

> Shizofrenija je dedna bolezen, katere simptomi so halucinacije, apatija, pomanjkanje čustev, slabo socialno funkcioniranje ter kognitivne motnje. Študije so pokazale povezavo med ekspresijo proteina s podanim mRNA zaporedjem in povečanjem tveganja za nastanek shizofrenije.

>mRNA zaporedje (*Homo sapiens*)

```
ATGGCCGGGCAGCACCTCCCGGTACCCCGGCTGGAGGGCGTTTCTCGGGAGCAGTTCATGC
AGCACCTCTACCCACAGAGAAAACCTCTTGTGTTGGAAGGGATTGATTGTTGGGGCCATGTACAAG
CAAATGGACAGTGGATTACCTAAGCCAAGTTGGAGGGAAGAAAGAAGTAAAGATTCATGTTGCTG
CAGTTGCACAGATGGACTTCATTAGTAAGAACTTTGTATATAGAACTTTACCTTTTGACCAGTTGGTC
CAGAGGGCAGCTGAAGAGAAACATAAAGAATTCTTTGTTTCAGAGGATGAGAAATACTACTTACG
GTCACCTTGAGAAGACCCTAGAAAGGATGTTGCAGATATCAGAAAGCAGTTTCCTTTGTTGAAAG
GAGATATTAAGTTTCCAGAATTCTTCAAAGAGGAACAGTTCTTTCCAGTGTTTTTCGAATTAGTTCA
CCAGGATTACAACCTATGGACTCATTATGATGTAATGGATAATTTGTTAATACAAGTGACAGGAAAAA
AGCGTGTGTACTCTTCAGTCCTCGAGATGCCAGTATTTATATTTAAAGGTACTAAATCAGAAGT
ACTGAATATAGATAACCCAGACTTGGCTAAATATCCACTTTTTTCCAAGGCTAGAAGATATGAATGT
TCCCTTGAAGCTGGTGATGTATTATTCATTCCTGCTTTATGGTTCATAATGTAATTTCTGAAGAGTTT
GGAGTGGGAGTGAATATCTTTGGAAGCACCTTCCATCTGAATGCTATGATAAACAGATACCTAT
GGAAACAAAGATCCTACAGCAGCATCAAGAGCTGCACAAATTCTGGACAGAGCCTTGAAAACA
CTGGCCGAGTTACCAGAGGAATATAGGGACTTCTATGCACGACGAATGGTCCTACACATTCAAG
ACAAAGCCTACAGCAAGAACTCTGAGTAA
```

1)

1.a) Najdi zapis zaporedja v GenBank.

NIH National Library of Medicine
National Center for Biotechnology Information

Nucleotide Nucleotide Search

GenBank Send to: Change region shown

Homo sapiens tRNA-yW synthesizing protein 5 (TYW5), transcript variant 1, mRNA

NCBI Reference Sequence: NM_001039693.3

FASTA Graphics

Go to: (v)

LOCUS	NM_001039693	5123 bp	mRNA	linear	PRI 28-APR-2025
DEFINITION	Homo sapiens tRNA-yW synthesizing protein 5 (TYW5), transcript variant 1, mRNA.				
ACCESSION	NM_001039693				
VERSION	NM_001039693.3				
KEYWORDS	RefSeq; MANE Select.				
SOURCE	Homo sapiens (human)				
ORGANISM	Homo sapiens				
	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo.				
REFERENCE	1 (bases 1 to 5123)				
AUTHORS	Zhang, C., Li, X., Zhao, L., Liang, R., Deng, W., Guo, W., Wang, Q., Hu, X., Du, X., Sham, P. C., Luo, X. and Li, T.				
TITLE	Comprehensive and integrative analyses identify TYW5 as a schizophrenia risk gene				
JOURNAL	BMC Med 20 (1), 169 (2022)				
PUBMED	35527273				
REMARK	COMMENT: Comprehensive and integrative analyses identify TYW5 as a				

Articles about the TYW5 gene

- Scalable multiplex co-fractionation/mass spectrometry platform for ac [Nat Commun. 2022]
- Comprehensive and integrative analyses identify TYW5 as a schizophrenia risk gene [BMC Med. 2022]
- Regulatory variants at 2q33.1 confer schizophrenia risk by modulating dis [Brain. 2022]

See all...

