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Welcome to

genomics aotearoa

Level 1 NCEA Genomics workshop for CB1.3

Auckland region

Karakia

Tukua te wairua kia rere ki ngā taumata	*Allow one's spirit to exercise its potential*
Hai ārahi i ā tātou mahi **Me tā tātou whai i ngā tikanga a rātou mā**	*To guide us in our work as well as in our pursuit of our ancestral traditions*
Kia mau kia ita **Kia kore ai e ngaro** **Kia pupuri** **Kia whakamaua**	*Take hold and preserve it* *Ensure it is never lost* *Hold fast.* *Secure it.*
Kia tina! TINA! Hui e! TĀIKI E!	*Draw together! Affirm!*

CB1.3 Essentials and Beyond: Teach Today and for Tomorrow

Demonstrate understanding of genetic variation in relation to an identified characteristic

Dr Wilda Laux

CB 1.3 / AS 92022 Demonstrate understanding of genetic variation in relation to an identified characteristic

Genetic variation:

What, why

Source = how: mutation, meiosis, fertilisation, small population size, different rates of survival, migration, non-random mating

Gene tracking methods:

Why: Genetic relationships (gene therapy, agriculture, species conservation…)

How: use genetic marker / specific DNA sequences to produce pedigree, phylogenetic tree

AS92022 Chem & Bio 1.3 changes

Te Poutāhū
Curriculum Centre

From …

Explanatory Note 3

For the purpose of this achievement standard, a *gene tracking methodology* identifies the presence or absence of a gene, genetic marker, or DNA sequence within an individual or population.

Examples of a gene tracking methodology include:

- Phylogenetic trees or pedigree charts
- genetic markers
- specific DNA sequences
- Punnett squares.

To …

Explanatory Note 3

For the purpose of this achievement standard, a gene tracking methodology identifies the presence or absence of one or more genes, genetic markers, or DNA sequences within an individual or population.

TODAY

Genetic variation

What When How Where *Why*

Source and nature of Genetic variation
For individuals/populations

Gene tracking methods

What When How Where *Why*

Between individuals/populations
Determine relationships

Level 1

TOMORROW

Gene expression

Genetic transfer

Human evolution

Speciation

Level 2-3

TEACH CONCEPTS

Teach for Tomorrow

Contexts and Connect

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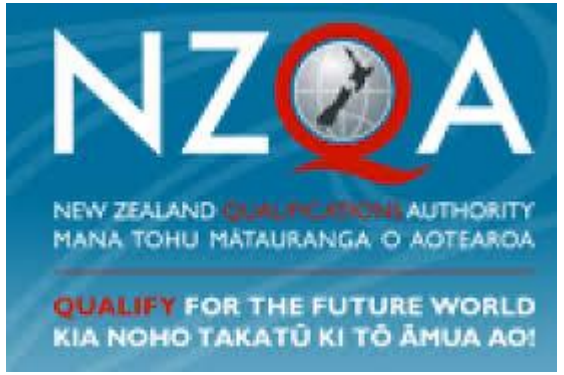
Lesson	Lesson title	Learning outcomes	Strategies	Suggested Resources
1	**Introduction to living things**	Introduction to genetics Review the different characteristics of an organism	Motivational video: while the video runs, comment/pause on areas of Biology and keywords used in this unit throughout the video and focus on the main ideas/keywords related to genetics/genomics Review 'atom to organism' in the context of a chosen species that you will use for later lessons, e.g., Tara iti (atom, molecule, cell, tissue, organ, organ system, organism = Tara iti) Review MRSGREN, the 7 characteristics of living things **(reiterate that respiration is not breathing)**	https://www.youtube.com/watch?v=B_PQ8qYtUL0&t=75s (introduction to genetics) https://chem.libretexts.org/Under_Construction/iLearn_Collaborative/Copy_of_DCW-Biology-Semester-2_Curated.imscc/01%3A_Course_Content/03%3A_Unit_9%3A_Humans/00%3A_Week_10%3A_Homeostasis/01%3A_Levels_of_Organization (atom to organism flowchart) Complete task 3 of Appendix 1
2	**Suggestion: Looking at animal vs plant cells**	Differentiate between animal and plant cells	PRACTICAL session: microscopy with onion cell vs animal slides	Microscopes, onions, slides, coverslips, iodine, water, forceps, prepared animal tissue slides https://hi-static.z-dn.net/files/d5d/69a5d971d7970df3681c933cd0df845c.doc WS_Onion cell lab.doc


BEANZ
Biology Educators' Association of New Zealand
Te Rōpū Whakaako Koirora o Aotearoa


NZASE
New Zealand Association of Science Educators


NZQA
NEW ZEALAND QUALIFICATIONS AUTHORITY
MANA TOHU MĀTAURANGA O AOTEAROA
QUALIFY FOR THE FUTURE WORLD
KIA NOHO TAKATŪ KI TŌ ĀMUA AO!


genomics aotearoa


GZ SCIENCE
Science and Chemistry Resources for New Zealand Secondary Schools


scipad


MINISTRY OF EDUCATION
TE TĀHUHU O TE MĀTAURANGA


ep
Education Perfect


UNIVERSITY OF AUCKLAND
Waipapa Taumata Rau
NEW ZEALAND


Welcome to No Brain Too Small
Revision Notes, Flash Cards,
Test Yourself Activities, Powerpoints & much more
No Brain Too Small

GENETICS TO GENOMICS

Individual genes
Passing down from parents to offspring (Punnett squares, pedigree, Mendelian principles)

Applications: medicine, agriculture, evolutionary biology

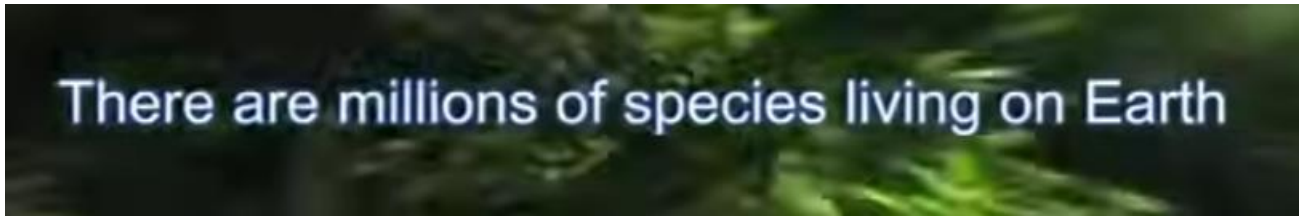
All genes (coding vs non-coding DNA, regulatory sequences) and their interactions with each other and the environment (DNA sequencing, bioinformatics, comparative genomics)

Applications: agriculture, gene therapy, forensics, evolution, …)
COLLABORATION WITH UoA

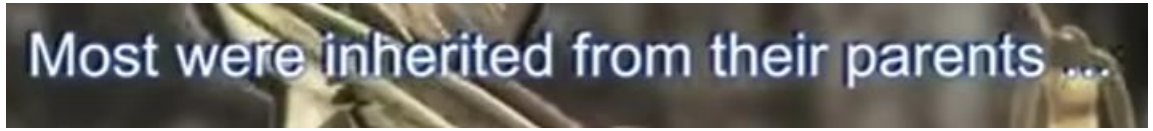
CB1.3 Essentials and Beyond: Teach Today and for Tomorrow

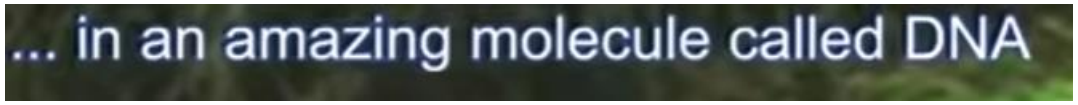
GENETICS

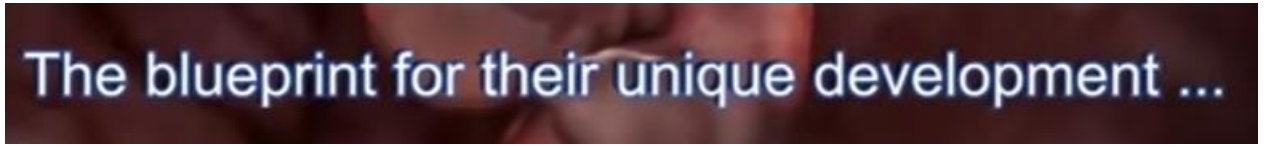
https://www.youtube.com/watch?v=B_PQ8qYtUL0&t=75s



Species live in communities within an ecosystem







CB1.3 Essentials and Beyond: Teach Today and for Tomorrow

Motivational video to introduce Biology
Atom to organism – organisms of choice (local curriculum), e.g., Tara iti, Gentoo penguin (BEANZ)
Living vs non-living (MRSGREN). Use chosen organism vs non-living. (Opportunity to do microscopy)
Classification systems in Biology (dichotomous keys, Taxonomy –apply to chosen organism) (L3)
Levels of organisation in an ecosystem (L3)
Relationships within the ecosystem (L3)
Adaptations of organisms (use chosen organism, adaptations to meet its niche) (L2, L3)
The wonders of DNA (karyotype, DNA, chromosome, gene, allele, base pairing rule)

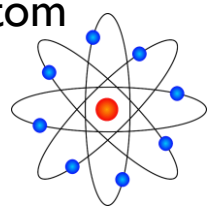
CB1.3 Essentials and Beyond: Teach Today and for Tomorrow

The central dogma of Biology (DNA to protein, codon table)
Types of variation (inherited vs acquired)
Sources of variation (meiosis (vs mitosis), fertilisation, mutation) in sexual reproduction (L2)
Looking through the Genbank database (L3)
Predict inheritance (Punnett squares)
Other sources of variation affecting populations: natural selection, genetic drift, migration, non random mating (L2, L3)
Pedigree charts (tracking through generations)
Use genetic information to compare sequences between species to make a phylogenetic tree
Gene tracking for different purposes, e.g., forensics, conservation, gene therapy…)

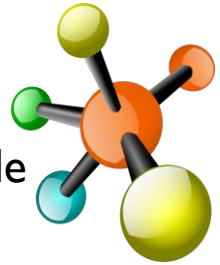
Dr Wilda Laux, 2024

The story of life!

