

Profile: Open Gargion

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Annotation by organism



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[HomoGene](#) [SHIELD](#) [DfG](#) [PhosphoSite](#)

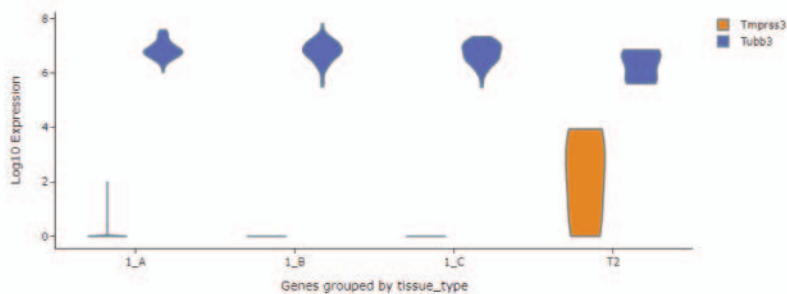
Deatness gene annotation



Functional annotation - transmembrane protease, serine 3

scRNA-seq Dataset (Shrestha, et. al, 2018)

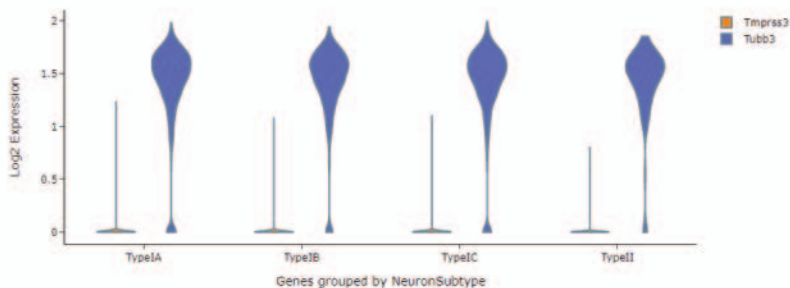
Log10 Gene Expression



P25-P27

scRNA-seq Dataset (Sun, et. al, 2018)

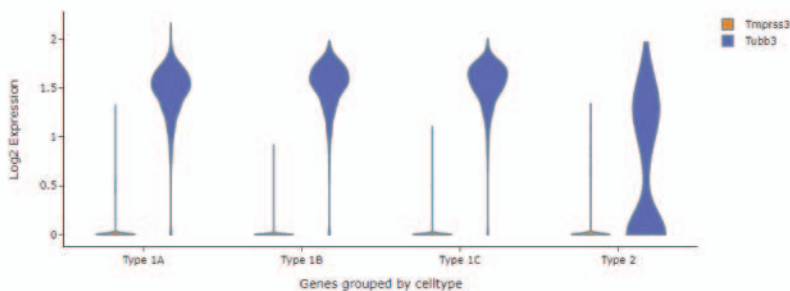
Log2 Gene Expression



9 week

scRNA-seq Dataset (Milon, et. al, 2021)

