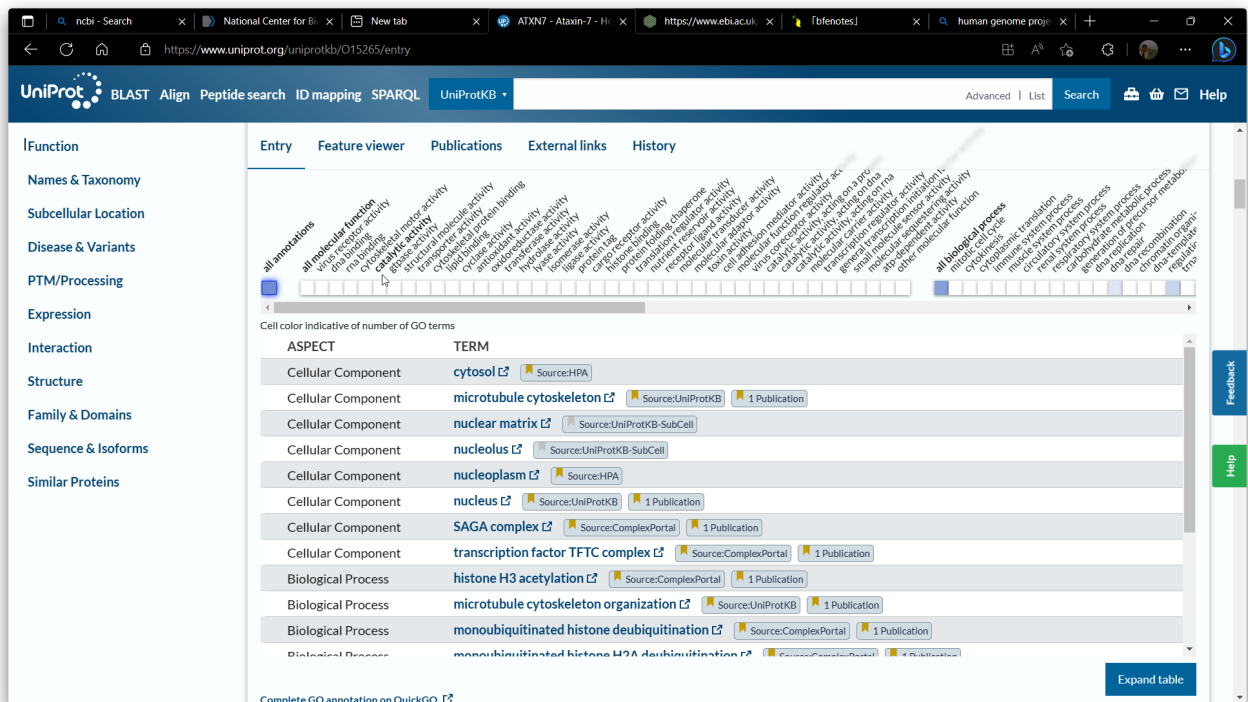


# BFE

- human genome project
- <https://www.uniprot.org/uniprotkb?query=ATXN7>
- select 4 random proteins
- align em
- Homolog?



- STRING: functional protein association networks ([string-db.org](http://string-db.org))
- PTM Post Translational Modification

## Disease & Variants<sup>i</sup>

### Involvement in disease<sup>i</sup>

#### Spinocerebellar ataxia 7 (SCA7)

1 Publication

**Note** | The disease is caused by variants affecting the gene represented in this entry

**Description** | Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show poor gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with brainstem and spinal cord. SCA7 belongs to the autosomal dominant cerebellar ataxias type II (ADCA II) which are associated with ataxia with retinal degeneration and pigmentary macular dystrophy.

