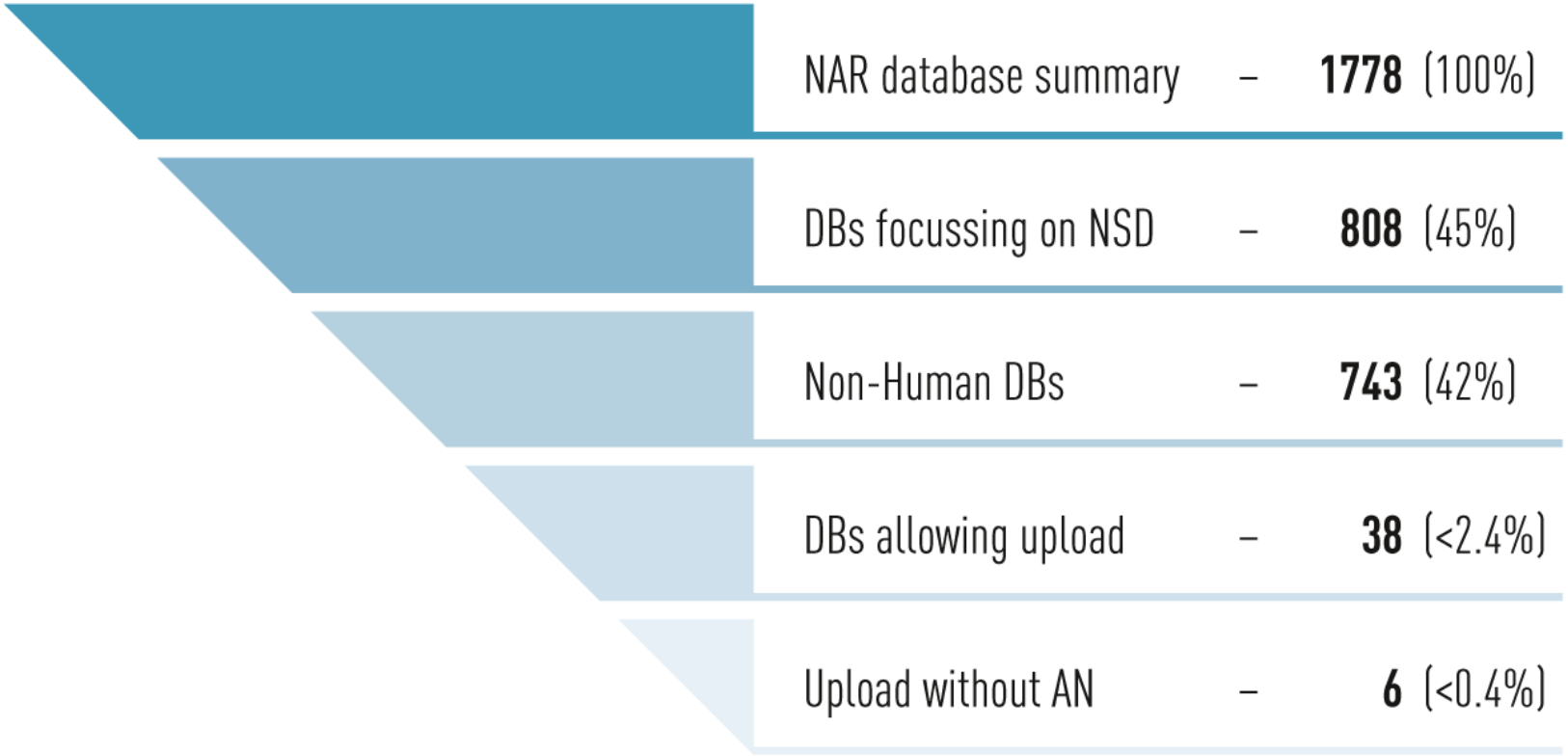
Drysdale *et al.* (2020) The ELIXIR Core Data Resources: fundamental infrastructure for the life sciences. *Bioinformatics*, 2020, 1–7; http://doi.org/10.1093/bioinformatics/btz959

