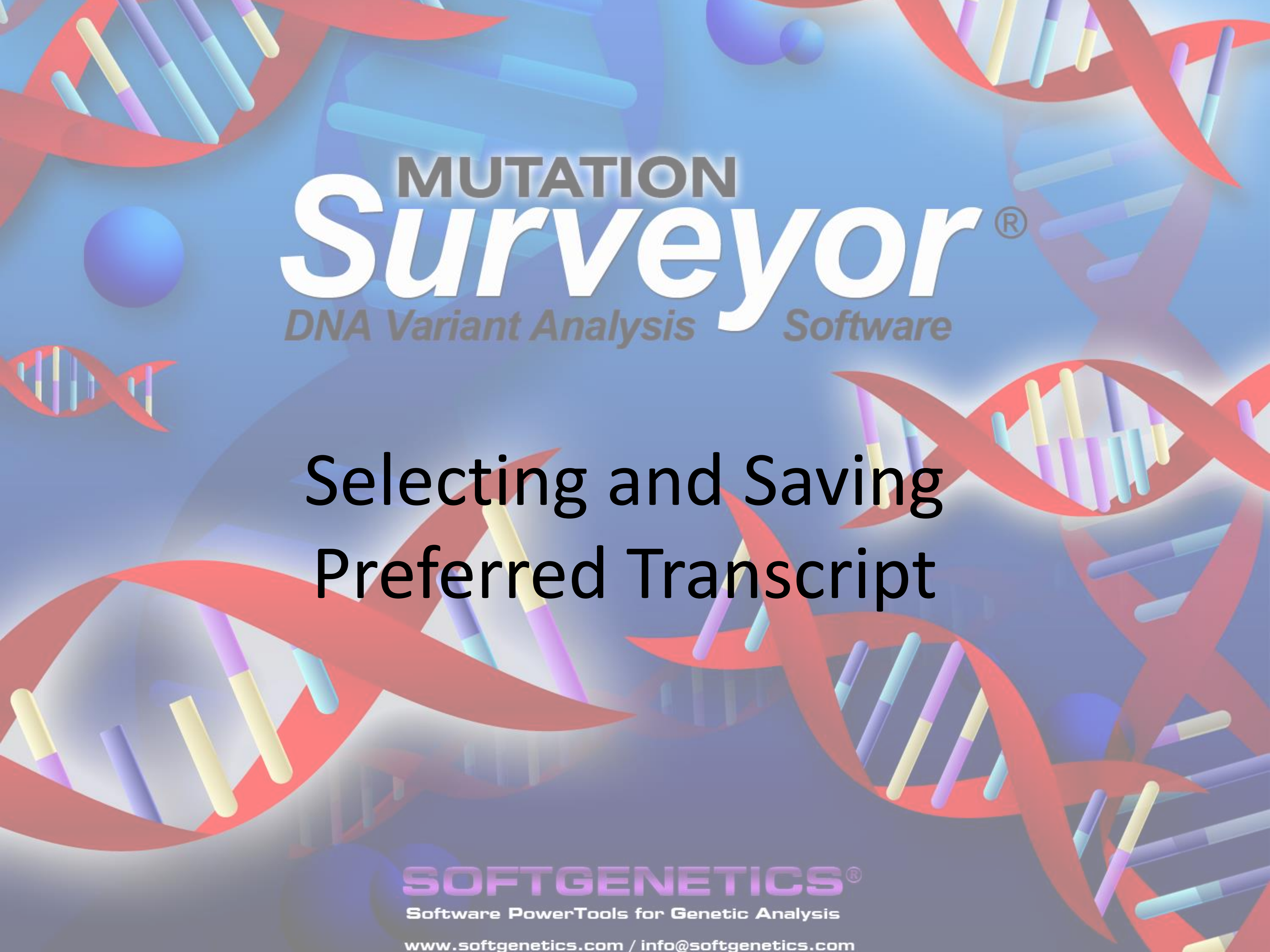


MUTATION

Surveyor®

DNA Variant Analysis Software

Selecting and Saving Preferred Transcript

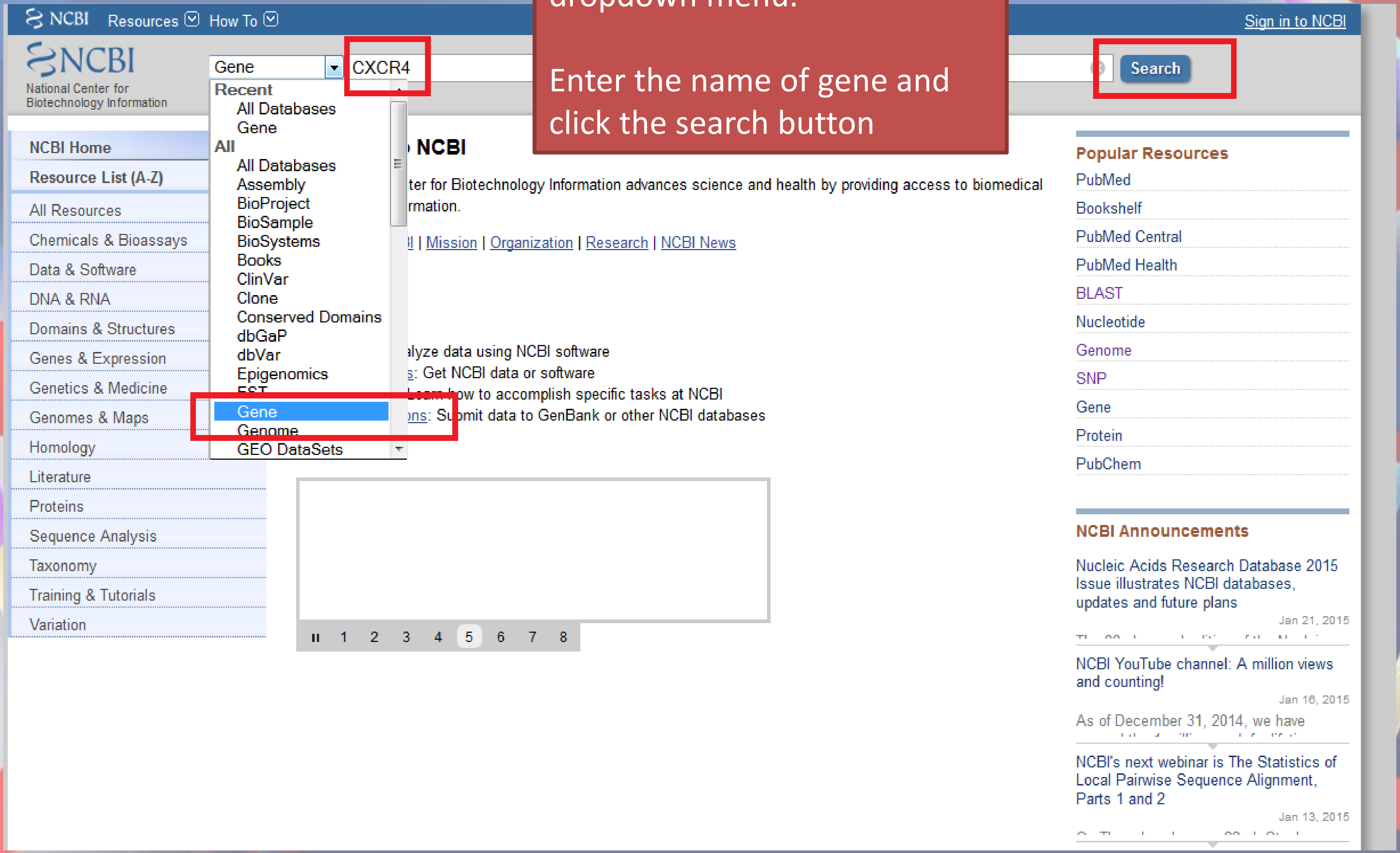
SOFTGENETICS®

Software PowerTools for Genetic Analysis

www.softgenetics.com / info@softgenetics.com

Select "Gene" from the dropdown menu.

Enter the name of gene and click the search button



Gene

CXCR4

...ter for Biotechnology Information advances science and health by providing access to biomedical ...rmation.

...alyze data using NCBI software

...s: Get NCBI data or software