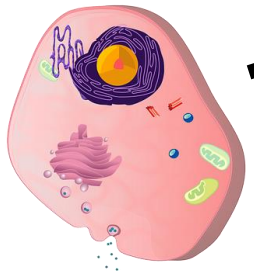
atom

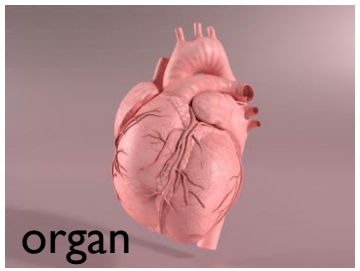


molecule

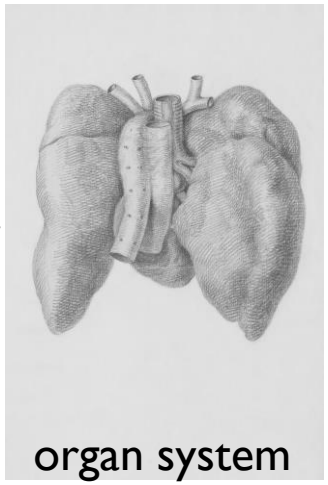


cell

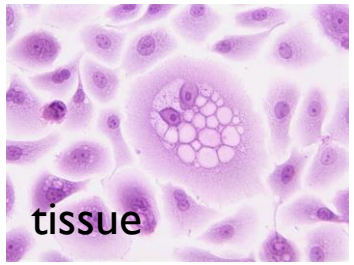




organ



organ system



tissue

Practical: Microscopy

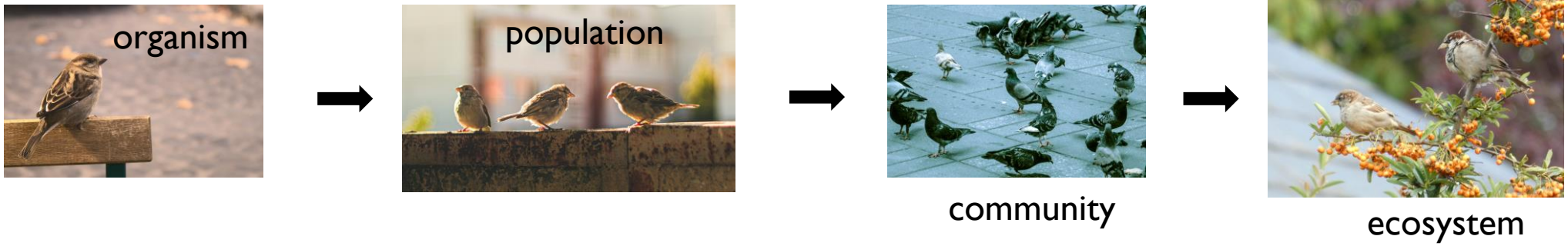
organism

Local curriculum: use a local organism

M R S G R E N

More on classification:
Dichotomous keys
Taxonomy
Use local organisms

The story of life!

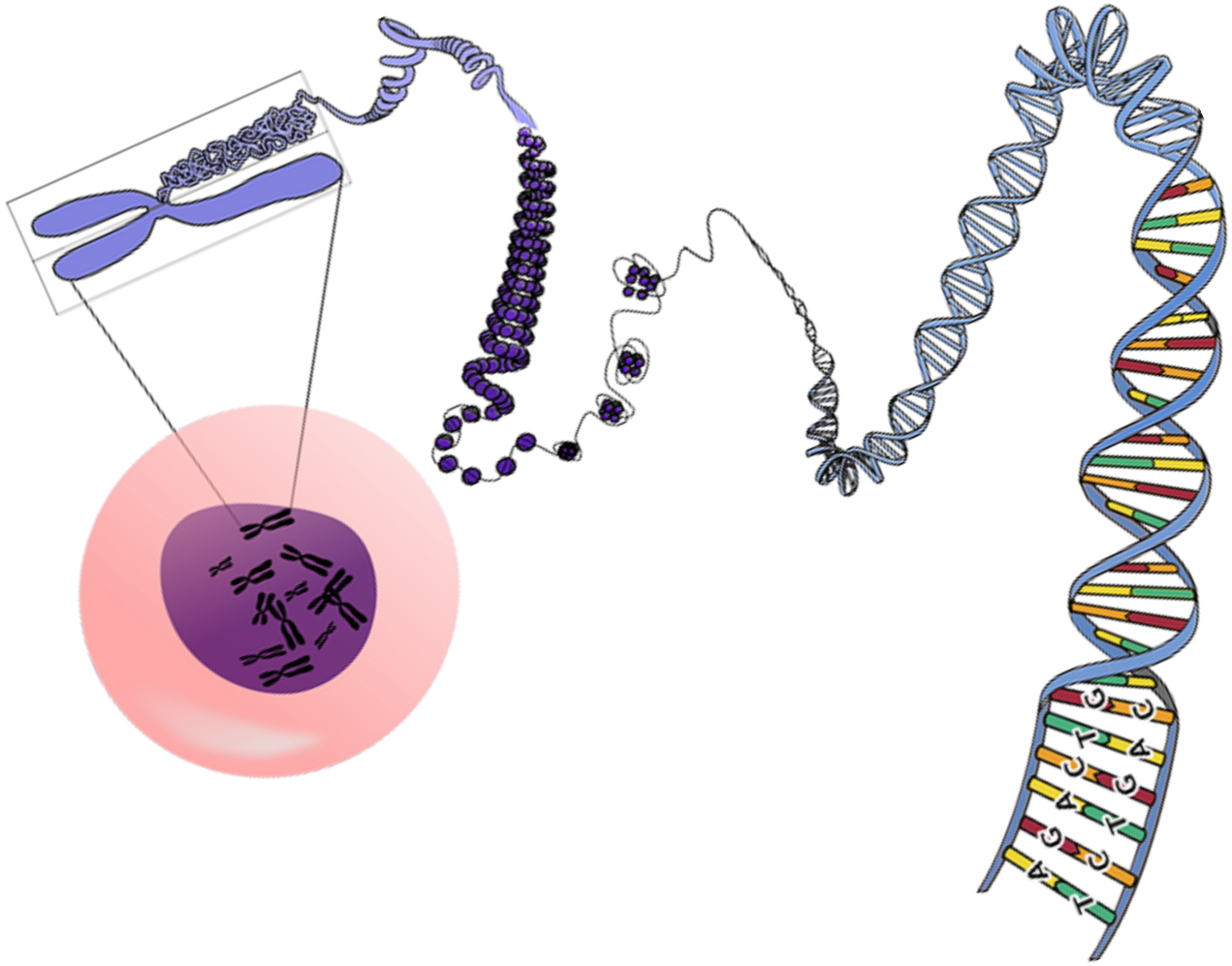


Relationships within the ecosystem (use context of local curriculum)
Adaptations lead to changes in DNA which are inherited

DNA structure

DNA, gene, alleles, chromosome, trait, nucleotide

https://pixabay.com/illustrations/dna-genetics-molecule-biology-5695421/



https://pixabay.com/vectors/genetics-chromosomes-rna-dna-156404/

genomics aotearoa

Each species has different numbers of chromosomes

Each human cell:
3.2 billions (3200 millions) nucleotides (human genome)

DNA is about 2 m long

About 20 000 protein-coding genes (1.5% of the entire genome)

