

# SLC1A3 variant M128R

## Overview of the raw variants

Raw variant 1: substitution at 859

GGAATGCGAGCTGTAGTCTATTATA T GACTACCACCATCATTGCTGTGGTG  
GGAATGCGAGCTGTAGTCTATTATA G GACTACCACCATCATTGCTGTGGTG

[NM\\_004172.4:n.859T>G](#)

## Affected transcripts - 1

[NM\\_004172.4\(SLC1A3\\_v001\):c.383T>G](#)

## Affected proteins - 1

NM\_004172.4(SLC1A3\_i001):p.(Met128Arg)

## Reference protein

1	MTKSNGEETPK	MGGRMERFQQ	GVKRTLLAK	KKVQNITKED	VKSYLFRNAF	VLLTVTAVIV
61	GTILGFTLRP	YRMSYREVKY	FSFPGELLMR	MLQMLVLPLI	ISSLVTGMAA	LDSKASGKMG
121	MRAVVYYMTT	TIIAVVIGII	IVIIIHPGKG	TKENMHREGK	IVRVTAADAF	LDLIRNMFPP
181	NLVEACFKQF	KTNYEKRSFK	VPIQANETLV	GAVINNVSEA	METLTRITEE	LVPVPGSVNG
241	VNALGLVVFS	MCFGFVIGNM	KEQGQALREF	FDSLNEAIMR	LVAVIMWYAP	VGILFLIAGK
301	IVEMEDMGVI	GGQLAMYTVT	VIVGLLIHAV	IVLPLLYFLV	TRKNPWVFIG	GLLQALITAL
361	GTSSSSATLP	ITFKCLEENN	GVDKRVTRFV	LPVGATINMD	GTALYEALAA	IFIAQVNNFE
421	LNFGQIITIS	ITATAASIGA	AGIPQAGLVT	MVIVLTSVGL	PTDDITLIIA	VDWFLDRLRT
481	TTNVLGDSL	AGIVEHLRSH	ELKNRDVEMG	NSVIEENEMK	KPYQLIAQDN	ETEKPIDSET
541	KM*					

## Protein predicted from variant coding sequence

1	MTKSNGEETPK	MGGRMERFQQ	GVKRTLLAK	KKVQNITKED	VKSYLFRNAF	VLLTVTAVIV
61	GTILGFTLRP	YRMSYREVKY	FSFPGELLMR	MLQMLVLPLI	ISSLVTGMAA	LDSKASGKMG
121	MRAVVYYRTT	TIIAVVIGII	IVIIIHPGKG	TKENMHREGK	IVRVTAADAF	LDLIRNMFPP
181	NLVEACFKQF	KTNYEKRSFK	VPIQANETLV	GAVINNVSEA	METLTRITEE	LVPVPGSVNG
241	VNALGLVVFS	MCFGFVIGNM	KEQGQALREF	FDSLNEAIMR	LVAVIMWYAP	VGILFLIAGK
301	IVEMEDMGVI	GGQLAMYTVT	VIVGLLIHAV	IVLPLLYFLV	TRKNPWVFIG	GLLQALITAL
361	GTSSSSATLP	ITFKCLEENN	GVDKRVTRFV	LPVGATINMD	GTALYEALAA	IFIAQVNNFE
421	LNFGQIITIS	ITATAASIGA	AGIPQAGLVT	MVIVLTSVGL	PTDDITLIIA	VDWFLDRLRT
481	TTNVLGDSL	AGIVEHLRSH	ELKNRDVEMG	NSVIEENEMK	KPYQLIAQDN	ETEKPIDSET
541	KM*					

## Effects on Restriction sites

### Raw variant Created Deleted

1

## Experimental services

Genomic description: 859T>G

# SLC1A3 variant C186S

## Overview of the raw variants

Raw variant 1: substitution at 1032

GTTCCCTCCAAATCTGGTAGAAGCC T GCTTTAAACAGTTTAAAACCAACTA  
GTTCCCTCCAAATCTGGTAGAAGCC A GCTTTAAACAGTTTAAAACCAACTA

[NM\\_004172.4:n.1032T>A](#)