...earn how to accomplish specific tasks at NCBI

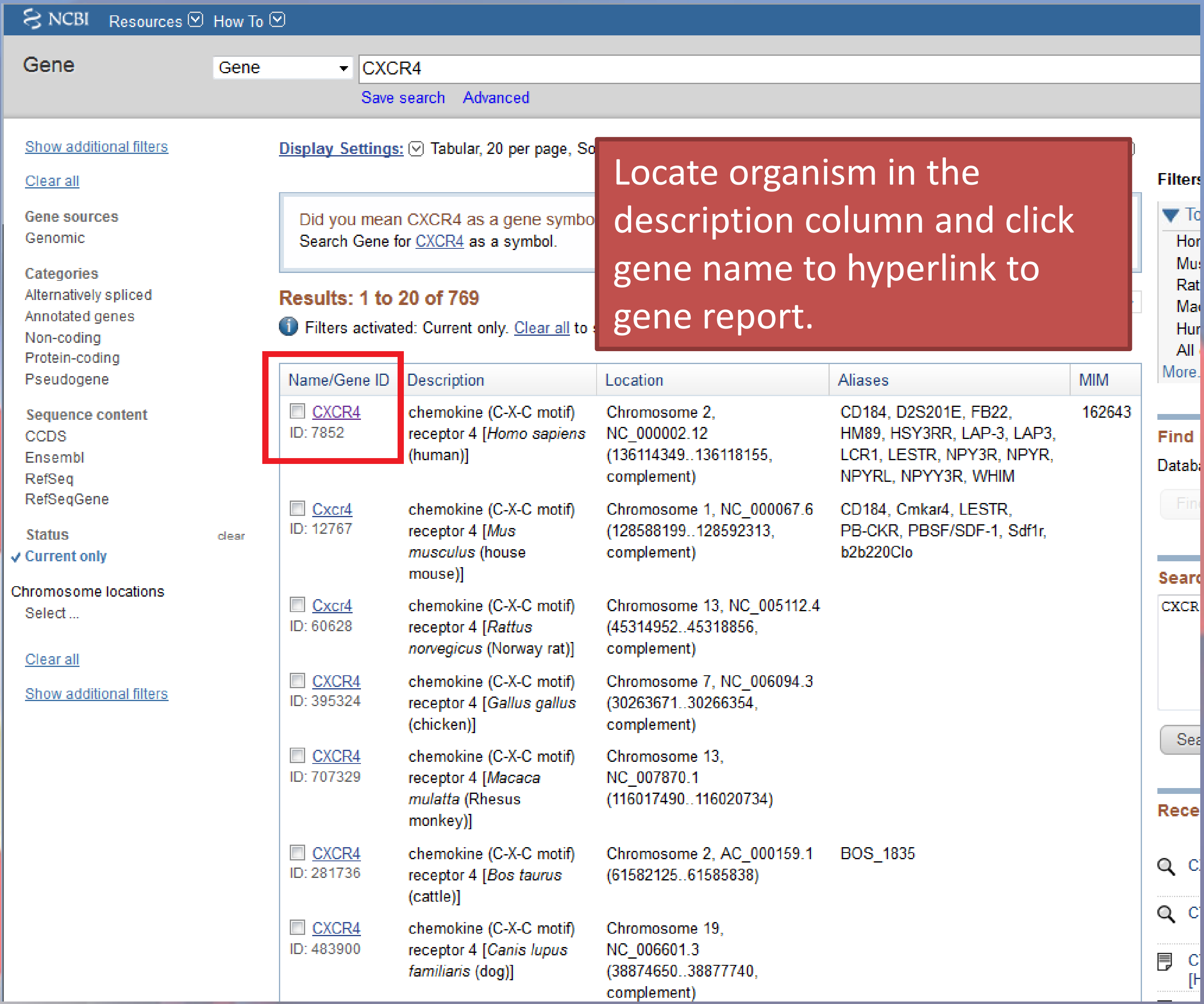
...ons: Submit data to GenBank or other NCBI databases

Nucleic Acids Research Database 2015 Issue illustrates NCBI databases, updates and future plans
Jan 21, 2015

NCBI YouTube channel: A million views and counting!
Jan 16, 2015

As of December 31, 2014, we have

NCBI's next webinar is The Statistics of Local Pairwise Sequence Alignment, Parts 1 and 2
Jan 13, 2015



Display Settings: Tabular, 20 per page, So

Did you mean CXCR4 as a gene symbo
Search Gene for CXCR4 as a symbol.