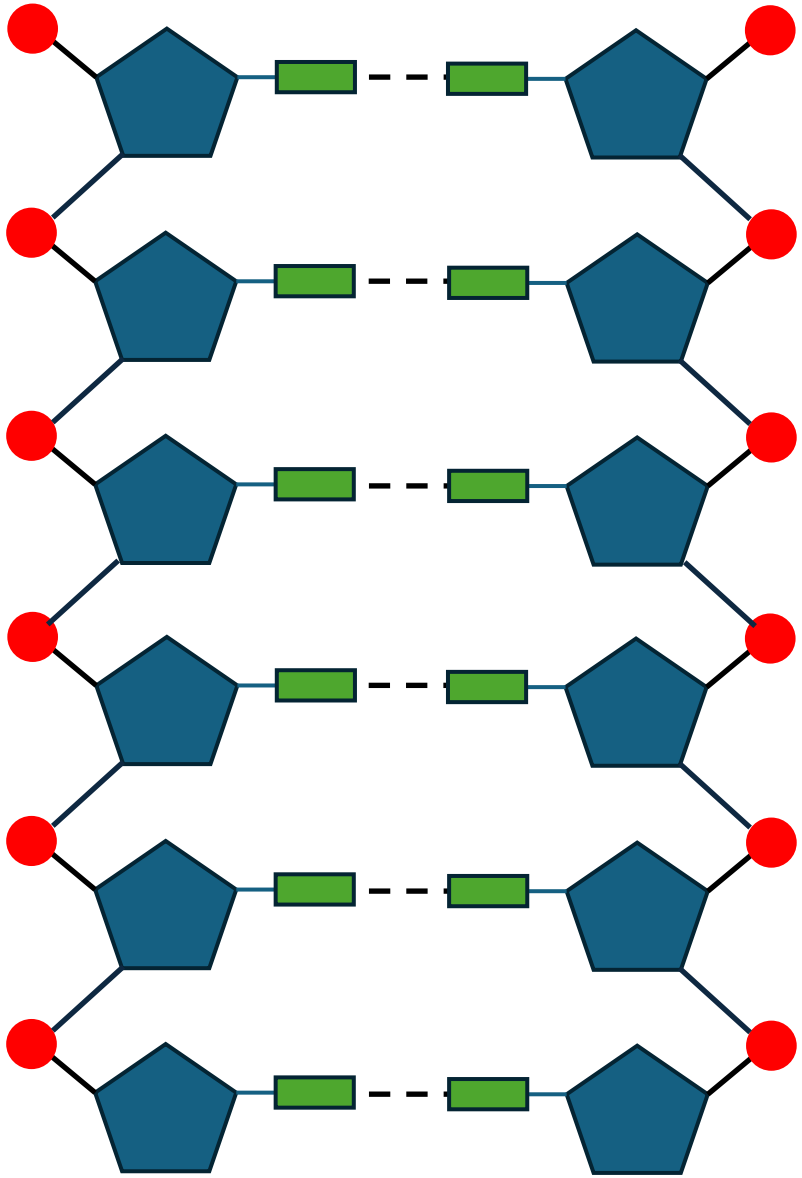
Photos from pixabaycom

Nucleotide

phosphate

Nitrogenous base

Sugar = deoxyribose



DNA to protein – the central dogma

protein

↑ translation

mRNA

↑ transcription

DNA

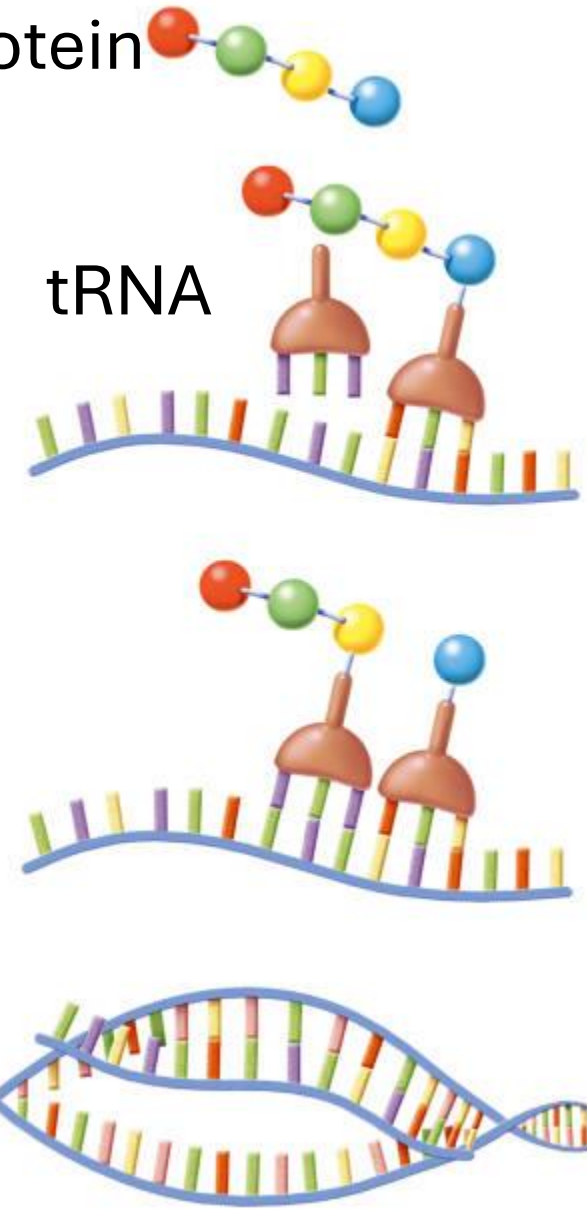
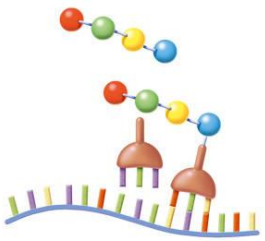
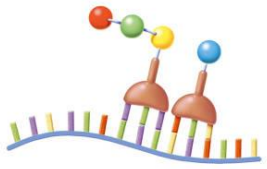


Image from Gettyimages

protein





mRNA

DNA

Gene - section of DNA
Genome – all genes contained in an organism

Gene 1

DNA

Exon 1 | **Intron 1** | **Exon 2** | **Intron 2** | **Exon 3** | **Intron 3**

DNA

promoter

transcription +polyA tail at 3’ and CAP at 5’

5’ CAP

polyA tail 3’