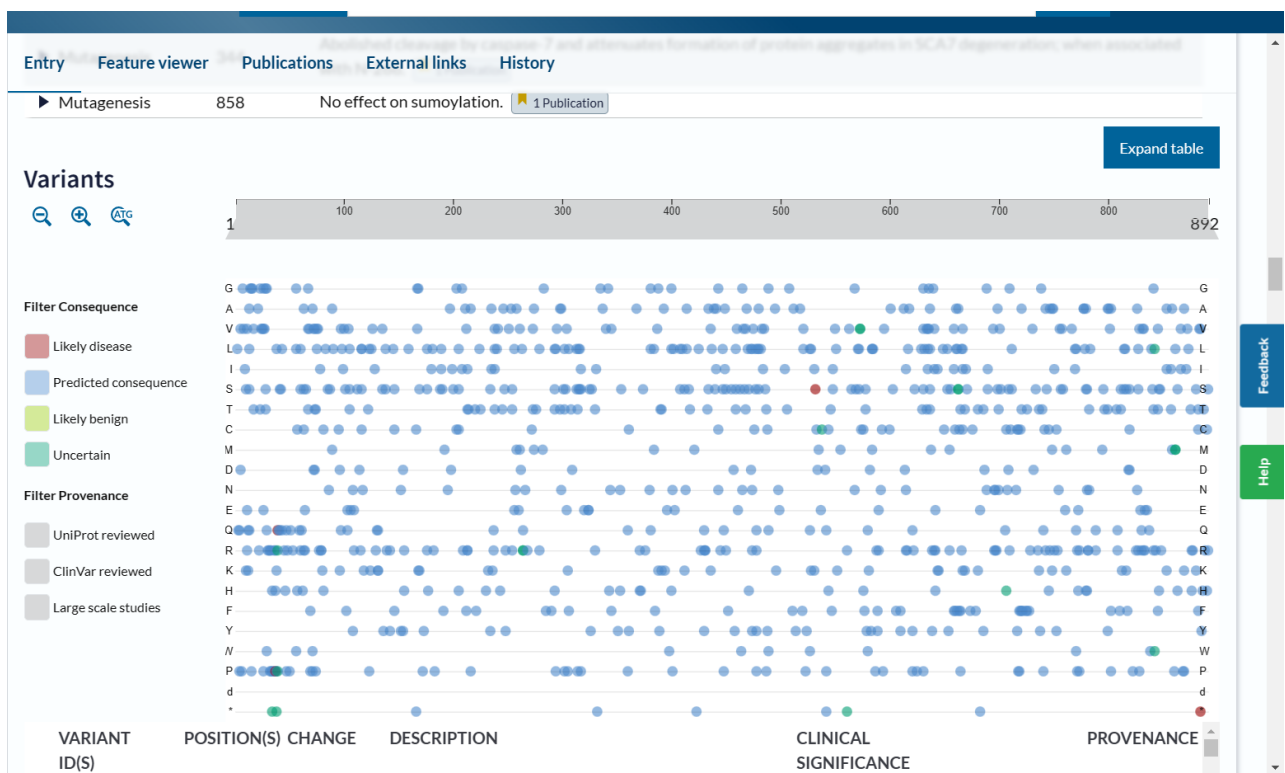
**See also** | MIM:164500 [↗](#)

### Features

Showing features for mutagenesis<sup>i</sup>.

•

- brain disease?



•

retrieve data from database

question relevant to class content

5'----->3'

left to right

**Class generated**

CLUSTAL O(1.2.4) multiple sequence alignment

```
sp|Q14CW9|AT7L3_HUMAN      -----
-----                      0
sp|O15265|ATX7_HUMAN
MSERAADDVRGEPRAAAAAAGGAAAAAARQQQQQQQQQPPPPQPQRQQHPPPPPRRTRP      60
sp|Q8R4I1|ATX7_MOUSE      MSERAADDVRGEPRAAGGA---AA--ARQ-----
QQQQPQLQPQR---QHPPLRRPRA      47
sp|Q9NPA8|ENY2_HUMAN      -----
-----                      0
```

```
sp|Q14CW9|AT7L3_HUMAN      -----
MKMEEMSLSGLDNSKLEAIAQEIY      24
sp|O15265|ATX7_HUMAN
EDGGPGAASTSAAAMATVGERRPLPSPEVMLGQSWNLWVEASKLPGKDGTELDESFKFEG      120
sp|Q8R4I1|ATX7_MOUSE
EDGGTGDTTTSAAAMATVGERRPLPSPEAMLGQSWNLWVEASKLPGKDGTELDESFKFEG      107
sp|Q9NPA8|ENY2_HUMAN      -----MV-----
VSKMNKDAQMRAAINQKLIETG      24
```

```
                                :. .
.   :  ::      *
```

```
sp|Q14CW9|AT7L3_HUMAN
ADLVEDSCLGFCFEVHRAVKCGYFFLDDTDPDSMKDFEIVDQPGLDIFGQVFNQWSKEC      84
sp|O15265|ATX7_HUMAN      ---KNREVMGLCRE-----
DMPIFGFCPAHDDFYLV      148
sp|Q8R4I1|ATX7_MOUSE      ---KNREVMGLCRE-----
DMPIFGLCPAHDDFYLV      135
sp|Q9NPA8|ENY2_HUMAN      ---ERERLKELLRA-----
KLIECGWK---D---Q      45
```

```
                                .   :
:   *
```