Results: 1 to 20 of 769

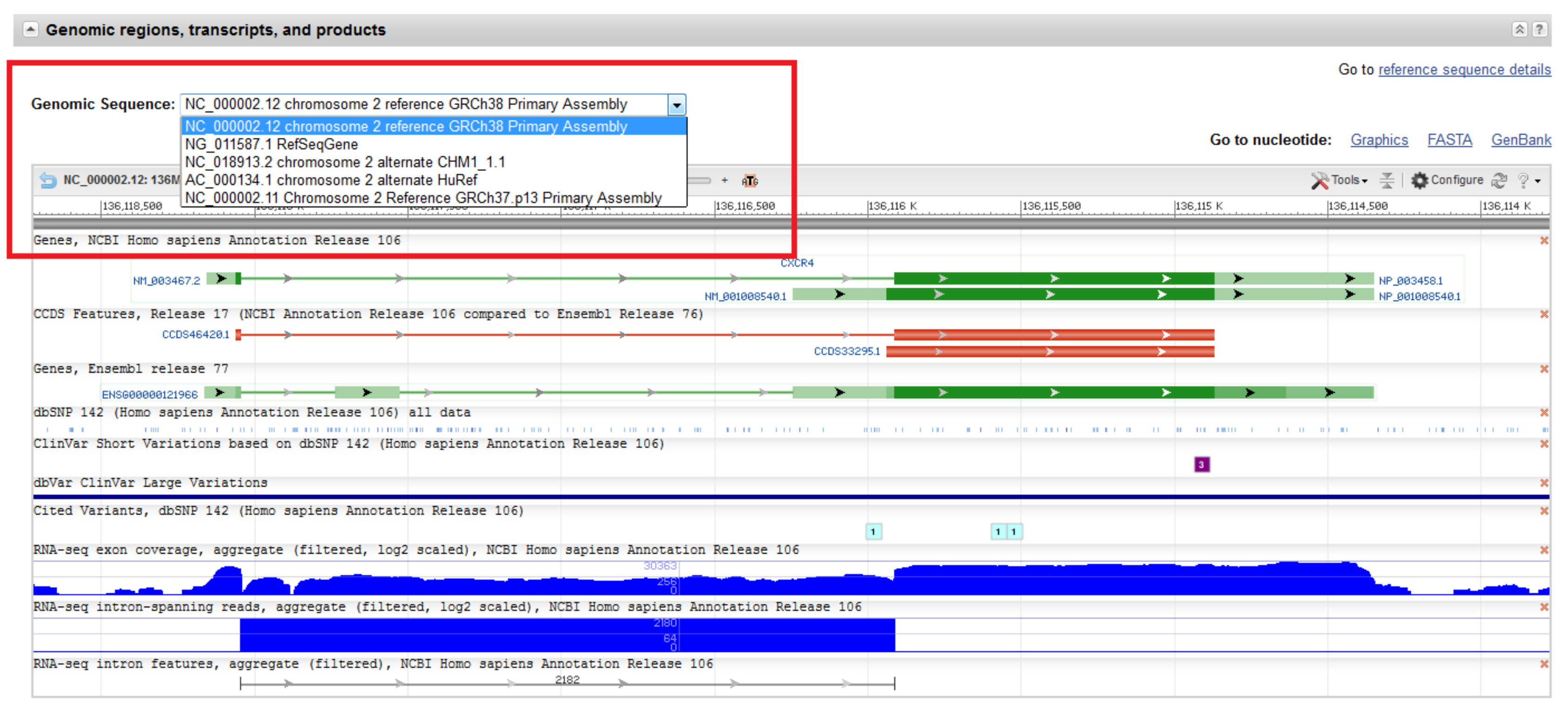
Filters activated: Current only. Clear all to

Name/Gene ID	Description	Location	Aliases	MIM
☐ CXCR4 ID: 7852	chemokine (C-X-C motif) receptor 4 [*Homo sapiens* (human)]	Chromosome 2, NC_000002.12 (136114349..136118155, complement)	CD184, D2S201E, FB22, HM89, HSY3RR, LAP-3, LAP3, LCR1, LESTR, NPY3R, NPYR, NPYRL, NPYY3R, WHIM	162643
☐ Cxcr4 ID: 12767	chemokine (C-X-C motif) receptor 4 [*Mus musculus* (house mouse)]	Chromosome 1, NC_000067.6 (128588199..128592313, complement)	CD184, Cmkar4, LESTR, PB-CKR, PBSF/SDF-1, Sdf1r, b2b220Clo	
☐ Cxcr4 ID: 60628	chemokine (C-X-C motif) receptor 4 [*Rattus norvegicus* (Norway rat)]	Chromosome 13, NC_005112.4 (45314952..45318856, complement)		
☐ CXCR4 ID: 395324	chemokine (C-X-C motif) receptor 4 [*Gallus gallus* (chicken)]	Chromosome 7, NC_006094.3 (30263671..30266354, complement)		
☐ CXCR4 ID: 707329	chemokine (C-X-C motif) receptor 4 [*Macaca mulatta* (Rhesus monkey)]	Chromosome 13, NC_007870.1 (116017490..116020734)		
☐ CXCR4 ID: 281736	chemokine (C-X-C motif) receptor 4 [*Bos taurus* (cattle)]	Chromosome 2, AC_000159.1 (61582125..61585838)	BOS_1835	
☐ CXCR4 ID: 483900	chemokine (C-X-C motif) receptor 4 [*Canis lupus familiaris* (dog)]	Chromosome 19, NC_006601.3 (38874650..38877740, complement)		

Locate organism in the description column and click gene name to hyperlink to gene report.

Select transcript from “Genomic regions, transcripts, and products” section.

NC accession files contain multiple transcripts/isoforms.