Exon 1 | **Intron 1** | **Exon 2** | **Intron 2** | **Exon 3** | **Intron 3**

Pre mRNA

splicing

CAP+ ATG

TAA/TGA/TAG+polyA tail

Exon 1 | **Exon 2** | **Exon 3**

mRNA

translation

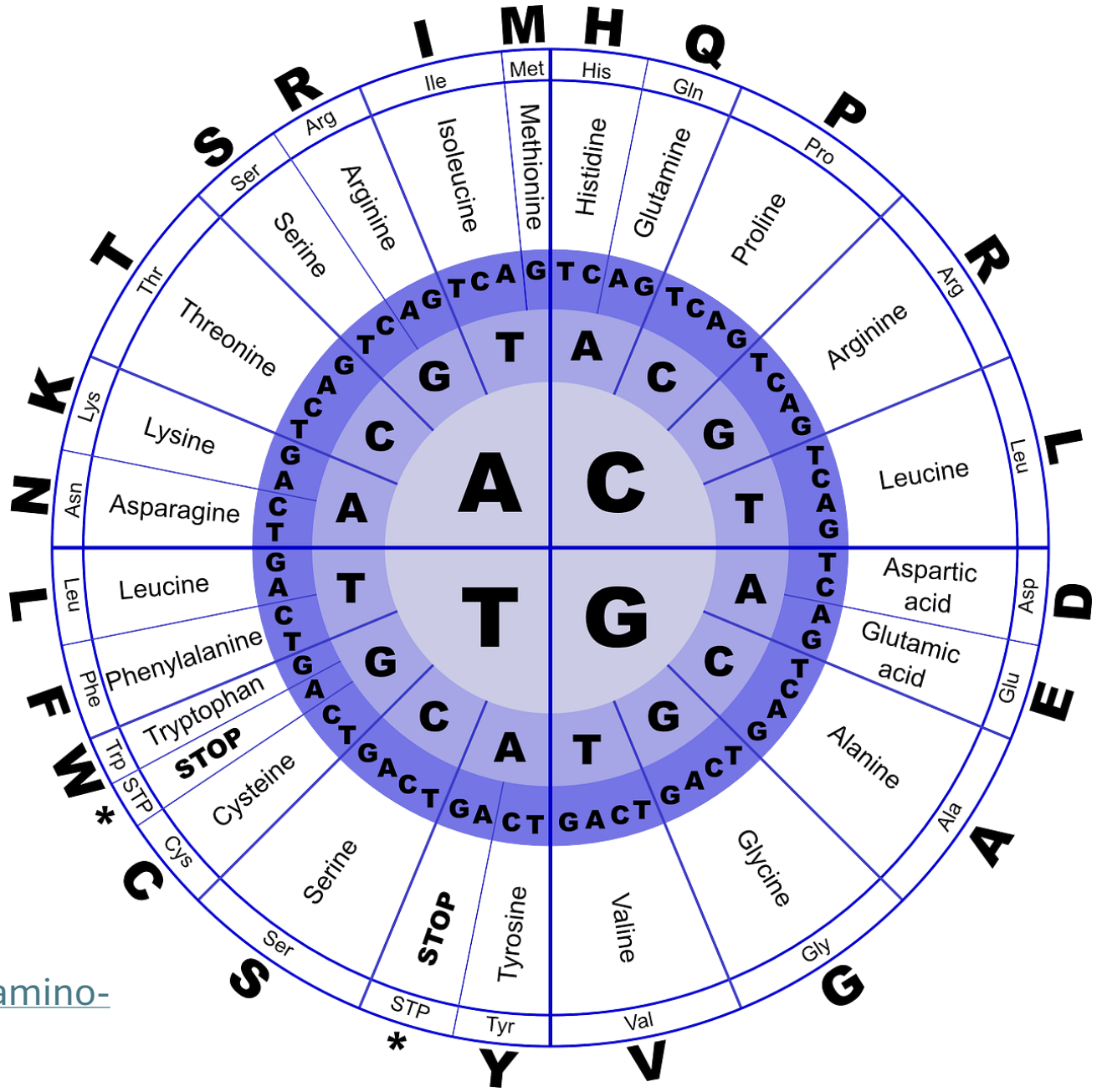
protein

Codon chart

5’ ACA CAT GAG TGC GCC ACC 3’ DNA

RNA

protein



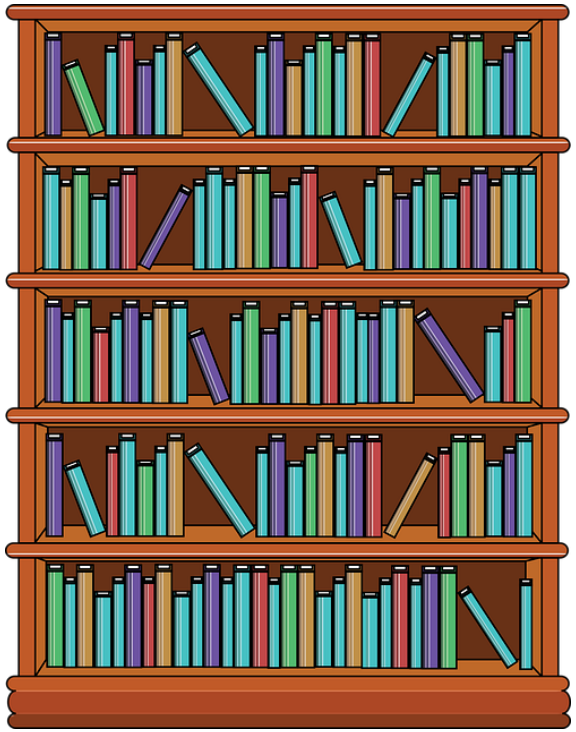
https://pixabay.com/vectors/dna-amino-acids-biology-code-152135/

Variations

- Sources: (meiosis, fertilisation, mutation) in sexual reproduction
- Where do we store all the various genes we discover?

Understanding gene versus genome and where their information is stored

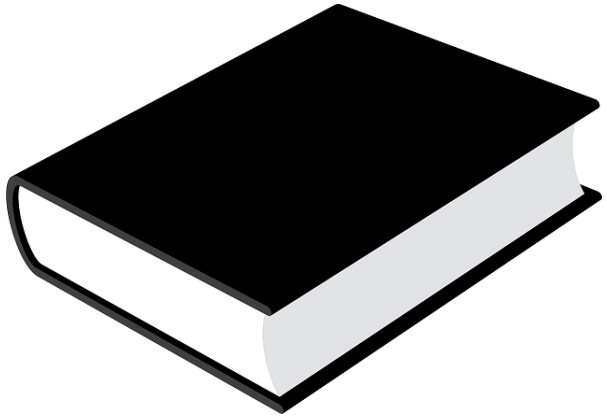
Genbank

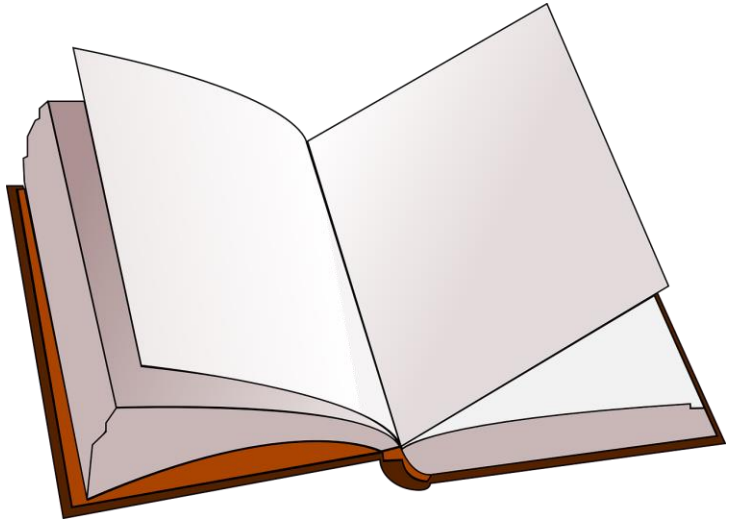


One chapter = one chromosome

One book = Organism = genome

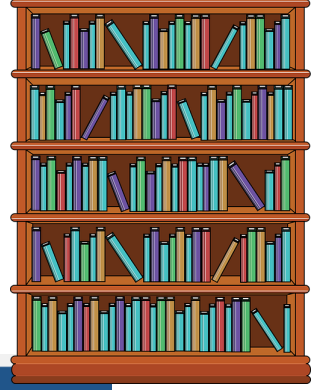
Complete set of genes in cell/organism





One page = one gene

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



National Library of Medicine
National Center for Biotechnology Information

GenBank | Nucleotide | Search

GenBank | Submit | Genomes | WGS | Metagenomes | TPA | TSA | INSDC | Documentation | Other

GenBank Overview

What is GenBank?

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

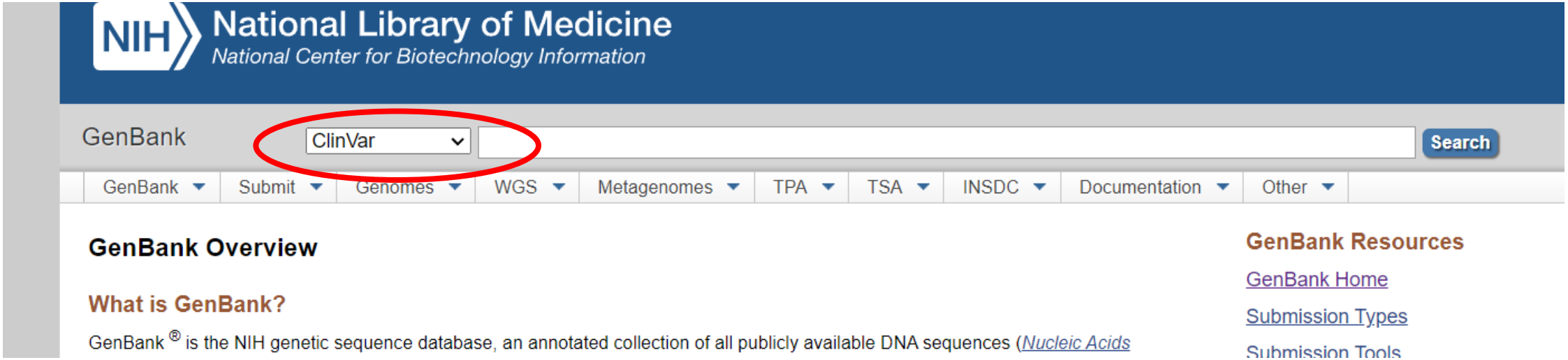
A GenBank release occurs every two months and is available from the ftp site. The release notes for the current version of GenBank provide detailed information about the release and notifications of upcoming changes to GenBank. Release notes for previous GenBank releases are also available. GenBank growth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

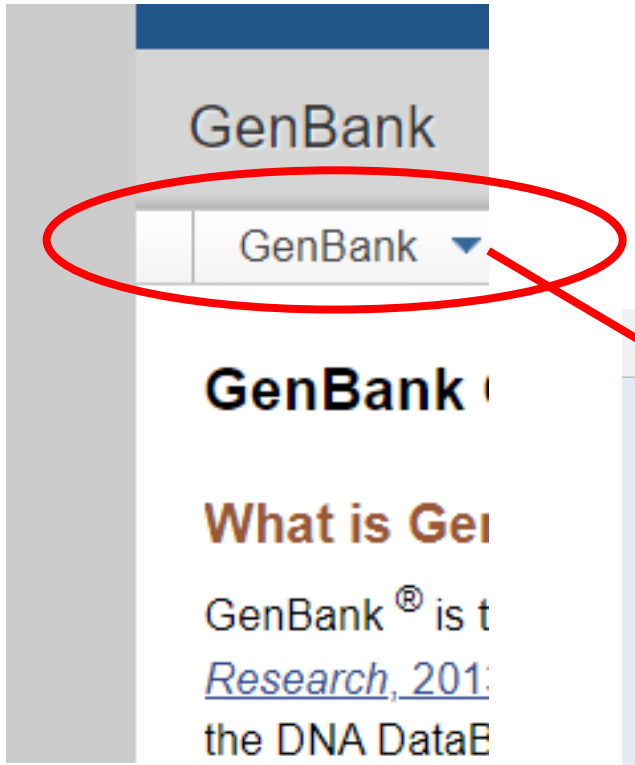
An annotated sample GenBank record for a *Saccharomyces cerevisiae* gene demonstrates many of the features of the GenBank flat file

GenBank Resources

GenBank Home
Submission Types
Submission Tools
Search GenBank
Update GenBank Records

Bank of variants' data





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

BLAST+ 2.15.0 is here!

