Sequence Alignment

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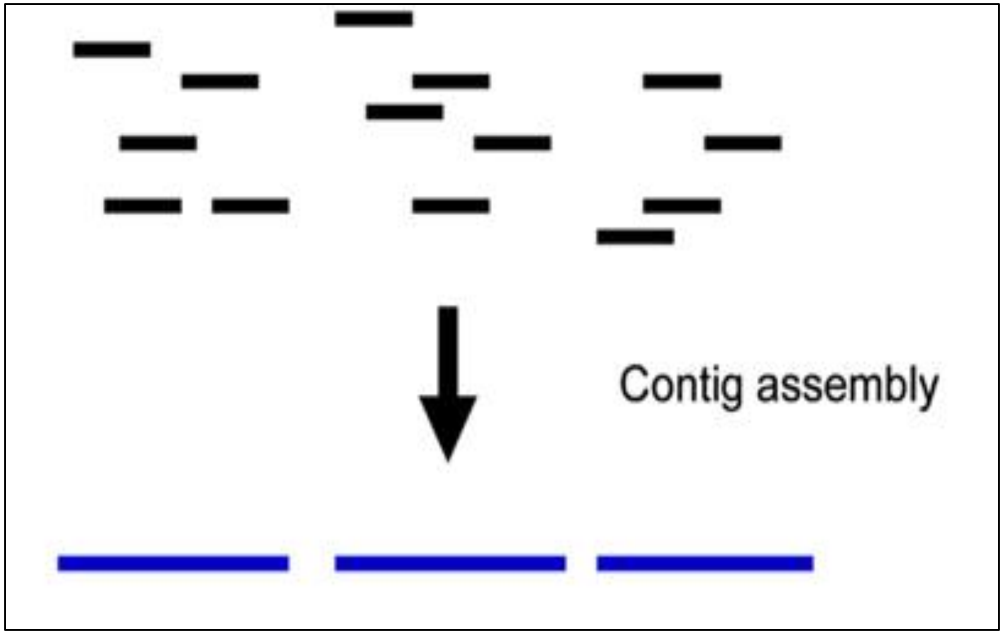
What is Sequence Alignment ?

A sequence alignment is a way of arranging the sequences of DNA , RNA, or protein to identify regions of similarity.

Comparable ?

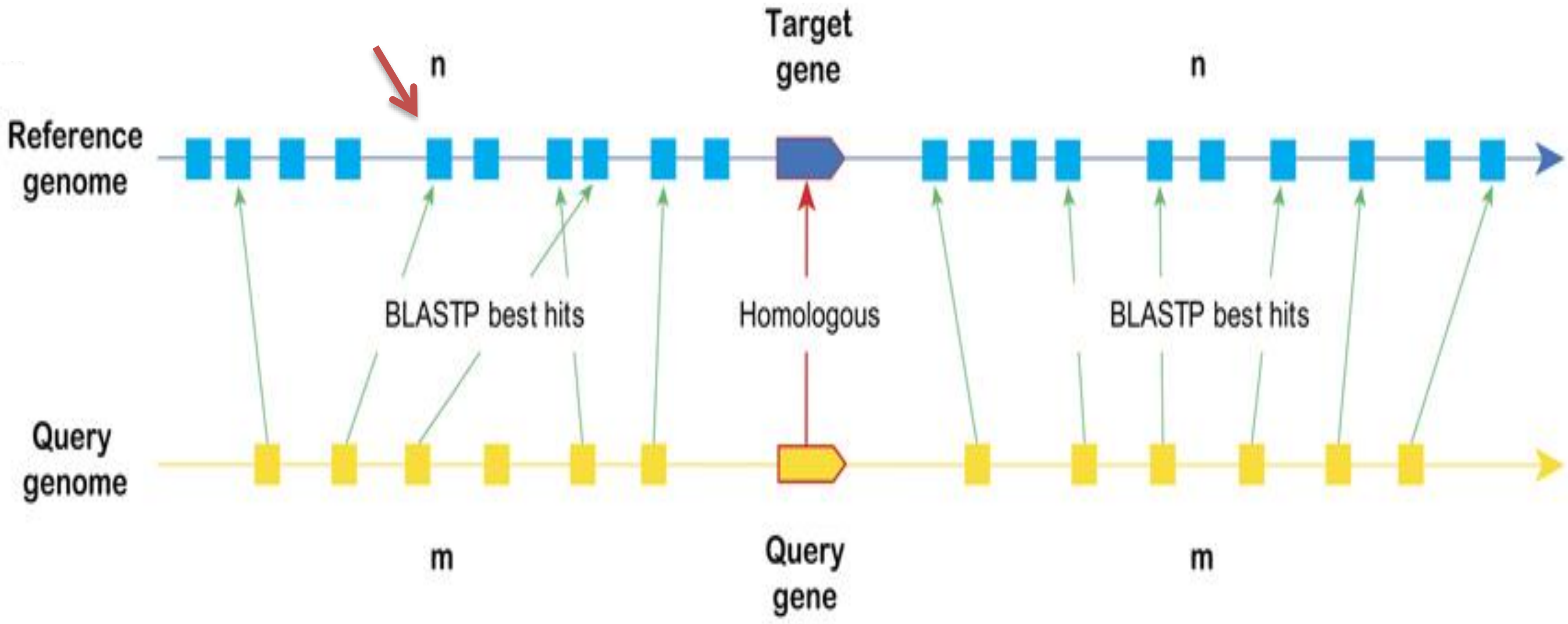
Sequence Alignment :Uses (1)

- **Sequence Assembly** : Genome sequence are assembled by using the sequence alignment methods to find the overlap between many short pieces of DNA .



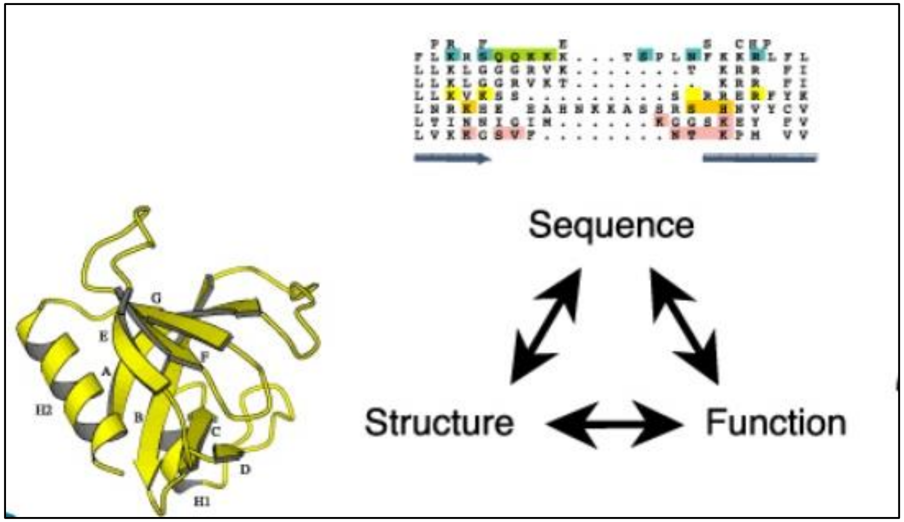
Sequence Alignment :Uses (2)

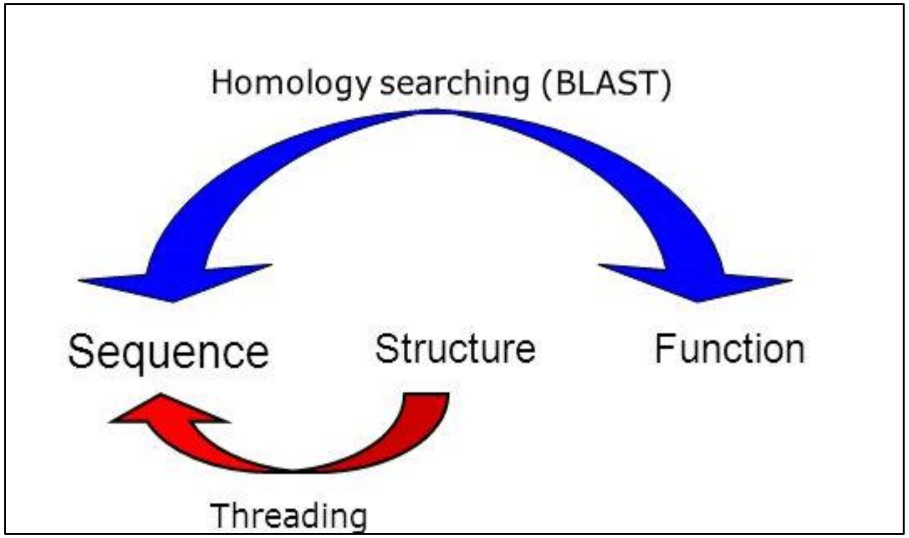
- **Gene Finding : Sequence similarity could help us to find the gene prediction just by doing comparison against the other set of sequences.**



Sequence Alignment :Uses (3)