Genomic regions, transcripts, and products

Go to reference sequence details

Genomic Sequence: NC_000002.12 chromosome 2 reference GRCh38 Primary Assembly

- NC_000002.12 chromosome 2 reference GRCh38 Primary Assembly
- NG_011587.1 RefSeqGene
- NC_018913.2 chromosome 2 alternate CHM1_1.1
- AC_000134.1 chromosome 2 alternate HuRef
- NC_000002.11 Chromosome 2 Reference GRCh37.p13 Primary Assembly

Go to nucleotide: Graphics FASTA GenBank

Genes, NCBI Homo sapiens Annotation Release 106

CCDS Features, Release 17 (NCBI Annotation Release 106 compared to Ensembl Release 76)

Genes, Ensembl release 77

dbSNP 142 (Homo sapiens Annotation Release 106) all data

ClinVar Short Variations based on dbSNP 142 (Homo sapiens Annotation Release 106)

dbVar ClinVar Large Variations

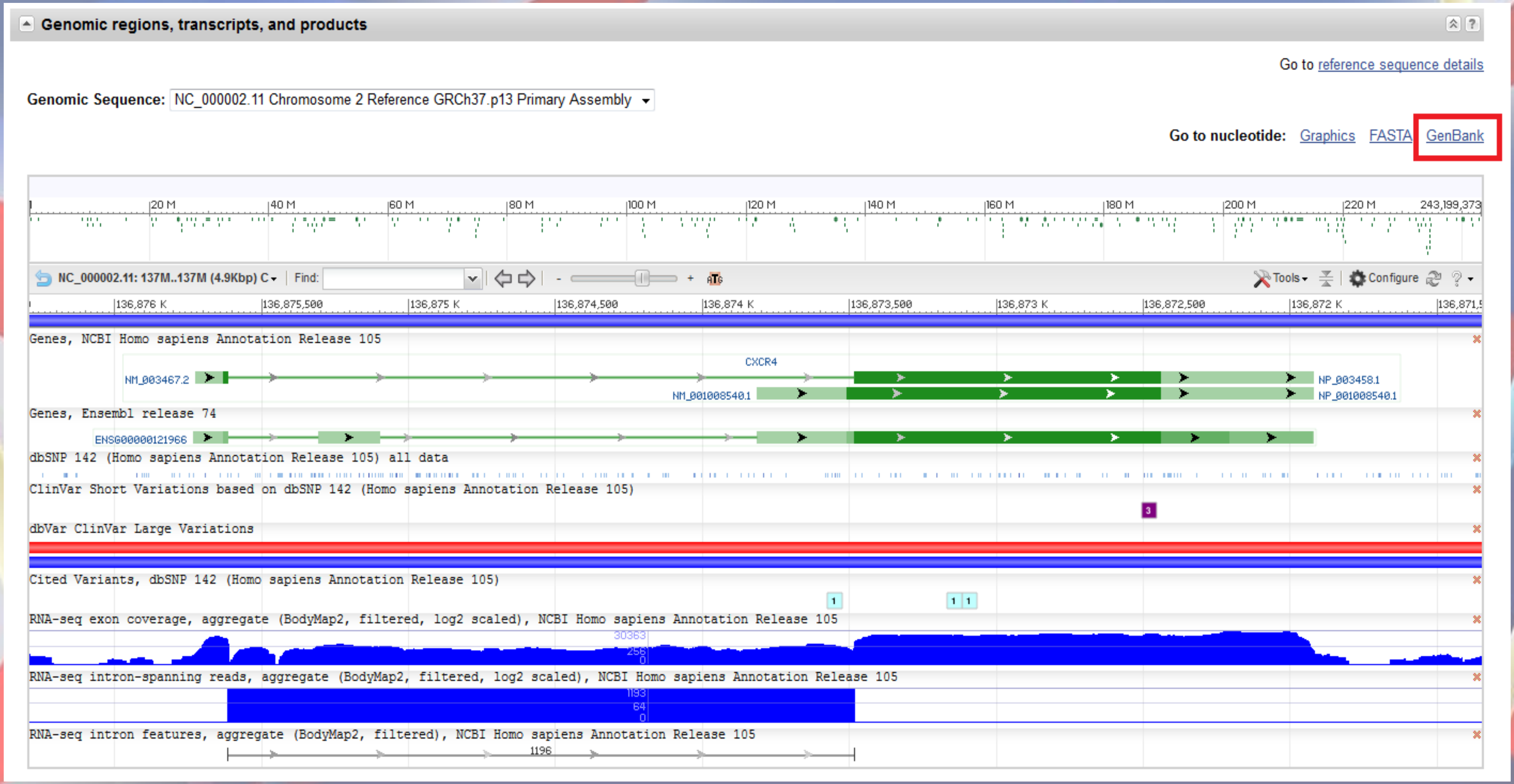
Cited Variants, dbSNP 142 (Homo sapiens Annotation Release 106)

RNA-seq exon coverage, aggregate (filtered, log2 scaled), NCBI Homo sapiens Annotation Release 106

RNA-seq intron-spanning reads, aggregate (filtered, log2 scaled), NCBI Homo sapiens Annotation Release 106

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation Release 106

After transcript is selected, click the GenBank option to the right of the page.



