



UNIVERSIDAD  
DE GRANADA

# Práctica 2

## Herramientas bioinformáticas en Genómica

Biología Molecular - Grado en Medicina  
Curso 2020/21

Profesora: Marisol Benítez Cantos



# ¿Qué vamos a hacer?

Vamos a usar bases de datos y herramientas bioinformáticas para conocer mejor un gen que origina una enfermedad.

**Durante la práctica** trabajamos con un gen de ejemplo.

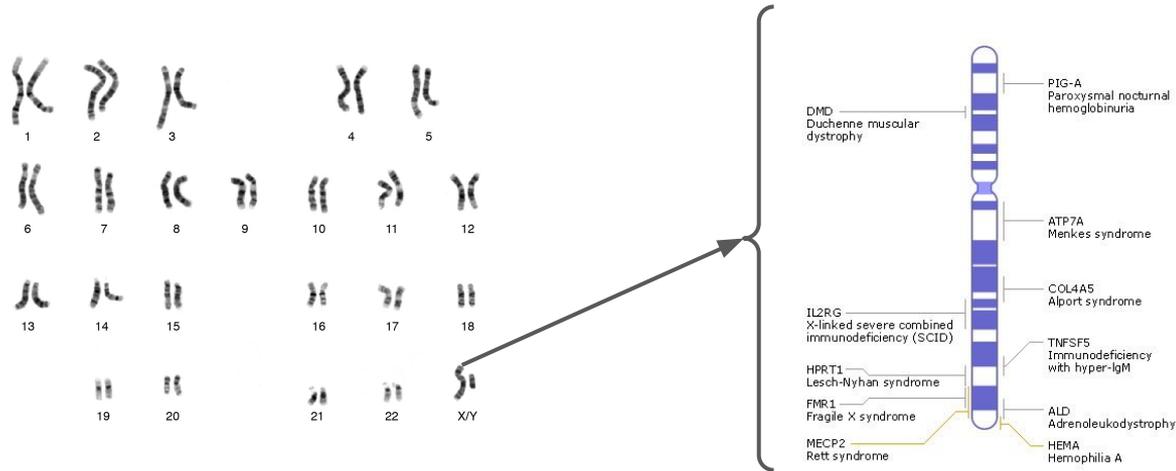
**En casa** elegid otra enfermedad monogénica y repetid la práctica para rellenar la tabla de la derecha.

**En el examen** habrá preguntas **sencillas** tipo test de opción múltiple.

PRÁCTICA II: BIOINFORMÁTICA	
Nombre y apellidos: María Soledad Benítez Cantos	
<b>Enfermedad:</b>	
Neoplasia endocrina múltiple de tipo 1	
Nombre oficial del gen responsable	<i>MEN1</i>
Alias	menin
Evidencia de la asociación gen-enfermedad	Frase: "We conclude that many "hyperplastic" parathyroid tumors in familial MEN-1 are in fact monoclonal and may progress or even begin to develop by inactivation of the MEN-1 gene (at 11q13) in a precursor cell." Enlace a publicación: <a href="https://pubmed.ncbi.nlm.nih.gov/2568586/">https://pubmed.ncbi.nlm.nih.gov/2568586/</a>
Ensembl ID del gen	ENSG00000133895
Localización cromosómica (cromosoma, coordenadas y hebra)	Chromosome 11: 64,803,510-64,811,294 reverse strand
Genes adyacentes	MAP4K2, SF1, RASGRP2
dbSNP ID de una variante patogénica	rs104894266
Consecuencia de la variante	Codón de parada prematuro
Número de transcritos	19
Ensembl ID del transcrito seleccionado	ENST00000450708.7
Longitud del transcrito (pb)	3162
Número de exones	10
Longitud de la proteína (aa)	615
Secuencia del transcrito en formato FASTA	>ENST00000450708.7 MEN1-214 cDNA:protein_coding GTGCGCCGCGGTGCCTAGTGTGGGATGTAAGCGCG GAGGCCGCCGCCACCGCCCGCCGC (...)
Primers diseñados	Forward: CGTGAGCTGGTGAAGAAGGT Reverse: GCTGTCCAATTTGGTGCCCTG
Identificador del miRNA que lo regula	hsa-miR-6877-5p
Secuencia del miRNA maduro	agggccgaagguggaagcugc
Laboratorio que realiza el diagnóstico genético de la enfermedad (preferiblemente en España)	Laboratorio de Genética Clínica SL, LabGenetics (Madrid)

# ¿Qué es la Genómica?

Es el campo de la Biología Molecular dedicado al estudio de **todo el material genético** de un organismo. Con técnicas de Biología Molecular y Bioinformática, se estudia la **estructura, mapeo, función**, evolución y edición de los genomas.

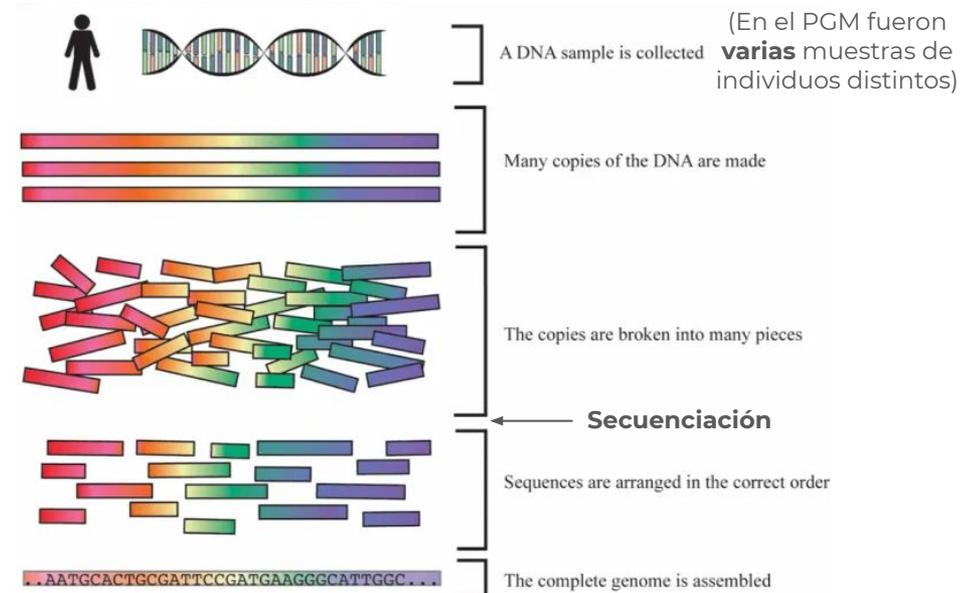


# Proyecto Genoma Humano (1999-2003)

**Secuenciar** el genoma humano (3 millones de pares de bases) y **publicar** los datos:

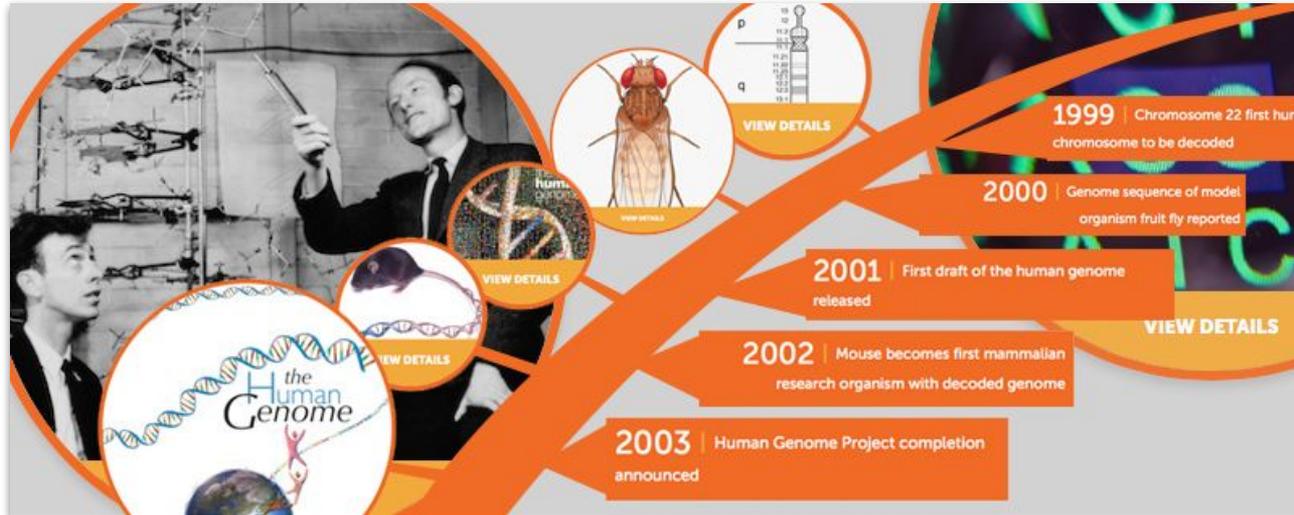
- Identificar genes y regiones regulatorias importantes
- Entender su papel en las enfermedades
- Investigar nuestros orígenes

## Proceso de secuenciación de un genoma



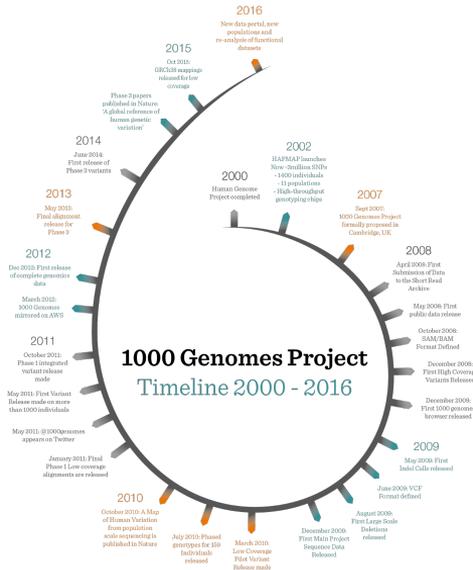
# Proyecto Genoma Humano (1999-2003)

- El ADN secuenciado procedía de **varios individuos** donantes anónimos.
- La secuenciación se llevó a cabo entre **20 laboratorios colaboradores** de Estados Unidos, Reino Unido, Francia, Alemania, Japón y China y se completó en **4 años**.

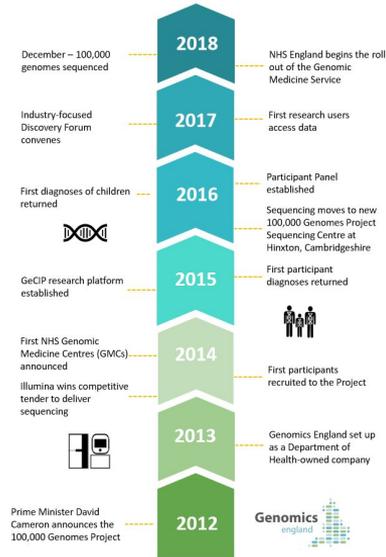


# Nuevos proyectos genómicos

Secuenciar a humanos **individualmente** para conocer la **variación genética** humana.