Log2 Gene Expression



9 week

HGVSc

NM_024022.4:c.1343T>C

NM_024022.4:c.1343T>C

NM_024022.4:c.1306C>T

NM_024022.4:c.1306C>G

NM_024022.4:c.1286A>G

NM_024022.4:c.1276G>A

NM_024022.4:c.1273T>C

NM_024022.4:c.1250G>A

NM_024022.4:c.1219T>C

NM_024022.4:c.1211C>T

NM_024022.4:c.1204G>A

NM_024022.4:c.1195-1G>C

NM_024022.4:c.1192C>T

NM_024022.4:c.1181_1187delinsATCACTGATCACCCACTCTGCACTGATCT

NM_024022.4:c.1159G>A

NM_024022.4:c.1129G>A

NM_024022.4:c.1048+1G>A

NM_024022.4:c.1039G>T

NM_024022.4:c.1025G>A

NM_024022.4:c.999del

NM_024022.4:c.989del

NM_024022.4:c.974T>A

NM_024022.4:c.953-5A>G

NM_024022.4:c.916G>A

NM_024022.4:c.913A>T

NM_024022.4:c.783-1G>T

NM_024022.4:c.783-1G>A

NM_024022.4:c.782+7_782+8insT

NM_024022.4:c.782+2T>A

NM_024022.4:c.771C>G

NM_024022.4:c.753G>C

NM_024022.4:c.743C>T

NM_024022.4:c.727G>A

NM_024022.4:c.718del

NM_024022.4:c.677C>A

NM_024022.4:c.647G>T

NM_024022.4:c.647G>A

NM_024022.4:c.646C>T

NM_024022.4:c.616G>T

HGVSp

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NP_076927.1:p.Met448Thr

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NP_076927.1:p.Arg436Gly

NP_076927.1:p.Asn429Ser

NP_076927.1:p.Ala426Thr

NP_076927.1:p.Cys425Arg

NP_076927.1:p.Gly417Glu

NP_076927.1:p.Cys407Arg

NP_076927.1:p.Pro404Leu

NP_076927.1:p.Gly402Arg

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NP_076927.1:p.Gln398Ter

NP_076927.1:p.Val394AspfsTer3

NP_076927.1:p.Ala387Thr

NP_076927.1:p.Gly377Ser

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NP_076927.1:p.Glu347Ter

NP_076927.1:p.Gly342Glu

NP_076927.1:p.Asp334MetfsTer24

NP_076927.1:p.Glu330GlyfsTer28

NP_076927.1:p.Leu325Gln

-

NP_076927.1:p.Ala306Thr

NP_076927.1:p.Ile305Phe

-

-

-

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NP_076927.1:p.His257Gln

NP_076927.1:p.Trp251Cys

NP_076927.1:p.Thr248Met

NP_076927.1:p.Gly243Arg

NP_076927.1:p.His240ThrfsTer35

NP_076927.1:p.Ser226Ter

NP_076927.1:p.Arg216Leu

NP_076927.1:p.Arg216His

NP_076927.1:p.Arg216Cys

NP_076927.1:p.Ala206Ser

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NM_024022.4:c.595G>A
NM_024022.4:c.582T>A
NM_024022.4:c.581G>T

NM_024022.4:c.579dup
NM_024022.4:c.554A>T

NM_024022.4:c.551T>C
NM_024022.4:c.436G>A
NM_024022.4:c.432del

NM_024022.4:c.413C>A
NM_024022.4:c.399G>C
NM_024022.4:c.326G>A

NM_024022.4:c.325C>T

NM_024022.4:c.323-6G>A
NM_024022.4:c.316C>T
NM_024022.4:c.310G>T
NM_024022.4:c.308A>G
NM_024022.4:c.271C>T

NM_024022.4:c.208del
NM_024022.4:c.115C>T

NM_024022.4:c.46C>T
NM_024022.4:c.36dup
NM_024022.4:c.36del

NP_076927.1:p.Gln203Ter
NP_076927.1:p.Val199Met
NP_076927.1:p.Cys194Ter
NP_076927.1:p.Cys194Phe

NP_076927.1:p.Cys194MetfsTer17
NP_076927.1:p.His185Leu

NP_076927.1:p.Leu184Ser
NP_076927.1:p.Gly146Ser
NP_076927.1:p.Gln144HisfsTer8

NP_076927.1:p.Ala138Glu
NP_076927.1:p.Trp133Cys
NP_076927.1:p.Arg109Gln

NP_076927.1:p.Arg109Trp

-
NP_076927.1:p.Arg106Cys
NP_076927.1:p.Glu104Ter
NP_076927.1:p.Asp103Gly
NP_076927.1:p.Arg91Ter

NP_076927.1:p.His70ThrfsTer19
NP_076927.1:p.Gln39Ter

NP_076927.1:p.Arg16Ter
NP_076927.1:p.Phe13LeufsTer10
NP_076927.1:p.Phe13SerfsTer12