## Affected transcripts - 1

[NM\\_004172.4\(SLC1A3\\_v001\):c.556T>A](#)

## Affected proteins - 1

NM\_004172.4(SLC1A3\_i001):p.(Cys186Ser)

## Reference protein

1	MTKSNGE	EPK	MGGRMER	FQQ	GVRKR	TLLAK	KKVQN	ITKED	VKSYL	FRNAF	VLLTV	TAVIV
61	GTILGF	TLRP	YRMSY	REVKY	FSFPG	ELLMR	MLQML	VLPLI	ISSLV	TGMAA	LDSKAS	GKMG
121	MRAVV	YYMTT	TIIAV	VIGII	IVIII	IHPGK	G	TKENM	HREGK	IVRVTA	ADAF	LDLIRN
181	NLVEA	CFKQF	KTNYE	KRSFK	VPIQA	NETLV	GAVINN	VSEA	METL	TRITEE	LVPVP	GSVNG
241	VNALG	LVVFS	MCFGF	VIGNM	KEQGQ	ALREF	FDSLNE	AIMR	LVAVI	MWYAP	VGILF	LIAGK
301	IVEMED	MGVI	GGQLA	MYTVT	VIVGL	LIHAV	IVLPL	LYFLV	TRKNP	PWVFI	G	LLQAL
361	GTSSSS	SATLP	ITFKC	LEENN	GVDKR	VTRFV	LPVGAT	INMD	GTALY	EALAA	IFIAQ	VNNFE
421	LNFGQ	IITIS	ITATA	AASIG	AGIPQ	AGLVT	MVIVL	TSVGL	PTDDI	TLIIA	VDWFL	DRLRT
481	TTNVL	GDSL	G	AGIVE	HLSRH	ELKNR	DVEM	G	NSVIE	ENEMK	KPYQL	IAQDN
541	KM	*										

## Protein predicted from variant coding sequence

1	MTKSNGE	EPK	MGGRMER	FQQ	GVRKR	TLLAK	KKVQN	ITKED	VKSYL	FRNAF	VLLTV	TAVIV
61	GTILGF	TLRP	YRMSY	REVKY	FSFPG	ELLMR	MLQML	VLPLI	ISSLV	TGMAA	LDSKAS	GKMG
121	MRAVV	YYMTT	TIIAV	VIGII	IVIII	IHPGK	G	TKENM	HREGK	IVRVTA	ADAF	LDLIRN
181	NLVEA	SFKQF	KTNYE	KRSFK	VPIQA	NETLV	GAVINN	VSEA	METL	TRITEE	LVPVP	GSVNG
241	VNALG	LVVFS	MCFGF	VIGNM	KEQGQ	ALREF	FDSLNE	AIMR	LVAVI	MWYAP	VGILF	LIAGK
301	IVEMED	MGVI	GGQLA	MYTVT	VIVGL	LIHAV	IVLPL	LYFLV	TRKNP	PWVFI	G	LLQAL
361	GTSSSS	SATLP	ITFKC	LEENN	GVDKR	VTRFV	LPVGAT	INMD	GTALY	EALAA	IFIAQ	VNNFE
421	LNFGQ	IITIS	ITATA	AASIG	AGIPQ	AGLVT	MVIVL	TSVGL	PTDDI	TLIIA	VDWFL	DRLRT
481	TTNVL	GDSL	G	AGIVE	HLSRH	ELKNR	DVEM	G	NSVIE	ENEMK	KPYQL	IAQDN
541	KM	*										

## Effects on Restriction sites

### Raw variant Created Deleted

1	AluI
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## Experimental services

Genomic description: 1032T>A

# SLC1A3 variant T318A

## Overview of the raw variants

Raw variant 1: substitution at 1428

GATTGGGGGGCAGCTTGCCATGTAC A CCGTGACTGTCATTGTTGGCTTACT  
GATTGGGGGGCAGCTTGCCATGTAC G CCGTGACTGTCATTGTTGGCTTACT

[NM\\_004172.4:n.1428A>G](#)

