

A decorative network diagram in the top-left corner of the slide. It features a complex web of interconnected nodes and edges. The nodes are represented by small circles, some of which are solid blue, some are solid grey, and some are hollow blue. The edges are thin grey lines. The overall structure is a dense, interconnected mesh.

Bioinformática

Trabajo Práctico Final

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A decorative network diagram in the bottom-right corner of the slide. It features a complex web of interconnected nodes and edges. The nodes are represented by small circles, some of which are solid blue, some are solid grey, and some are hollow blue. The edges are thin grey lines. The overall structure is a dense, interconnected mesh.



Agenda

⊙ Introducción

- Síndrome de Charcot - Marie - Tooth
- Gen GJB1
- La proteína GJB1

⊙ Procesamiento

- Tooling
- Secuenciamiento de mRNA
- BLASTp, MAS y árbol filogenético
- Comprobación de los resultados

⊙ Conclusiones



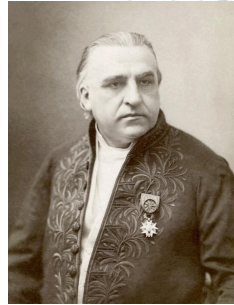
A decorative network diagram in the top-left corner, featuring a complex web of interconnected nodes and lines. The nodes are represented by circles of varying sizes, some with concentric rings, and the lines are thin and grey. The diagram is partially cut off by the left edge of the frame.

Introducción

A decorative network diagram in the bottom-right corner, similar to the one in the top-left. It shows a cluster of interconnected nodes and lines, with nodes represented by circles of varying sizes. The diagram is also partially cut off by the right edge of the frame.

Síndrome de Charcot - Marie - Tooth

- © Descrita en 1888
 - Jean-Martin Charcot
 - Pierre Marie
 - Howard Henry Tooth
- © Neuropatía motora y sensorial hereditaria




Síndrome de Charcot - Marie - Tooth

Tipo	Nombre	Genes
CMT1	Desmielinizante	PMP22 MPZ LITAF EGR2, NEFL
CMT2	Axonal	KIF1B MFN2 LMNA GARS NEFL y otros...
CMT3	Dejerine-Sottas	MPZ, EGR2, PMP22 y PRX
CMT4	Espinal	GDAP SBF1 SBF2 PRX FIG4 y otros...
CMT5	Piramidal	4q34.3–q35.2
CMT6	Con atrofia óptica	MFN2
CMTDI	Dominancia Incompleta	10q24.1–q25.1 DNM2 YARS MPZ INF2 GNB4
CMTRI	Recesivo Incompleto	GDAP1 KARS
CMTX	Ligada al sexo	GJB1 CMTX2 CMTX3 NAMSD PRPS1 PDK3



Gen GJB1

- ◎ Codifica la proteína GJB1
 - ◎ Otros nombres
 - CMTX
 - CMTX1
 - CX32
 - ◎ Se encuentra en Xq13.1
- 

Gen GJB1

- ◎ 10% de las mutaciones son patogénicas y probablemente patogénicas
- ◎ Incluyen
 - Mutación sin sentido, con cambios de sentido y doble cambio de sentido
 - Borrado de aminoácidos
 - Cambio de marco de lectura
 - Inserciones o borrados

Proteína GJB1

- ◎ Proteína Transmembranal
- ◎ Se suele encontrar en:
 - Hígado
 - Riñones
 - Páncreas
 - Sistema Nervioso
- ◎ Se produce en las células de Schwann
- ◎ Facilita la transmisión de nutrientes, iones y moléculas

A decorative network diagram in the top-left corner, featuring a complex web of interconnected nodes and lines. The nodes are represented by small circles, some of which are larger and have concentric circles, suggesting different levels of connectivity or importance. The lines are thin and gray, creating a mesh-like structure.


Procesamiento

A decorative network diagram in the bottom-right corner, similar to the one in the top-left. It shows a cluster of nodes connected by lines, with some nodes being larger and more prominent than others. The overall style is minimalist and technical.



Tooling

Software

- ◎ Perl 5.18 + CPAN + BioPerl 1.16
 - ◎ Bash
 - ◎ NCBI BLAST+ 2.2.28
 - ◎ EMBOSS 6.6.0.0
- 

Tooling

Web

- ◎ ensembl.org
- ◎ CLUSTAL OMEGA <http://www.ebi.ac.uk/Tools/msa/clustalo/>
- ◎ HomoloGene
- ◎ ClinVar
- ◎ dbSNP

Secuenciamiento de mRNA

1. Descarga de la secuencia en formato GenBank del Gen
2. Uso de BioPerl::SeqIO para traducir los reading frames
3. Uso del tamaño del reading frame para buscar el más probable

BLASTp+, MAS y árbol filogenético

1. Uso de BioPerl::RemoteBlast
 - a. Se debían buscar uno a uno los matches
2. Uso de BioPerl::StandAloneBlastPlus
 - a. Más directo, output derecho a archivo
3. Uso de bash e instalación local de BLAST+
 - a. Porque BioPerl no instala apropiadamente StandAloneBlastPlus y BLASTp de NCBI estaba caído

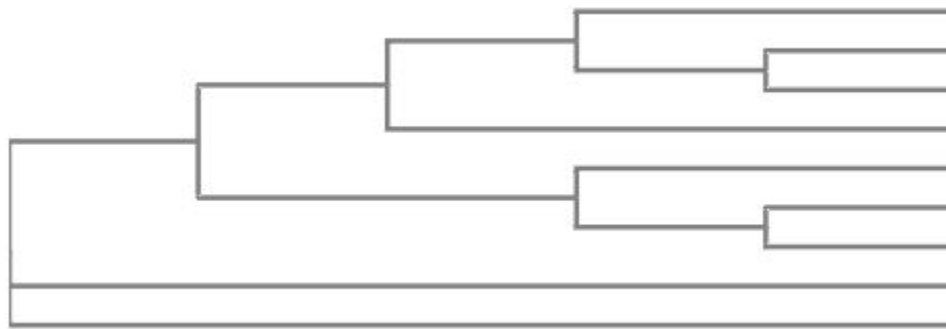
BLASTp+, MAS y árbol filogenético

1. Se intentó con el algoritmo MUSCLE usando el ejemplo de

<http://www.bioinfopoint.com/index.php/code/3-multiple-sequence-alignment-with-bioperl-and-muscle>

2. Se intentó con herramientas on-line
 - a. Galaxy (MUSCLE)
 - b. NCBI (COBALT)
 - c. EBI (CLUSTAL)

BLASTp+, MAS y árbol filogenético



O93533.1 0.11416
A2VE67.1 0.02243
Q8MIT9.1 0.02181
P08983.2 0.13654
P08033.1 0.00634
P08034.1 0
Q60HF7.1 0.00359
O18968.1 0.00534
Q6WGK6.1 0.00526

Comprobación de Resultados

1. Instalación de EMBOSS local y script de BASH
2. Instalación de base PROSITE en EMBOSS
3. ***getorf*** para obtener el ORF
 - a. filtrando por secuencias con más de 600 nucleótidos
4. ***patmatmotifs*** para obtener la proteína

A decorative network diagram in the top-left corner, consisting of interconnected nodes and lines, with some nodes highlighted in blue and others in grey.

Conclusión

A decorative network diagram in the bottom-right corner, consisting of interconnected nodes and lines, with some nodes highlighted in blue and others in grey.