Genomic regions, transcripts, and products

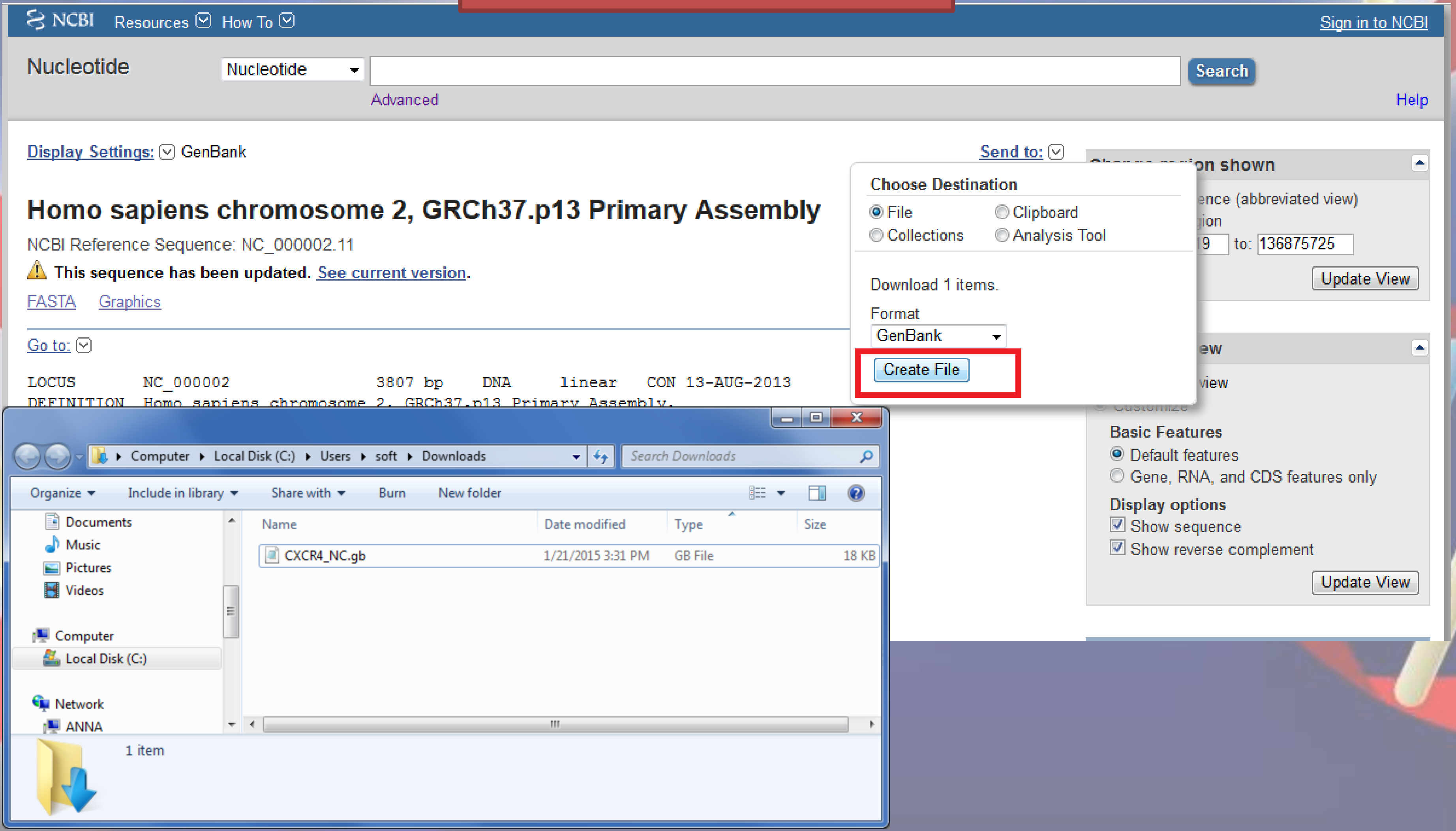
Go to reference sequence details

Genomic Sequence: NC_000002.11 Chromosome 2 Reference GRCh37.p13 Primary Assembly

Go to nucleotide: Graphics FASTA GenBank

NC_000002.11: 137M..137M (4.9Kbp)

1. Navigate to “Send to”
2. Select File
3. Select GenBank format
4. Select Create file
5. Save file to computer



Nucleotide: Nucleotide

Display Settings: GenBank

Send to:

Choose Destination

● File ○ Clipboard
○ Collections ○ Analysis Tool

Download 1 items.

Format: GenBank

Homo sapiens chromosome 2, GRCh37.p13 Primary Assembly

NCBI Reference Sequence: NC_000002.11

⚠ **This sequence has been updated. See current version.**

FASTA Graphics

Go to:

```
LOCUS       NC_000002              3807 bp    DNA     linear   CON 13-AUG-2013
DEFINITION  Homo sapiens chromosome 2, GRCh37.p13 Primary Assembly.
```

ence (abbreviated view)

ion

9 to: 136875725

Basic Features

● Default features
○ Gene, RNA, and CDS features only

Display options

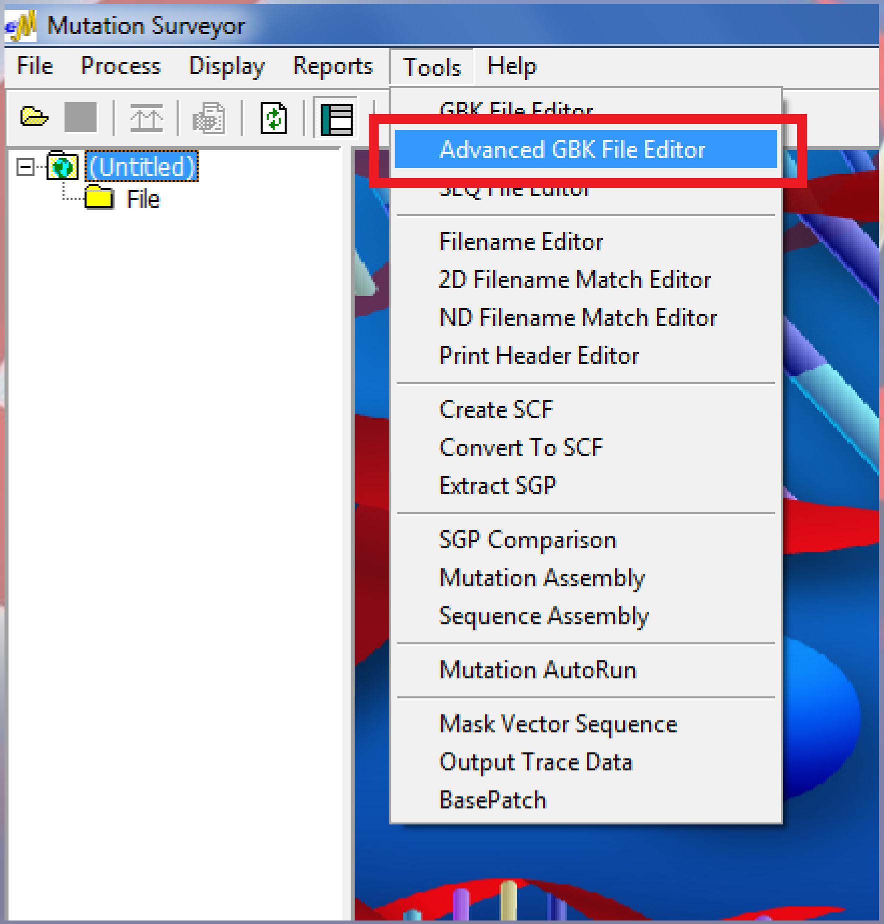
☑ Show sequence
☑ Show reverse complement