Proyecto **1000** genomas



Proyecto **100000** genomas

# Bases de datos

**Nucleic Acids  
Research** 

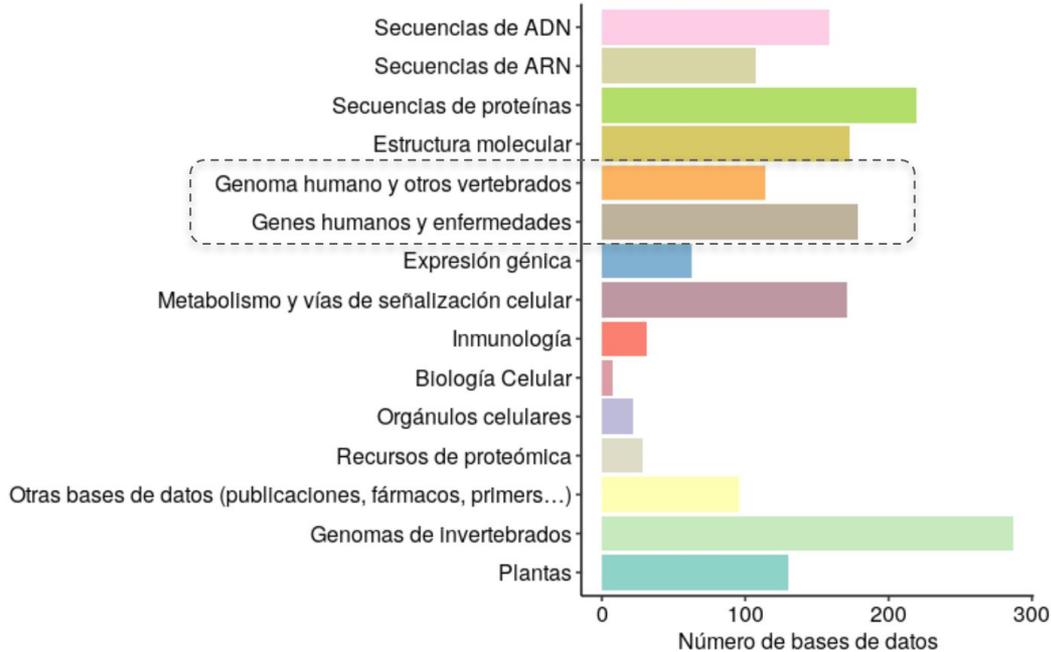
*Nucleic Acids Research*, 2021, Vol. 49, Database issue **D1–D9**  
doi: 10.1093/nar/gkaa1216

## **The 2021 *Nucleic Acids Research* database issue and the online molecular biology database collection**

Daniel J. Rigden<sup>1,\*</sup> and Xosé M. Fernández<sup>2</sup>

- La revista *Nucleic Acids Research* recopila anualmente todas las bases de datos de Biología Molecular que existen (actualmente **1641**)
- En 2021 se han añadido **90** bases de datos nuevas, **7** de ellas sobre COVID-19 y SARS-CoV-2.
- Puedes explorar el catálogo completo [aquí](#).

# Bases de datos



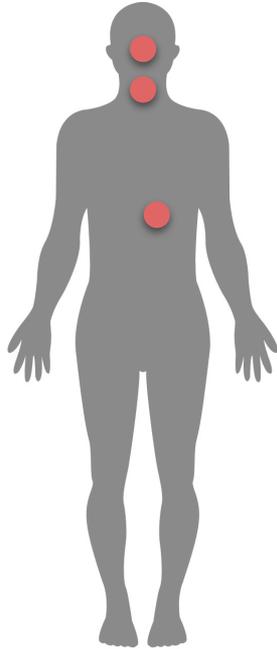
En esta práctica vamos a usar las siguientes bases de datos y herramientas bioinformáticas:



Primer-BLAST



# Neoplasia endocrina múltiple de tipo 1



Síndrome autosómico dominante causado por mutaciones en el gen supresor de tumores ***MEN1***.

## Criterios para el diagnóstico

- **Clínico:** 2 o más tumores primarios endocrinos en las glándulas **p**aratiroides, **p**ituitaria anterior y/o islotes **p**ancreáticos (las 3 P's).
- **Familiar:** ocurrencia de un tumor asociado a *MEN1* en un familiar de primer grado de un paciente diagnosticado.
- **Genético:** identificación de una mutación germinal en *MEN1* en un individuo asintomático.

# Antes de empezar

Consideraciones para seguir mejor la práctica:

- Los **rectángulos rojos** de las diapositivas indican dónde hay que escribir o hacer click en las páginas web.
- Abre todos los enlaces en **pestañas nuevas**.
- Si en algún momento te pierdes (¡es totalmente normal!), levanta la mano.



# ¿Hay algún gen asociado a esta enfermedad?

Respuesta:  DisGeNET

[Enlace](#)

# Buscar en DisGeNet

The screenshot shows the DisGeNet website interface. At the top, there is a navigation bar with links: Home, About, Search, Browser, API, Downloads, Cytoscape, RDF, disgenet2r, Help, and COVID-19. The DisGeNET logo is in the top right corner. Below the navigation bar, there are three radio buttons for search criteria: 'diseases' (selected), 'genes', and 'variants'. A search input field contains the text 'multiple endocrine neoplasia type 1', which is highlighted with a red box. To the right of the input field is a search button with a magnifying glass icon and a close button with an 'x' icon. Below the search results, there is a section titled 'General links' containing a list of four links:

- [Summary of All Gene-Disease Associations.](#)
- [All Evidences supporting the Gene-Disease Associations.](#)
- [Summary of All Variant-Disease Associations.](#)
- [All Evidences supporting the Variant-Disease Associations.](#)

**Buscamos el nombre de la enfermedad**

# Asociaciones gen-enfermedad

Home About Search Browser API Downloads Cytoscape RDF disgenet2r Help COVID-19 DisGeNET

Multiple Endocrine Neoplasia Type 1

**Name:** Multiple Endocrine Neoplasia Type 1  
**UMLS CUI:** C0025267  
**Type:** disease  
**MeSH Class:** Congenital, Hereditary, and Neonatal Diseases and Abnormalities; Neoplasms; Endocrine System Diseases  
**MeSH:** D018761  
**OMIM:** 131100  
**Semantic Type:** Neoplastic Process  
**Phenotypic abnormality:** None  
**Disease Ontology:** genetic disease

Similar diseases

Summary of Gene-Disease Associations

**Evidences for Gene-Disease Associations**

Summary of Variant-Disease Associations

Evidences for Variant-Disease Associations

Summary of Disease-Disease Associations

Disease Mappings

**Vemos las evidencias sobre asociaciones gen-enfermedad**

# Asociaciones gen-enfermedad

Gen asociado	Puntuación de la asociación (máx. = 1)*	Frase que apoya la asociación gen-enfermedad				Enlace a la publicación (PubMed)	
Gene	Score <sub>gda</sub>	Association Type	Type	Original DB	Sentence supporting the association	PMID	PMID Year
✓ MEN1	0.900	GeneticVariation	disease	BEFREE	Both mouse models developed well-differentiated (WD) G1/G2 PanNETs at a much shorter latency than <a href="#">M</a> ...	31160716	2020
✓ MEN1	0.900	GeneticVariation	disease	BEFREE	Fourteen patients with a clinical diagnosis (n=13) or suspicion (n=1) of <a href="#">MEN1</a> who had negative genetic sc...	31658439	2020
✓ MEN1	0.900	Biomarker	disease	BEFREE	<a href="#">MEN1</a> is caused by inactivating mutations of the tumor suppressor gene <a href="#">MEN1</a> which encodes the protein <a href="#">menin</a> .	31263451	2019
✓ MEN1	0.900	GeneticVariation	disease	BEFREE	Genetic analysis of <a href="#">MEN1</a> and other ACC associated genes, loss of heterozygosity (LOH) of MEN1 locus, ...	30721134	2019
✓ MEN1	0.900	AlteredExpression	disease	BEFREE	We reported that inactivation of <a href="#">menin</a> (the protein product of <a href="#">MEN1</a> ) increases activity of Dnmt1 and medi...	30190513	2019
✓ MEN1	0.900	Biomarker	disease	BEFREE	<a href="#">Menin</a> , a protein encoded by the <a href="#">MEN1</a> gene, suppresses cancers associated with <a href="#">multiple endocri...</a>	31497350	2019
✓ MEN1	0.900	Biomarker	disease	BEFREE	To date, a large number of frameshift, nonsense and missense mutations of <a href="#">MEN1</a> have been identified to...	31652443	2019
✓ MEN1	0.900	GeneticVariation	disease	BEFREE	<a href="#">Multiple endocrine neoplasia type 1 (MEN1)</a> is an autosomal dominant disease caused by mutations in the <a href="#">M</a> ...	30339208	2019
✓ MEN1	0.900	GeneticVariation	disease	BEFREE	Based on the array data, we concluded that the deletion was limited to the <a href="#">MEN1</a> gene and that the father...	30481156	2019
✓ MEN1	0.900	GeneticVariation	disease	BEFREE	Pituitary adenomas (PAs) may rarely occur in well-defined hereditary conditions, like <a href="#">multiple endocrine ne...</a>	30630164	2019
✓ MEN1	0.900	Biomarker	disease	BEFREE	True <a href="#">MEN1</a> or phenocopy? Evidence for geno-phenotypic correlations in <a href="#">MEN1 syndrome</a> .	31044390	2019
✓ MEN1	0.900	Biomarker	disease	BEFREE	<a href="#">Menin</a> is the protein mutated in patients with <a href="#">multiple endocrine neoplasia type 1 (MEN1) syndrome</a> and th...	31211356	2019
✓ MEN1	0.900	Biomarker	disease	BEFREE	All <a href="#">MEN1</a> positive ( <a href="#">MEN1</a> <sup>+</sup> n = 63) and <a href="#">MEN1</a> negative ( <a href="#">MEN1</a> <sup>-</sup> n = 75) de...	30268878	2019

\*Esta tabla aparece ordenada por Score descendente por defecto, por lo que las mejores asociaciones siempre aparecen al principio.

# ¿Qué características tiene el gen?

Respuesta:  *e!Ensembl*

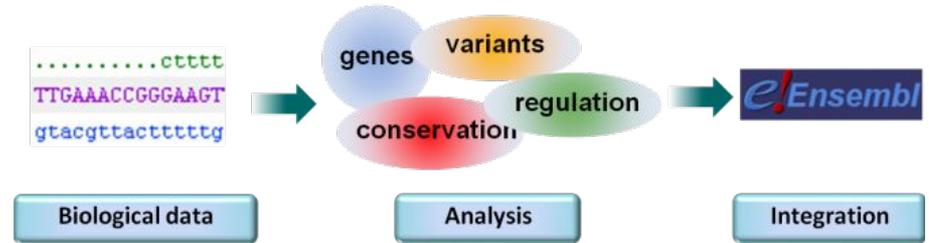
[Enlace](#)

# Ensembl

Es un **navegador genómico** que actúa como punto de acceso único a datos procedentes de múltiples bases de datos y proyectos bioinformáticos.

Contiene información de genes y transcritos a nivel de genoma, gen y proteína:

- Secuencias (ADN, ARN y proteína)
- Isoformas
- Variación genética
- Dominios proteicos
- Homología entre especies
- Elementos reguladores



Otro navegador genómico interesante es [UCSC Genome Browser](#).

# Buscar en Ensembl

**Ensembl** BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog Login/Register

**Tools** [BioMart >](#) [BLAST/BLAT >](#) [Variant Effect Predictor >](#)

[All tools](#) Export custom datasets from Ensembl with this data-mining tool Search our genomes for your DNA or protein sequence Analyse your own variants and predict the functional consequences of known and unknown variants

**Gen MEN1 de humano**

Search  
Human for  
MEN1

e.g. BRCA2 or rat 5:62797383-63627669 or rs699 or coronary heart disease

**Ensembl Release 103 (February 2021)**

- New mouse genome: GRCm39.
- Variant Recorder tool to convert between different ways of representing genetic variants.
- MANE Plus Clinical to identify clinically important transcripts not represented by MANE Select.

[More release news](#) on our blog

**Se puede buscar por:**

- Nombres de genes
- Identificadores (ID) únicos de gen, transcrito, variante, elemento regulador...
- Coordenadas genómicas
- Fenotipos

**All genomes** -- Select a species --

**Pig breeds**  
Pig reference genome and 12 additional breeds  
[View full list of all species](#)

**Favourite genomes**

- Human** GRCh38.p13  
[Still using GRCh37?](#)
- Mouse** GRCm39
- Zebrafish** GRCz11

# Buscar en Ensembl

**e!Ensembl** BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog Login/Register

**New Search** Search Human...

**Current selection:**  
< all Species  
Only searching Human

**Restrict category to:**  
Gene 11  
Transcript 28  
Phenotype 2  
GeneTree 1  
GenomicAlignment 2  
Clones & Regions 1

**Per page:**  
10 25 50 100

**Layout:**  
Standard Table

**Tip:**  
Help and Documentation can be searched from the homepage! Just type in a term you want to know more about, like non-synonymous SNP.