sp|Q14CW9|AT7L3\_HUMAN VCPNCSRSIAASRFAPHLEKC-----  
----LGMGRNSSRI-AN 117

sp|O15265|ATX7\_HUMAN  
VCNDCNQVVKPQAFQSHYERRHSSSSKPPLAVPPTSVFSFFPSLSKSKGGSASGSNRSSS 208

sp|Q8R4I1|ATX7\_MOUSE  
VCNDCNQVVKPQAFQSHYERRHSSSSKPALAVPHTSVFSLLPSLSKSKGSGAGGSSRPPS 195

sp|Q9NPA8|ENY2\_HUMAN LKAHCKEVIKEKGLEHVTVD-----  
----- 65  
: .\*. : . :

sp|Q14CW9|AT7L3\_HUMAN RR-IAN---SNNMNKSESD-----  
QEDNDDINDNDWSYGSEK----K-----AK 153

sp|O15265|ATX7\_HUMAN  
GGVLSASSSSSKLLKSPKEKLQLRGNTRPMHPIQQSRVPHGRIMTPSVKVEKIHPKMDGT 268

sp|Q8R4I1|ATX7\_MOUSE GGVLCAS-  
SSSKLLRLPKEKLPLRGNMKPMHPVQQIKVPHGRVMTSPSVKVEKMHPKMDGT 254

sp|Q9NPA8|ENY2\_HUMAN -----  
DLVAEITPKGRALVPDSVK----- 84  
: \*

sp|Q14CW9|AT7L3\_HUMAN KRKSDKNPNSPRRSKS-----  
LKHKNGELSNSDPF-----KYNNSTG 190

sp|O15265|ATX7\_HUMAN  
LLKSAVGPTCPATVSSLVKPGLNCPSIPKPTLPSPGQILNGKGLPAPPTLEKKPEDNSNN 328

sp|Q8R4I1|ATX7\_MOUSE  
LLKSTVGPACPATMSSAVKPGLNCPSIPKPTLPSPGQILNGKGLPAMPTLEKKSEDSSNN 314

sp|Q9NPA8|ENY2\_HUMAN -----  
----- 84

sp|Q14CW9|AT7L3\_HUMAN  
ISYETLGPEELRSLTTQCGVISEHTKKMCTRSRLRCPQHTDEQRRTVRIYFLGPSAVLPE 250

sp|O15265|ATX7\_HUMAN

RKFLNKRLSEREFDPDIHCGVIDLDTKKPCTRSLTCKTHSLTQRRAVQGRRKRFDVLLAE 388

sp|Q8R4I1|ATX7\_MOUSE

RKFLNKRLSEREFDPDIHCGVIDLDTKKPCTRSLTCKTHSLTQRRAVQGRRKRFDVLLAE 374

sp|Q9NPA8|ENY2\_HUMAN

----KELLQRIRTFDAQ 97

: ..\* :

sp|Q14CW9|AT7L3\_HUMAN VESSLDND-

SFDMTDSQALISRLQWDGSSDLSPSDSGSSKTSENQGWGLGTNSSESRTK 309

sp|O15265|ATX7\_HUMAN HKNKTREKELIRHPDSQQPPQPLR-----

DPHPAPPRTSQEPHQNPBGVIPSESKPFVAS 443

sp|Q8R4I1|ATX7\_MOUSE HKNKAREKELIRHD-SQQVPHPLR-----

DPHPTPPRTPQEPQ-----LPAESKPFLAS 422

sp|Q9NPA8|ENY2\_HUMAN

-----HASL----- 101

.