We have included two exciting new features in the latest BLAST+ release

Tue, 28 Nov 2023

More BLAST news...

Web BLAST

Nucleotide BLAST
nucleotide ▶ nucleotide

blastx
translated nucleotide ▶ protein

tblastn
protein ▶ translated nucleotide

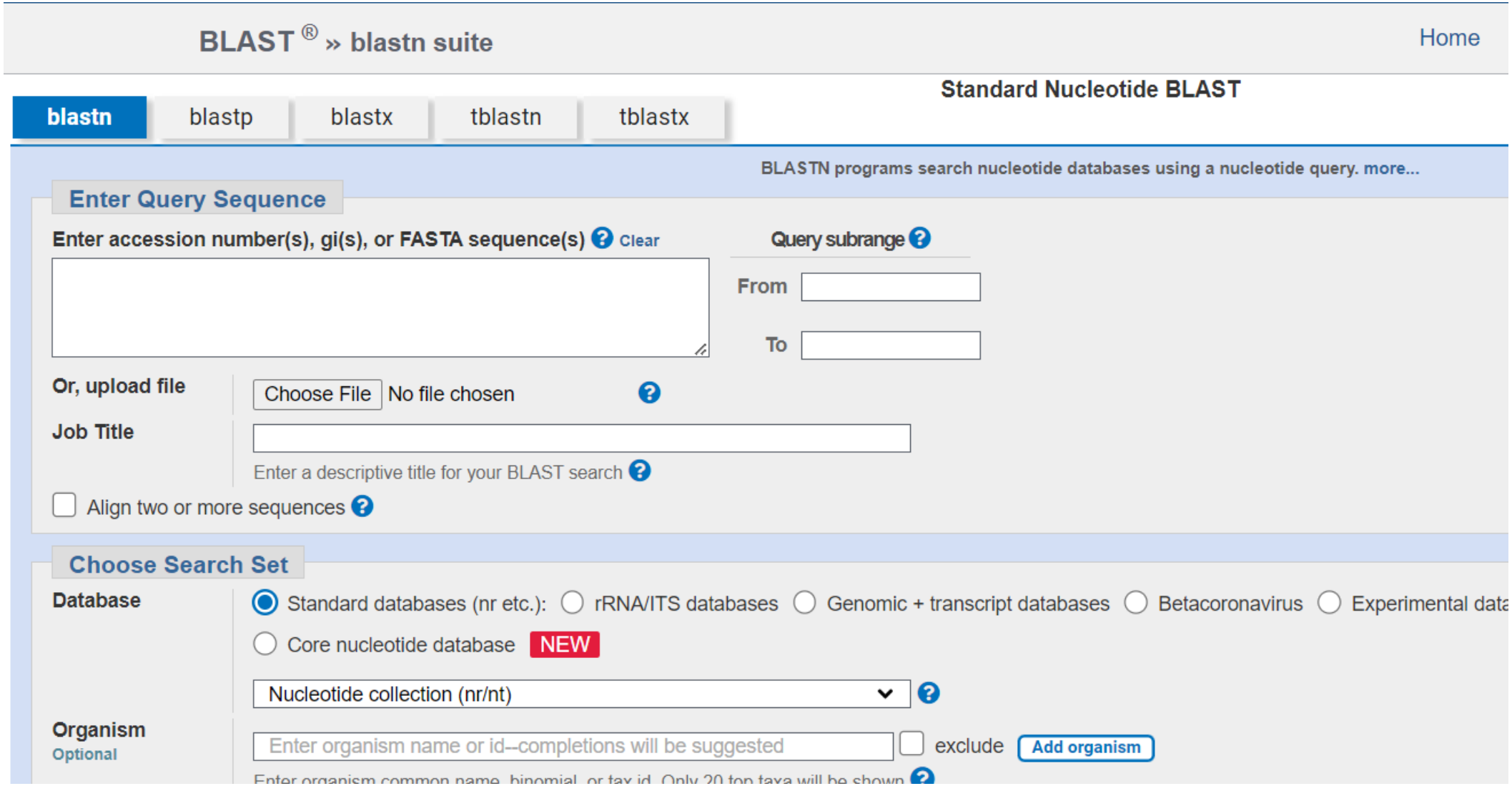
Protein BLAST
protein ▶ protein

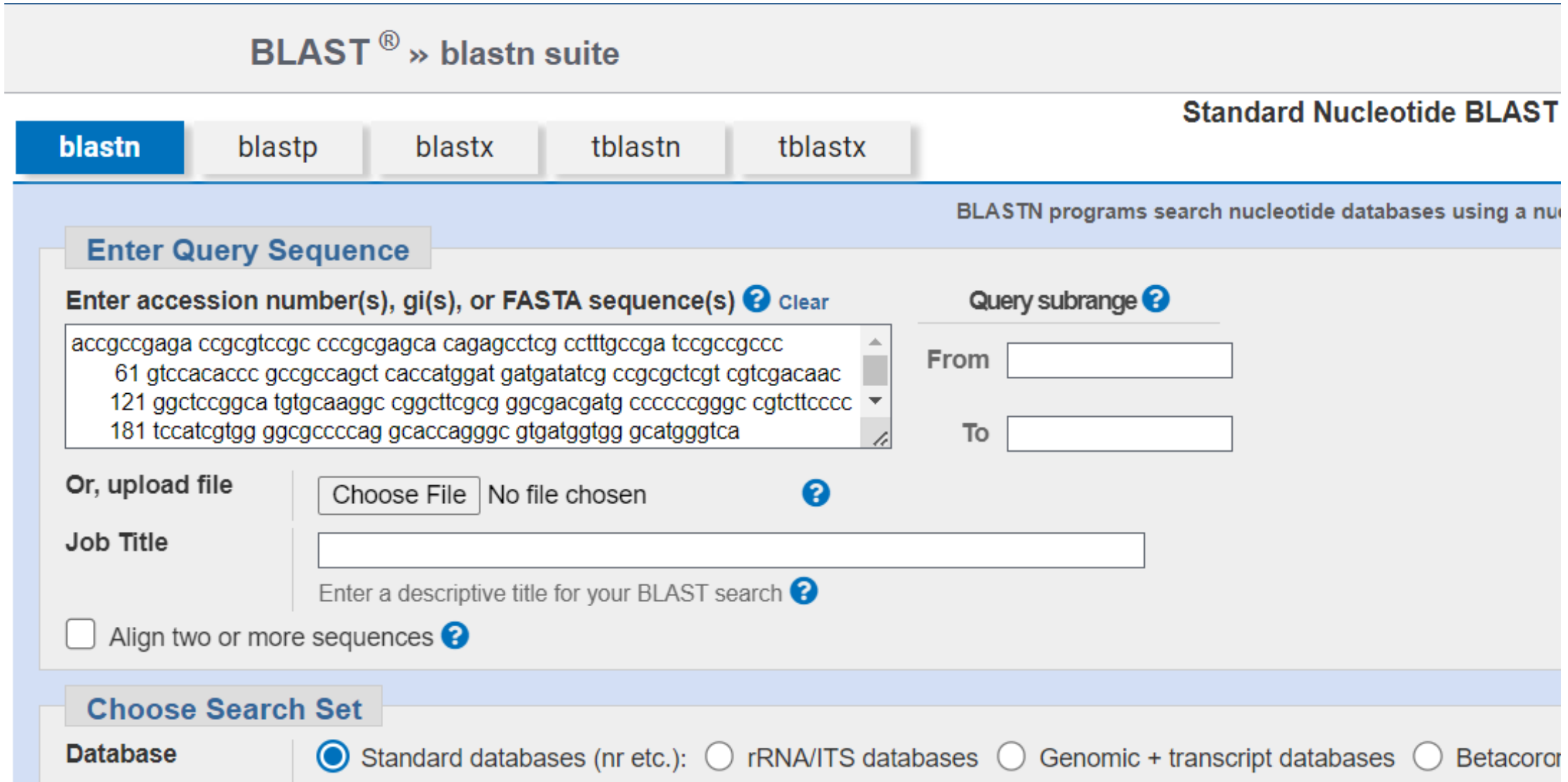
BLAST Genomes

Enter organism common name, scientific name, or tax id

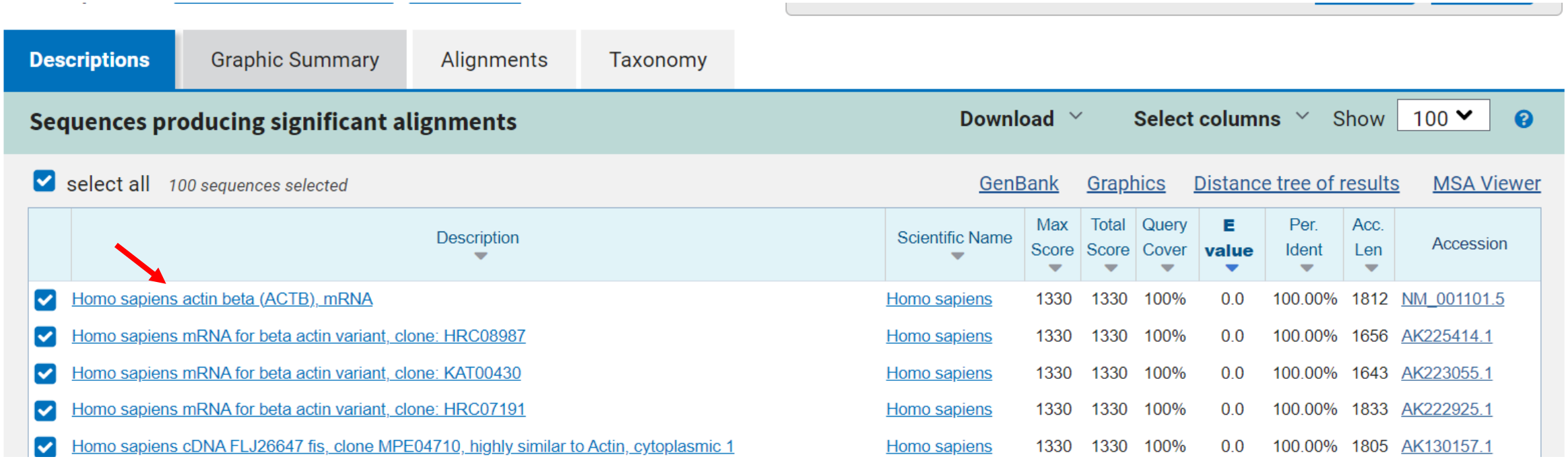
Search

Human Mouse Rat Microbes


BLAST ® » blastn suite
Home
Standard Nucleotide BLAST
blastn
blastp
blastx
tblastn
tblastx
BLASTN programs search nucleotide databases using a nucleotide query. more...
Enter Query Sequence
Enter accession number(s), gi(s), or FASTA sequence(s)
Clear
Query subrange
From
To
Or, upload file
Choose File
No file chosen
Job Title
Enter a descriptive title for your BLAST search
Align two or more sequences
Choose Search Set
Database
Standard databases (nr etc.):
rRNA/ITS databases
Genomic + transcript databases
Betacoronavirus
Experimental data
Core nucleotide database
NEW
Nucleotide collection (nr/nt)
Organism
Optional
Enter organism name or id--completions will be suggested
exclude
Add organism
Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown


BLAST ® » blastn suite
Standard Nucleotide BLAST
blastn
blastp
blastx
tblastn
tblastx
BLASTN programs search nucleotide databases using a nu
Enter Query Sequence
Enter accession number(s), gi(s), or FASTA sequence(s)
Clear
accgccgaga ccgcgtccgc cccgcgagca cagagcctcg cctttgccga tccgccgccc
61 gtccacaccc gccgccagct caccatggat gatgatatcg ccgcgctcgt cgtcgacaac
121 ggctccggca tgtgcaaggc cggcttcgcg ggcgacgatg ccccccgggc cgtcttcccc
181 tccatcgtgg ggcgccccag gcaccagggc gtgatggtgg gcatgggtca
Query subrange
From
To
Or, upload file
Choose File
No file chosen
Job Title
Enter a descriptive title for your BLAST search
Align two or more sequences
Choose Search Set
Database
Standard databases (nr etc.):
rRNA/ITS databases
Genomic + transcript databases
Betacoron

Result of sequence alignment



Descriptions | Graphic Summary | Alignments | Taxonomy

Sequences producing significant alignments Download Select columns Show 100

select all *100 sequences selected* GenBank Graphics Distance tree of results MSA Viewer

Description	Scientific Name	Max Score	Total Score	Query Cover	E value	Per. Ident	Acc. Len	Accession
Homo sapiens actin beta (ACTB), mRNA	Homo sapiens	1330	1330	100%	0.0	100.00%	1812	NM_001101.5
Homo sapiens mRNA for beta actin variant, clone: HRC08987	Homo sapiens	1330	1330	100%	0.0	100.00%	1656	AK225414.1
Homo sapiens mRNA for beta actin variant, clone: KAT00430	Homo sapiens	1330	1330	100%	0.0	100.00%	1643	AK223055.1
Homo sapiens mRNA for beta actin variant, clone: HRC07191	Homo sapiens	1330	1330	100%	0.0	100.00%	1833	AK222925.1
Homo sapiens cDNA FLJ26647 fis, clone MPE04710, highly similar to Actin, cytoplasmic 1	Homo sapiens	1330	1330	100%	0.0	100.00%	1805	AK130157.1