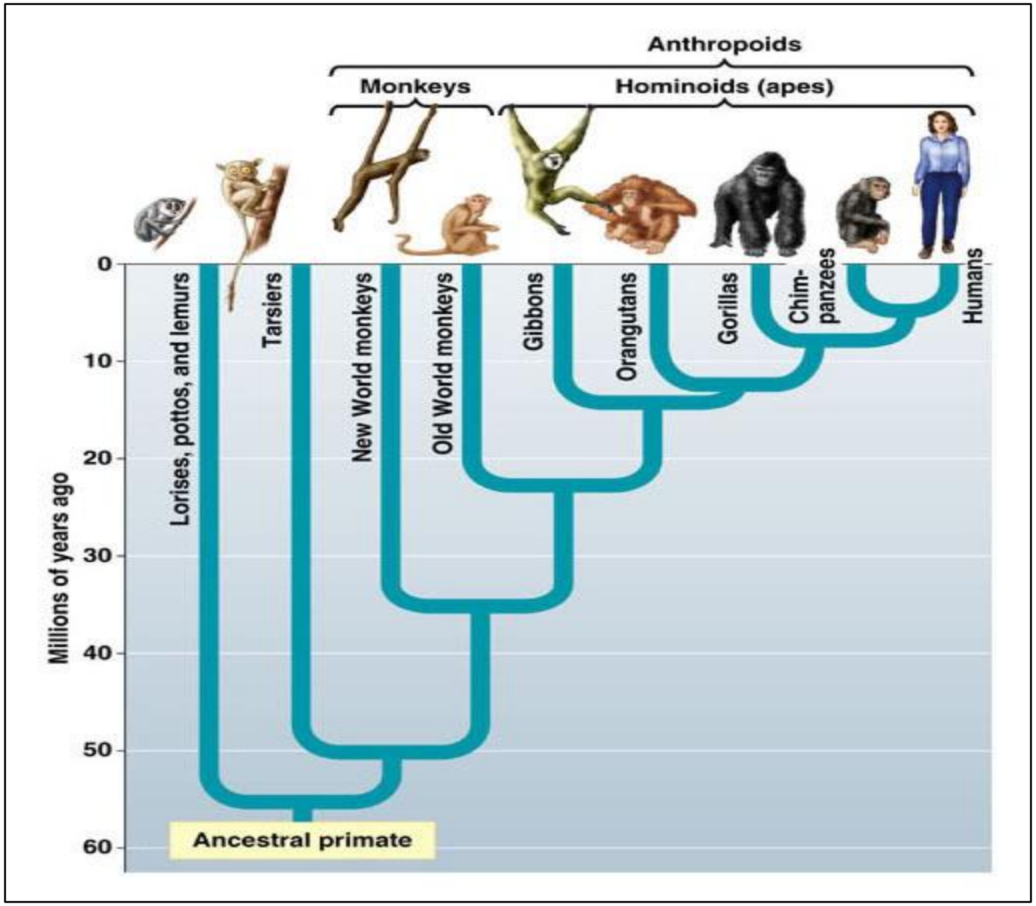
- **Function prediction : Function of any unknown sequence could be predicted by comparing with other known sequence .**





Sequence Alignment :Uses (4)

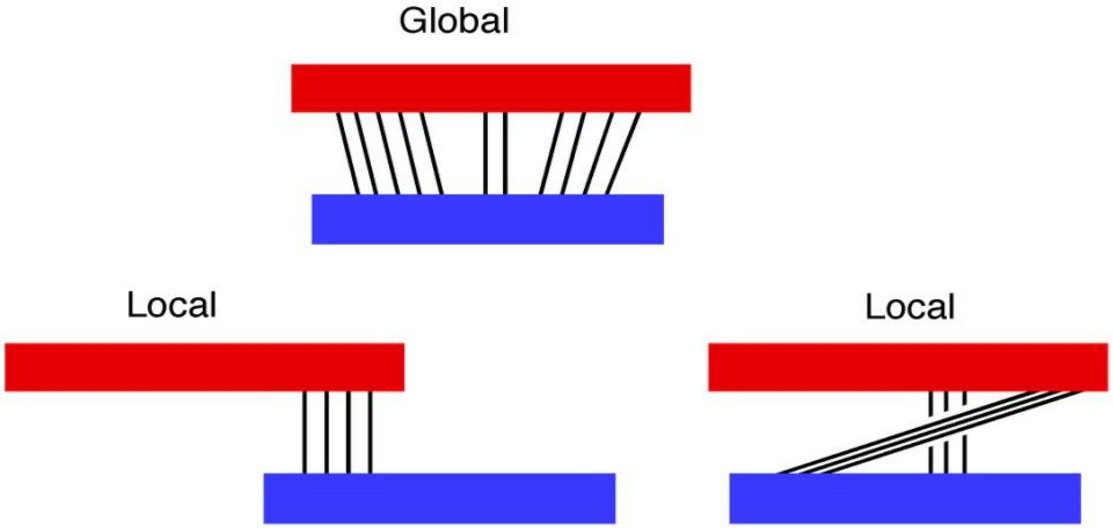
- Sequence Divergence : Amount of sequence similarity (10%, 20%,30% ...sometimes 90 %) between sequences tell us how closely they are related



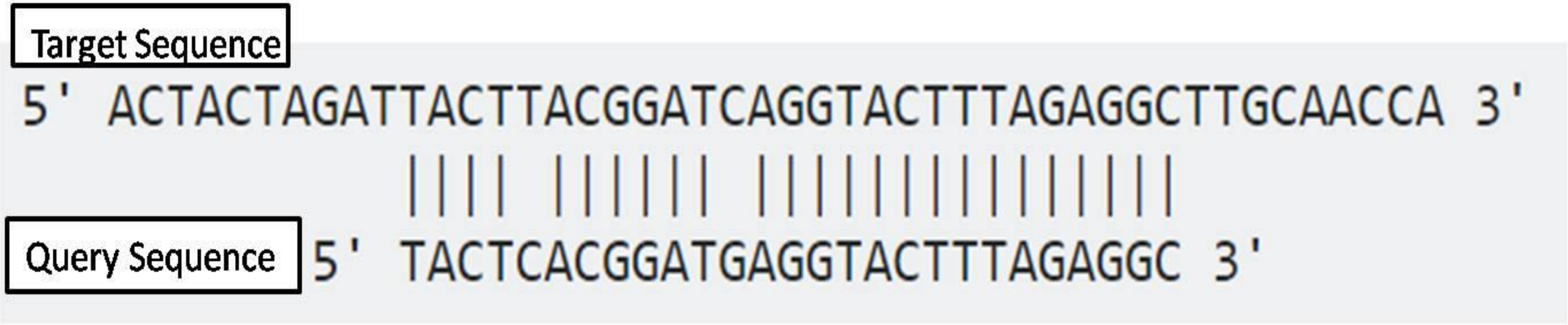
Types of Alignments

- **Global : This attempt to align every residue in every sequence.**

- **Local: It is more useful for dissimilar sequences that are suspected to contain regions of similarity or similar sequence motifs within their larger sequence context.**



Local Alignment

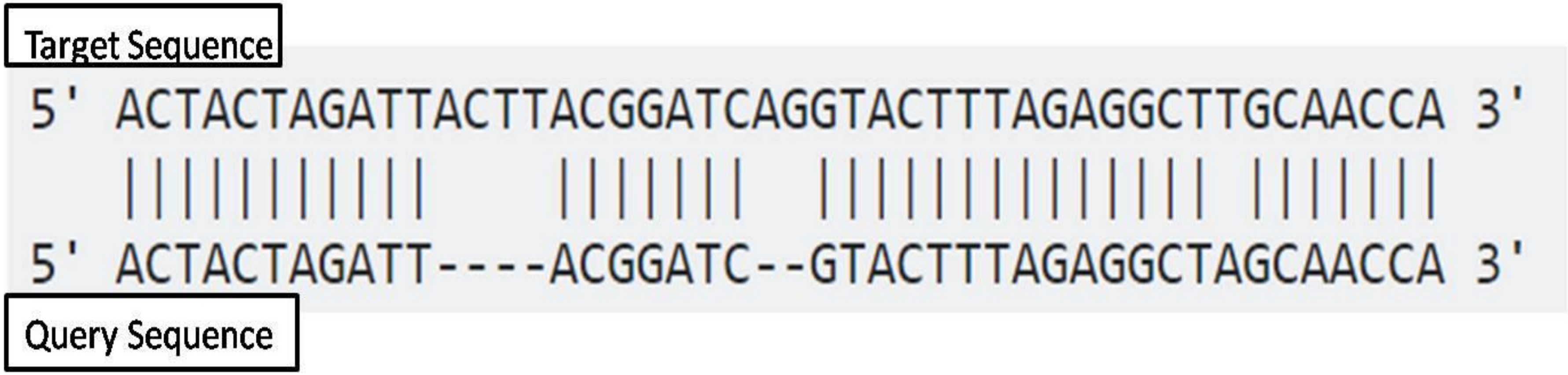


Target Sequence

```
5' ACTACTAGATTACTTACGGATCAGGTACTTTAGAGGCTTGCAACCA 3'
             |||| |||||| |||||||||||||||
          5' TACTCACGGATGAGGTACTTTAGAGGC 3'
```

Query Sequence

Global Alignment



Target Sequence

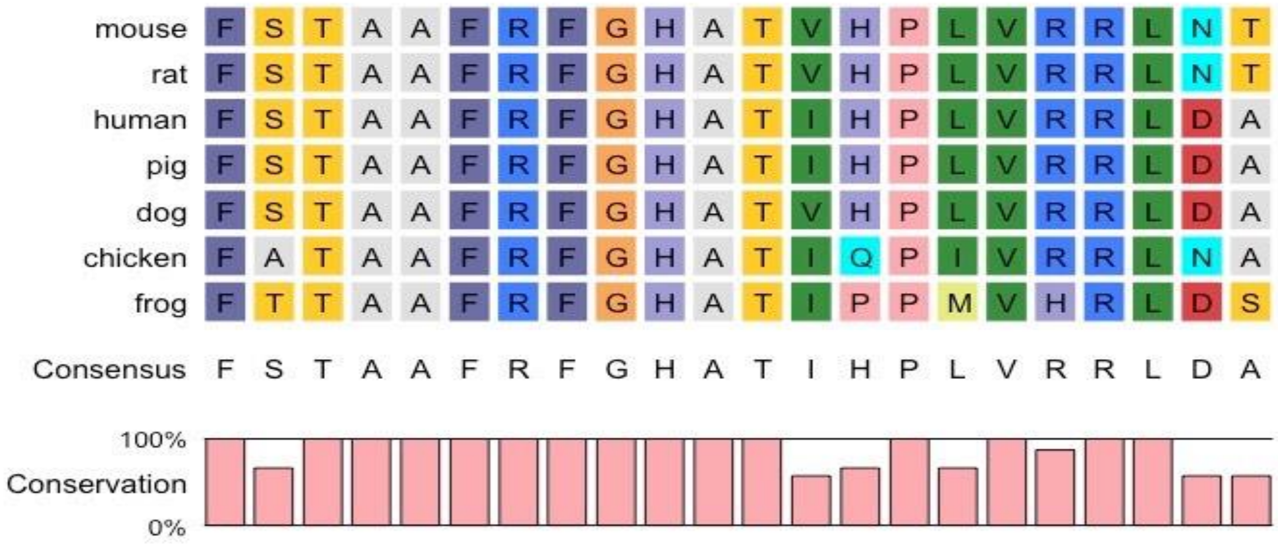
```
5' ACTACTAGATTACTTACGGATCAGGTACTTTAGAGGCTTGCAACCA 3'
   |||||||||||    |||||||  |||||||||||||| |||||||
5' ACTACTAGATT----ACGGATC--GTACTTTAGAGGCTAGCAACCA 3'
```

Query Sequence

Types of Alignments: Based on number of sequences

- **Pair wise Sequence Alignment** : This alignments can only be used between two sequences at a time.