## Affected transcripts - 1

[NM\\_004172.4\(SLC1A3\\_v001\):c.952A>G](#)

## Affected proteins - 1

NM\_004172.4(SLC1A3\_i001):p.(Thr318Ala)

## Reference protein

1	MTKSNGE	EPK	MGGRMER	FQQ	GVRKR	TLLAK	KKVQN	ITKED	VKSYL	FRNAF	VLLTV	TAVIV
61	GTILGF	TLRP	YRMSY	REVKY	FSFPG	ELLMR	MLQML	VLPLI	ISSLV	TGMAA	LDSKAS	GKMG
121	MRAVV	YYMTT	TIIAV	VIGII	IVIII	HPGKG	TKENM	HREGK	IVRV	TAADAF	LDLIR	NMFPP
181	NLVEA	CFKQF	KTNYE	KRSFK	VPIQA	NETLV	GAVINN	VSEA	METL	TRITEE	LVPVP	GSVNG
241	VNALG	LVVFS	MCFGF	VIGNM	KEQGQ	ALREF	FDSL	NEAIMR	LVAV	IMWYAP	VGILF	LIAGK
301	IVEMED	MGVI	GGQLA	MYTVT	VIVGL	LIHAV	IVLPL	LYFLV	TRKN	PWVFIG	GLLQA	LITAL
361	GTSSSS	SATLP	ITFKC	LEENN	GVDKR	VTRFV	LPVG	ATINMD	GTAL	YEALAA	IFIAQ	VNNFE
421	LNFGQ	IITIS	ITATA	AASIG	AGIPQ	AGLVT	MVIV	LTSVGL	PTDD	ITLIIA	VDWFL	DRLRT
481	TTNVL	GDSL	G	AGIVE	HLSRH	ELKNR	DVEMG	NSVIE	ENEMK	KPYQL	IAQDN	ETEKPIDSET
541	KM*											

## Protein predicted from variant coding sequence

1	MTKSNGE	EPK	MGGRMER	FQQ	GVRKR	TLLAK	KKVQN	ITKED	VKSYL	FRNAF	VLLTV	TAVIV
61	GTILGF	TLRP	YRMSY	REVKY	FSFPG	ELLMR	MLQML	VLPLI	ISSLV	TGMAA	LDSKAS	GKMG
121	MRAVV	YYMTT	TIIAV	VIGII	IVIII	HPGKG	TKENM	HREGK	IVRV	TAADAF	LDLIR	NMFPP
181	NLVEA	CFKQF	KTNYE	KRSFK	VPIQA	NETLV	GAVINN	VSEA	METL	TRITEE	LVPVP	GSVNG
241	VNALG	LVVFS	MCFGF	VIGNM	KEQGQ	ALREF	FDSL	NEAIMR	LVAV	IMWYAP	VGILF	LIAGK
301	IVEMED	MGVI	GGQLA	MYAVT	VIVGL	LIHAV	IVLPL	LYFLV	TRKN	PWVFIG	GLLQA	LITAL
361	GTSSSS	SATLP	ITFKC	LEENN	GVDKR	VTRFV	LPVG	ATINMD	GTAL	YEALAA	IFIAQ	VNNFE
421	LNFGQ	IITIS	ITATA	AASIG	AGIPQ	AGLVT	MVIV	LTSVGL	PTDD	ITLIIA	VDWFL	DRLRT
481	TTNVL	GDSL	G	AGIVE	HLSRH	ELKNR	DVEMG	NSVIE	ENEMK	KPYQL	IAQDN	ETEKPIDSET
541	KM*											