Cook *et al.* (2020) The European Bioinformatics Institute in 2020: building a global infrastructure of interconnected data resources for the life sciences. *Nucleic Acids Research* 48:D17-D23; http://doi.org/10.1093/nar/gkz1033

Divers



Public database inventory



Rohden F, Huang S, Dröge G, Hartman Scholz A, and contributing authors (2019). Combined study in DSI in public and private databases and DSI traceability. https://www.cbd.int/abs/DSI-peer/Study-Traceability-databases.pdf

EMBL-EBI

Conditions d'utilisation

Data Services

1. The online data services and databases of EMBL-EBI are generated in part from data contributed by the community who remain the data owners.

2. When you contribute scientific data to a database through our website or other submission tools this information will be released at a time and in a manner consistent with the scientific data and we may store it permanently.

3. EMBL-EBI itself places no additional restrictions on the use or redistribution of the data available via its online services other than those provided by the original data owners.

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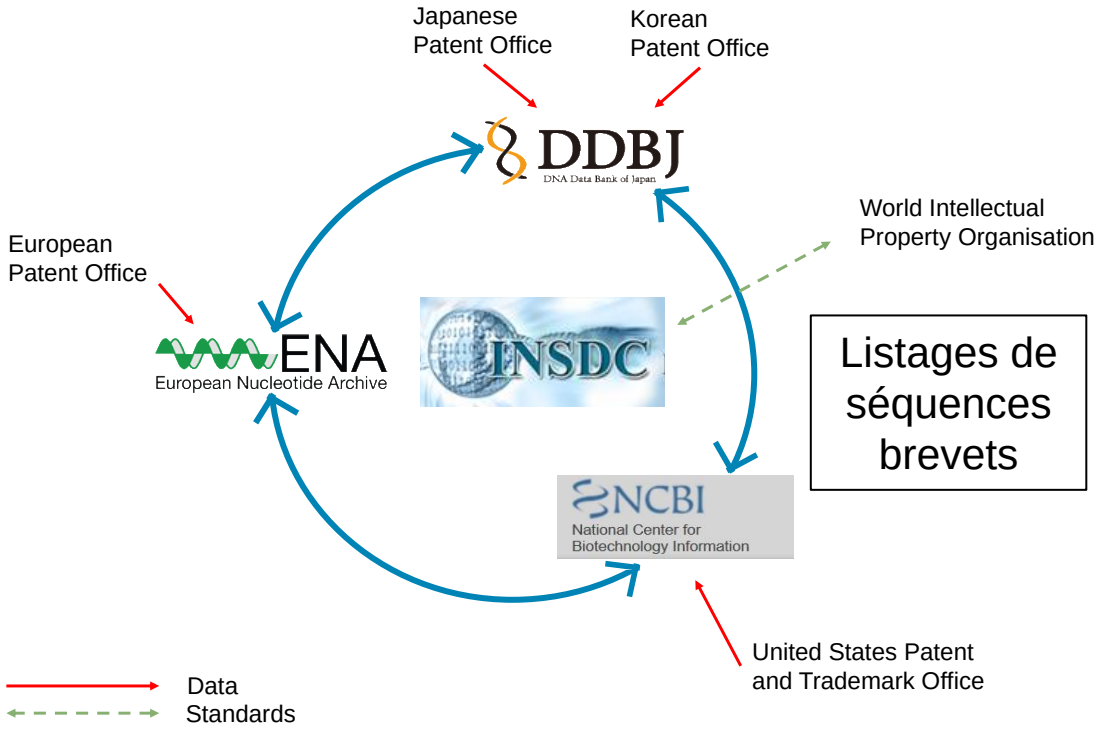
5. The original data may be subject to rights claimed by third parties, including but not limited to, patent, copyright, other intellectual property rights, biodiversity-related access and benefit-sharing rights. For the specific case of the EGA database and human data consented for biomedical research, these rights may be formalised in Data Access Agreements. It is the responsibility of users of EMBL-EBI services to ensure that their exploitation of the data does not infringe any of the rights of such third parties.