- **Multiple Sequence Alignment** : This alignments can only be used between more than two sequences at a time.



mouse	F	S	T	A	A	F	R	F	G	H	A	T	V	H	P	L	V	R	R	L	N	T
rat	F	S	T	A	A	F	R	F	G	H	A	T	V	H	P	L	V	R	R	L	N	T
human	F	S	T	A	A	F	R	F	G	H	A	T	I	H	P	L	V	R	R	L	D	A
pig	F	S	T	A	A	F	R	F	G	H	A	T	I	H	P	L	V	R	R	L	D	A
dog	F	S	T	A	A	F	R	F	G	H	A	T	V	H	P	L	V	R	R	L	D	A
chicken	F	A	T	A	A	F	R	F	G	H	A	T	I	Q	P	I	V	R	R	L	N	A
frog	F	T	T	A	A	F	R	F	G	H	A	T	I	P	P	M	V	H	R	L	D	S
Consensus	F	S	T	A	A	F	R	F	G	H	A	T	I	H	P	L	V	R	R	L	D	A

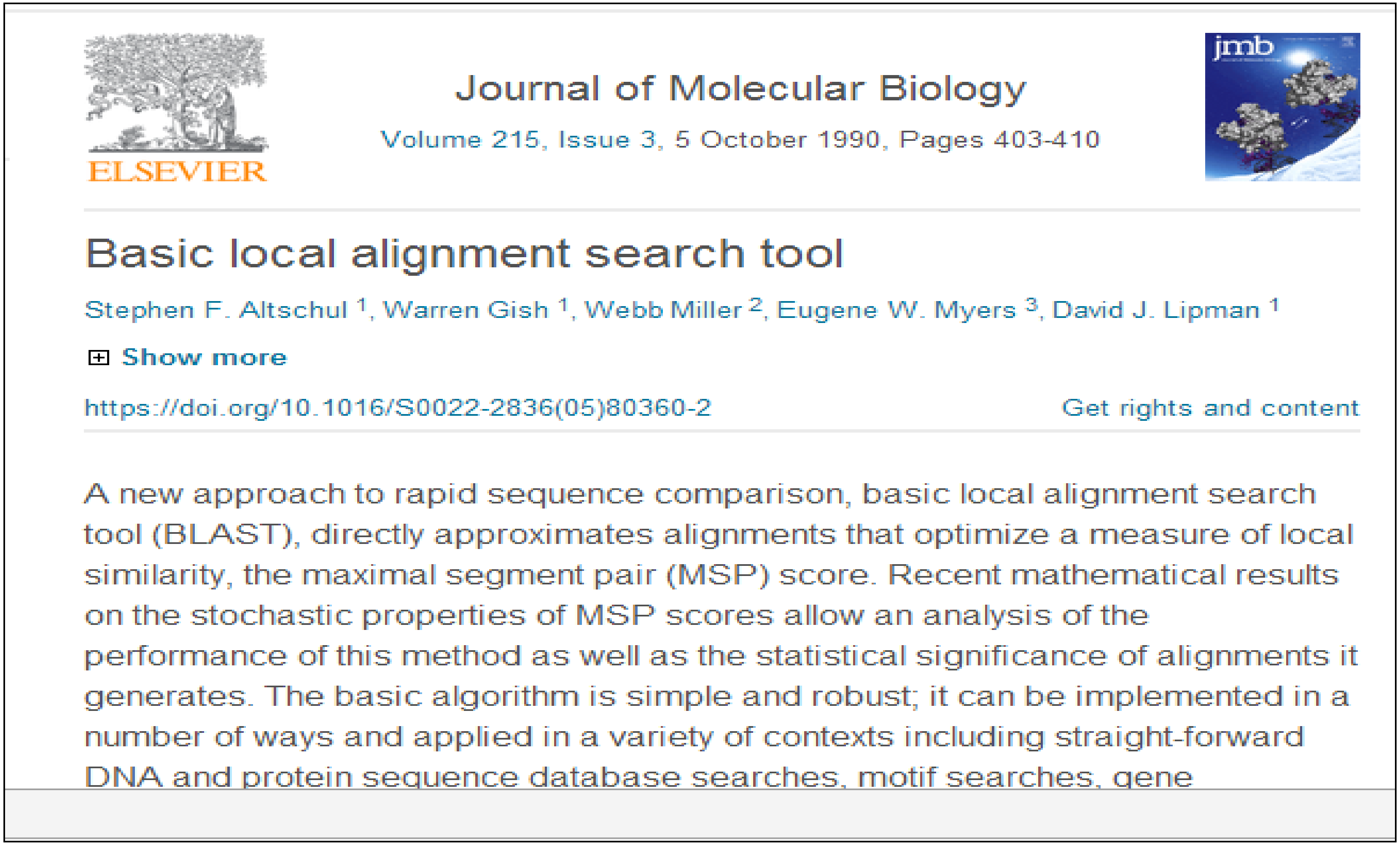
Tools for Sequence Alignments

There are many tools for sequence Alignment. In this session, we will discuss about

- **BLAST**
- **BLAT**
- **CLUSTALW**

Sequence Alignment : BLAST

- **BLAST** stands for **B**asic **L**ocal **A**lignment **S**earch **T**ool



ELSEVIER

Journal of Molecular Biology

Volume 215, Issue 3, 5 October 1990, Pages 403-410

Basic local alignment search tool

Stephen F. Altschul[1], Warren Gish[1], Webb Miller[2], Eugene W. Myers[3], David J. Lipman[1]

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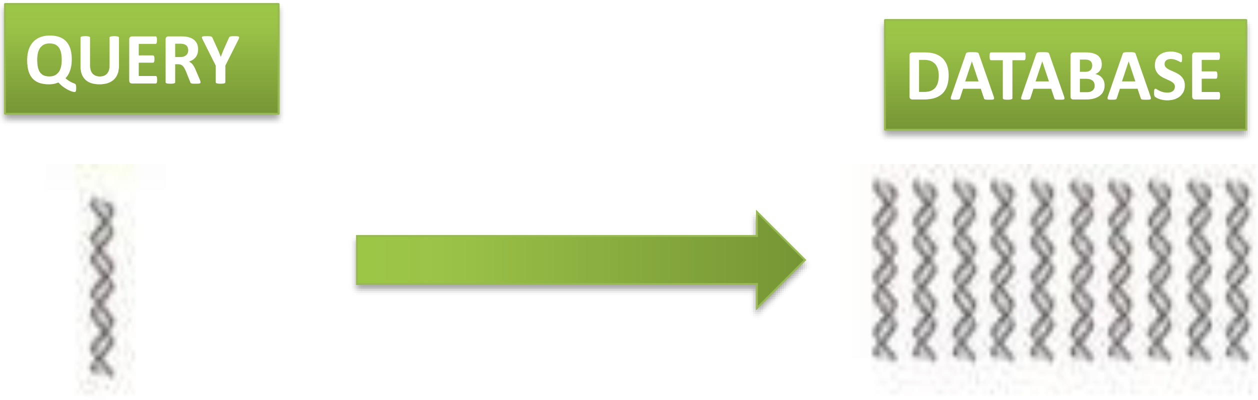
A new approach to rapid sequence comparison, basic local alignment search tool (BLAST), directly approximates alignments that optimize a measure of local similarity, the maximal segment pair (MSP) score. Recent mathematical results on the stochastic properties of MSP scores allow an analysis of the performance of this method as well as the statistical significance of alignments it generates. The basic algorithm is simple and robust; it can be implemented in a number of ways and applied in a variety of contexts including straight-forward DNA and protein sequence database searches, motif searches, gene

- **Blast was developed by Stephan Altschul and colleagues at NCBI in 1990.**

- **BLAST is an algorithm for comparing primary biological sequence information, such as the amino-acid sequences of proteins or the nucleotides of DNA sequences.**

- **Blast is most used bioinformatics program (cited >60000 times).**

- A BLAST search enables a researcher to compare a query sequence with a library or databases of sequences, and identify library sequences that resemble the query sequence above a certain threshold.



QUERY

DATABASE

Types of BLAST (1)

- **BLASTN : search nucleotide databases using a nucleotide query**

  (A)Query : ATGCATCGATC

  (B) Database : ATCGATGATCGACATCGATCAGCTACG

- **BLASTP : search protein databases using a protein query**

  (A)Query : VIVALASVEGAS

  (B) DATABASE : TARDEFGGAVIVADAVISASTILHGGQWLC

- **BLASTX : search protein databases using a translated nucleotide query**

  (A)Query : ATGCATCGATC

  (B)DATABASE : TARDEFGGAVIVADAVISASTILHGGQWLC

Types of BLAST (2)