Name	Date modified	Type	Size
CXCR4_NC.gb	1/21/2015 3:31 PM	GB File	18 KB

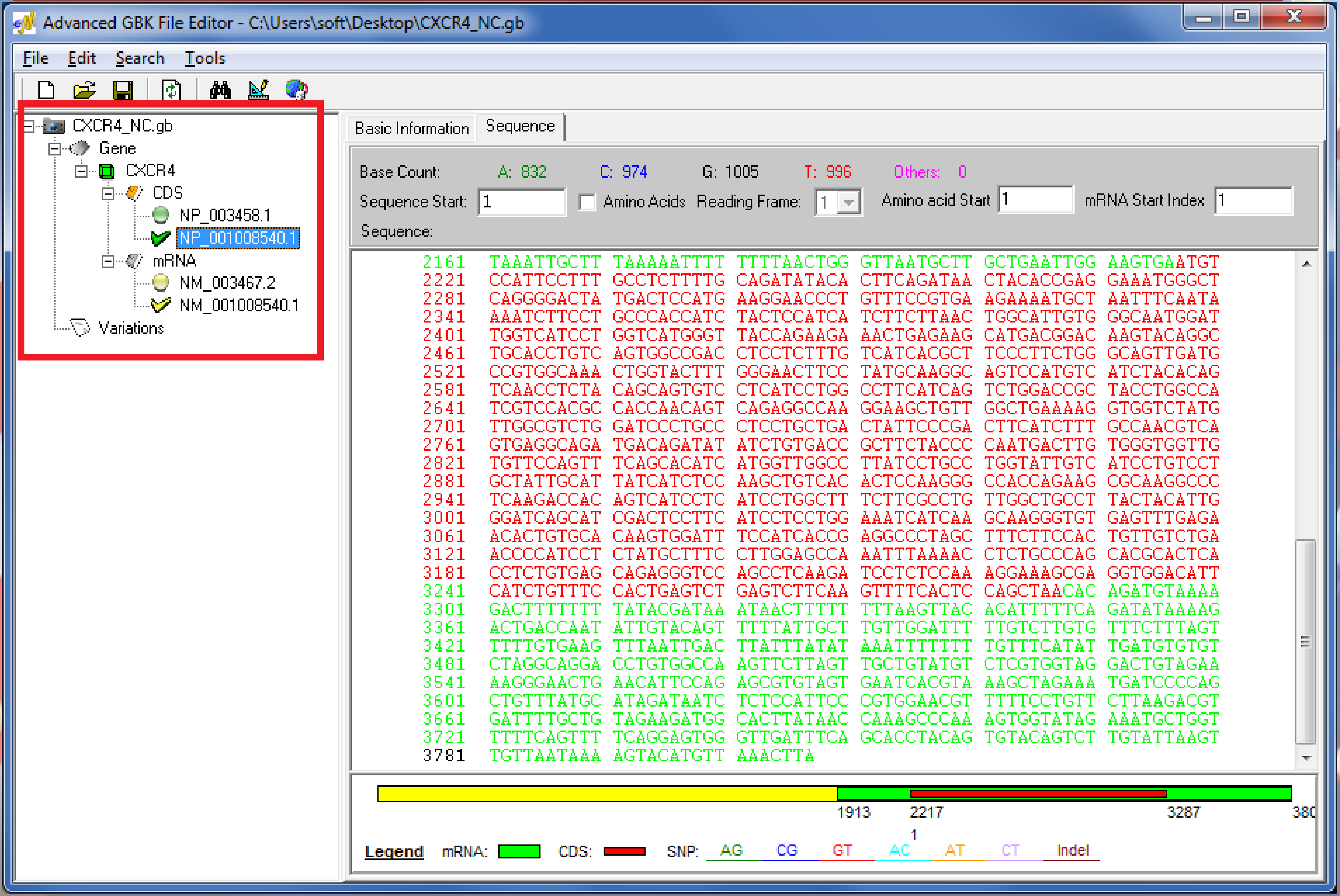
1 item

Launch Mutation Surveyor

Navigate to Tools > Advanced GBK File Editor



Open GenBank file
Isoform information will populate in left hand column
Click NP number to select isoform- a check mark will indicate that it is selected
Click the “Sequence” tab to review transcript