Homo sapiens actin beta (ACTB), mRNA

Sequence ID: NM_001101.5 **Length:** 1812 **Number of Matches:** 1

Range 1: 1 to 720 GenBank Graphics ▼ Next Match ▲ P

Score	Expect	Identities	Gaps	Strand
1330 bits(720)	0.0	720/720(100%)	0/720(0%)	Plus/Plus

```
Query  1    ACCGCCGAGACCGCGTCCGCCCCGCGAGCACAGAGCCTCGCCTTTGCCGATCCGCCGCCC  60
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  1    ACCGCCGAGACCGCGTCCGCCCCGCGAGCACAGAGCCTCGCCTTTGCCGATCCGCCGCCC  60

Query  61   GTCCACACCCGCCGCCAGCTCACCATGGATGATGATATCGCCGCGCTCGTCGTCGACAAC  120
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  61   GTCCACACCCGCCGCCAGCTCACCATGGATGATGATATCGCCGCGCTCGTCGTCGACAAC  120

Query  121  GGCTCCGGCATGTGCAAGGCCGGCTTCGCGGGCGACGATGCCCCCCGGGCCGTCTTCCCC  180
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  121  GGCTCCGGCATGTGCAAGGCCGGCTTCGCGGGCGACGATGCCCCCCGGGCCGTCTTCCCC  180

Query  181  TCCATCGTGGGGCGCCCCAGGCACCAGGGCGTGATGGTGGGCATGGGTCAGAAGGATTCC  240
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  181  TCCATCGTGGGGCGCCCCAGGCACCAGGGCGTGATGGTGGGCATGGGTCAGAAGGATTCC  240

Query  241  TATGTGGGCGACGAGGCCCAGAGCAAGAGAGGCATCCTCACCCTGAAGTACCCCATCGAG  300
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  241  TATGTGGGCGACGAGGCCCAGAGCAAGAGAGGCATCCTCACCCTGAAGTACCCCATCGAG  300

Query  301  CACGGCATCGTCACCAACTGGGACGACATGGAGAAAATCTGGCACCACACCTTCTACAAT  360
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  301  CACGGCATCGTCACCAACTGGGACGACATGGAGAAAATCTGGCACCACACCTTCTACAAT  360

Query  361  GAGCTGCGTGTGGCTCCCGAGGAGCACCCCGTGCTGCTGACCGAGGCCCCCCTGAACCCC  420
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  361  GAGCTGCGTGTGGCTCCCGAGGAGCACCCCGTGCTGCTGACCGAGGCCCCCCTGAACCCC  420

Query  421  AAGGCCAACCGCGAGAAGATGACCCAGATCATGTTTGAGACCTTCAACACCCCAGCCATG  480
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  421  AAGGCCAACCGCGAGAAGATGACCCAGATCATGTTTGAGACCTTCAACACCCCAGCCATG  480

Query  481  TACGTTGCTATCCAGGCTGTGCTATCCCTGTACGCCTCTGGCCGTACCACTGGCATCGTG  540
            ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
```

Other sources of variations

Variations affecting populations:

- natural selection,
- genetic drift,
- migration,
- non-random mating

CB 1.3 / AS 92022 Demonstrate understanding of genetic variation in relation to an identified characteristic

Genetic variation:

What, why

Source = how: mutation, meiosis, fertilisation, small population size, different rates of survival, migration, non-random mating

Gene tracking methods:

Why: To identify genetic relationships (several contexts)

How: use genetic marker / specific DNA sequences to produce pedigree, phylogenetic tree, …

TRACKING genes and comparing genomes

- Forensics
- Genetic relationships (Pedigrees, Phylogenetic trees)
- Genetic testing/screening
- Gene therapy
- Precision medicine
- Conservation

GENOMICS in CONTEXT

Genetic testing to beat cancer

Made by Aotearoa Science Agency and funded by Genomics Aotearoa.