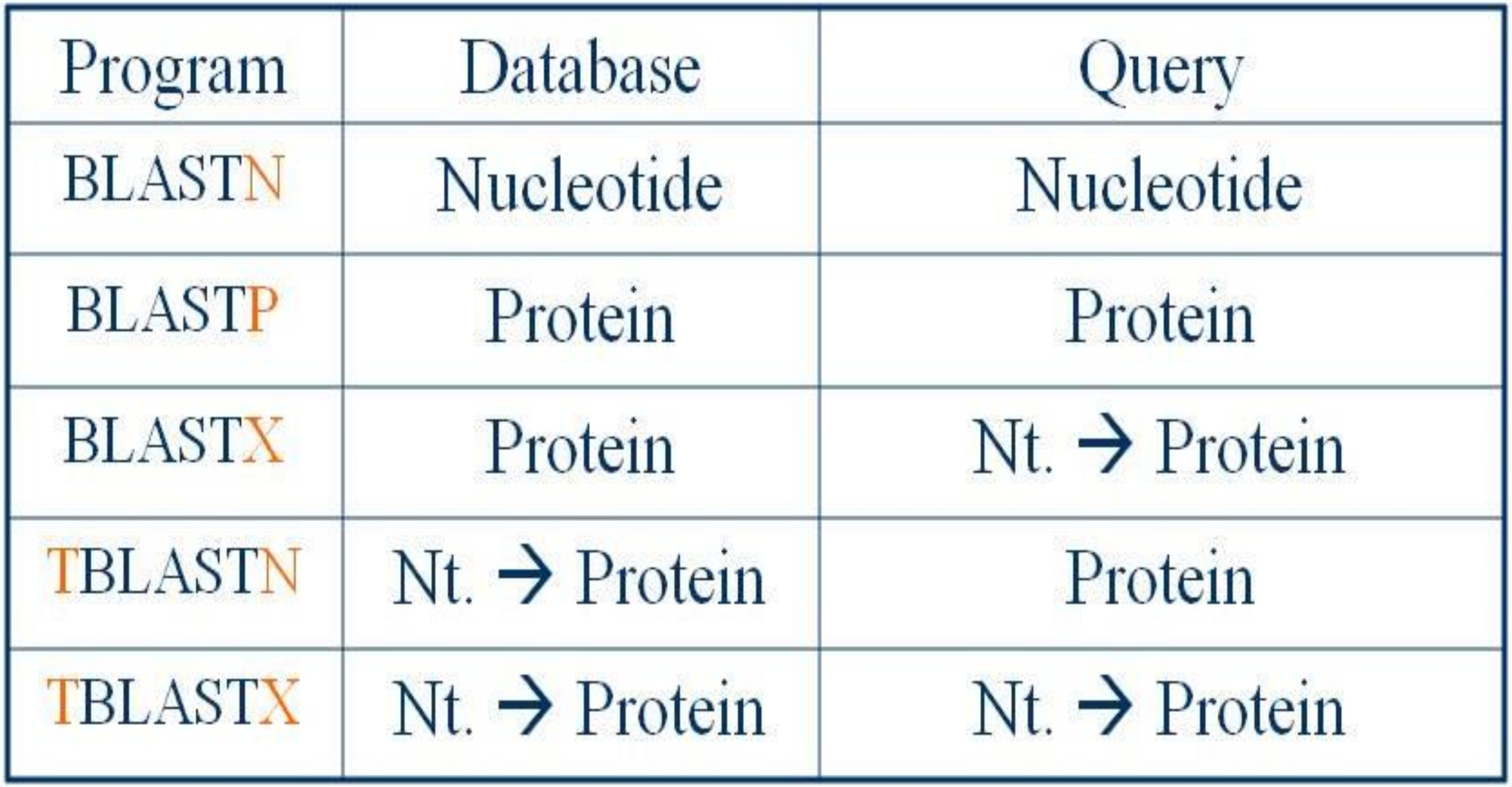
- **TBLASTN : search translated nucleotide databases using a protein query**

  (A)Query : TARDEFGGAVI
  (B)DATABASE : ATCGATGATCGACATCGATCAGCTACG

- **TBLASTX : search translated nucleotide databases using a translated nucleotide query**

  (A)Query : CGATGATCG
  (B)DATABASE : ATCGATGATCGACATCGATCAGCTACG

Types of BLAST : ALL

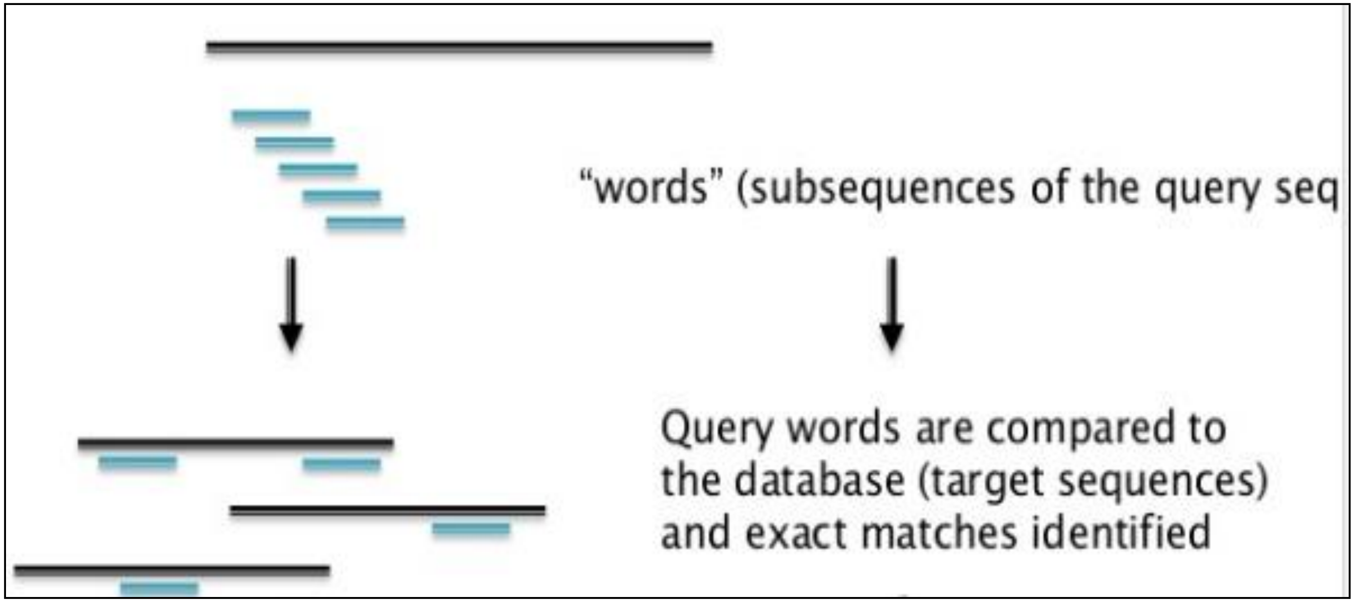


Program	Database	Query
BLASTN	Nucleotide	Nucleotide
BLASTP	Protein	Protein
BLASTX	Protein	Nt. → Protein
TBLASTN	Nt. → Protein	Protein
TBLASTX	Nt. → Protein	Nt. → Protein

How does BLAST Works?

- Construct a dictionary of all words in the query

- Initiate a local alignment for each word match between query and DB



"words" (subsequences of the query seq
Query words are compared to the database (target sequences) and exact matches identified

BLAST: Global Alignment

- It compares the whole sequence with another sequence.

- So, output of Global is one to one comparison of two sequences.

- This method is useful if you have small group of sequences.

BLAST: Local Alignment

- Local method uses the subset of sequence and attempts to align against the subset of another sequence.

- So, output of local alignment gives the subset of regions which are highly similar.

- Example : Compare two sequence A and B

```
(A) GCATTACTAATATATTAGTAAATCAGAGTAGTA
           |||||||||
(B) AAGCGAATAATATATTTATACTCAGATTATTGCGCG
```

BLAST: Input Format

Many program for sequence alignment expect sequences to be in FASTA format

Example 1 :

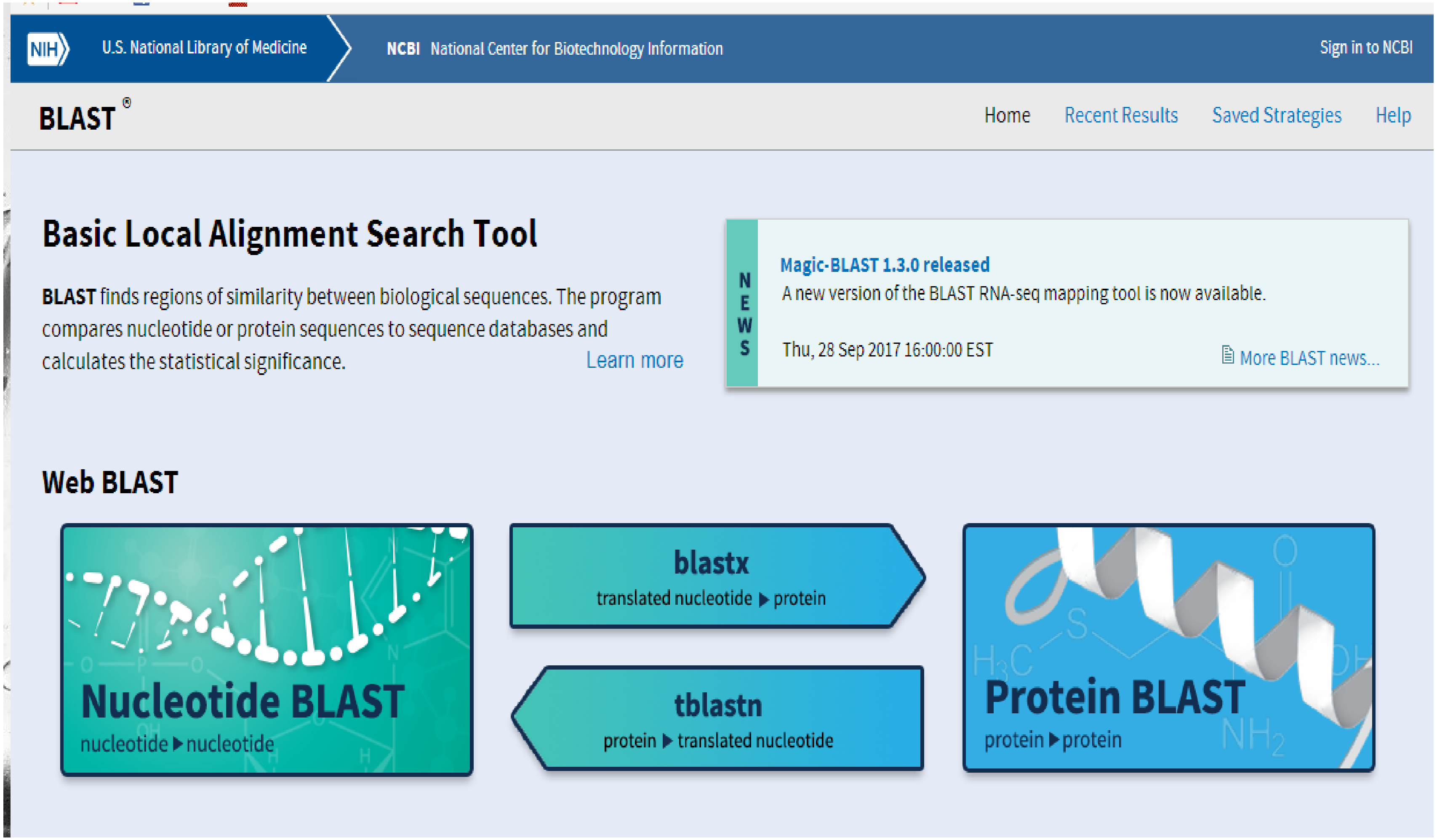
```
>L37107.1 Canis familiaris p53 mRNA, partial cds
GTTCCGTTTGGGGTTCCTGCATTCCGGGACAGCCAAGTCTGTTACTTGGACGTACTCCCCTCTCCTCAAC
AAGTTGTTTTGCCAGCTGGCGAAGACCTGCCCCGTGCAGCTGTGGGTCAGCTCCCCACCCCCACCCAATA
CCTGCGTCCGCGCTATGGCCATCTATAAGAAGTCGGAGTTCGTGACCGAGGTTGTGCGGCGCTGCCCCCA
CCATGAACGCTGCTCTGACAGTAGTGACGGTCTTGCCCCTCCTCAGCATCTCATCCGAGTGGAAGGAAAT
TTGCGGGCCAAGTACCTGGACGACAGAAACACTTTTCGACACAGTGTGGTGGTGCCTTATGAGCCACCCG
AGGTTGGCTCTGACTATACCACCATCCACTACAACTACATGTGTAACAGTTCCTGCATGGGAGGCATGAA
CCGGCGGCCCATCCTCACTATCATCACCCTGGAAGACTCCAGTGGAAACGTGCTGGGACGCAACAGCTTT
GAGGTACGCGTTTGTGCCTGTCCCGGGAGAGACCGCCGGACTGAGGAGGAGAATTTCCACAAGAAGGGGG
AGCCTTGTCCTGAGCCACCCCCCGGGAGTACCAAGCGAGCACTGCCTCCCAGCACCAGCTCCTCTCCCCC
GCAAAAGAAGAAGCCACTAGATGGAGAATATTTCACCCTTCAGATCCGTGGGCGTGAACGCTATGAGATG
TTCAGGAATCTGAATGAAGCCTTGGAGCTGAAGGATGCCCAGAGTGGAAAGGAGCCAGGGGGAAGCAGGG
CTCACTCCAGCCACCTGAAGGCAAAGAAGGGGCAATCTACCTCTCGCCATAAAAAACTGATGTTCAAGAGAGAA
```