https://www.ebi.ac.uk/about/terms-of-use



Listages de séquences brevets

EXAMPLES OF PREPUBLICATION DATA-RELEASE GUIDELINES

Project type	Prepublication data release recommended	Prepublication data release optional
Genome sequencing	Whole-genome or mRNA sequence(s) of a reference organism or tissue	Sequences from a few loci for cross-species comparisons in a limited number of samples
Polymorphism discovery	Catalogue of variants from genomic and/or transcriptomic samples in one or more populations	Variants in a gene, a gene family or a genomic region in selected pedigrees or populations
Genetic association studies	Genomewide association analysis of thousands of samples	Genotyping of selected gene candidates
Somatic mutation discovery	Catalogue of somatic mutations in exomes or genomes of tumour and non-tumour samples	Somatic mutations of a specific locus or limited set of genomic regions
Microbiome studie	Whole-genome sequence of microbial	Sequencing of target locus in a limited me samples
RNA p		pression profiles of al system(s)
Proteomic studies	Mass spectrometry data sets from large panels of normal and disease tissues	Mass spectrometry data sets from a well-defined and limited set of tissues
Metabolomic studies	Catalogue of metabolites in one or more tissues of an organism	Analyses of metabolites induced in a perturbed biological system(s)
RNAi or chemical library screen	Large-scale screen of a cell line or organism analysed for standard phenotypes	Focused screens used to validate a hypothetical gene network
3D-structure elucidation	Large-scale cataloguing of 3D structures of proteins or compounds	3D structure of a synthetic protein or compound elucidated in the context of a focused project

Principes de Toronto

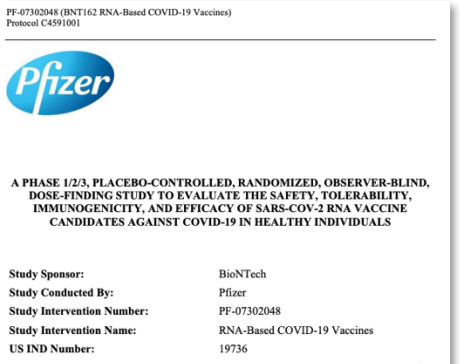
Nature **volume 461**, pages168–170(2009)

Obligations relatives à l'APA dans les accords existants

Données ouvertes relatives au COVID-19

Viral Sequences	Host Sequences	Expression	Proteins	Biochemistry	Literature
All (262,317)	All (1,046)	All (82)	All (712)	All (2,255)	All (212,276)
Sequences (27,649)	Human studies (controlled access) (4)	Gene expression (4)	Protein sequences (57)	Pathways (16)	Coronaviruses (109,172)
Raw reads (125,159)	Human reads (consented for full access) (784)	Gene expression experiments (23)	Protein families (126)	Interactions (1,812)	Diseases (96,222)
Sequenced samples (96,632)	Other species reads (250)	Single cell expression (4)	Protein structures - Knowledge Base (7)	Complexes (29)	Related viruses and diseases (2,109)
Studies (163)	Association studies (8)	Single cell expression experiments (19)	Protein structures (260)	Compound document (8)	Genes, receptors and antibodies (4,773)
Genes (22)		Protein expression experiments (32)	Electron microscopy density maps (250)	Drug targets (390)	
Browser (1)			Electron microscopy public image archive (12)		
Variants (12,691)					