## Effects on Restriction sites

Raw variant	Created	Deleted
1	BceAI, MwoI	BsrGI, Hpy166II

## Experimental services

Genomic description: 1428A>G

# SLC1A3 variant A329T

## Overview of the raw variants

Raw variant 1: substitution at 1461

TGTCATTGTTGGCTTACTCATTAC G CAGTCATCGTCTTGCCACTCCTCTA  
TGTCATTGTTGGCTTACTCATTAC A CAGTCATCGTCTTGCCACTCCTCTA

[NM\\_004172.4:n.1461G>A](#)

## Affected transcripts - 1

[NM\\_004172.4\(SLC1A3\\_v001\):c.985G>A](#)

## Affected proteins - 1

NM\_004172.4(SLC1A3\_i001):p.(Ala329Thr)

## Reference protein

1	MTKSNGE	EPK	MGGRMER	FQQ	GV	RKRTLLAK	KKVQNIT	KED	VKS	YLFRNAF	VLLTVTAVIV
61	GTILGF	TLRP	YRMSYRE	VKY	FS	FPGELLMR	MLQMLVL	PLI	ISS	LVTGMAA	LDSKASGKMG
121	MRAVVY	YMTT	TIIAVVIG	II	IV	IIHHPGKG	TKENMH	REGK	IV	RVTAADAF	LDLIRNMFPP
181	NLVEAC	FKQF	KTNYEK	RSFK	VPI	QANETLV	GAVINN	VSEA	MET	LTRITEE	LVPVPGSVNG
241	VNALGL	VVFS	MC	FGFVIGNM	KE	QGQALREF	FDSL	NEAIMR	LV	AVIMWYAP	VGILFLIAGK
301	IVEMED	MGVI	GGQLAM	YTVT	VIV	GLLIHAV	IVL	PLLYFLV	TR	KNPWVFIG	GLLQALITAL
361	GTSSSS	SATLP	ITFKC	LEENN	GVD	KRVTRFV	LP	VGATINMD	GT	ALYEALAA	IFIAQVNNFE
421	LNFGQI	ITIS	ITATAA	SIGA	AGIP	QAGLVT	MV	IVLTSVGL	PT	DDITLIIA	VDWFLDRLRT
481	TTNVLG	DSL	LG	AGIVEH	LSRH	ELK	NRDVEMG	NS	VIEENEMK	KPYQLIAQDN	ETEKPIDSET
541	KM	*									

## Protein predicted from variant coding sequence

1	MTKSNGE	EPK	MGGRMER	FQQ	GV	RKRTLLAK	KKVQNIT	KED	VKS	YLFRNAF	VLLTVTAVIV
61	GTILGF	TLRP	YRMSYRE	VKY	FS	FPGELLMR	MLQMLVL	PLI	ISS	LVTGMAA	LDSKASGKMG
121	MRAVVY	YMTT	TIIAVVIG	II	IV	IIHHPGKG	TKENMH	REGK	IV	RVTAADAF	LDLIRNMFPP
181	NLVEAC	FKQF	KTNYEK	RSFK	VPI	QANETLV	GAVINN	VSEA	MET	LTRITEE	LVPVPGSVNG
241	VNALGL	VVFS	MC	FGFVIGNM	KE	QGQALREF	FDSL	NEAIMR	LV	AVIMWYAP	VGILFLIAGK
301	IVEMED	MGVI	GGQLAM	YTVT	VIV	GLLIHAV	IVL	PLLYFLV	TR	KNPWVFIG	GLLQALITAL
361	GTSSSS	SATLP	ITFKC	LEENN	GVD	KRVTRFV	LP	VGATINMD	GT	ALYEALAA	IFIAQVNNFE
421	LNFGQI	ITIS	ITATAA	SIGA	AGIP	QAGLVT	MV	IVLTSVGL	PT	DDITLIIA	VDWFLDRLRT
481	TTNVLG	DSL	LG	AGIVEH	LSRH	ELK	NRDVEMG	NS	VIEENEMK	KPYQLIAQDN	ETEKPIDSET
541	KM	*									