Example 2 :

```
>NM_033360.3 Homo sapiens KRAS proto-oncogene, GTPase (KRAS), transcript
variant a, mRNA
TCCTAGGCGGCGGCCGCGGCGGCGGAGGCAGCAGCGGCGGCGGCAGTGGCGGCGGCGAAGGTGGCGGCGG
CTCGGCCAGTACTCCCGGCCCCCGCCATTTCGGACTGGGAGCGAGCGCGGCGCAGGCACTGAAGGCGGCG
GCGGGGCCAGAGGCTCAGCGGCTCCCAGGTGCGGGAGAGAGGCCTGCTGAAAATGACTGAATATAAACTT
GTGGTAGTTGGAGCTGGTGGCGTAGGCAAGAGTGCCTTGACGATACAGCTAATTCAGAATCATTTTGTGG
ACGAATATGATCCAACAATAGAGGATTCCTACAGGAAGCAAGTAGTAATTGATGGAGAAACCTGTCTCTT
GGATATTCTCGACACAGCAGGTCAAGAGGAGTACAGTGCAATGAGGGACCAGTACATGAGGACTGGGGAG
GGCTTTCTTTGTGTATTTGCCATAAATAATACTAAATCATTTGAAGATATTCACCATTATAGAGAACAAA
TTAAAAGAGTTAAGGACTCTGAAGATGTACCTATGGTCCTAGTAGGAAATAAATGTGATTTGCCTTCTAG
```

NCBI BLAST SERVER

Open the website : https://blast.ncbi.nlm.nih.gov/Blast.cgi



NIH U.S. National Library of Medicine

NCBI National Center for Biotechnology Information

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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

Magic-BLAST 1.3.0 released

A new version of the BLAST RNA-seq mapping tool is now available.

Thu, 28 Sep 2017 16:00:00 EST

More BLAST news...

Web BLAST

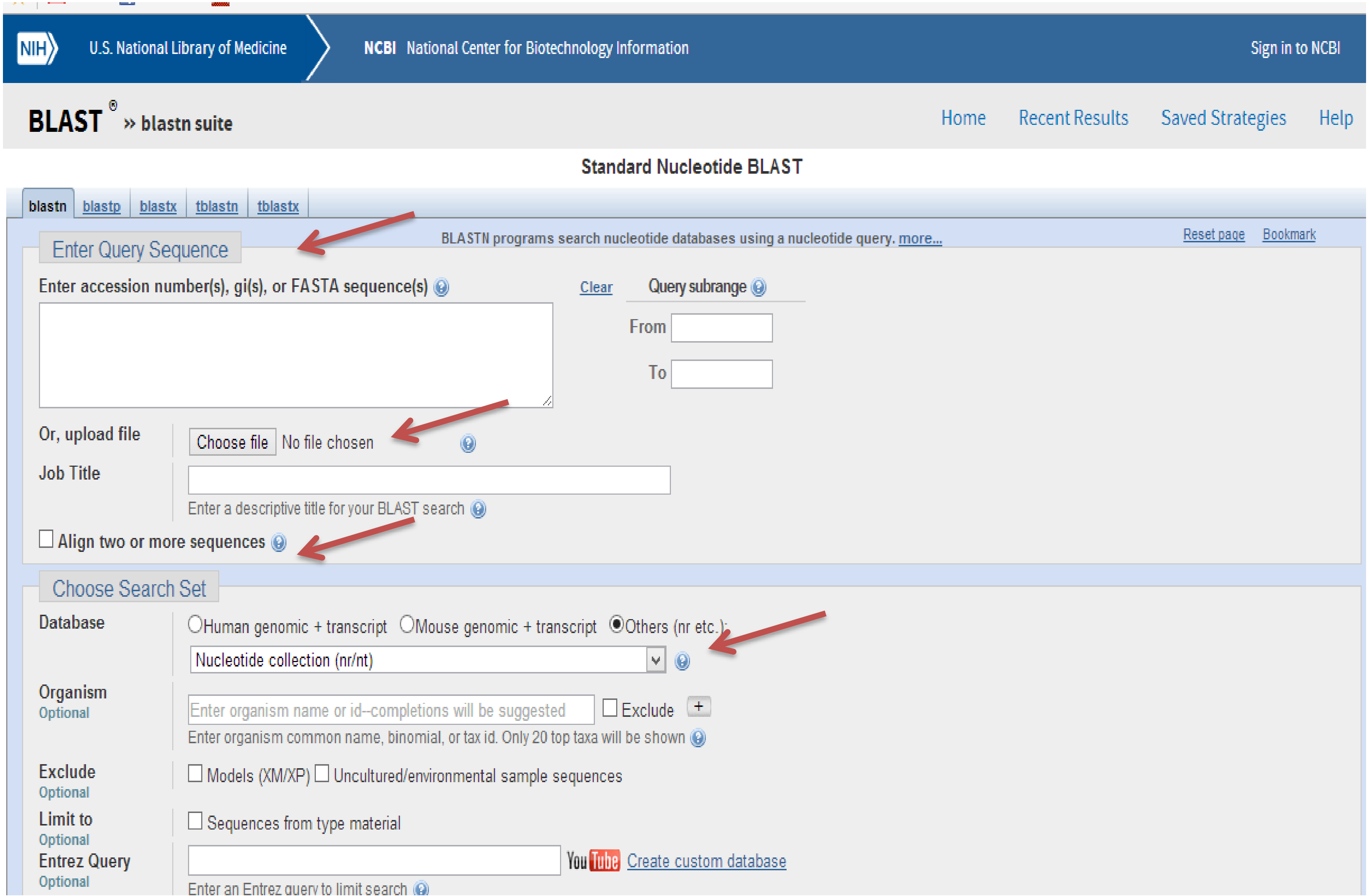
Nucleotide BLAST
nucleotide ▶ nucleotide

blastx
translated nucleotide ▶ protein

tblastn
protein ▶ translated nucleotide

Protein BLAST
protein ▶ protein

Window of BLASTN



NIH
U.S. National Library of Medicine
NCBI National Center for Biotechnology Information
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Standard Nucleotide BLAST

BLASTN programs search nucleotide databases using a nucleotide query. more...

Enter Query Sequence

Enter accession number(s), gi(s), or FASTA sequence(s)

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: ○Human genomic + transcript ○Mouse genomic + transcript ●Others (nr etc.):

Nucleotide collection (nr/nt)