Only searching Human **MEN1**   
45 results match MEN1 when restricted to species: Human

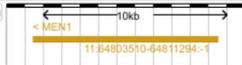
**MEN1 (Human Gene)**  
**ENSG00000133895** 11:64803510-64811294:-1  
Menin 1 [Source:HGNC Symbol;Acc:HGNC:7010]  
LRG\_509 (LRG display in Ensembl gene record; description: Locus Reference Genomic record for **MEN1**.) is an external reference matched to Gene ENSG00000133895  
[Variant table](#) • [Phenotypes](#) • [Location](#) • [External Refs.](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

**MEN1-204 (Human Transcript)**  
**ENST00000377313** 11:64803514-64810132:-1  
Menin 1 [Source:HGNC Symbol;Acc:HGNC:7010].  
[Location](#) • [External Refs.](#) • [cDNA seq.](#) • [Exons](#) • [Variant table](#) • [Protein seq.](#) • [Population](#) • [Protein summary](#)

**MEN1-205 (Human Transcript)**  
**ENST00000377316** 11:64803516-64810485:-1  
Menin 1 [Source:HGNC Symbol;Acc:HGNC:7010].  
[Location](#) • [External Refs.](#) • [cDNA seq.](#) • [Exons](#) • [Variant table](#) • [Protein seq.](#) • [Population](#) • [Protein summary](#)

**MEN1-206 (Human Transcript)**  
**ENST00000377321** 11:64803516-64810558:-1  
Menin 1 [Source:HGNC Symbol;Acc:HGNC:7010]  
LRG\_509i2 (LRG display in Ensembl transcript record; description: Locus Reference Genomic record for **MEN1**.) is an external reference matched to Transcript ENST00000377321  
[Location](#) • [External Refs.](#) • [cDNA seq.](#) • [Exons](#) • [Variant table](#) • [Protein seq.](#) • [Population](#) • [Protein summary](#)

**MEN1-207 (Human Transcript)**  
**ENST00000377326** 11:64803516-64810608:-1  
Menin 1 [Source:HGNC Symbol;Acc:HGNC:7010].  
[Location](#) • [External Refs.](#) • [cDNA seq.](#) • [Exons](#) • [Variant table](#) • [Protein seq.](#) • [Population](#) • [Protein summary](#)

**Best gene match**  
Human Gene **MEN1**  
HGNC Symbol; Acc:HGNC:7010  
  
Protein coding gene  
*menin 1*

**Suggestions**  
mien1 den1 hen1 mea1 mek1 men mena mend ment  
met1 ren1 sen1 ten1 mecd1 mecl1 mecp1 mect1 meg1  
meg1 meig1 meis1 mel1b mel1c memo1 men2a men2b menin  
menke meox1 mep1a mep1b merm1 merp1 mesh1 mesp1 meth1  
psen1 plen1 mea mea2 meaf meaf meaf meaf meaf meaf meaf  
meat meax meb mec

Gen **MEN1** de humano



# Características del gen

**Ensembl ID**

**Alias**

**Localización (cromosoma, inicio, fin, hebra)**

**Número de isoformas**

**Enlace a UniProtKB: función de la proteína**

**Human (GRCh38.p13)**

Location: 11:64,803,510-64,811,294

Gene: MEN1

**Gene-based displays**

- Summary
  - Splice variants
  - Transcript comparison
  - Gene alleles
- Sequence
  - Secondary Structure
- Comparative Genomics
  - Genomic alignments
  - Gene tree
  - Gene gain/loss tree
  - Orthologues
  - Paralogues
  - Ensembl protein families
- Ontologies
  - GO: Cellular component
  - GO: Biological process
  - GO: Molecular function
- Phenotypes
- Genetic Variation
  - Variant table
  - Variant image
  - Structural variants
- Gene expression
- Pathway
- Regulation
- External references
- Supporting evidence
- ID History
  - Gene history

**Gene: MEN1** ENSG00000133895

**Description** menin 1 [Source:HGNC Symbol;Acc:HGNC:7010]

**Location** [Chromosome 11: 64,803,510-64,811,294](#) reverse strand.  
GRCh38:CM000673.2

**About this gene** This gene has 19 transcripts ([splice variants](#)), [180 orthologues](#) and is associated with [105 phenotypes](#).

**Transcripts** [Show transcript table](#)

**Summary**

**Name** [MEN1](#) (HGNC Symbol)

**CCDS** This gene is a member of the Human CCDS set: [CCDS31600.1](#), [CCDS8083.1](#)

**UniProtKB** This gene has proteins that correspond to the following UniProtKB identifiers: [Q00255](#)

**RefSeq** This Ensembl/Gencode gene contains transcript(s) for which we have [selected identical RefSeq transcript\(s\)](#). If there are other RefSeq transcripts available they will be in the [External references](#) table

**LRG** [LRG 509](#) provides a stable genomic reference framework for describing sequence variants for this gene

**Ensembl version** ENSG00000133895.17

**Other assemblies** This gene maps to [64,570,982-64,578,766](#) in GRCh37 coordinates.  
View this locus in the GRCh37 archive: [ENSG00000133895](#)

**Gene type** Protein coding

**Annotation method** Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see [article](#).

# Función del gen (UniProtKB)

UniProtKB es una base de datos de secuencias de proteínas y sus anotaciones.

UniProtKB - O00255 (MEN1\_HUMAN)

Display [Help video](#) [BLAST](#) [Align](#) [Format](#) [Add to basket](#) [History](#) [Add a publication](#) [Feedback](#)

Entry

Publications

Feature viewer

Feature table

None

Function

Names & Taxonomy

Subcellular location

Pathology & Biotech

Protein | **Menin**

Gene | **MEN1**

Organism | *Homo sapiens (Human)*

Status | Reviewed - Annotation score: ●●●●● - Experimental evidence at protein level<sup>1</sup>

### Function<sup>i</sup>

Essential component of a MLL/SET1 histone methyltransferase (HMT) complex, a complex that specifically methylates 'Lys-4' of histone H3 (H3K4). Functions as a transcriptional regulator. Binds to the TERT promoter and represses telomerase expression. Plays a role in TGFB1-mediated inhibition of cell-proliferation, possibly regulating SMAD3 transcriptional activity. Represses JUND-mediated transcriptional activation on AP1 sites, as well as that mediated by NFkB subunit RELA. Positively regulates HOXC8 and HOXC6 gene expression. May be involved in normal hematopoiesis through the activation of HOXA9 expression (By similarity). May be involved in DNA repair. [By similarity](#) [6 Publications](#)

**Función de la proteína**

# Genes vecinos

Volvemos a Ensembl:

The screenshot shows the Ensembl genome browser interface for the MEN1 gene. The top navigation bar includes the Ensembl logo, links for BLAST/BLAT, VEP, Tools, BioMart, Downloads, Help & Docs, and Blog, along with a search bar and a Login/Register link. The main header displays 'Human (GRCh38.p13)' and 'Pestaña de región cromosómica'. A red box highlights the 'Location: 11:64,803,510-64,811,294' field. The left sidebar contains a tree view of 'Gene-based displays' with categories like Summary, Sequence, Comparative Genomics, Ontologies, Genetic Variation, Gene expression, Pathway, Regulation, External references, Supporting evidence, and ID History. The main content area shows the 'Gene: MEN1 ENSG00000133895' page with a 'Summary' section and various fields: Description, Location, About this gene, Transcripts, Name, CCDS, UniProtKB, RefSeq, LRG, Ensembl version, Other assemblies, Gene type, and Annotation method.

Ensembl BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog Login/Register

Search Human...

Human (GRCh38.p13) ▼ **Pestaña de región cromosómica**

Location: 11:64,803,510-64,811,294 Gene: MEN1

Gene-based displays

- Summary
  - Splice variants
  - Transcript comparison
  - Gene alleles
- Sequence
  - Secondary Structure
- Comparative Genomics
  - Genomic alignments
  - Gene tree
  - Gene gain/loss tree
  - Orthologues
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  - Phenotypes
- Genetic Variation
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- Regulation
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View this locus in the GRCh37 archive: [ENSG00000133895](#)

**Gene type** Protein coding

**Annotation method** Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see [article](#).

# Genes vecinos

Ensembl BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog Login/Register

Human (GRCh38.p13) Search Human...

Location: 11:64,803,510-64,811,294 Gene: MEN1

### Chromosome 11: 64,803,510-64,811,294

Assembly exceptions Chr. 11

### Region in detail

Scroll: Track height: Drag/Select:

**Chromosome bands**  
Contigs  
Genes  
(Comprehensive set from GENCODE 37)

**Regulatory Build**

**Gene Legend**

- Ensembl protein coding (Anotación automatizada)
- processed transcript
- RNA gene
- CTCF
- Open Chromatin
- Promoter Flank
- merged Ensembl/Havana pseudogene (Anotación automatizada + manual)
- Enhancer
- Promoter
- Transcription Factor Binding Site

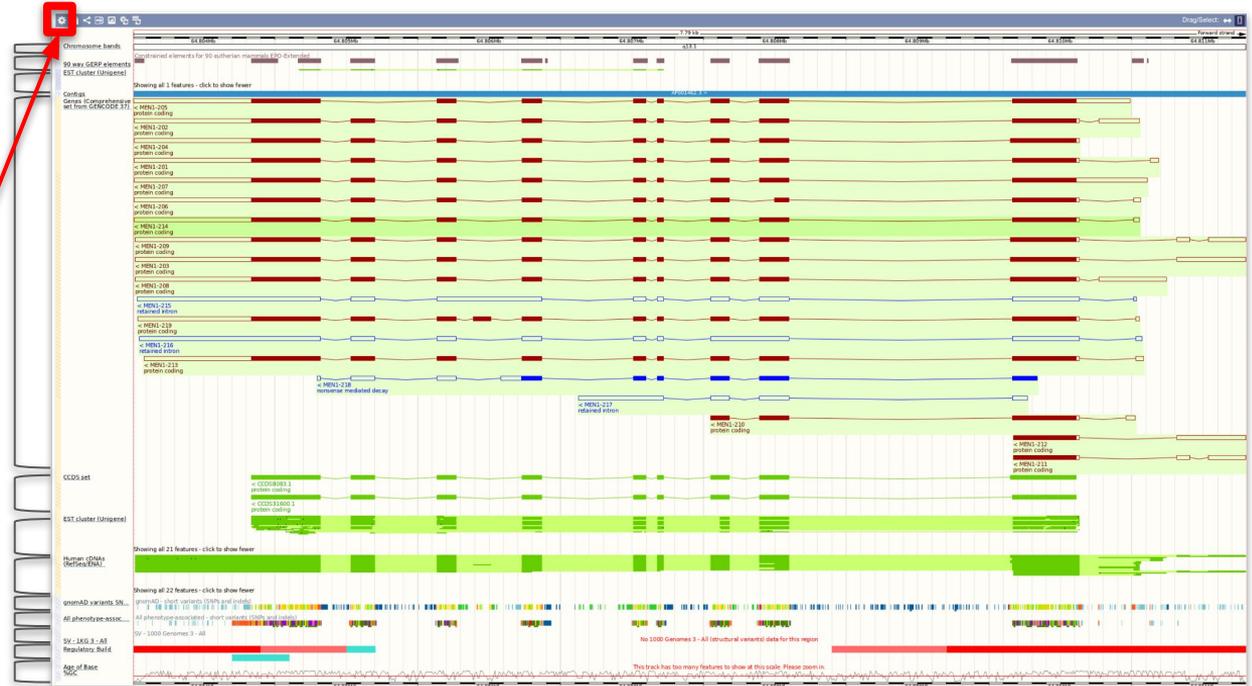
**Regulation Legend**

Legenda de colores de los genes

# Pistas de Ensembl

Hacemos scroll en la pantalla anterior hasta llegar a este otro visualizador genómico:

- Información procedente de distintas bases de datos
- Podemos ver isoformas, variantes (SNPs e indels), regiones de regulación...
- Son **customizables**: permiten mostrar sólo la información que nos interesa



# Variantes patogénicas

Queremos visualizar sólo las isoformas y las variantes:

**Apagamos  
todas estas  
pistas (9)**

The screenshot shows the UCSC Genome Browser's track configuration interface. The 'Active tracks' section is expanded, showing a list of tracks organized into categories. The following tracks are highlighted with red boxes:

- Genes and transcripts:** Comprehensive Gene Annotations from GENCODE 37
- mRNA and protein alignments:** mRNA and protein alignments
- Variation:** All phenotype-associated - short variants (SNPs and indels)
- Regulation:** Regulatory Build
- Information and decorations:** %GC

Callouts from yellow boxes point to these tracks:

- Pista de isoformas:** Points to 'Comprehensive Gene Annotations from GENCODE 37'.
- Pista de variantes:** Points to 'All phenotype-associated - short variants (SNPs and indels)'.