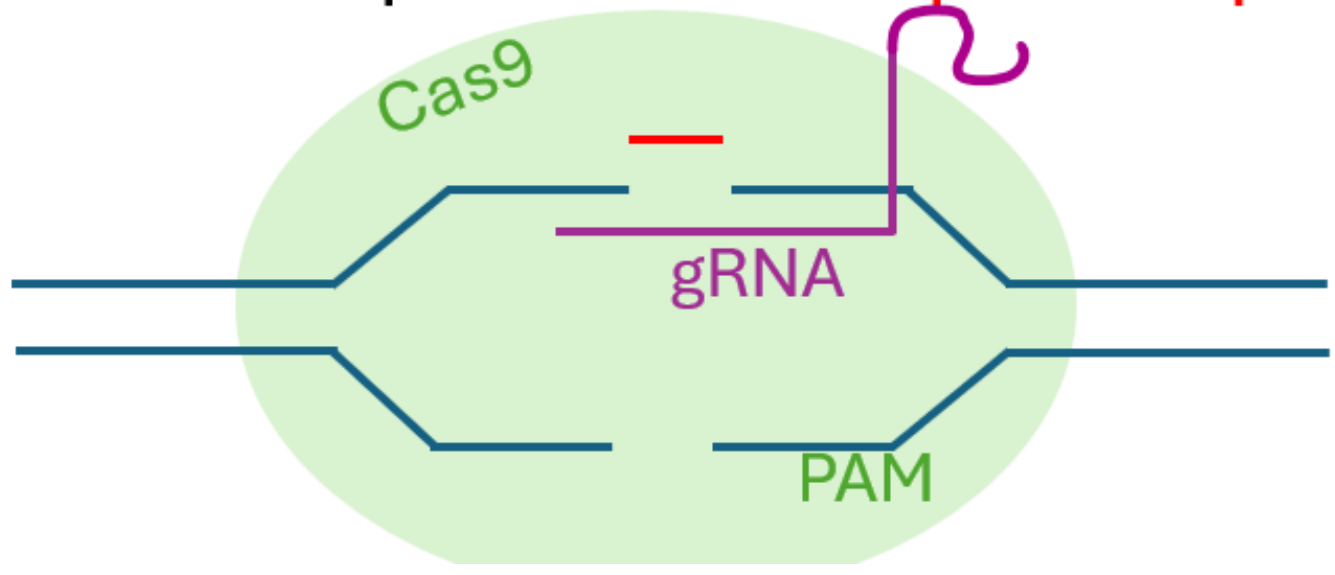
- https://www.rnz.co.nz/programmes/what-if-genomics-in-aotearoa/story/2018952807/what-if-we-can-use-genetic-testing-to-beat-cancer

GENE THERAPY

- Genomics - whole genome sequencing
- Normal vs variants
- Knowledge of variants – correct, replace, shutdown expression….
- Viral vectors vs CRISPR/Cas 9 system vs RNAi (RNA interference)

CRISPR-Cas 9 system

- Gene editing tool used to edit the genome, so you can correct or introduce a mutation
- Cas9 enzyme, guide RNA (gRNA) complementary to target genomic region and takes Cas 9 there, genomic region of interest, repair template (short sequence)
- Homology-directed repair with the repair template



Gene therapy – Boston Children's hospital

https://www.childrenshospital.org/programs/gene-therapy-program

SICKLE CELL ANEMIA (SCA)

- Casgevy, a cell-based gene therapy, approved for the treatment of SCA in patients 12 y+ with recurrent vaso-occlusive crises.
- First FDA-approved therapy utilizing CRISPR/Cas9 targeted genome editing technology.
- Patients' hematopoietic (blood) stem cells are modified by CRISPR/Cas9 technology (remove, add, replace).
- Modified blood stem cells are transplanted back into the patient where they attach and multiply within the bone marrow and increase the production of fetal hemoglobin (HbF)

https://www.gov.uk/government/news/mhra-authorises-world-first-gene-therapy-that-aims-to-cure-sickle-cell-disease-and-transfusion-dependent-thalassemia

Facioscapulohumeral muscular dystrophy (FSHD)

- muscle-wasting disease
- Overexpression of DUX4 gene coding for human double homeobox 4 (DUX4) protein, toxic to muscle
- No currently approved therapy
- RNAi will knockdown translation of DUX4

Kiwi patient first in world to test new gene therapy

1 July 2024 | Health and medicine, Faculty of Medical and Health Sciences

The first person in the world has just been dosed in a New Zealand trial of a revolutionary new genetic therapy.

https://www.auckland.ac.nz/en/news/2024/07/01/First-person-gets-new-gene-therapy.html

Phylogenetic trees

Diagram representing the evolutionary relationships among many species. It shows how different organisms are related through common ancestors and shows the branching patterns of evolution.

Generated using information, e.g., genetic information that is common and different between species.

Classifies species into groups based on evolutionary relationships rather than physical characteristics

Generated using genetic, morphological or fossil evidence

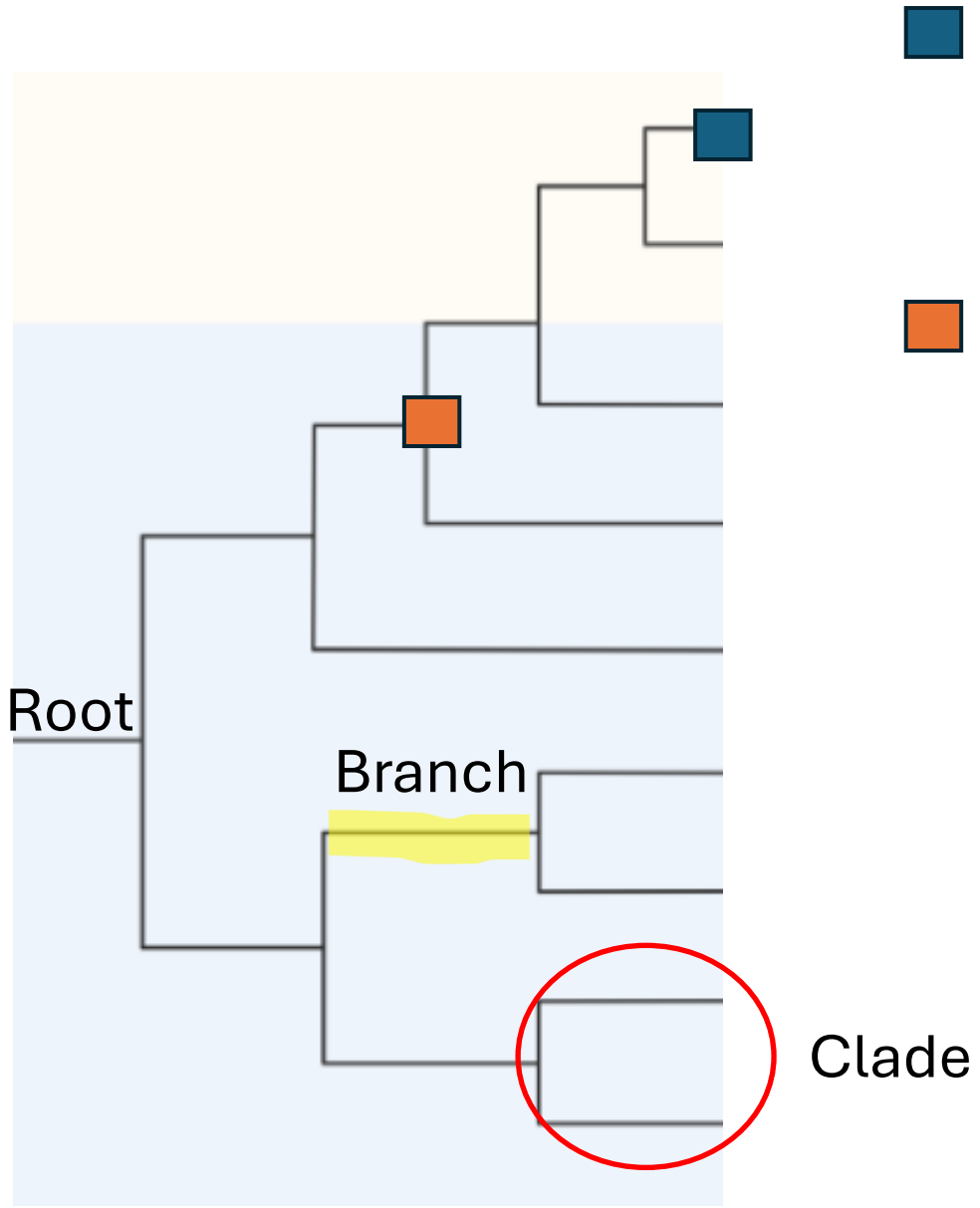
Phylogenetic tree - terminology

ROOT: connects the original most common ancestor among all species

BRANCH: when root undergoes speciation

NODES: Points of common ancestors for the following branches

CLADE: groups of organisms believed to have come from a common ancestor

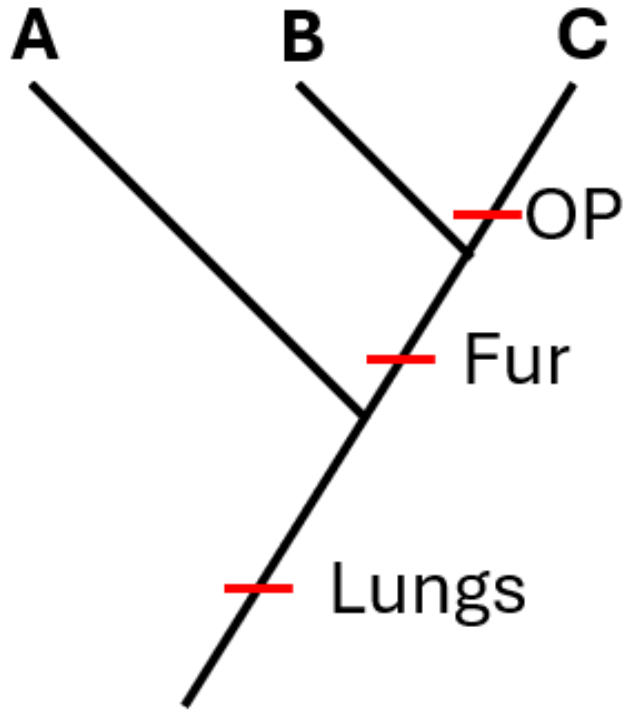


Cladogram – simple version of a phylogenetic tree

Shows the branching order of species without necessarily indicating the exact timing of evolutionary events.