sp|Q14CW9|AT7L3\_HUMAN KKKSHLSLV----GTASGLGSNK-----

KKKPKPPAPPTPS---IYDDIN----- 347

sp|O15265|ATX7\_HUMAN

KPKPHTPSLPRPPGCPAQGGGAPIDPPPVHESHPPLPATEPASRLSSEEGEGDDKEES 503

sp|Q8R4I1|ATX7\_MOUSE

KPKPQTPSLPRPPGCPAQGGGSTPIDPPPGQESHPPLPATEPASRLSSEEGEGDDREES 482

sp|Q9NPA8|ENY2\_HUMAN

----- 101

sp|Q14CW9|AT7L3\_HUMAN

----- 347

sp|O15265|ATX7\_HUMAN

VEKLDCHYSGHHPQPASFCTFGSRQIGRGYYVFDSRWNRLRCALNLMVEKHLNAQLWKKI 563

sp|Q8R4I1|ATX7\_MOUSE

VEKLDCHYSGRHPQPASFCTFGSRQIGRGYYVFDSRWNRLRCALNLMVEKHLNAQLWKKI 542

```
sp|Q9NPA8|ENY2_HUMAN -----
----- 101

sp|Q14CW9|AT7L3_HUMAN -----
----- 347

sp|O15265|ATX7_HUMAN
PPVPSTTSPISTRIPHRTNSVPTSQCGVSYLAAATVSTSPVLLSSTCISPNSKSVPAHGT 623
sp|Q8R4I1|ATX7_MOUSE
PPVPCTTSPVSARVPHRTNSVPTSQGGISYLAATTVSAPPVLLSSTCISPNSKSVPAHGT 602
sp|Q9NPA8|ENY2_HUMAN -----
----- 101

sp|Q14CW9|AT7L3_HUMAN -----
----- 347

sp|O15265|ATX7_HUMAN
TLNAQPAASGAMDPVCSMQSRQVSSSSSSPSTPSGLSSVPSSPMSRKPKQLKSSKSLRPK 683
sp|Q8R4I1|ATX7_MOUSE
TLNAQPAGSGAMDPVCSVQSRQVSASSSPSTPSGLSSVPSSPLSRKPQKWKPSKSIRPK 662
sp|Q9NPA8|ENY2_HUMAN -----
----- 101

sp|Q14CW9|AT7L3_HUMAN -----
----- 347

sp|O15265|ATX7_HUMAN ESSGNSTNCQNASSSTSGGSGKKRKNSSPLL VHSSSSS-
SSSSSSSHSMESFRKNCVAHS 742
sp|Q8R4I1|ATX7_MOUSE
ESSALSTNCHNASSSTSGGSGKKRKNSSPLLVPSSSSSSSSSSSSSHSVNSFRKNCVAHS 722
sp|Q9NPA8|ENY2_HUMAN -----
----- 101
```

```

sp|Q14CW9|AT7L3_HUMAN      -----
-----                      347
sp|O15265|ATX7_HUMAN
GPPYPSTVTSSHSIGLNCVTNKANAVNVRHDQSGRGPPTGSPAESIKRMSVMVNSSDSTL      802
sp|Q8R4I1|ATX7_MOUSE
GTPYLSTAPSSHSIGLNCVTNKTHSVSLRHEQAGRGPAGVSSAEPKRMMSVMVNSSDSTL      782
sp|Q9NPA8|ENY2_HUMAN      -----
-----                      101


sp|Q14CW9|AT7L3_HUMAN      -----
-----                      347
sp|O15265|ATX7_HUMAN
SLGPFIHQSNELPVNSHGFSHSHTPLDKLIGKKRKCSPPSSSSINNSSSKPTKVAKVPAV      862
sp|Q8R4I1|ATX7_MOUSE      SLGPFIHQASELPVNP-----
HSHTPLDKLIGKKRKCSPPSSSTVGNSGSKPTKVAKLPAM 837
sp|Q9NPA8|ENY2_HUMAN      -----
-----                      101


sp|Q14CW9|AT7L3_HUMAN      -----                      347
sp|O15265|ATX7_HUMAN      NNVHMKHTGTIPGAQGLMNSSLLHQPKARP      892
sp|Q8R4I1|ATX7_MOUSE      NNVHMKHTGNISGAQGLTNNSSLLHQPKARP      867
sp|Q9NPA8|ENY2_HUMAN      -----                      101

```

google list of genes in humans  
information retrieval

1. Gnee name