On the right side of the track list, there are icons for each track: a star for visibility and an information icon (i) for details. A callout box labeled 'Información sobre la pista' points to the information icon for the 'Comprehensive Gene Annotations from GENCODE 37' track.

Información  
sobre la pista

Pista de isoformas

Pista de variantes

# Variantes patológicas

Configure Region Image | Configure Overview Image | Configure Chromosome Image | Personal Data

Find a track

Select from available configurations: Current unsaved [Save current configuration](#)

### Variation

**Enable/disable all Phenotype/disease variants by source**

- All ClinVar variant annotations - short variants (SNPs and indels)
- ClinVar variants described as being probable-pathogenic, pathogenic, drug-response or histocompatibility - short variants (SNPs and indels)
- All phenotype-associated - short variants (SNPs and indels)
- HGMD-PUBLIC - short variants (SNPs and indels)
- NHGRI-EBI GWAS catalog phenotype - short variants (SNPs and indels)
- OMIM phenotype - short variants (SNPs and indels)
- PhenCode - short variants (SNPs and indels)

**Enable/disable all LSDB-associated variants**

- All LSDB-associated - short variants (SNPs and indels)
- HbVar - short variants (SNPs and indels)
- Infervers - short variants (SNPs and indels)
- KAT6BDB - short variants (SNPs and indels)
- LMDD - short variants (SNPs and indels)
- LSDB - short variants (SNPs and indels)
- OIVD - short variants (SNPs and indels)
- PAHdb - short variants (SNPs and indels)
- dbPEX - short variants (SNPs and indels)

**Enable/disable all Phenotype annotations**

- Phenotype annotations (all types)
- Phenotype annotations (Variations)
- Phenotype annotations (StructuralVariations)
- Phenotype annotations (Genes)

Looking for more data? Search the [Trackhub Registry](#) for external sources of annotation

**Key**

- Track style
- External data
- Forward strand
- User-added track

**Active tracks**

- Genome Reference Consortium Issues (0/17)
- Sequence and assembly (3/18)
  - Sequence (2/4)
  - Markers (0/1)
  - GRC alignments (1/2)
  - Simple features (0/4)
  - Clones & misc. regions (0/7)
- Genes and transcripts (1/77)
  - Genes (1/5)
  - Long reads (0/14)
  - Prediction transcripts (0/1)
  - LRG (0/1)
  - RNASeq models (0/56)
- mRNA and protein alignments (0/8)
  - mRNA alignments (0/4)
  - EST alignments (0/1)
  - Protein alignments (0/3)
- Variation (3/83)
  - Structural variants (1/26)
  - Phenotype, disease and curated variants (1/20)
  - Repeat and other (0/17)
  - Failed variants (0/1)
  - Structural variants (1/26)
- Somatic mutations (0/5)
  - Somatic variants (0/2)
  - Somatic structural variants (0/3)
- Regulation (0/467)
  - Regulatory Build (0/1)
  - Features by Cell/Tissue (0/415)
  - DNA methylation (0/47)
  - Other regulatory regions (0/4)
- Comparative genomics (0/203)
  - Multiple alignments (0/5)
  - Conservation regions (0/5)
  - BLASTz/LASTz alignments (0/193)
- Genome targeting (0/1)
- Oligo probes (0/37)

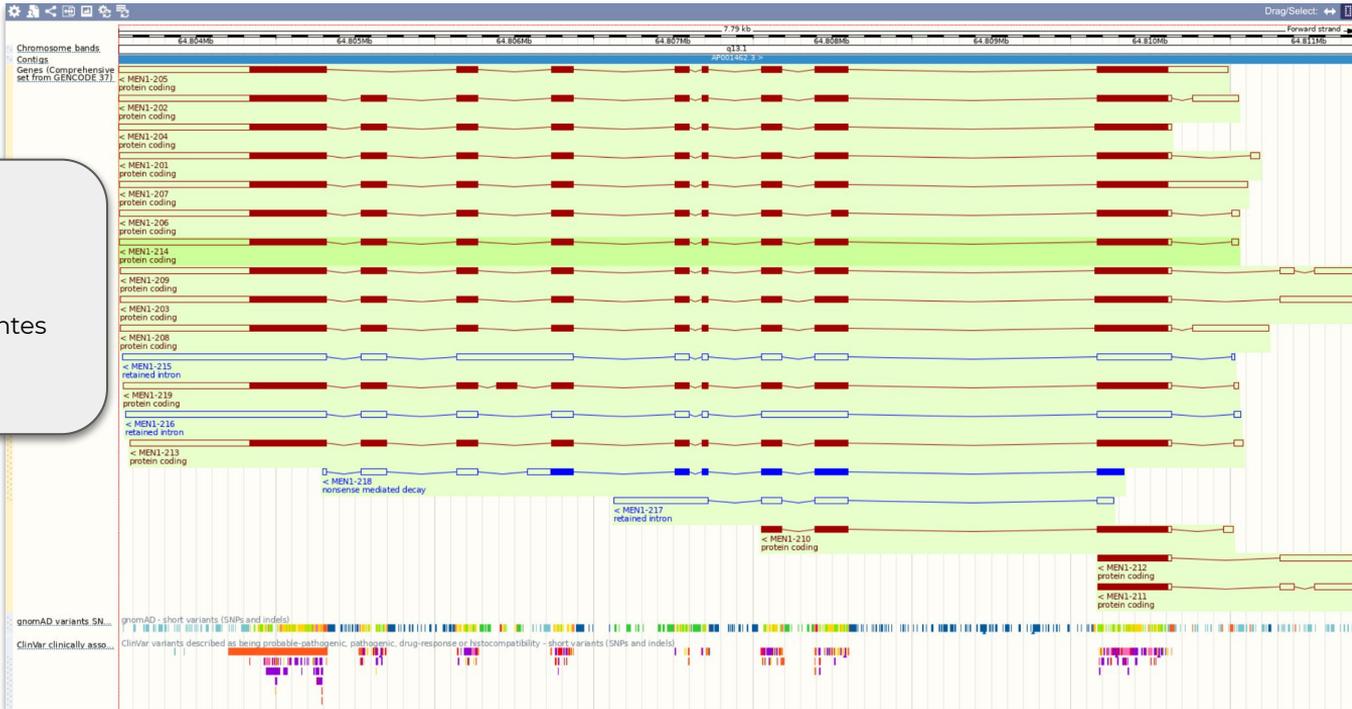
Activamos la pista  
de variantes  
patológicas

(en modo Normal)

Pista de variantes  
patológicas

# Variantes patogénicas

Hay variantes en todo el gen, pero las **patogénicas** están principalmente en **exones**.



## Leyenda:

- UTRs
- Exones codificantes
- ∨ Intrones

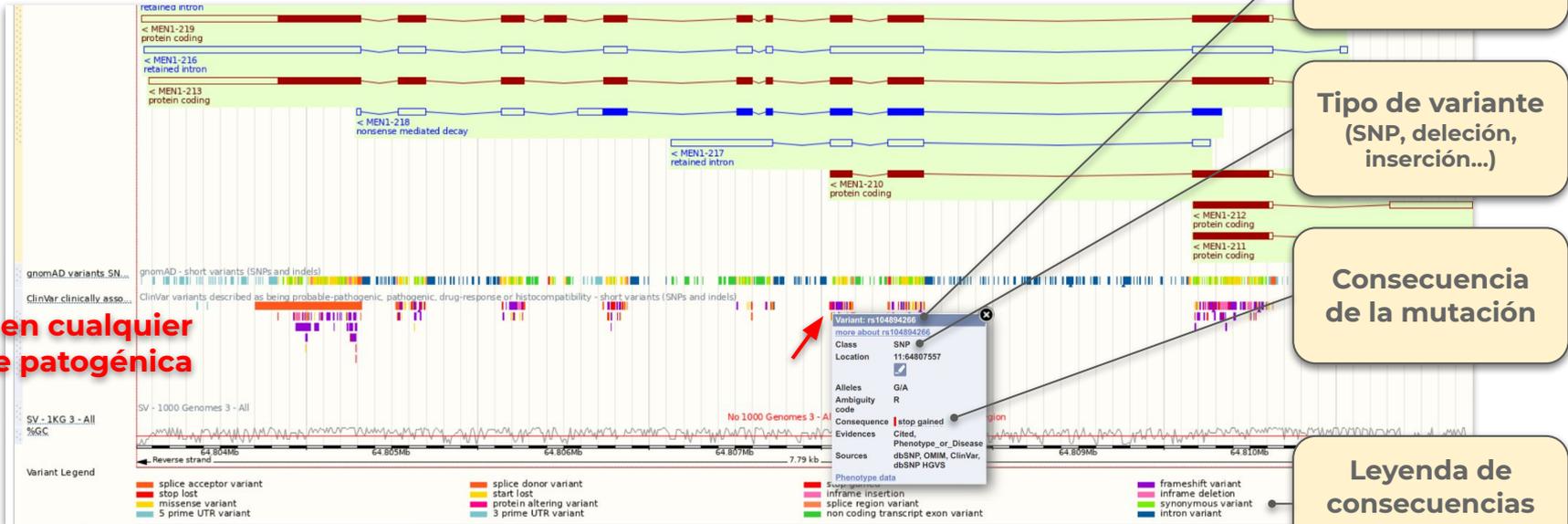
Pista de isoformas

Variantes

Variantes patogénicas

# Variantes patogénicas

Click en cualquier variante patogénica



Identificador de variante

Tipo de variante (SNP, deleción, inserción...)

Consecuencia de la mutación

Leyenda de consecuencias de las variantes

# Isoformas

Human (GRCh38.p13) **Volvemos a la pestaña del gen**

Location: 11:64,803,510-64,811,294 **Gene: MEN1**

**Longitud del ARNm (pb)**

**Ensembl ID del transcrito**

**Longitud de la proteína (aa)**

**Tipo de transcrito**

**Tabla de isoformas**

**Elegimos una isoforma \***

Gene: MEN1 ENSG00000133895

Description: menin 1 [Source:HGNC Symbol;Acc:HGNC:7010]

Location: Chromosome 11: 64,803,510-64,811,294 reverse strand. GRCh38:CM000673.2

About this gene: This gene has 19 transcripts (splice variants), 180 orthologues and is associated with 105 phenotypes.