Organism
Optional

Enter organism name or id--completions will be suggested □ Exclude +

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

Exclude
Optional

□ Models (XM/XP) □ Uncultured/environmental sample sequences

Limit to
Optional

□ Sequences from type material

Entrez Query
Optional

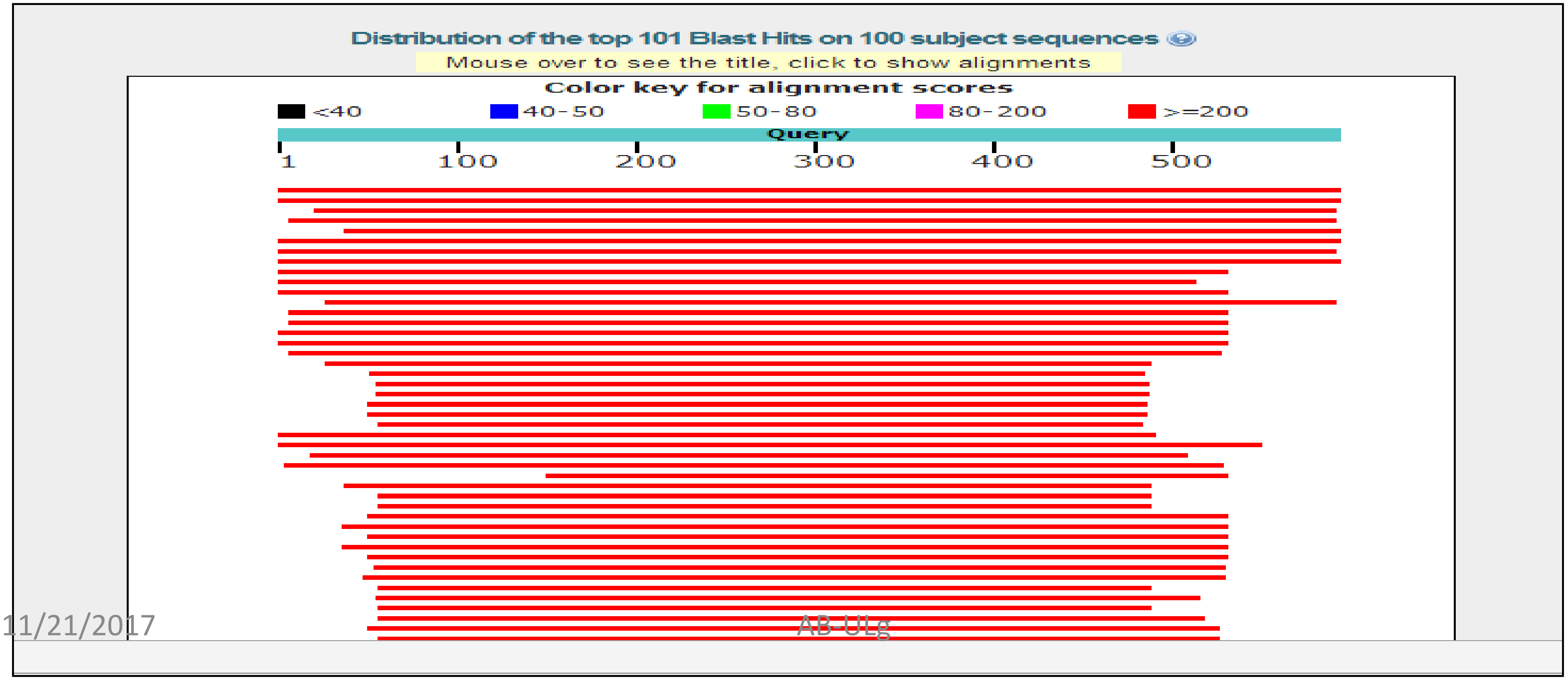
Enter an Entrez query to limit search

Let us work on BLASTN

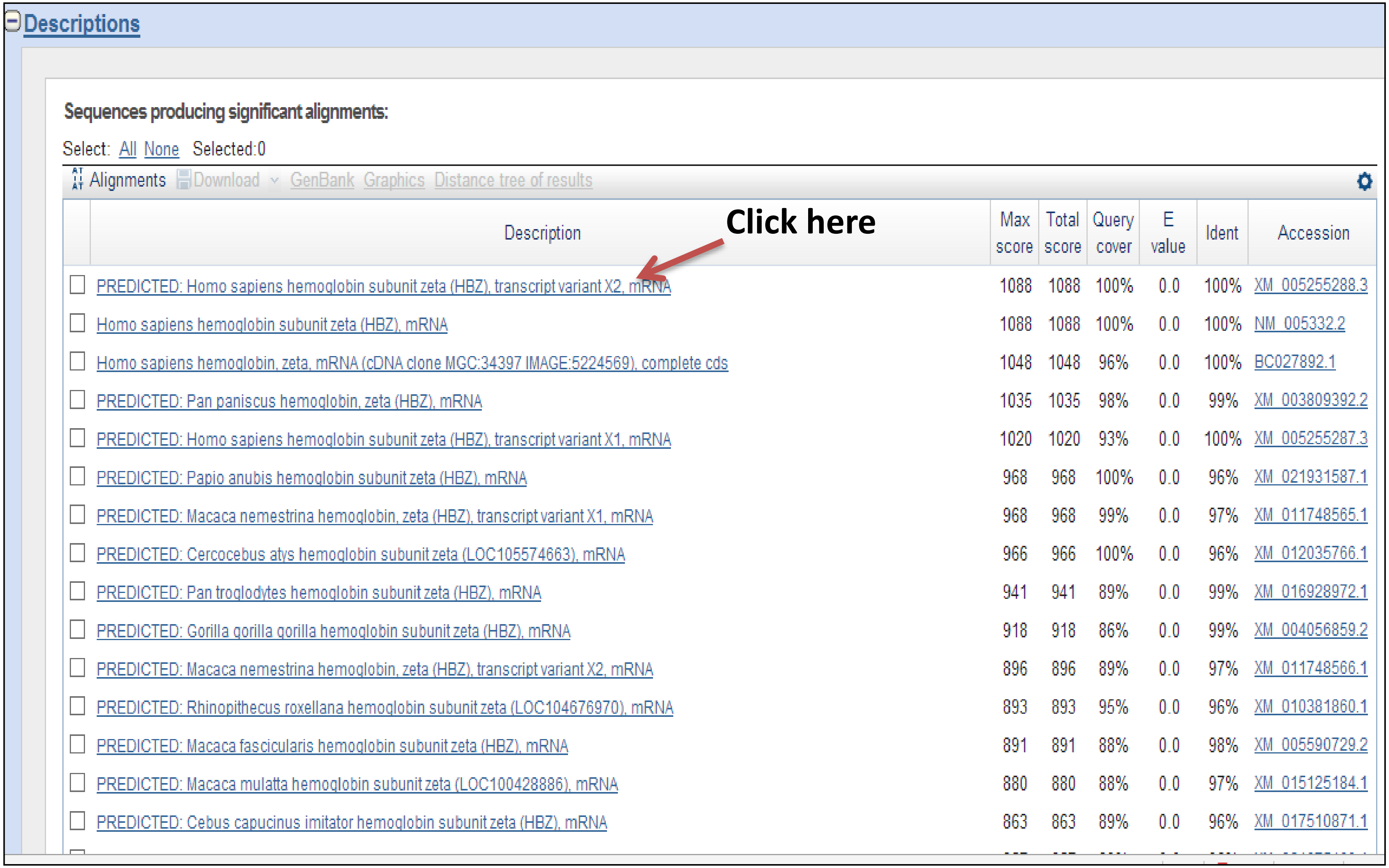
- Select following sequence and give input into NCBI BLASTN query section

```
>Seq1
ACCAAGGCCAGTCCTGAGCAGGCCCAACTCCAGTGCAGCTGCCCACCCTGCCGCCATGTCTCTGACCAAG
ACTGAGAGGACCATCATTGTGTCCATGTGGGCCAAGATCTCCACGCAGGCCGACACCATCGGCACCGAGA
CTCTGGAGAGGCTCTTCCTCAGCCACCCGCAGACCAAGACCTACTTCCCGCACTTCGACCTGCACCCGGG
GTCCGCGCAGTTGCGCGCGCACGGCTCCAAGGTGGTGGCCGCCGTGGGCGACGCGGTGAAGAGCATCGAC
GACATCGGCGGCGCCCTGTCCAAGCTGAGCGAGCTGCACGCCTACATCCTGCGCGTGGACCCGGTCAACT
TCAAGCTCCTGTCCCACTGCCTGCTGGTCACCCTGGCCGCGCGCTTCCCCGCCGACTTCACGGCCGAGGC
CCACGCCGCCTGGGACAAGTTCCTATCGGTCGTATCCTCTGTCCTGACCGAGAAGTACCGCTGAGCGCCG
CCTCCGGGACCCCCAGGACAGGCTGCGGCCCCTCCCCCGTCCTGGAGGTTCCCCAGCCCCACTTACCGCG TAATGCGCCAATAAACCAATGAACGAAGC
```

- You will get list of Hits


Distribution of the top 101 Blast Hits on 100 subject sequences
Mouse over to see the title, click to show alignments
Color key for alignment scores
<40
40-50
50-80
80-200
>=200
Query
1
100
200
300
400
500

- **You will see statistic of alignments (Identity, E value)**



Descriptions

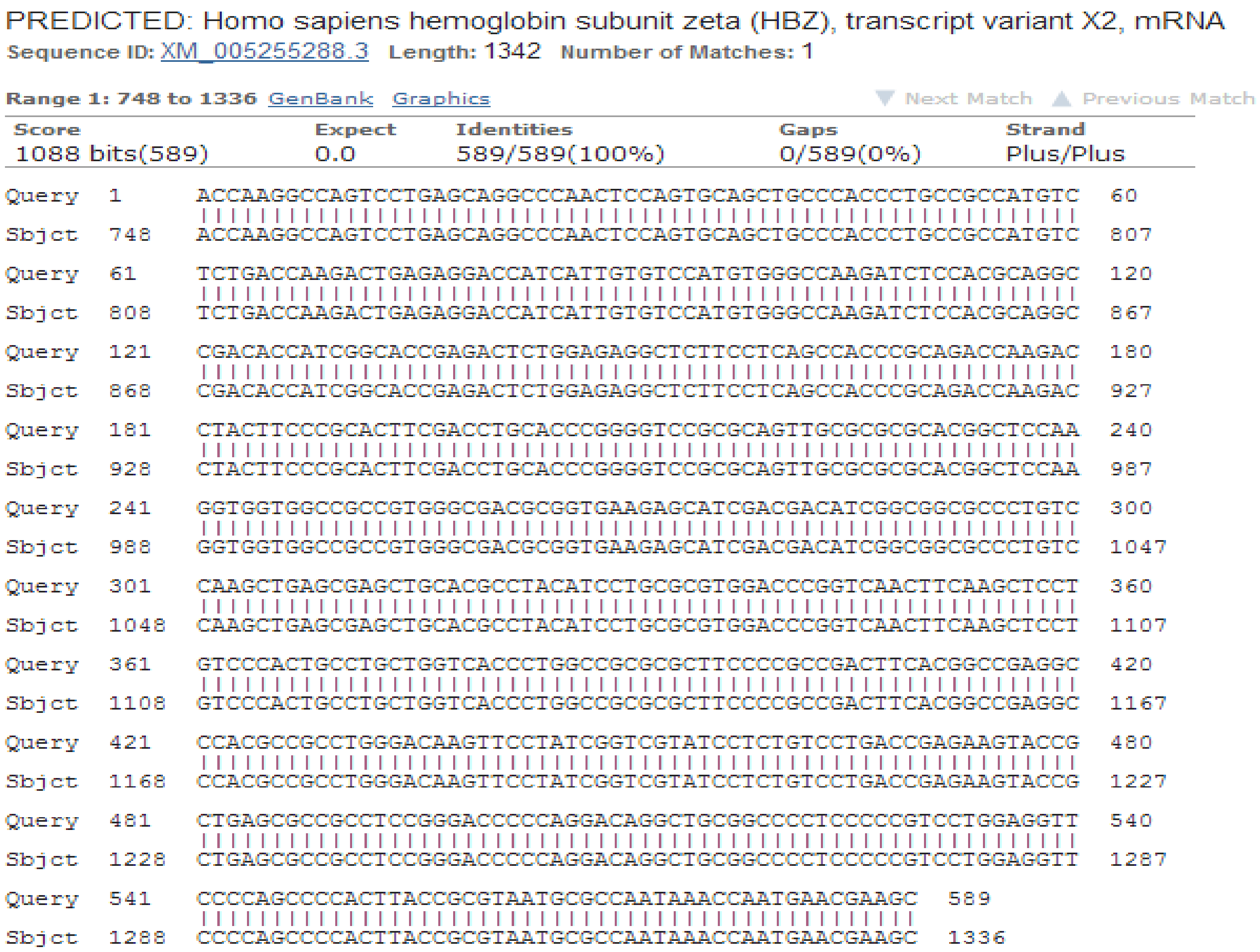
Sequences producing significant alignments:

Select: All None Selected:0

Click here

	Description	Max score	Total score	Query cover	E value	Ident	Accession
□	PREDICTED: Homo sapiens hemoglobin subunit zeta (HBZ), transcript variant X2, mRNA	1088	1088	100%	0.0	100%	XM_005255288.3
□	Homo sapiens hemoglobin subunit zeta (HBZ), mRNA	1088	1088	100%	0.0	100%	NM_005332.2
□	Homo sapiens hemoglobin, zeta, mRNA (cDNA clone MGC:34397 IMAGE:5224569), complete cds	1048	1048	96%	0.0	100%	BC027892.1
□	PREDICTED: Pan paniscus hemoglobin, zeta (HBZ), mRNA	1035	1035	98%	0.0	99%	XM_003809392.2
□	PREDICTED: Homo sapiens hemoglobin subunit zeta (HBZ), transcript variant X1, mRNA	1020	1020	93%	0.0	100%	XM_005255287.3
□	PREDICTED: Papio anubis hemoglobin subunit zeta (HBZ), mRNA	968	968	100%	0.0	96%	XM_021931587.1
□	PREDICTED: Macaca nemestrina hemoglobin, zeta (HBZ), transcript variant X1, mRNA	968	968	99%	0.0	97%	XM_011748565.1
□	PREDICTED: Cercocebus atys hemoglobin subunit zeta (LOC105574663), mRNA	966	966	100%	0.0	96%	XM_012035766.1
□	PREDICTED: Pan troglodytes hemoglobin subunit zeta (HBZ), mRNA	941	941	89%	0.0	99%	XM_016928972.1
□	PREDICTED: Gorilla gorilla gorilla hemoglobin subunit zeta (HBZ), mRNA	918	918	86%	0.0	99%	XM_004056859.2
□	PREDICTED: Macaca nemestrina hemoglobin, zeta (HBZ), transcript variant X2, mRNA	896	896	89%	0.0	97%	XM_011748566.1
□	PREDICTED: Rhinopithecus roxellana hemoglobin subunit zeta (LOC104676970), mRNA	893	893	95%	0.0	96%	XM_010381860.1
□	PREDICTED: Macaca fascicularis hemoglobin subunit zeta (HBZ), mRNA	891	891	88%	0.0	98%	XM_005590729.2
□	PREDICTED: Macaca mulatta hemoglobin subunit zeta (LOC100428886), mRNA	880	880	88%	0.0	97%	XM_015125184.1
□	PREDICTED: Cebus capucinus imitator hemoglobin subunit zeta (HBZ), mRNA	863	863	89%	0.0	96%	XM_017510871.1

▪ How well alignment is ? : Bad, Good, Very Good?



PREDICTED: Homo sapiens hemoglobin subunit zeta (HBZ), transcript variant X2, mRNA

Sequence ID: XM_005255288.3 **Length:** 1342 **Number of Matches:** 1

Range 1: 748 to 1336

Score	Expect	Identities	Gaps	Strand
1088 bits(589)	0.0	589/589(100%)	0/589(0%)	Plus/Plus

```
Query  1     ACCAAGGCCAGTCCTGAGCAGGCCCAACTCCAGTGCAGCTGCCCACCCTGCCGCCATGTC  60
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  748   ACCAAGGCCAGTCCTGAGCAGGCCCAACTCCAGTGCAGCTGCCCACCCTGCCGCCATGTC  807

Query  61    TCTGACCAAGACTGAGAGGACCATCATTGTGTCCATGTGGGCCAAGATCTCCACGCAGGC  120
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  808   TCTGACCAAGACTGAGAGGACCATCATTGTGTCCATGTGGGCCAAGATCTCCACGCAGGC  867

Query  121   CGACACCATCGGCACCGAGACTCTGGAGAGGCTCTTCCTCAGCCACCCGCAGACCAAGAC  180
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  868   CGACACCATCGGCACCGAGACTCTGGAGAGGCTCTTCCTCAGCCACCCGCAGACCAAGAC  927

Query  181   CTACTTCCCGCACTTCGACCTGCACCCGGGGTCCGCGCAGTTGCGCGCGCACGGCTCCAA  240
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  928   CTACTTCCCGCACTTCGACCTGCACCCGGGGTCCGCGCAGTTGCGCGCGCACGGCTCCAA  987

Query  241   GGTGGTGGCCGCCGTGGGCGACGCGGTGAAGAGCATCGACGACATCGGCGGCGCCCTGTC  300
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  988   GGTGGTGGCCGCCGTGGGCGACGCGGTGAAGAGCATCGACGACATCGGCGGCGCCCTGTC  1047

Query  301   CAAGCTGAGCGAGCTGCACGCCTACATCCTGCGCGTGGACCCGGTCAACTTCAAGCTCCT  360
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  1048  CAAGCTGAGCGAGCTGCACGCCTACATCCTGCGCGTGGACCCGGTCAACTTCAAGCTCCT  1107

Query  361   GTCCCACTGCCTGCTGGTCACCCTGGCCGCGCGCTTCCCCGCCGACTTCACGGCCGAGGC  420
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  1108  GTCCCACTGCCTGCTGGTCACCCTGGCCGCGCGCTTCCCCGCCGACTTCACGGCCGAGGC  1167

Query  421   CCACGCCGCCTGGGACAAGTTCCTATCGGTCGTATCCTCTGTCCTGACCGAGAAGTACCG  480
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  1168  CCACGCCGCCTGGGACAAGTTCCTATCGGTCGTATCCTCTGTCCTGACCGAGAAGTACCG  1227

Query  481   CTGAGCGCCGCCTCCGGGACCCCCAGGACAGGCTGCGGCCCCTCCCCCGTCCTGGAGGTT  540
             ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  1228  CTGAGCGCCGCCTCCGGGACCCCCAGGACAGGCTGCGGCCCCTCCCCCGTCCTGGAGGTT  1287

Query  541   CCCCAGCCCCACTTACCGCGTAATGCGCCAATAAACCAATGAACGAAGC  589
             |||||||||||||||||||||||||||||||||||||||||||||||||
Sbjct  1288  CCCCAGCCCCACTTACCGCGTAATGCGCCAATAAACCAATGAACGAAGC  1336
```

RESULT INTERPRETATION

1. **How many sequences crossed the threshold E value ???**

2. **How many sequences show > 50 % identity with database ??**

3. **How many sequences show > 90 % identity with database ??**

4. **Prepare tabular output for BLASTP and BLASTN results.**

QUESTIONS

Blastx : Let us run

1 . Perform the blastx

2. How many sequences shows 90% identity against the database

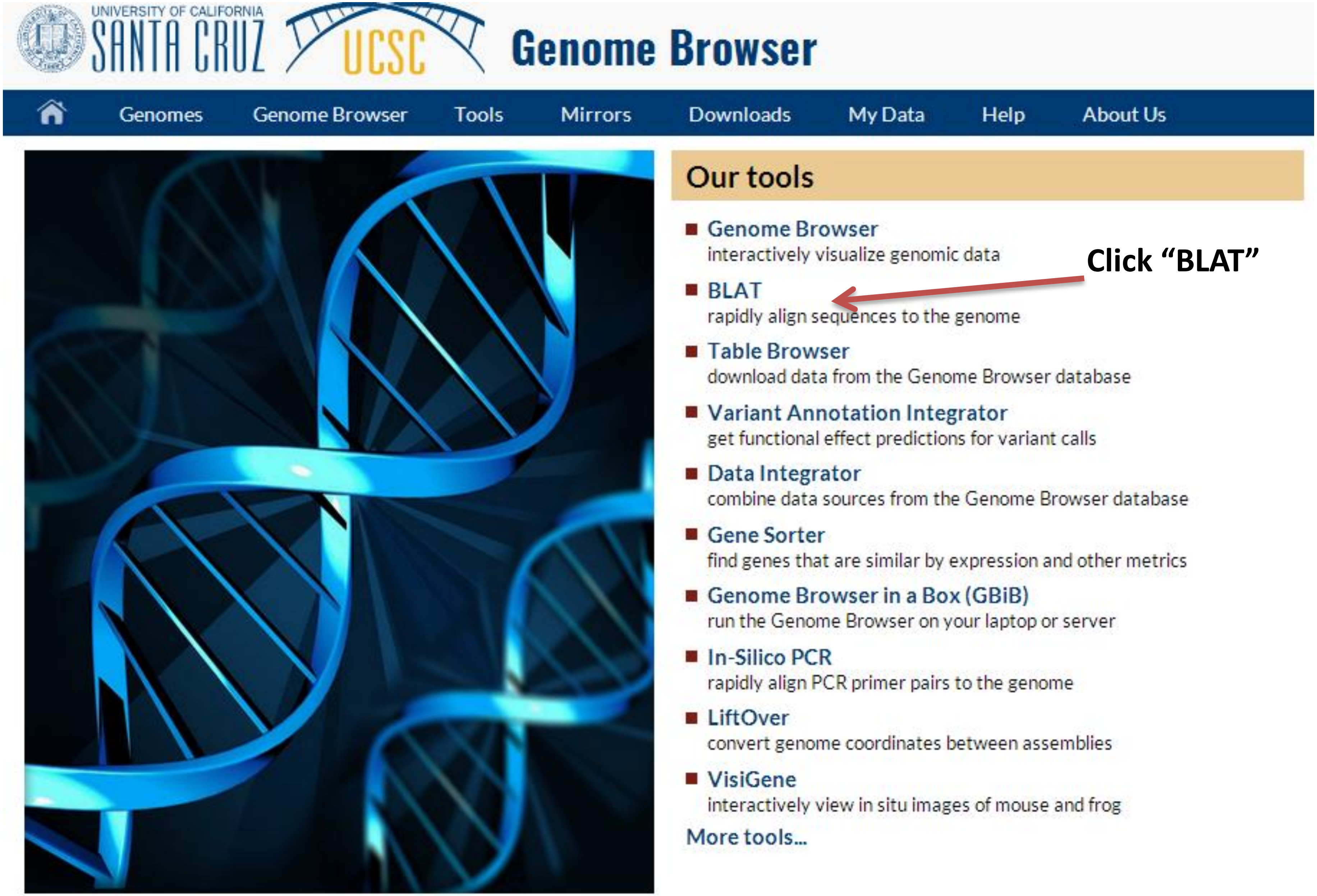
3. What is their e-value ??