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Sequence Start: 1

☐ Amino Acids

Reading Frame: 1

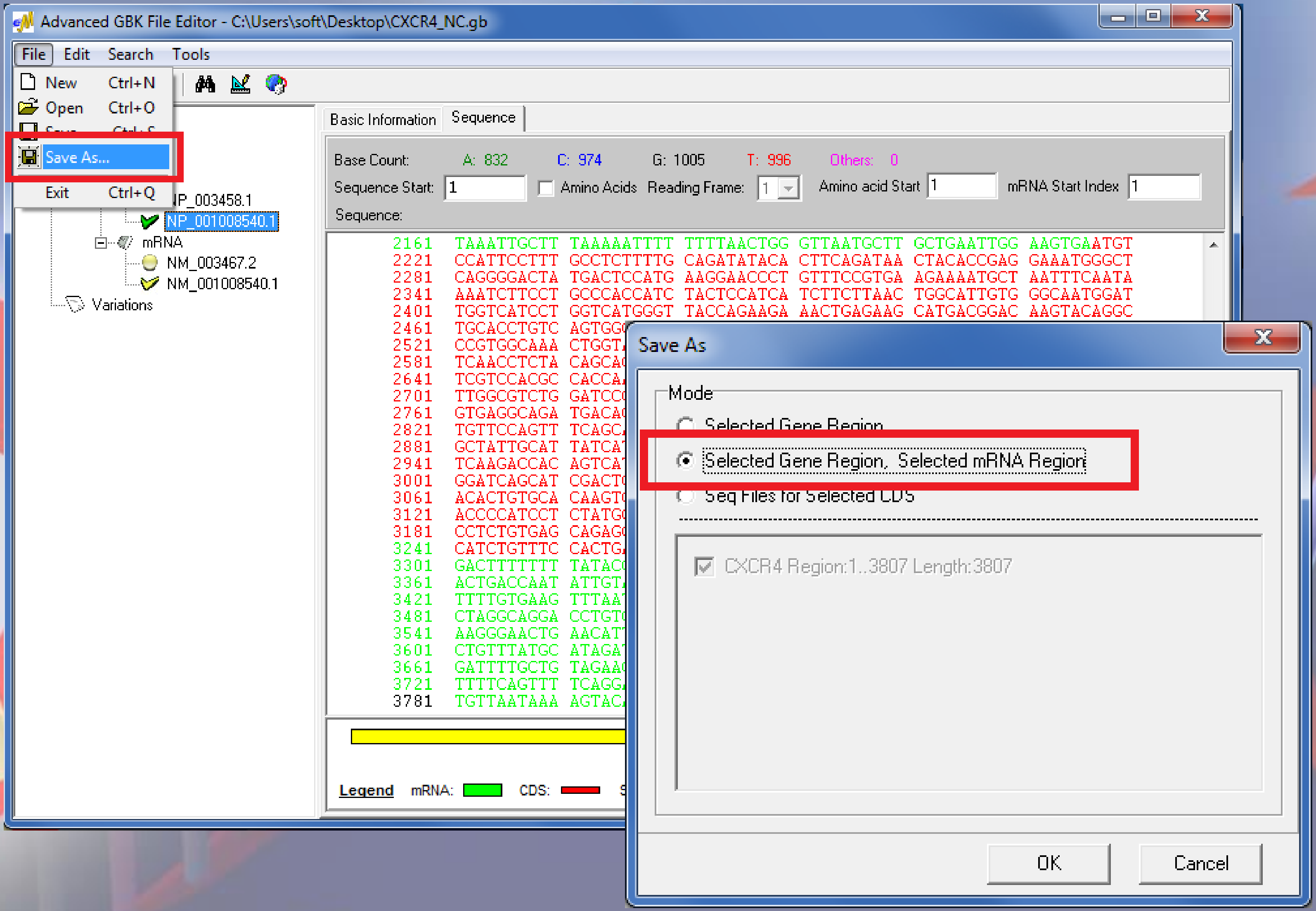
Amino acid Start: 1

mRNA Start Index: 1

Sequence:

```
2161   TAAATTGCTT TAAAAATTTT TTTTAACTGG GTTAATGCTT GCTGAATTGG AAGTGAATGT
2221   CCATTCCTTT GCCTCTTTTG CAGATATACA CTTCAGATAA CTACACCGAG GAAATGGGCT
2281   CAGGGGACTA TGACTCCATG AAGGAACCCT GTTTCCGTGA AGAAAATGCT AATTTCAATA
2341   AAATCTTCCT GCCCACCATC TACTCCATCA TCTTCTTAAC TGGCATTGTG GGCAATGGAT
2401   TGGTCATCCT GGTCATGGGT TACCAGAAGA AACTGAGAAG CATGACGGAC AAGTACAGGC
2461   TGCACCTGTC AGTGGCCGAC CTCCTCTTTG TCATCACGCT TCCCTTCTGG GCAGTTGATG
2521   CCGTGGCAAA CTGGTACTTT GGGAACTTCC TATGCAAGGC AGTCCATGTC ATCTACACAG
2581   TCAACCTCTA CAGCAGTGTC CTCATCCTGG CCTTCATCAG TCTGGACCGC TACCTGGCCA
2641   TCGTCCACGC CACCAACAGT CAGAGGCCAA GGAAGCTGTT GGCTGAAAAG GTGGTCTATG
2701   TTGGCGTCTG GATCCCTGCC CTCCTGCTGA CTATTCCCGA CTTCATCTTT GCCAACGTCA
2761   GTGAGGCAGA TGACAGATAT ATCTGTGACC GCTTCTACCC CAATGACTTG TGGGTGGTTG
2821   TGTTCCAGTT TCAGCACATC ATGGTTGGCC TTATCCTGCC TGGTATTGTC ATCCTGTCCT
2881   GCTATTGCAT TATCATCTCC AAGCTGTCAC ACTCCAAGGG CCACCAGAAG CGCAAGGCCC
2941   TCAAGACCAC AGTCATCCTC ATCCTGGCTT TCTTCGCCTG TTGGCTGCCT TACTACATTG
3001   GGATCAGCAT CGACTCCTTC ATCCTCCTGG AAATCATCAA GCAAGGGTGT GAGTTTGAGA
3061   ACACTGTGCA CAAGTGGATT TCCATCACCG AGGCCCTAGC TTTCTTCCAC TGTTGTCTGA
3121   ACCCCATCCT CTATGCTTTC CTTGGAGCCA AATTTAAAAC CTCTGCCCAG CACGCACTCA
3181   CCTCTGTGAG CAGAGGGTCC AGCCTCAAGA TCCTCTCCAA AGGAAAGCGA GGTGGACATT
3241   CATCTGTTTC CACTGAGTCT GAGTCTTCAA GTTTTCACTC CAGCTAACAC AGATGTAAAA
3301   GACTTTTTTT TATACGATAA ATAACTTTTT TTTAAGTTAC ACATTTTTCA GATATAAAAG
3361   ACTGACCAAT ATTGTACAGT TTTTATTGCT TGTTGGATTT TTGTCTTGTG TTTCTTTAGT
3421   TTTTGTGAAG TTTAATTGAC TTATTTATAT AAATTTTTTT TGTTTCATAT TGATGTGTGT
3481   CTAGGCAGGA CCTGTGGCCA AGTTCTTAGT TGCTGTATGT CTCGTGGTAG GACTGTAGAA
3541   AAGGGAACTG AACATTCCAG AGCGTGTAGT GAATCACGTA AAGCTAGAAA TGATCCCCAG
3601   CTGTTTATGC ATAGATAATC TCTCCATTCC CGTGGAACGT TTTTCCTGTT CTTAAGACGT
3661   GATTTTGCTG TAGAAGATGG CACTTATAAC CAAAGCCCAA AGTGGTATAG AAATGCTGGT
3721   TTTTCAGTTT TCAGGAGTGG GTTGATTTCA GCACCTACAG TGTACAGTCT TGTATTAAGT
3781   TGTTAATAAA AGTACATGTT AAACTTA
```

Select File-Save As
Select the option to save “Selected Gene Region, Selected mRNA Region”
Rename file and select the directory to save the file



Base Count: A: 832 C: 974 G: 1005 T: 996 Others: 0

Sequence Start: 1 ☐ Amino Acids Reading Frame: 1 Amino acid Start 1 mRNA Start Index 1