Hide transcript table

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt Match	RefSeq Match	Flags
MEN1-203	<a href="#">ENST00000337652.5</a>	3162	615aa	Protein coding	<a href="#">CCDS8083</a>	<a href="#">000255-1</a>	-	TSL:5 GENCODE basic
MEN1-208	<a href="#">ENST00000394374.6</a>	3155	615aa	Protein coding	<a href="#">CCDS8083</a>	<a href="#">000255-1</a>	-	TSL:5 GENCODE basic
MEN1-207	<a href="#">ENST00000377326.7</a>	3150	610aa	Protein coding	<a href="#">CCDS31600</a>	<a href="#">000255-2</a>	-	TSL:5 GENCODE basic APPRIS P1
MEN1-209	<a href="#">ENST00000394376.5</a>	3034	615aa	Protein coding	<a href="#">CCDS8083</a>	<a href="#">000255-1</a>	-	TSL:5 GENCODE basic
MEN1-202	<a href="#">ENST00000315422.9</a>	2960	610aa	Protein coding	<a href="#">CCDS31600</a>	<a href="#">000255-2</a>	-	TSL:5 GENCODE basic APPRIS P1
MEN1-201	<a href="#">ENST00000312049.11</a>	2731	610aa	Protein coding	<a href="#">CCDS31600</a>	<a href="#">000255-2</a>	-	TSL:1 GENCODE basic APPRIS P1
MEN1-214	<a href="#">ENST00000450708.7</a>	2712	610aa	Protein coding	<a href="#">CCDS31600</a>	<a href="#">000255-2</a>	<a href="#">NM_001370259.2</a>	TSL:5 GENCODE basic APPRIS P1 <b>MANE Select v0.92</b>
MEN1-204	<a href="#">ENST00000377313.6</a>	2691	615aa	Protein coding	<a href="#">CCDS8083</a>	<a href="#">000255-1</a>	-	TSL:2 GENCODE basic
MEN1-213	<a href="#">ENST00000440873.6</a>	2661	610aa	Protein coding	<a href="#">CCDS31600</a>	<a href="#">000255-2</a>	-	TSL:5 APPRIS P1
MEN1-205	<a href="#">ENST00000377316.6</a>	2868	555aa	Protein coding	-	<a href="#">E7EN32</a>	-	TSL:5 GENCODE basic
MEN1-219	<a href="#">ENST00000672304.1</a>	2800	652aa	Protein coding	-	<a href="#">A0A5F9ZH33</a>	-	GENCODE basic
MEN1-206	<a href="#">ENST00000377321.5</a>	2614	575aa	Protein coding	-	<a href="#">000255-3</a>	-	TSL:1 GENCODE basic
MEN1-212	<a href="#">ENST00000429702.5</a>	944	146aa	Protein coding	-	<a href="#">Q9GZQ5</a>	-	CDS 3' incomplete TSL:1
MEN1-210	<a href="#">ENST00000413626.1</a>	870	261aa	Protein coding	-	<a href="#">E7ENS2</a>	-	CDS 3' incomplete TSL:3
MEN1-211	<a href="#">ENST00000424912.1</a>	816	146aa	Protein coding	-	<a href="#">Q9GZQ5</a>	-	CDS 3' incomplete TSL:1
MEN1-218	<a href="#">ENST00000672079.1</a>	1246	259aa	Nonsense mediated decay	-	<a href="#">A0A5F9Z168</a>	-	CDS 5' incomplete
MEN1-215	<a href="#">ENST00000478548.2</a>	3132	No protein	Retained intron	-	-	-	TSL:2

\* Preferiblemente la isoforma con la etiqueta "MANE Select v0.92" (columna *Flags*), ya que ha sido escogida como isoforma de referencia del gen.

# Información de la isoforma

**Ensembl ID**

**Número de exones**

**Ocultamos la tabla de isoformas**

**Legenda:**

- UTRs
- Exones codificantes
- Intrones

**“General identifiers”:** enlaces de este transcrito en otras bases de datos.



# Secuencia del ARNm

En la sección  
Sequence  
seleccionamos  
cDNA

**Human (GRCh38.p13)**

Location: 11:64,803,510-64,811,294    Gene: MEN1    Transcript: MEN1-214

**Transcript-based displays**

- Summary
- Sequence
  - Exons
  - cDNA**
  - Protein
- Protein Information
  - Protein summary
  - Domains & features
  - Variants
  - 3D Protein model
- Genetic Variation
  - Variant table
  - Variant image
  - Haplotypes
  - Population comparison
  - Comparison image
- External References
  - General identifiers
  - Oligo probes
  - Supporting evidence
- ID History
  - Transcript history
  - Protein history

**Transcript: MEN1-214** ENST00000450708.7

**Description** menin 1 [Source:HGNC Symbol;Acc:HGNC:7010]

**Location** [Chromosome 11: 64,803,516-64,810,551 reverse strand.](#)

**About this transcript** This transcript has [10 exons](#), is annotated with [11 domains and features](#), is associated with [4111 variant alleles](#) and maps to [501 oligo probes](#).

**Gene** This transcript is a product of gene [ENSG00000133895.17](#) [Show transcript table](#)

**Summary**

< MEN1-214 protein coding  
← Reverse strand → 7.04 kb

**Statistics** Exons: 10, Coding exons: 9, Transcript length: 2,712 bps, Translation length: 610 residues

**CCDS** This transcript is a member of the Human CCDS set: [CCDS31600](#)

**Uniprot** This transcript corresponds to the following Uniprot identifiers: [O00255](#)

**Transcript Support Level (TSL)** [TSL:5](#)

**Version** ENST00000450708.7

**Type** Protein coding

**Annotation Method** Manual annotation (determined on a case-by-case basis) from the Havana project.

**Annotation Attributes** RNA-Seq supported partial [Definitions](#)

**GENCODE basic gene** This transcript is a member of the [Genecode basic](#) gene set.

Configure this page  
Custom tracks  
Export data  
Share this page  
Bookmark this page

# Secuencia del ARNm

Human (GRCh38.p13) ▾

Location: 11:64,803,510-64,811,294 Gene: MEN1 Transcript: MEN1-214

Transcript-based displays

- Summary
- Sequence
  - Exons
  - cDNA
  - Protein
- Protein Information
  - Protein summary
  - Domains & features
  - Variants
  - 3D Protein model
- Genetic Variation
  - Variant table
  - Variant image
  - Haplotypes
  - Population comparison
  - Comparison image
- External References
  - General identifiers
  - Oligo probes
- Supporting evidence
  - ID History
  - Transcript history
  - Protein history

Configure this page

Custom tracks

Export data

Share this page

Bookmark this page

Transcript: MEN1-214 ENST00000450708.7

Description: menin 1 [Source:HGNC Symbol;Acc:HGNC:7010-7]

Location: Chromosome 11: 64,803,516-64,810,551 reverse strand.

About this transcript: This transcript has 10 exons, is annotated with 11 domains and features, is associated with 4111 variant alleles and maps to 501 oligo probes.

Gene: This transcript is a product of gene ENSG00000133895.17 [Hide transcript table](#)

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt Match	RefSeq Match	Flags
MEN1-203	ENST00000337652.5	3162	615aa	Protein coding	CCDS8083	Q00255-1	-	TSL:5 GENCODE basic
MEN1-208	ENST00000394374.6	3155	615aa	Protein coding	CCDS8083	Q00255-1	-	TSL:5 GENCODE basic
MEN1-207	ENST00000377326.7	3150	610aa	Protein coding	CCDS31600	Q00255-2	-	TSL:5 GENCODE basic APPRIS P1
MEN1-209	ENST00000394376.5	3034	615aa	Protein coding	CCDS8083	Q00255-1	-	TSL:5 GENCODE basic
MEN1-202	ENST00000315422.9	2960	610aa	Protein coding	CCDS31600	Q00255-2	-	TSL:5 GENCODE basic APPRIS P1
MEN1-201	ENST00000312049.11	2731	610aa	Protein coding	CCDS31600	Q00255-2	-	TSL:1 GENCODE basic APPRIS P1
MEN1-214	ENST00000450708.7	2712	610aa	Protein coding	CCDS31600	Q00255-2	NM_001370259.2	TSL:5 GENCODE basic APPRIS P1 MANE
MEN1-204	ENST0000037313.6	2691	615aa	Protein coding	CCDS8083	Q00255-1	-	TSL:2 GENCODE basic
MEN1-213	ENST00000440873.6	2661	610aa	Protein coding	CCDS31600	Q00255-2	-	TSL:5 APPRIS P1
MEN1-205	ENST00000377316.6	2868	555aa	Protein coding	-	EZENS2	-	TSL:5 GENCODE basic
MEN1-219	ENST00000672304.1	2800	652aa	Protein coding	-	A0ASF9ZHS3	-	GENCODE basic
MEN1-206	ENST00000377321.5	2614	575aa	Protein coding	-	Q00255-3	-	TSL:1 GENCODE basic
MEN1-212	ENST00000429702.5	944	146aa	Protein coding	-	Q9GZQ5	-	CDS 3' incomplete TSL:1
MEN1-210	ENST00000413626.1	870	261aa	Protein coding	-	EZENS2	-	CDS 3' incomplete TSL:3
MEN1-211	ENST00000424912.1	816	146aa	Protein coding	-	Q9GZQ5	-	CDS 3' incomplete TSL:1
MEN1-218	ENST00000672079.1	1246	259aa	Nonsense mediated decay	-	A0ASF9ZHS3	-	CDS 3' incomplete
MEN1-215	ENST00000478548.2	3132	No protein	Retained intron	-	-	-	TSL:2
MEN1-216	ENST00000671939.1	2891	No protein	Retained intron	-	-	-	-
MEN1-217	ENST00000671965.1	1037	No protein	Retained intron	-	-	-	-

cDNA sequence

[Download sequence](#) [BLAST this sequence](#)

Codons Alternating codons Alternating codons

Exons An exon Another exon

Download sequence

File name: Homo\_sapiens\_MEN1-214\_sequence

File format: FASTA

[Preview](#) [Download](#) [Download Compressed](#)

Settings

Included sequences:

- Select/deselect all
- cDNA (transcripts)
- Coding sequences (CDS)
- Amino acid sequences
- 5' UTRs
- 3' UTRs
- Exons
- Introns
- Genomic sequence

Guide to file formats

FASTA

Text sequence(s): DNA and/or amino acids

```
>11:64803516-64810551:reverse:menin1
ATTCAGCAACAAAAGCAACACGGG
GAGTCTCTCCACAAACATGGGCAI
TCTTAGGAGTGAATAATTGATGC
TTTGGGAAGAGGACCTCCG
AGGCCCCACAACTCCGCAAGTC
TTTTCGTTCCGCACTGGGCACTC
GCTGGCCATCGTAGGCTGATGCTI
```

RTF

Marked-up sequence, with or without variants

Descargamos la  
secuencia

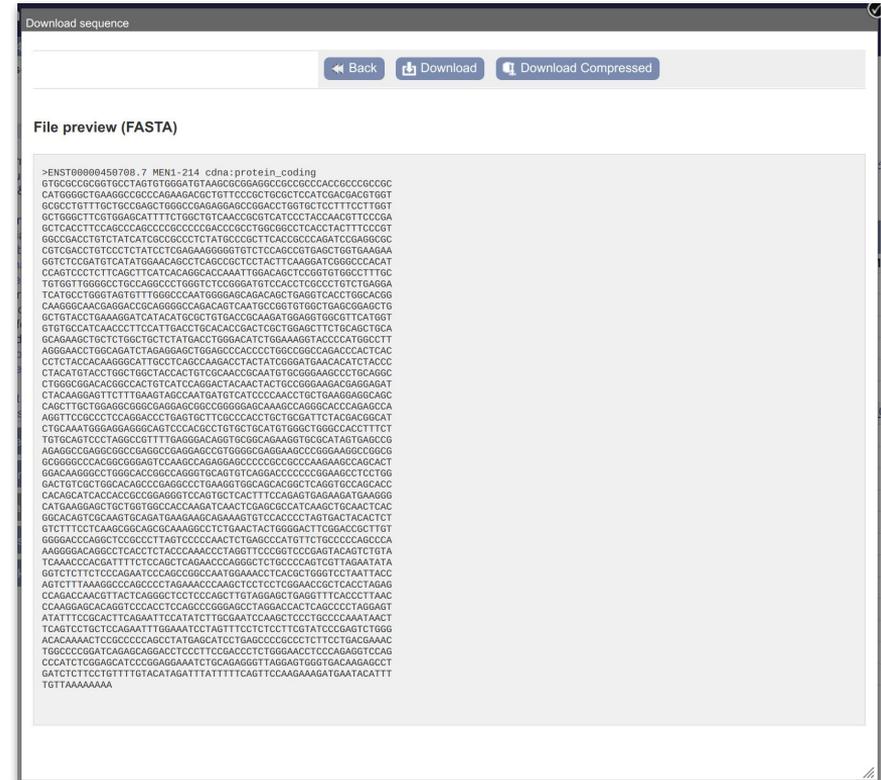
Seleccionamos  
sólo cDNA (ARNm)  
y le damos a  
Preview

# Secuencia del ARNm

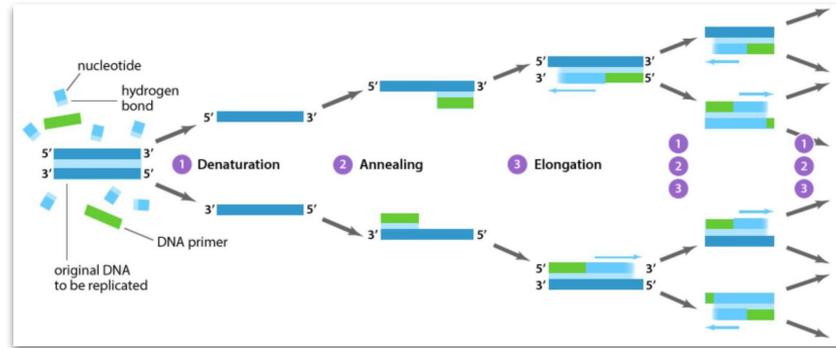
## Formato FASTA

Formato para representar secuencias de nucleótidos y aminoácidos.