https://www.covid19dataportal.org/


PF-07302048 (BNT162 RNA-Based COVID-19 Vaccines)
Protocol C4591001

Pfizer

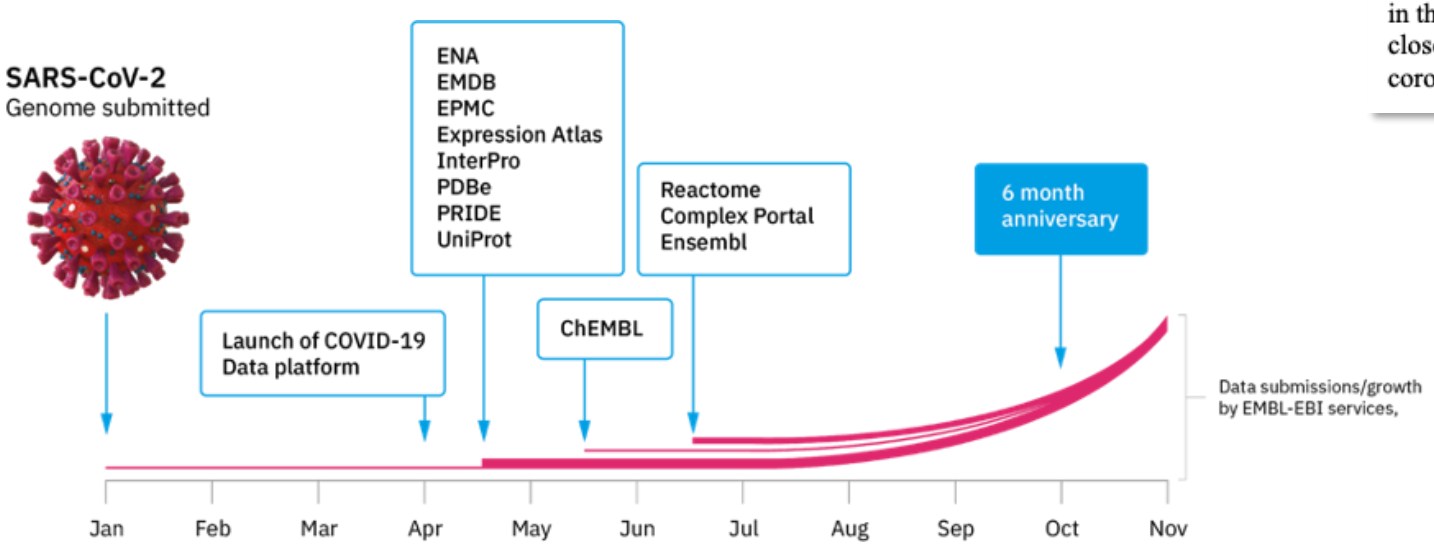
A PHASE 1/2/3, PLACEBO-CONTROLLED, RANDOMIZED, OBSERVER-BLIND, DOSE-FINDING STUDY TO EVALUATE THE SAFETY, TOLERABILITY, IMMUNOGENICITY, AND EFFICACY OF SARS-COV-2 RNA VACCINE CANDIDATES AGAINST COVID-19 IN HEALTHY INDIVIDUALS

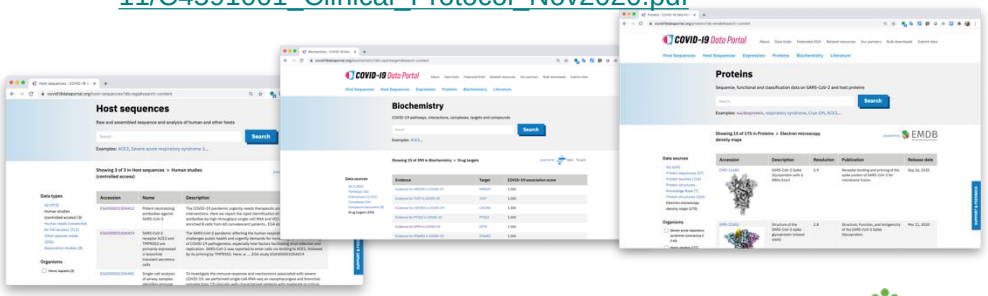
Study Sponsor:	BioNTech
Study Conducted By:	Pfizer
Study Intervention Number:	PF-07302048
Study Intervention Name:	RNA-Based COVID-19 Vaccines
US IND Number:	19736

2.2. Background

In December 2019, a pneumonia outbreak of unknown cause occurred in Wuhan, China. In January 2020, it became clear that a novel coronavirus (2019-nCoV) was the underlying cause. Later in January, the genetic sequence of the 2019-nCoV became available to the World Health Organization (WHO) and public (MN908947.3), and the virus was categorized in the *Betacoronavirus* subfamily. By sequence analysis, the phylogenetic tree revealed a closer relationship to severe acute respiratory syndrome (SARS) virus isolates than to another coronavirus infecting humans, the Middle East respiratory syndrome (MERS) virus.