ACMG Codes	Classification	PMID
PVS1_moderate, PM3, PP3, PM2	Likely Pathogenic	PMID: 30242206
PM3_VeryStrong, PP1, PP3, PM2	Pathogenic	PMID:24033266,28566687
PM3_Strong, PP1_moderate, PP3, PM2_Supporting	Likely Pathogenic	PMID:26969326,28566687,24033266
PM3, PM5, PP3, PM2	Likely Pathogenic	PMID:28566687
PM3, PP1, PP3, PM2	Likely Pathogenic	PMID:26969326
PM3_Strong, PP1_Strong, PS3_Supporting, PP3	Pathogenic	PMID:21786053,11907649,20301607,25741868,12920079,24033266,28566687,28695016,29196752,30487145
PM3_Supporting, PP1_Strong PP3, PM2	Likely Pathogenic	PMID:21534946
PM3, PP1, PP3, PM2	Likely Pathogenic	PMID:28695016
PM3_supporting, PP1_Strong, PP3, PM2	Likely Pathogenic	PMID:11424922,12920079
PM3_Moderate, PP1_Strong, PP3, PS3_Supporting, PM2	Pathogenic	PMID:11462234,16021470,31850270,12920079
PM3, PP1, PP3, PM2	Likely Pathogenic	PMID:28695016
PVS1_Stong, PP1_moderate, PP3, PM2	Pathogenic	PMID:21726435,25525159
PVS1_Stong, PP1_moderate, PM2	Pathogenic	PMID:16021470,24033266,25525159
PVS1_Strong, PP1_moderate, PM2	Pathogenic	PMID:11137999
PM3, PP1, PP3, PM2	Likely Pathogenic	PMID:24130743,25770132
PM3_supporting, PP1_moderate, PP3, PM2	Likely Pathogenic	PMID:26226137
PVS1_Strong, PP1, PM2	Pathogenic	PMID:29889784
PVS1_Strong, PP1, PM2	Pathogenic	PMID:31045651
PM3_moderate, PP1_Moderate, PP3, PM2	Likely Pathogenic	PMID:20301607,21117948,24033266,25525159
PVS1_Strong, PM3_supporting, PM2	Likely Pathogenic	PMID:28566687
PVS1_Strong, PP1_strong, PM2	Pathogenic	PMID:16460646
PM3, PP1, PP3, PM2	Likely Pathogenic	PMID:28566687
PVS1_Strong, PM3, PP3, PM2	Pathogenic	PMID:28566687
PM3_VeryStrong, PP1_Strong, PM2_Supporting, PP3, PS3_Supporting.	Pathogenic	PMID:24033266,17551081,21786053,23958653,24526180,28695016,23208854,28246597,31045651
PM3_supporting, PP1_moderate, PP3, PM2	Likely Pathogenic	PMID:26226137
PVS1_Strong, PM3_supporting, PP1, PM2	Likely Pathogenic	PMID:30303587
PVS1_Strong, PS1, PM2	Likely Pathogenic	PMID:29072634
PVS1_moderate, PM3_supporting, PP1, PP3, PM2	Likely Pathogenic	PMID:15447792
PVS1_Strong, PM3_supporting, PP3, PM2	Likely Pathogenic	PMID:28566687
PM3, PM1, PP3, PM2	Likely Pathogenic	PMID:25770132
PS1_Strong, PM2_Moderate, PM5_Moderate, PP3	Pathogenic	PMID:11462234,12920079
PM3_Supporting, PS3_supporting, PP3, PM2	Likely Pathogenic	PMID:24526180
PM3_supporting, PP1_moderate, PP3, PM2	Likely Pathogenic	PMID:24416283,20301607,24033266
PVS1_Strong, PP1, PM2	Likely Pathogenic	PMID:30622556
PVS1_Strong, PM3_supporting, PM2	Likely Pathogenic	PMID:24853665
PS3_supporting, PP3, PP1, PM2	Likely Pathogenic	PMID:16021470
PM5, PP3, PS3_Supporting, PM2	Likely Pathogenic	PMID:12920079,17551081,16021470,24033266,26445815
PM5, PP3, PS3_Supporting, PM2	Likely Pathogenic	PMID:17551081
PM3_supporting, PP1_strong, PP3, PM2	Likely Pathogenic	PMID:26226137

PVS1_Strong, PM3_Supporting, PP1, PM2	Pathogenic	PMID:24130743,25770132
PM3_Strong, PP3, PM2	Likely Pathogenic	PMID:21786053
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PM3_supporting, PP1_Moderate, PP3, PM2	Likely Pathogenic	PMID:11424922,12920079
PVS1_Strong, PP1, PM2	Likely Pathogenic	PMID:24033266,26036852,26408194,2856687,30622556
PM3_Supporting, PM5, PP3, PM2	Likely Pathogenic	PMID:23967202
PS3_supporting, PM3_moderate, PP1, PP3, PM2	Likely Pathogenic	PMID:31016883,31379920,32235586,32235586
PM3_supporting, PP1_moderate, PP3, PM2	Likely Pathogenic	PMID:26226137
PVS1_Strong, PM3_supporting, PP1, PM2	Pathogenic	PMID:31379920
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PM3_Strong, PP1_moderate, PP3, PM2	Likely Pathogenic	PMID:26226137
PM3_supporting, PP1_moderate, PP3, PM2	Likely Pathogenic	PMID:26226137
PM3_supporting, PP1, PM5, PP3, PM2	Likely Pathogenic	PMID:24853665
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PM3_VeryStrong, PP1, PS3_Supporting, PP3	Pathogenic	PMID:11137999,24033266,24416283,28695016,30622556
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PP1, PS3_Supporting, PP3, PM2	Likely Pathogenic	PMID:11907649,12920079
PVS1_Strong, PM3_supporting, PP1, PM2	Pathogenic	PMID:26226137
		PMID:21534946,26036852,11907649,12920079,24033266,26408194,28263784,30311386,15447792,25741868,17981648,19170735,21786053,28566687,29293505
PVS1_Strong, PM3_Verystrong, PP1_Strong	Pathogenic	PMID:26969326
PVS1_Strong, PM3_supporting, PM2	Likely Pathogenic	PMID:26969326
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PVS1_Strong, PM3, PP1, PM2	Pathogenic	PMID:27344577,30245029,22133722
PVS1_Strong, PM3_supporting, PP1, PM2	Likely Pathogenic	PMID:23226338
PVS1_Strong, PM3_supporting, PM2	Likely Pathogenic	PMID:28246597