- Línea de **encabezado** que comienza por ">". Contiene información sobre la secuencia (ID, nombre...)
- Líneas de **secuencia**



# ¿Cómo haría una PCR de este gen?

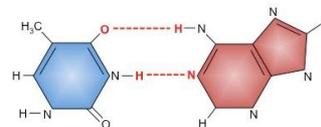


Respuesta: **Primer-BLAST**

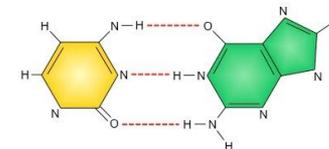
[Enlace](#)

# Diseño de primers para PCR

- **Longitud:** 18-24 pb de longitud
- **Contenido en G/C:**
  - Contenido en G/C del 40-60%
  - 1-2 pares G/C al principio y al final
- **Temperatura de desnaturalización (Tm):**
  - 50-60°C
  - Menos de 5°C de diferencia entre primers
- **Secuencia:**
  - Sin regiones complementarias entre ellos:**dímeros de primers**
  - Evitar regiones que formen estructuras secundarias internas
  - Evitar hibridaciones inespecíficas con otras partes del genoma

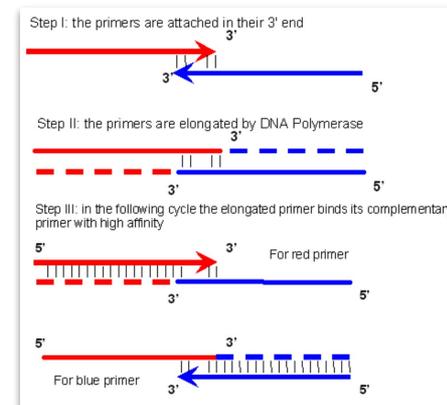


Thymine / Uracil pairs with Adenine  
(2 hydrogen bonds)



Cytosine pairs with Guanine  
(3 hydrogen bonds)

## Formación de dímeros de primers





# Primer-Blast

**Exon/intron selection**

A refseq mRNA sequence as PCR template input is required for options in the section ?

Exon junction span:  ?

Exon junction match: Min 5' match:  Min 3' match:  Max 3' match:

Minimal and maximal number of bases that must anneal to exons at the 5' or 3' side of the junction ?

Intron inclusion:  Primer pair must be separated by at least one intron on the corresponding genomic DNA ?

Intron length range: Min:  Max:  ?

Note: Parameter values that differ from the default are highlighted in yellow

**Primer Pair Specificity Checking Parameters**

Specificity check:  Enable search for primer pairs specific to the intended PCR template ?

Search mode:  ?

Database:  ?

Exclusion:  Exclude predicted Refseq transcripts (accession with XM, XR prefix)  Exclude uncultured/environmental sample sequences ?

Organism:  [Add organism](#) ?

Enter an organism name (or organism group name such as enterobacteriaceae, rodents), taxonomy id or select from the suggestion list as you type. ?

Entrez query (optional):  ?

Primer specificity stringency: Primer must have at least  total mismatches to unintended targets, including at least  mismatches within the last  bps at the 3' end. ?

Ignore targets that have  or more mismatches to the primer. ?

Max target amplicon size:  ?

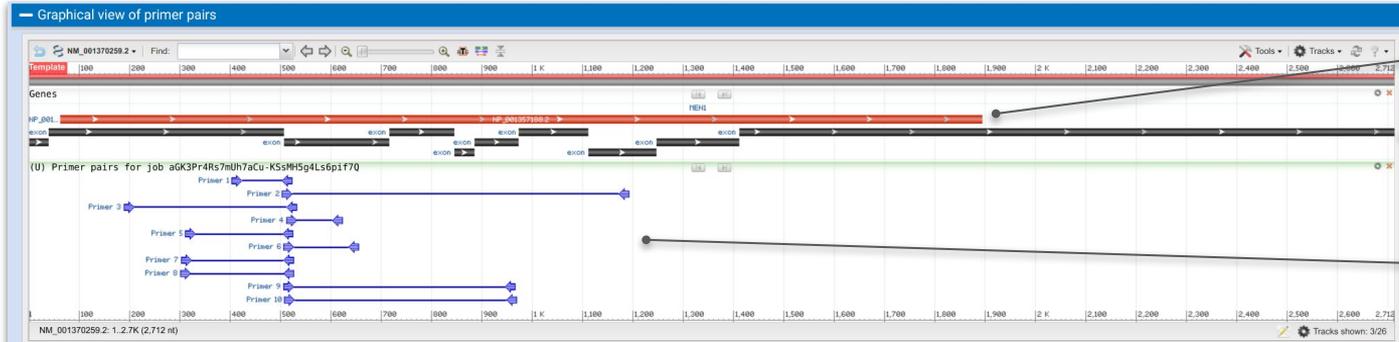
Allow splice variants:  Allow primer to amplify mRNA splice variants (requires refseq mRNA sequence as PCR template input) ?

Show results in a new window  Use new graphic view ?

**Get Primers**

Primer-Blast tiene en cuenta las secuencias del **resto de transcritos de humano** para no generar primers con hibridaciones **inespecíficas**

# Primer-Blast



Región codificante

ARNm (codificante + UTRs)

Parejas de primers diseñados

Detailed primer reports

You can re-search for specific primers by accepting some of the unintended targets, check the box(es) next to the ones you accept and try again to re-search for specific primers  ?

Primer pair 1

	Sequence (5'→3')	Template strand	Length	Start	Stop	Tm	GC%	Self complementarity	Self 3' complementarity
Forward primer	CGTGAGCTGGTGAAGAAGGT	Plus	20	404	423	59.97	55.00	4.00	0.00
Reverse primer	GCTGTCCAATTTGGTGCCTG	Minus	20	523	504	60.04	55.00	7.00	1.00
Product length	120								

Products on intended targets

>NM\_001370259.2 Homo sapiens menin 1 (MEN1), transcript variant 4, mRNA

product length = 120

Forward primer 1 CGTGAGCTGGTGAAGAAGGT 20  
 Template 404 ..... 423

Reverse primer 1 GCTGTCCAATTTGGTGCCTG 20  
 Template 523 ..... 504

Products on potentially unintended templates

> NM\_001370262.1 Homo sapiens menin 1 (MEN1), transcript variant 7, mRNA

Secuencias de los primers *forward* y *reverse*

Longitud de la secuencia amplificada (pb)

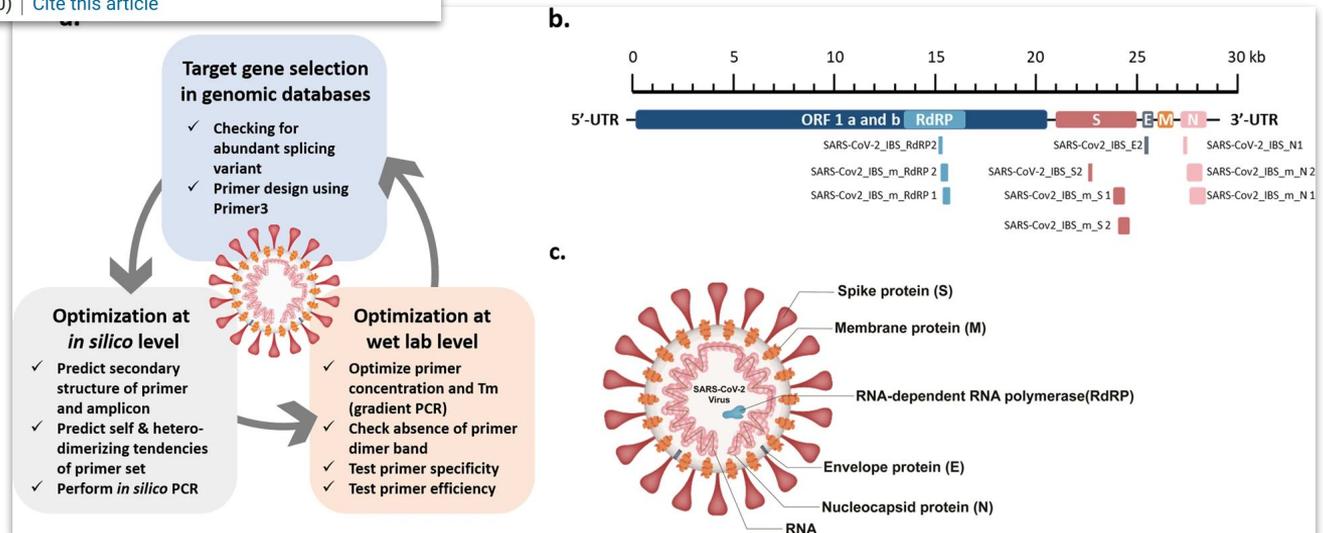
# Diseño de primers para SARS-CoV-2

Article | [Open Access](#) | Published: 16 June 2020

## Optimization of primer sets and detection protocols for SARS-CoV-2 of coronavirus disease 2019 (COVID-19) using PCR and real-time PCR

Myungsun Park, Joungha Won, Byung Yoon Choi & C. Justin Lee 

*Experimental & Molecular Medicine* 52, 963–977(2020) | [Cite this article](#)



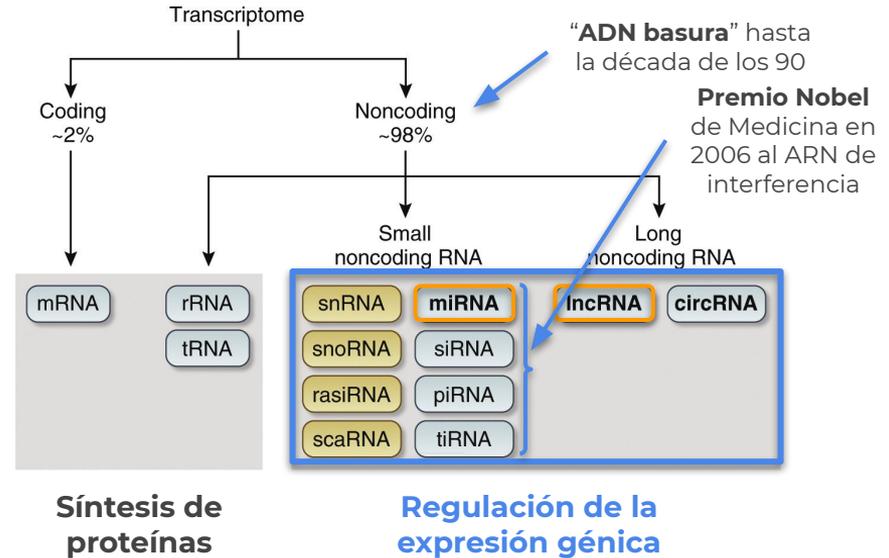
# ¿Existe algún microARN que regule al gen?