https://pfe-pfizercom-d8-prod.s3.amazonaws.com/2020-11/C4591001_Clinical_Protocol_Nov2020.pdf





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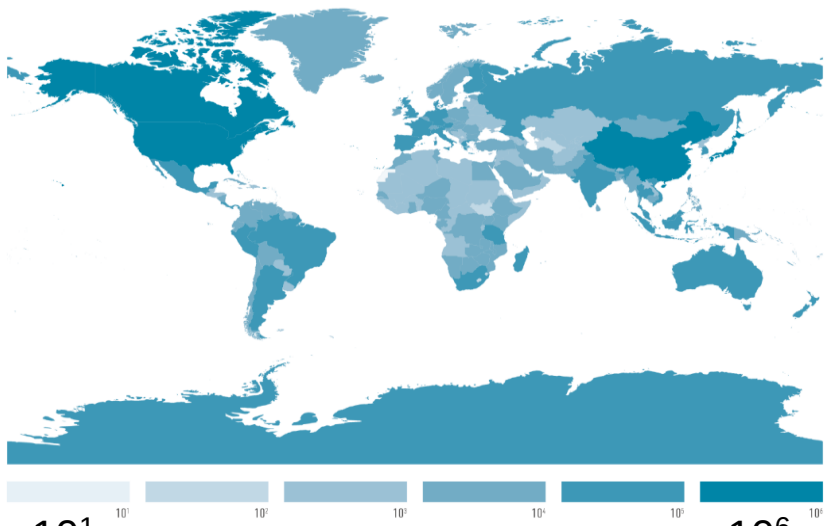
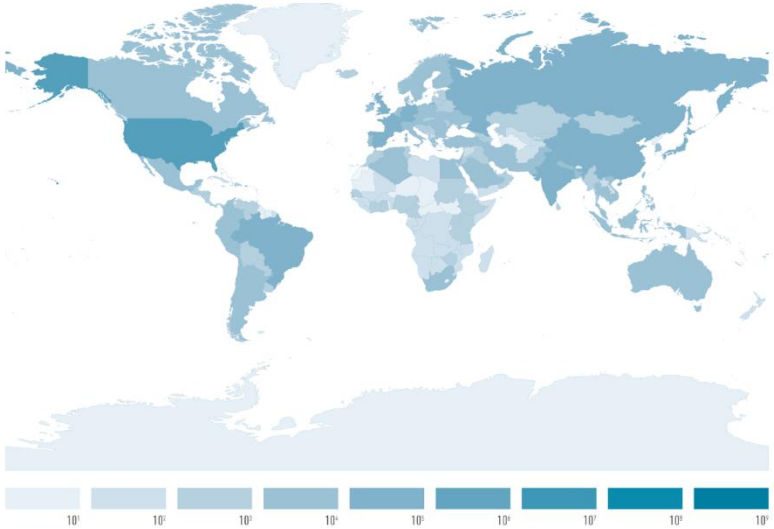
Sens du flux des avantages non monétaires

What is the country of origin for non-human NSD?

1.	China	18.23 %
2.	United States	17.39 %
3.	Canada	9.10 %
4.	Japan	7.24 %
5.	India	3.46 %
6.	Australia	2.66 %
7.	Mexico	2.54 %
8.	Brazil	2.30 %
9.	Germany	1.83 %
10.	Spain	1.58 %



Pays d'origine de l'ISN

Pays utilisateurs de l'ISN

1.	United States	22.69 %
2.	China	15.42 %
3.	India	6.16 %
4.	Japan	3.97 %
5.	Germany	3.67 %
6.	United Kingdom	3.45 %
7.	France	2.84 %
8.	Brazil	2.83 %
9.	Spain	2.31 %
10.	Russian Federation	2.25 %