Shows the relative recency of common ancestors and the emergence of traits.

Does not represent the actual time scale or the amount of evolutionary change.



The making of a cladogram

1. Make a table of characters, using the following key:

1 - shows that a species has the character under study

0 - shows that the species does not have the character under study

Derived characters	Species			
		A	B	C
	Lungs	1	1	1
	Fur	0	1	1
	Opposable thumbs	0	0	1

2. Use the principle of maximum parsimony to generate the simplest possible tree with the fewest number of evolutionary changes.

The making of a cladogram

Derived characters		Species		
		A	B	C
	Lungs	1	1	1
	Fur	0	1	1
	Opposable thumbs	0	0	1

3. Rearrange the table with the least common character at the top row and the most common character (oldest) at the bottom row.

Derived characters		Species		
		A	B	C
	Opposable thumbs	0	0	1
	Fur	0	1	1
	Lungs	1	1	1

The making of a cladogram

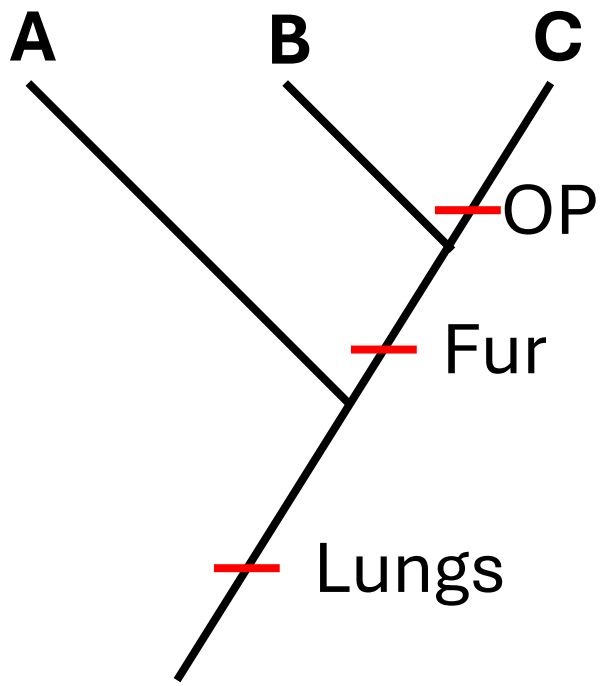
Derived characters	Species			
		A	B	C
	Opposable thumbs	0	0	1
	Fur	0	1	1
	Lungs	1	1	1

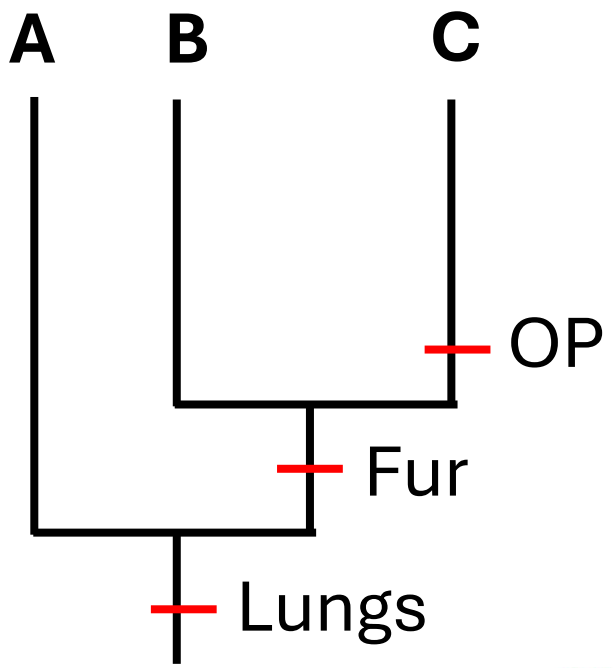
4. Start the tree with the most common character near the root and each new branch is the evolution of a new character. On the left, place the species with the least shared characters.

The making of a cladogram

Derived characters		Species		
		A	B	C
	Opposable thumbs	0	0	1
	Fur	0	1	1
	Lungs	1	1	1

Root = the most common character Each
Branch = evolution of a new character
Left = species with the least shared characters
Right = species with the most shared characters





OP = opposable thumbs

To make a cladogram

Derived characters	Species			
		Dog	Rabbit	Cat
	Retractable claws	0	0	1
	Fur	1	1	1
	Canines	1	0	1

1. Rearrange the table with the least common character at the top row and the most common character (oldest) at the bottom row.

Derived characters	Species			
		Rabbit	Dog	Cat
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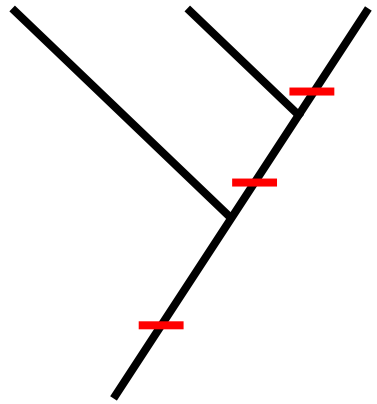

Making a cladogram

Derived characters	Species			
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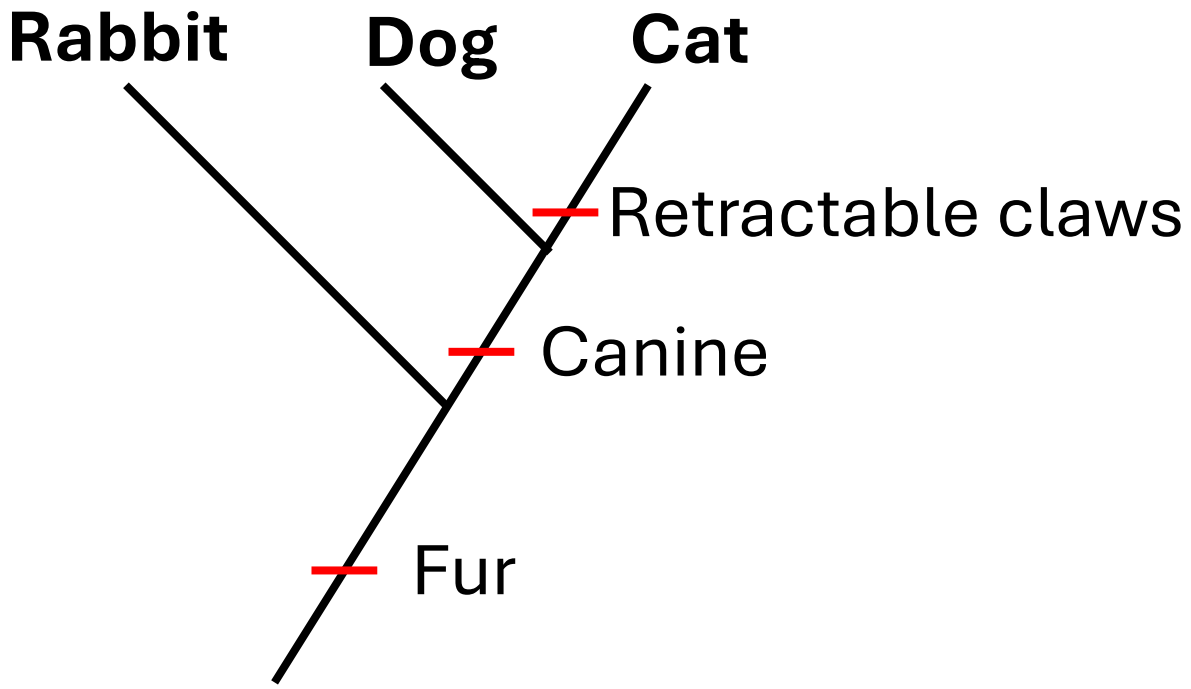

TEACH CONCEPTS

Teach for Tomorrow

Contexts and Connect

Food / Science 1 Oct 2024

What if... genomics could safeguard our food supply?

The rapid warming and periods of droughts as a result of climate change will impact our ability to grow food. Researchers at Plant and Food, AgResearch and the Bragato Research Institute are... Read more (VIDEO)

Science / Environment 28 Aug 2024

What if... genomics could help save an endangered species?

In 2018 the kākāpō became the first species in the world to have every individual's genome sequenced. How is the information being used today to help conservation? Read more (VIDEO)

https://www.rnz.co.nz/programmes/what-if-genomics-in-aotearoa

genomics aotearoa

thank you

Heather Cooper
Jayashree Panjabi
Debbie McCreath
Polona Le Quesne Stabej
Caroline Taripo-Keith
Nick Matzke

www.genomics-aotearoa.org.nz