Respuesta: 

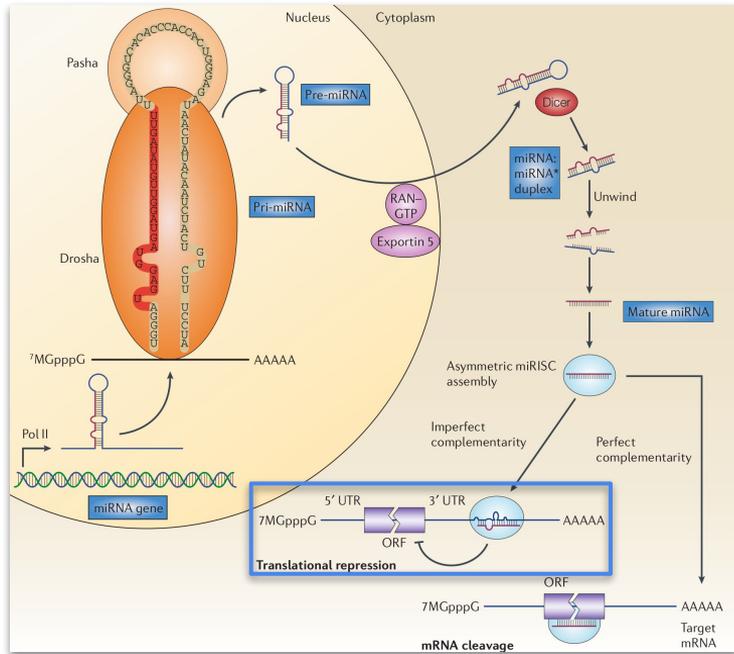
[Enlace](#)

# ARNs no codificantes: miRNA y lncRNA

- No traducidos a proteína
- Proceden de “genes de ARN” (color **morado** en Ensembl)
- **En síntesis de proteínas:** ARN ribosómico y de transferencia
- **En regulación de la expresión:** miRNA, lncRNA, entre otros.



# microRNAs

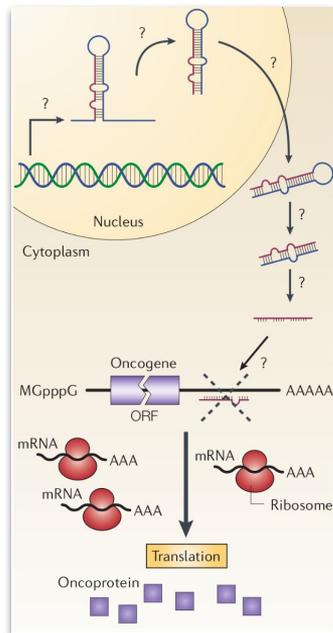


Biogénesis y actuación de miRNA

- ARN pequeños (18-20 nt)
- Regulan **negativamente** la expresión de genes
- Dos mecanismos de acción:
  - Inhiben la traducción del ARNm uniéndose a su **extremo 3'** (hibridación **imperfecta**, más frecuente)
  - Provocan la escisión del ARNm uniéndose a regiones **codificantes** (hibridación **perfecta**)
- Un miRNA puede controlar **varios genes** y un gen puede estar controlado por **varios miRNA**

# microRNAs y cáncer: Oncomirs

miRNA como **supresor tumoral**  
(silenciando un oncogen)

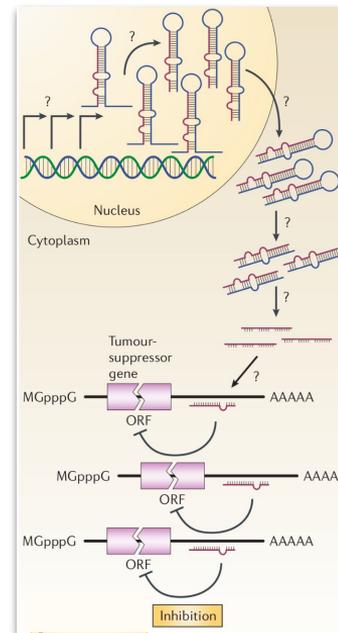


Si se infraexpresa

Si se sobreexpresa

**Formación de un tumor**  
↑ Proliferación  
↑ Invasión  
↑ Angiogénesis  
↓ Muerte celular

miRNA como **oncogen**  
(silenciando un supresor tumoral)



# miRWalk

Base de datos sobre relaciones entre miRNAs y genes.

**miRWalk**

HOME FAQ RESOURCES ABOUT

### News and Updates:

- Jan/2021 - **new\_update\_2021** - Annual update for the year 2021 was completed. At the request of numerous users, the duplex information has been integrated and can now be saved.
- Apr/2020 - **SERVER\_PROBLEMS** - Due to the high access rate, we are getting into some problems. We have decided to move the database to a stronger server. This can take some time and we apologize for the down-times. We will work on it as soon as possible. The miRWalk team.
- Mar/2020 - **genesets\_update** - All genesets (KEGG, GO and Reactome) were updated.
- Mar/2020 - **new\_update** - Data from all species were updated. Since a bug in the code some genes were missing in the last data update from december last year.
- Mar/2020 - **danio\_rerio** - new organism added. Zebrafish (Danio rerio) with all genes and transcripts is included into miRWalk database now.  
[read more...](#)

### New version of miRWalk

miRWalk is an improved version of the previous database (i.e. miRWalk). The new version of miRWalk stores predicted data obtained with a machine learning algorithm including experimentally verified miRNA-target interactions. The focus lies on accuracy, simplicity, user-friendly design and mostly up to date informations. More information can be obtained under [Frequently Asked Questions](#).

## Buscamos *MEN1* en el campo *Gene*

### Search for a single gene or miRNA

miRNAs: miRNA names (e.g. hsa-miR-214-3p) or Accession numbers (e.g. MIMAT0000271) based on current miRBase. While searching single miRNAs, also short names or family miRNA (e.g. let-7) belongs to several miRNAs are also acceptable. A list of miRNAs will be shown. mRNAs: Official Genesymbols (e.g. GAS2), EntrezIDs (e.g. 10608), Ensembl-IDs (e.g. ENSG00000148935 or ENST00000454564) and RefseqIDs (e.g. NM\_001143830) were accepted.

species **human** Gene **MEN1** miRNA search

# miRNA asociados al gen

Identificador del miRNA

Compuesto de 4 partes:

- Especie: hsa es *Homo sapiens*
- Procesamiento: miR es miRNA maduro (mir sería el gen o pre-miRNA)
- Identificador numérico
- Brazo: 5' o 3' (sólo para miRNA maduros)

## MEN1

Entrezid [4221](#)  
Genesymbol [MEN1](#)  
Alias MEAI;SCG2  
Description Homo sapiens merin 1 (MEN1), transcript variant 1, mRNA.  
Chromosome 11  
Ensemblid [ENSG00000133895](#)

### Transcripts:

Refseq	Ensemblid	Length	Startcds	Endcds
NM_001370251	ENST00000672304	2828	50	2008
NM_001370259	ENST00000450708	2712	62	1894
NM_001370260	ENST00000440873	2702	50	1882
NM_001370261	ENST00000315422	2960	308	2140
NM_001370263	ENST00000377321	2609	62	1789
NM_130799	ENST00000312049	2770	111	1943
NM_130801	ENST00000377313	2736	62	1909
NM_130802	ENST00000394374	3172	498	2345
NM_130803	ENST00000337652	3179	505	2352
NM_130804	ENST00000394376	3051	377	2224

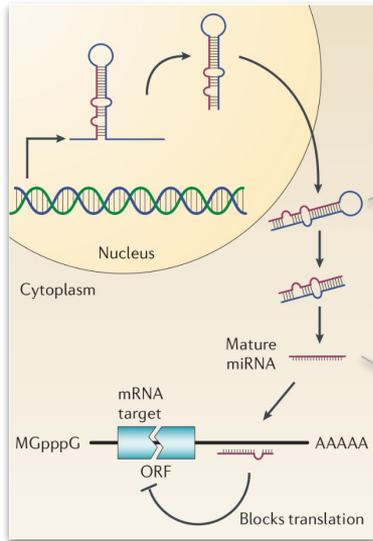
Región del gen donde se une el miRNA

## Seleccionamos un miRNA

### Interactions:

Mirna	Refseqid	Genesymbol	Duplex	Score	Position	Binding Site	Au	Me	N Pairings	Targets can	Mirdb	Mirtarbase
hsa-miR-6877-5p	NM_130799	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	1976,1996	0.32	-7.238	16	—	—	—
hsa-miR-6877-5p	NM_130801	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	1942,1962	0.32	-7.238	16	—	—	—
hsa-miR-6877-5p	NM_130802	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	2378,2398	0.32	-7.238	16	—	—	—
hsa-miR-6877-5p	NM_001370259	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	1927,1947	0.32	-7.238	16	—	—	—
hsa-miR-6877-5p	NM_001370260	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	1915,1935	0.32	-7.238	16	—	—	—
hsa-miR-6877-5p	NM_001370261	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	2173,2193	0.32	-7.238	16	—	—	—
hsa-miR-6877-5p	NM_001370263	MEN1	((( (((((((((( (&)))))))))))))	0.80	3UTR	1822,1842	0.32	-7.238	16	—	—	—
hsa-miR-125b-2-3p	NM_130799	MEN1	.....((((((((((( (&))))))))))	0.81	3UTR	2676,2693	0.59	-15.567	14	—	—	—
hsa-miR-520e-3p	NM_130799	MEN1	---((( (((((((((( (&))))))))))	0.81	3UTR	2321,2341	0.4	-9.685	16	—	—	—
hsa-miR-4280	NM_130799	MEN1	((( (((((((((( (&))))))))))	0.81	3UTR	2105,2129	0.48	-9.6	17	—	—	—

# Información del miRNA en miRBase



The screenshot shows the miRBase database entry for **hsa-miR-6877**. The page is divided into several sections:

- Stem-loop sequence hsa-miR-6877**: Shows the pre-miRNA hairpin structure with the sequence: 5' aguuc g ug a cu g and 3' gggcc uucc ca cga u cuu c. A "Get sequence" button is present.
- Deep sequencing**: Shows 373 reads, 0 reads per million, and 92 experiments. A bar chart displays the sequencing data.
- Confidence**: States "Annotation confidence: not enough data" and includes a feedback form with "Yes", "No", and "Leave comment" options.
- Genome context**: Provides coordinates for GRCh38: GCA\_000001405.15 on chromosome 9: 133051996-133052059 [+].
- Database links**: Lists HGNC: MIR6877 and miRBase Tracker: MI0022724.
- Mature sequence hsa-miR-6877-5p**: Shows accession MIMAT0027654 and sequence 6 - agggcgaagguggaagcugc - 27. A "Get sequence" button is present.
- Deep sequencing**: Shows 344 reads and 81 experiments.
- Evidence**: Lists "experimental; meta-analysis [1]".
- Predicted targets**: Lists TargetScanVert: hsa-miR-6877-5p and miRDB: hsa-miR-6877-5p.
- Mature sequence hsa-miR-6877-3p**: Shows accession MIMAT0027655 and sequence 40 - cagccucugccuuggccucc - 60.