QUESTIONS

- Is it possible to localise its position on human genome ?

- How to analysis its gene structure ?

- For this, Open the UCSC Browser available at https://genome.ucsc.edu/



Our tools

- **Genome Browser**
  interactively visualize genomic data
- **BLAT**
  rapidly align sequences to the genome
- **Table Browser**
  download data from the Genome Browser database
- **Variant Annotation Integrator**
  get functional effect predictions for variant calls
- **Data Integrator**
  combine data sources from the Genome Browser database
- **Gene Sorter**
  find genes that are similar by expression and other metrics
- **Genome Browser in a Box (GBiB)**
  run the Genome Browser on your laptop or server
- **In-Silico PCR**
  rapidly align PCR primer pairs to the genome
- **LiftOver**
  convert genome coordinates between assemblies
- **VisiGene**
  interactively view in situ images of mouse and frog

More tools...

Click “BLAT”

Difference Between BLAST and BLAT

- BLAT is an alignment tool like BLAST, but it is structured differently.

- BLAT works by keeping an index of an entire genome in memory.

- Thus, the target database of BLAT is not a set of GenBank sequences, but instead an index derived from the assembly of the entire genome.

Advantages of BLAT over BLAST

- Its Speed is very high (no queues, response in seconds).

- The ability to submit a long list of simultaneous queries in fasta format.

- A direct link into the UCSC browser.

- Alignment block details in natural genomic order.

- An option to launch the alignment later as part of a custom track.

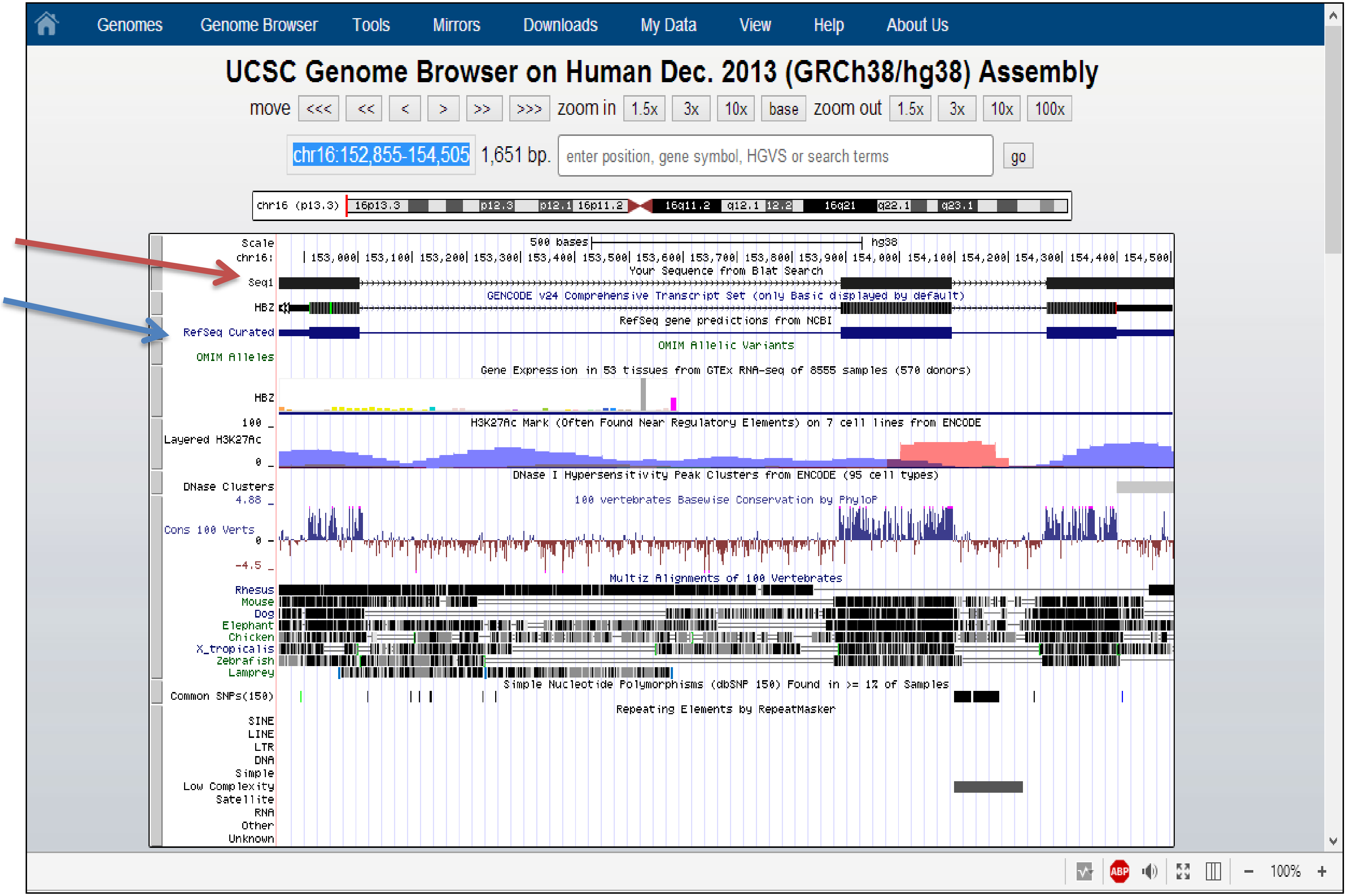
- **Paste following sequence into Query search Box and click Submit**

```
>Seq1
ACCAAGGCCAGTCCTGAGCAGGCCCAACTCCAGTGCAGCTGCCCACCCTGCCGCCATGTCT
CTGACCAAGACTGAGAGGACCATCATTGTGTCCATGTGGGCCAAGATCTCCACGCAGGCCG
ACACCATCGGCACCGAGACTCTGGAGAGGCTCTTCCTCAGCCACCCGCAGACCAAGACCTA
CTTCCCGCACTTCGACCTGCACCCGGGGTCCGCGCAGTTGCGCGCACGGCTCCAAGGTG
GTGGCCGCCGTGGGCGACGCGGTGAAGAGCATCGACGACATCGGCGGCGCCCTGTCCAAGC
TGAGCGAGCTGCACGCCTACATCCTGCGCGTGGACCCGGTCAACTTCAAGCTCCTGTCCCA
CTGCCTGCTGGTCACCCTGGCCGCGCTTCCCCGCCGACTTCACGGCCGAGGCCCACGCC
GCCTGGGACAAGTTCCTATCGGTCGTATCCTCTGTCCTGACCGAGAAGTACCGCTGAGCGC
CGCCTCCGGACCCCCAGGACAGGCTGCGGCCCCTCCCCCGTCCTGGAGGTTCCCCAGCCC
CACTTACCGCGTAATGCGCCAATAAACCAATGAACGAAGC
```

Which output did you see ??

Can you have a look at your sequence ? How ?

How many exons are present in your sequence ?



UCSC Genome Browser on Human Dec. 2013 (GRCh38/hg38) Assembly

chr16:152,855-154,505 1,651 bp.

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move <<< << < > >> >>>
zoom in 1.5x 3x 10x base
zoom out 1.5x 3x 10x 100x
enter position, gene symbol, HGVS or search terms
go
chr16 (p13.3)
16p13.3
p12.3
p12.1
16p11.2
16q11.2
q12.1
12.2
16q21
q22.1
q23.1
Scale
500 bases
hg38
chr16:
153,000
153,100
153,200
153,300
153,400
153,500
153,600
153,700
153,800
153,900
154,000
154,100
154,200
154,300
154,400
154,500
Your Sequence from Blat Search
Seq1
GENCODE v24 Comprehensive Transcript Set (only Basic displayed by default)
HBZ
RefSeq gene predictions from NCBI
RefSeq Curated
OMIM Allelic Variants
OMIM Alleles
Gene Expression in 53 tissues from GTEx RNA-seq of 8555 samples (570 donors)
HBZ
100 _
Layered H3K27Ac
0 _
H3K27Ac Mark (Often Found Near Regulatory Elements) on 7 cell lines from ENCODE
DNase I Hypersensitivity Peak Clusters from ENCODE (95 cell types)
DNase Clusters
4.88 _
100 vertebrates Basewise Conservation by PhyloP
Cons 100 Verts
0 -
-4.5 _
Multiz Alignments of 100 Vertebrates
Rhesus
Mouse
Dog
Elephant
Chicken
X_tropicalis
Zebrafish
Lamprey
Simple Nucleotide Polymorphisms (dbSNP 150) Found in >= 1% of Samples
Common SNPs(150)
Repeating Elements by RepeatMasker
SINE
LINE
LTR
DNA
Simple
Low Complexity
Satellite
RNA
Other
Unknown
ABP
100%

QUESTIONS