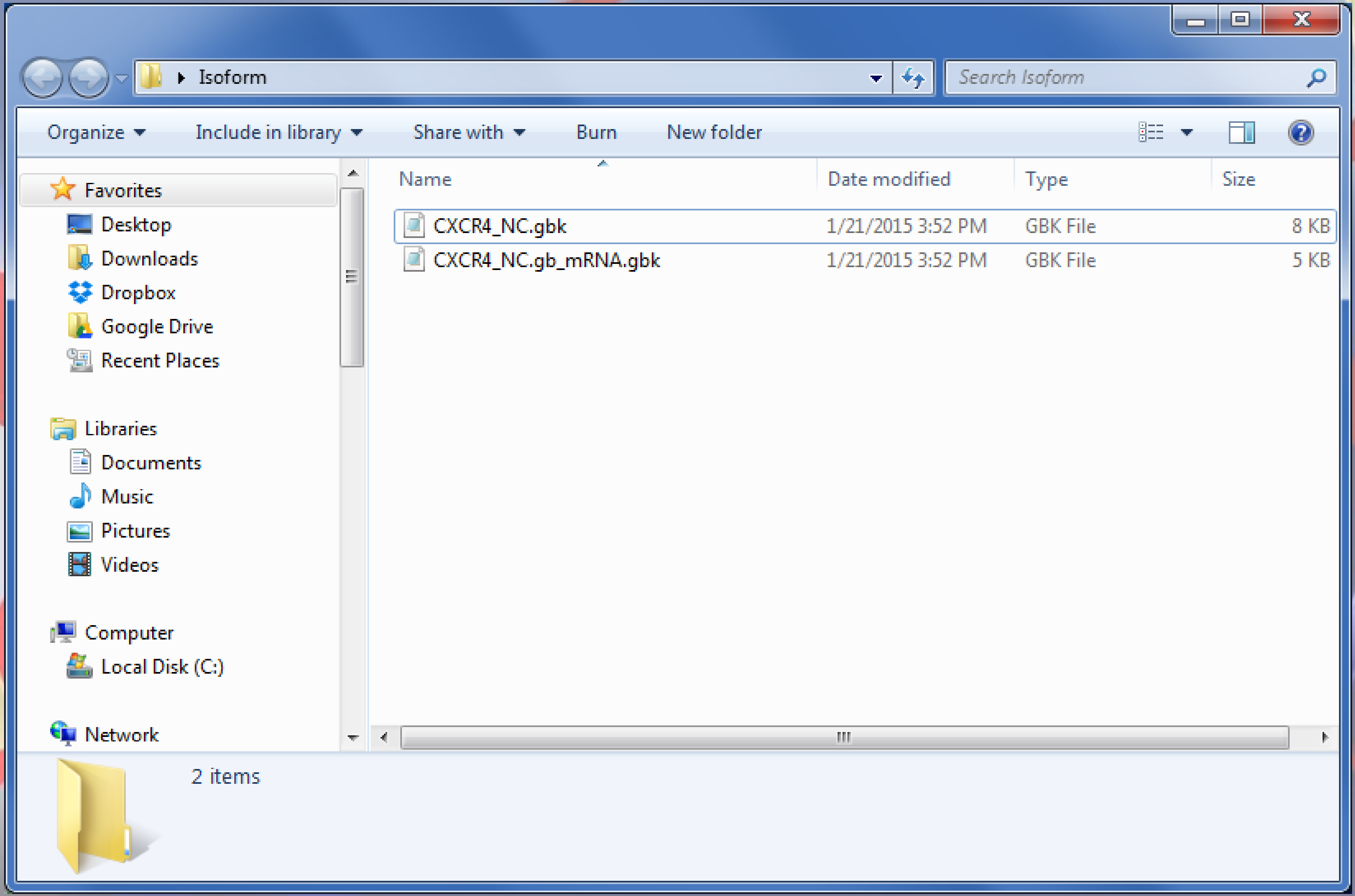
Sequence:

```
2161  TAAATTGCTT  TAAAAATTTT  TTTTAACTGG  GTTAATGCTT  GCTGAATTGG  AAGTGAATGT
2221  CCATTCCTTT  GCCTCTTTTG  CAGATATACA  CTTCAGATAA  CTACACCGAG  GAAATGGGCT
2281  CAGGGGACTA  TGACTCCATG  AAGGAACCCT  GTTTCCGTGA  AGAAAATGCT  AATTTCAATA
2341  AAATCTTCCT  GCCCACCATC  TACTCCATCA  TCTTCTTAAC  TGGCATTGTG  GGCAATGGAT
2401  TGGTCATCCT  GGTCATGGGT  TACCAGAAGA  AACTGAGAAG  CATGACGGAC  AAGTACAGGC
2461  TGCACCTGTC  AGTGG
2521  CCGTGGCAAA  CTGGT
2581  TCAACCTCTA  CAGCA
2641  TCGTCCACGC  CACCA
2701  TTGGCGTCTG  GATCC
2761  GTGAGGCAGA  TGACA
2821  TGTTCCAGTT  TCAGC
2881  GCTATTGCAT  TATCA
2941  TCAAGACCAC  AGTCA
3001  GGATCAGCAT  CGACT
3061  ACACTGTGCA  CAAGT
3121  ACCCCATCCT  CTATG
3181  CCTCTGTGAG  CAGAG
3241  CATCTGTTTC  CACTG
3301  GACTTTTTTT  TATAC
3361  ACTGACCAAT  ATTGT
3421  TTTTGTGAAG  TTTAA
3481  CTAGGCAGGA  CCTGT
3541  AAGGGAACTG  AACAT
3601  CTGTTTATGC  ATAGA
3661  GATTTTGCTG  TAGAA
3721  TTTTCAGTTT  TCAGG
3781  TGTTAATAAA  AGTAC
```

○ Selected Gene Region
● Selected Gene Region, Selected mRNA Region
○ Seq Files for Selected CDS

☑ CXCR4 Region:1..3807 Length:3807

Two files will save:
filename.gbk = Full GenBank file for selected isoform
*filename*_mRNA.gbk = mRNA file for selected isoform, introns removed



Name	Date modified	Type	Size
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CXCR4_NC.gb_mRNA.gbk	1/21/2015 3:52 PM	GBK File	5 KB