Si no se abre esta página de miRBase, copia el identificador del miRNA (**hsa-miR-6877-5p** en este caso), haz click [aquí](#) y pégalo en el buscador de la derecha ("Search by miRNA name or keyword")

**Estructura que forma el pre-miRNA**

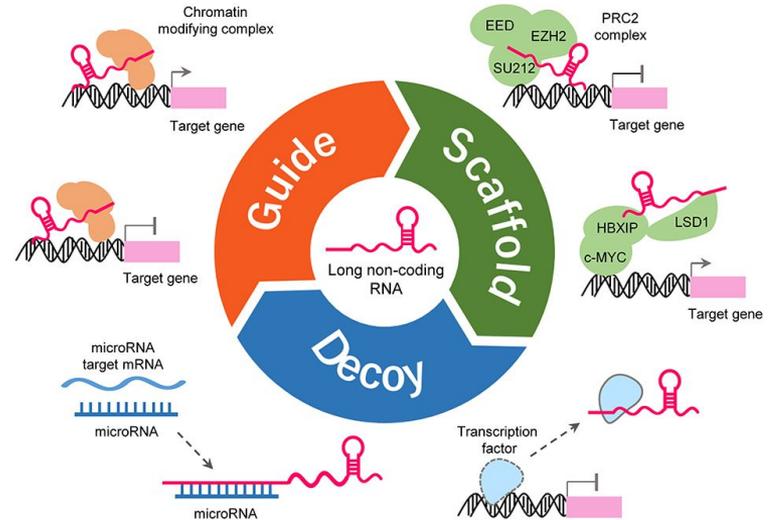
**Secuencia del brazo 5'**

(miRNA maduros)

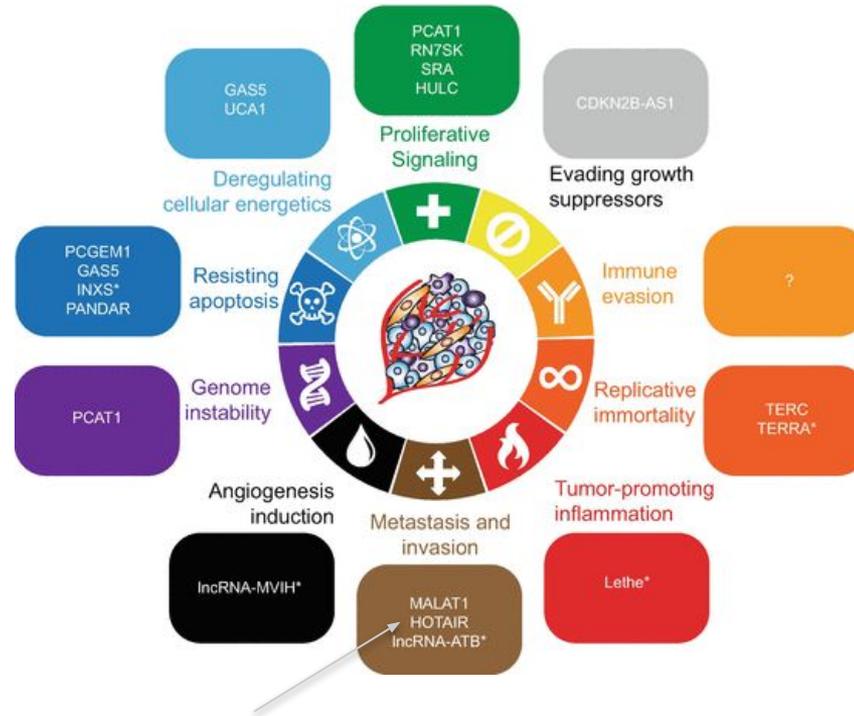
**Secuencia del brazo 3'**

# lncRNA

- *Long non-coding RNA*
- > **200** nucleótidos
- **Regulación genética** a varios niveles: epigenético, transcripción, post-transcripcional...
- Diversos mecanismos de acción:
  - **Guía:** unión a ADN
  - **Scaffold** (andamio): unión a complejos proteicos
  - **Esponja:** unión y secuestro de miRNAs, factores de transcripción...



# IncRNA y cáncer



Entrada del lncRNA *HOTAIR* en la base de datos RNAcentral ([enlace](#))

# ¿Qué laboratorios realizan el diagnóstico genético en España?

Respuesta:



[Enlace](#)

# Buscar en NCBI GTR



COVID-19 is an emerging, rapidly evolving situation.



[Public health information \(CDC\)](#) | [Research information \(NIH\)](#) | [SARS-CoV-2 data \(NCBI\)](#) | [Prevention and treatment information \(HHS\)](#)

## GTR: GENETIC TESTING REGISTRY

### Pruebas en humanos

All GTR

Human Tests

Microbe Tests

Conditions/Phenotypes

Genes

Labs

GeneReviews

Advanced search for tests

multiple endocrine neoplasia, type 1|

Search Human Tests

MEN1: Multiple endocrine neoplasia, type 1

Condition

Buscamos el nombre de la enfermedad

Find tests by searching test names, disease names, phenotypes, gene symbols and names, protein names, laboratory names, directors and locations.

YouTube GTR Tutorials

**IMPORTANT NOTE:** NIH does not independently verify information submitted to the GTR; it relies on submitters to provide information that is accurate and not misleading. NIH makes no endorsements of tests or laboratories listed in the GTR. GTR is not a substitute for medical advice. *Patients and consumers* with specific questions about a genetic test should contact a health care provider or a genetics professional.

# Tests genéticos de MEN1 en España

multiple endocrine neoplasia, type 1

Human Tests

Human tests (38) Microbe tests (0) Conditions (1) Genes (1) Laboratories (76)

Filters

Test type:  Clinical (38)

Test purpose:  Diagnosis (38)

Lab location:  Spain (38)

Results: 1 to 20 of 38

Tests names and labs	Conditions	Genes, analytes, and microbes	Methods
<a href="#">Oncology Genetic Panel, Panel Massive Sequencing (NGS) 90 Genes</a> Reference Laboratory Genetics Spain	126	90	C Sequence analysis of the entire coding region
<b>MULTIPLE ENDOCRINE NEOPLASIA TYPE 1 (MEN1)</b> Laboratorio de Genetica Clinica SL Spain	1	1	D Deletion/duplication analysis C Sequence analysis of the entire coding region
<a href="#">Multiple Endocrine Neoplasia Type 1, Deletions-Duplications (MLPA) MEN1 Gene</a> Reference Laboratory Genetics Spain	1	1	D Deletion/duplication analysis
<a href="#">Multiple Endocrine Neoplasia Type 1, Sequencing MEN1 Gene</a> Reference Laboratory Genetics Spain	1	1	C Sequence analysis of the entire coding region
<a href="#">Multiple endocrine neoplasia type 1</a> Bioarray Spain	1	1	C Sequence analysis of the entire coding region
<a href="#">Multiple endocrine Neoplasia type 1 (MEN1); MEN1 gene sequence analysis</a> Bioarray Spain	1	1	C Sequence analysis of the

Seleccionamos Human tests

Aplicamos 3 filtros

Seleccionamos una prueba

Enfermedades que diagnostica la prueba

Tipo de prueba

# Tests genéticos de MEN1 en España

NCBI Resources How To marisolbc@go.ugr.es My NCBI Sign Out

GTR: GENETIC TESTING REGISTRY

multiple endocrine neoplasia, type 1 Human Tests Search Advanced search for tests

GTR Home > Tests > MULTIPLE ENDOCRINE NEOPLASIA TYPE 1 (MEN1)

### MULTIPLE ENDOCRINE NEOPLASIA TYPE 1 (MEN1)

Clinical test for [Multiple endocrine neoplasia, type 1](#)  
Offered by [Laboratorio de Genetica Clinica SL](#)

GTR Test ID: GTR000558399.1  
Last updated: 2020-10-09  
[Test version history](#)

Reviews  
GeneReviews  
PubMed Clinical Queries  
Reviews in PubMed

Clinical resources  
MedGen  
OMIM  
et  
trials.gov

Practice guidelines  
ACMG, 2016  
ACMG/NSGC, 2015  
ACMG, 2015  
ACMG, 2013  
Thakker et al., 2012

Overview How To Order Indication Methodology Performance Characteristics Interpretation **Laboratory Contact**

**Laboratory information**  
Laboratorio de Genetica Clinica SL, LabGenetics

**Laboratory director(s)**  
Jorge Puente, Lab Director  
Angela Sesto, PhD, Scientific Director

**Laboratory contact**  
Calle Poeta Rafael Morales, 2. Planta 2  
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**Laboratory affiliations**  
Private center

Phone number: +34916592298  
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Email: [info@labgenetics.com.es](mailto:info@labgenetics.com.es)  
Website: <http://www.labgenetics.es/>

**Nombre del laboratorio**

**Datos de contacto**

# Entrega en PRADO: 30/05/2021 - 23:55h

Rellenad individualmente la tabla de la derecha con otra enfermedad monogénica a vuestra elección.

## Consejos:

- En Ensembl intentad elegir el transcrito con la etiqueta “MANE select v0.92” (transcrito de referencia). Podéis escoger cualquier variante patogénica.
- Elegid cualquier pareja de primers diseñados por Primer-Blast.
- Elegid cualquier miRNA en miRWalk.

Cualquier duda, escribidme a [marisolbc@ugr.es](mailto:marisolbc@ugr.es)

PRÁCTICA II: BIOINFORMÁTICA	
Nombre y apellidos: María Soledad Benítez Cantos	
<b>Enfermedad:</b>	
Neoplasia endocrina múltiple de tipo 1	
Nombre oficial del gen responsable	<i>MEN1</i>
Alias	menin
Evidencia de la asociación gen-enfermedad	Frase: "We conclude that many "hyperplastic" parathyroid tumors in familial MEN-1 are in fact monoclonal and may progress or even begin to develop by inactivation of the MEN-1 gene (at 11q13) in a precursor cell." Enlace a publicación: <a href="https://pubmed.ncbi.nlm.nih.gov/2568586/">https://pubmed.ncbi.nlm.nih.gov/2568586/</a>
Ensembl ID del gen	ENSG00000133895
Localización cromosómica (cromosoma, coordenadas y hebra)	Chromosome 11: 64,803,510-64,811,294 reverse strand
Genes adyacentes	MAP4K2, SF1, RASGRP2
dbSNP ID de una variante patogénica	rs104894266
Consecuencia de la variante	Codón de parada prematuro
Número de transcritos	19
Ensembl ID del transcrito seleccionado	ENST00000450708.7
Longitud del transcrito (pb)	3162
Número de exones	10
Longitud de la proteína (aa)	615
Secuencia del transcrito en formato FASTA	>ENST00000450708.7 MEN1-214 cDNA:protein_coding GTGCCCGCGGTGCTAGTGTGGGATGTAAGCGCG GAGGCCCGCCGCCACCGCCGCCGC (...)
Primers diseñados	Forward: CGTGAGCTGGTGAAGAAGGT Reverse: GCTGTCCAATTTGGTGCCTG
Identificador del miRNA que lo regula	hsa-miR-6877-5p
Secuencia del miRNA maduro	agggccgaagguggaagcugc
Laboratorio que realiza el diagnóstico genético de la enfermedad (preferiblemente en España)	Laboratorio de Genética Clínica SL, LabGenetics (Madrid)

# ¿Cómo escojo un gen?

Si no conoces ninguna enfermedad monogénica, DisGeNet tiene una [tabla](#) con todas las asociaciones gen-enfermedad publicadas.

Seleccionar un gen cuya columna “Score” sea igual a 1 (la asociación es 100% fiable).

Source: ALL

1 - 25 of 1134942 results

Download Share

Add/Remove filter

Results per page 25

Filter within current results:

Gene	Gene Full Name	DSI <sub>g</sub>	DPI <sub>g</sub>	pLI	Disease	Type	Score <sub>gda</sub>	EL <sub>gda</sub>	EI <sub>gda</sub>	N. PMIDs	N. SNPs <sub>gda</sub>	First Ref.	Last Ref.
ESR1	estrogen rece...	0.324	0.962	1.00	Malignant ne...	disease	1.000	None	0.967	3371	41	1983	2020
BRCA1	BRCA1 DNA ...	0.367	0.923	9.2E-29	Malignant ne...	disease	1.000	strong	0.956	2827	251	1992	2020
CFTR	CF transmem...	0.424	0.885	2.2E-58	Cystic Fibrosis	disease	1.000	None	0.979	2327	632	1989	2020
AR	androgen rec...	0.351	0.846	0.99	Malignant ne...	disease	1.000	imited	0.982	1885	25	1992	2020
BRAF	B-Raf proto-o...	0.319	0.846	1.00	melanoma	disease	1.000	None	0.983	1637	35	1986	2020
BRCA2	BRCA2 DNA ...	0.379	0.846	2.4E-25	Malignant ne...	disease	1.000	strong	0.951	1524	212	1994	2020

# Diagramas biológicos con BioRender

**BioRender** es una plataforma online que cuenta con más de 20000 iconos relacionados con la Biología y que permite crear ilustraciones científicas de forma sencilla.

Útil para trabajos del grado, TFG, TFM...

The screenshot shows the BioRender website interface. At the top, there is a navigation menu with links for Features, Webinars, Icon Library, Pricing, Learning Hub, Testimonials, and Sign in, along with a 'Sign up free' button. Below the navigation is a search bar with the text 'Search 1000s of icons'. The main content area displays a 3D molecular model of a T cell receptor complex, including components like MHC Class II, CD4, CD3, and TCR, with an arrow indicating the release of IFN-γ. The BioRender logo is visible in the top left corner.