1.b) Koliko eksonov vsebuje? Kje najdemo ta podatek?

>> Najdemo ga na GenBank, protein vsebuje 8 eksonov

1.a) Poišči UniProt kodo iskanega proteina:

>>A2RUC4

1.b) Protein je metaloproteaza, kateri ion veže? Koliko teh ionov veže celoten protein?

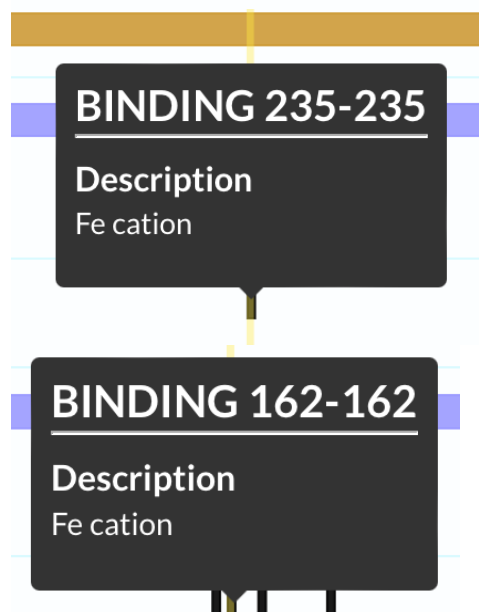
Kje se nahaja vezavno mesto?

>>Fe²⁺ ion, vezan je en ion na podenoto, torej celoten protein veže dav iona.

Cofactorⁱ

Fe²⁺ ([UniProtKB](#) | [Rhea](#)  | [CHEBI:29033](#) )  1 Publication

Note: Binds 1 Fe²⁺ ion per subunit.  1 Publication



1.d) Protein imamo v raztopini pri pH vrednosti izoelektrične točke celotnega proteina. Kako je pri takšnem pH nabit ostanek št. 133?

>> +1

dbSNP

SNP rs203772

Create alert Advanced

Validation Status
by-ALFA
by-cluster
by-frequency
Publication
PubMed Cited
PubMed Linked
Function Class
intron
Annotation
somatic
Global MAF
Custom range...
[Clear all](#)
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Display Settings: Summary, Sorted by SNP_ID
Search results
Items: 2
☐ **rs203772** [*Homo sapiens*]
1.

Variant type: SNV
Alleles: G>A,C [\[Show Flanks\]](#)
Chromosome: 2:200025965 (GRCh38)
2:200890688 (GRCh37)
Canonical SPDI: NC_000002.12:200025964:G:A,NC_000002.12:200025964:G:C
Gene: LOC124906112 ([Varview](#))
Functional Consequence: intron_variant
Validated: by frequency,by alfa,by cluster
MAF: A=0.308191/11085 ([ALFA](#))
A=0.165094/35 (Vietnamese)
A=0.194097/1243 (1000Genomes_30X)
...more
HGVS: NC_000002.12:g.200025965G>A, NC_000002.12:g.200025965G>C,
NC_000002.11:g.200890688G>A, NC_000002.11:g.200890688G>C
[PubMed](#)

1.e) Enonukleotidni polimorfizem rs203772 je povezan shizofrenijo, tveganjski alel pa je povezan z višjo transkripcijo gena TYW5 v prefrontalni skorji. V kateri NCBI bazi lahko najdemo podatke za ta polimorfizem?

>> To je glavna baza za SNP-je (Single Nucleotide Polymorphisms).

dbSNP

SNP rs203772

Create alert Advanced

Validation Status
by-ALFA
by-cluster
by-frequency
Publication
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PubMed Linked
Function Class
intron
Annotation
somatic
Global MAF
Custom range...
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Display Settings: Summary, Sorted by SNP_ID
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...more
HGVS: NC_000002.12:g.200025965G>A, NC_000002.12:g.200025965G>C,
NC_000002.11:g.200890688G>A, NC_000002.11:g.200890688G>C
[PubMed](#)

2.

2.a) Katere domene sestavljajo protein?

>>JmjC

2.b) Koliko člankov vsebuje ime te domene?

>>597

2.c) Koliko preglednih člankov je izšlo v zadnjih petih letih?

>>34

2.d) Domene JmjN in JmjC se pojavljajo skupaj in verjetno tvorijo eno samo funkcionalno enoto v zviti strukturi proteina, vendar so kasneje domeno JmjC odkrili tudi brez domene JmjN pri organizmih vse od bakterij do človeka. Poišči protein, ki vsebuje obe domeni.

Searching in

UniProtKB

Gene Name [GN]

YDJ1 Remove

AND Domain [FT] jmjC Evidence Any Remove

AND Domain [FT] jmjN Evidence Any Remove

AND All a4_human, P05067, cdc7 human Remove

Add Field

Cancel Search

>>

★ Q6B0I6 · KDM4D_HUMAN

Protein ⁱ	Lysine-specific demethylase 4D	Amino acids	523 (go to sequence)
Gene ⁱ	KDM4D	Protein existence ⁱ	Evidence at protein level
Status ⁱ	★ UniProtKB reviewed (Swiss-Prot)	Annotation score ⁱ	5/5
Organism ⁱ	Homo sapiens (Human)		

2.e) Isto domeno vsebuje tudi človeška lizinska histonska demetilaza KDM4. Ali se vezavna mesta pri našem iskanem proteinu in v proteinu KDM4 na katerih mestih pokrivajo?

>> ne, nikjer ne najdemo pokrivajočih-se vezavnih mest.

<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>-----MAG-----</div> <div>METMKSKANCAQNPNCNIMIFHPTKEEFNDFDKYIAYMESQGAHRAGLAKIIPPKEWKARETYDNISEI</div>	<div>3</div> <div>69</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>-----OHLVPVPRLEGVSRREQFMQHLYPQRK-PL</div> <div>L IATPLQQVASGRAGVFTQYHKKKKAMTVGEYRHLANSKKYQTPPHQNFEDLERKYWKNRIYNPSIYGA</div>	<div>30</div> <div>138</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>VLEGIDLGPC^TSKWTVDYLSQVGGKKEVKIHV--AAVA---QMDFISKNFVYRTLPFDQLVQRAAEKK</div> <div>DISGSLFDENTKQWNLGHLGTIQDLLKECGV^VIEGVNTPYLYFGMWKTTFAWHTEDMDLYS-----</div>	<div>93</div> <div>200</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>HKEFFVS^EDEKY^YLRSLGEDPRKDVADIRKQF^PLLKGD^IKFPE^FFKEEQFFSSVFR^ISSPGLQLWT^HY^D</div> <div>I^NYLHLGE^PK^TWYVVPPEHGQRLE-RLAREL^FPGSSR--G^GGAF^LR^HK^VALISPTVLKENGIPF-----</div>	<div>162</div> <div>261</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>VMD^NLLIQVTG^KK^RRV^VLFSPRDAQ^YLYLKGTK-----SEVL^NIDNPDLAKY^YPLFSKARRYE^CSLEAGD^V</div> <div>---NRITQEAGEFM-VT-----FPYGY^HAGFNHGFNCAE^AIN^FFATPRWIDY^YGKM---ASQ^SS^GGEARV</div>	<div>226</div> <div>317</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>L^FI^PALWF^HNVISEEFGVGN^IFWKH^LPSECY^DKTD^TY-----GNKD^PT-----AASRAAQ^ILD^RA</div> <div>T^FSM-----DAFVRI^LQPERY^DLWKRQDRAVVDHME^PRVPASQELSTQKEVQLPRRA</div>	<div>282</div> <div>370</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>LKT^LAELP^EEYRDFYARR^MVL-----HIQDKAYS^KNS^E----</div> <div>ALGLRQLP^SSHWARHSPWP^MAARSGTRCHTLVCSSLPRRSVSGTATQPRAAAVHSSK^KPSSTP^SSTPGP</div>	<div>315</div> <div>439</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>-----</div> <div>SAQIIHPSNGRRGRGRPPQKLRAQELTLQTPAKRPLL^AGGTTCTASGPEPEPLPEDGALMDKPVPLSPGL</div>	<div>315</div> <div>508</div>
A2RUC4:Binding site		
<div><div>sp A2RUC4 TYW5_HUMAN</div><div>sp Q6B0I6 KDM4D_HUMAN</div></div>	<div>-----</div> <div>QHPVKASGCSWAPVP</div>	<div>315</div> <div>523</div>
A2RUC4:Binding site		