## Effects on Restriction sites

Raw variant	Created	Deleted
1	HpyCH4III	

## Experimental services

Genomic description: 1461G>A

# SLC1A3 variant V393I

## Overview of the raw variants

Raw variant 1: substitution at 1653

GCGCGTCACCAGATTCGTGCTCCCC G TAGGAGCCACCATTAAACATGGATGG  
GCGCGTCACCAGATTCGTGCTCCCC A TAGGAGCCACCATTAAACATGGATGG

[NM\\_004172.4:n.1653G>A](#)

## Affected transcripts - 1

[NM\\_004172.4\(SLC1A3\\_v001\):c.1177G>A](#)

## Affected proteins - 1

NM\_004172.4(SLC1A3\_i001):p.(Val393Ile)

## Reference protein

1	MTKSNGEETPK	MGGRMERFQQ	GVKRTLLAK	KKVQNITKED	VKSYLFRNAF	VLLTVTAVIV
61	GTILGFTLRP	YRMSYREVKY	FSFPGELLMR	MLQMLVLPLI	ISSLVTGMAA	LDSKASGKMG
121	MRAVVYYMTT	TIIAVVIGII	IVIIIHGPGK	TKENMHREGK	IVRVTAADAF	LDLIRNMFPP
181	NLVEACFKQF	KTNYEKRSFK	VPIQANETLV	GAVINNVSEA	METLTRITEE	LVPVPGSVNG
241	VNALGLVVFS	MCFGFVIGNM	KEQGQALREF	FDSLNEAIMR	LVAVIMWYAP	VGILFLIAGK
301	IVEMEDMGVI	GGQLAMTYVT	VIVGLLIHAV	IVLPLLYFLV	TRKNPWVFIG	GLLQALITAL
361	GTSSSSATLP	ITFKCLEENN	GVDKRVTRFV	LPVGGATINMD	GTALYEALAA	IFIAQVNNFE
421	LNFGQIITIS	ITATAASIGA	AGIPQAGLVT	MVIVLTSVGL	PTDDITLIIA	VDWFLDRLRT
481	TTNVLGDSL	AGIVEHLRSH	ELKNRDVEMG	NSVIEENEMK	KPYQLIAQDN	ETEKPIDSET
541	KM*					

## Protein predicted from variant coding sequence

1	MTKSNGEETPK	MGGRMERFQQ	GVKRTLLAK	KKVQNITKED	VKSYLFRNAF	VLLTVTAVIV
61	GTILGFTLRP	YRMSYREVKY	FSFPGELLMR	MLQMLVLPLI	ISSLVTGMAA	LDSKASGKMG
121	MRAVVYYMTT	TIIAVVIGII	IVIIIHGPGK	TKENMHREGK	IVRVTAADAF	LDLIRNMFPP
181	NLVEACFKQF	KTNYEKRSFK	VPIQANETLV	GAVINNVSEA	METLTRITEE	LVPVPGSVNG
241	VNALGLVVFS	MCFGFVIGNM	KEQGQALREF	FDSLNEAIMR	LVAVIMWYAP	VGILFLIAGK
301	IVEMEDMGVI	GGQLAMTYVT	VIVGLLIHAV	IVLPLLYFLV	TRKNPWVFIG	GLLQALITAL
361	GTSSSSATLP	ITFKCLEENN	GVDKRVTRFV	LPVGGATINMD	GTALYEALAA	IFIAQVNNFE
421	LNFGQIITIS	ITATAASIGA	AGIPQAGLVT	MVIVLTSVGL	PTDDITLIIA	VDWFLDRLRT
481	TTNVLGDSL	AGIVEHLRSH	ELKNRDVEMG	NSVIEENEMK	KPYQLIAQDN	ETEKPIDSET
541	KM*					

## Effects on Restriction sites

### Raw variant Created Deleted

1

## Experimental services

Genomic description: 1653G>A

# SLC1A3 variant R454Q alias R499Q

## Overview of the raw variants

Raw variant 1: substitution at 1467

GCTGGGATTGTGGAGCACTTGTAC G ACATGAACTGAAGAACAGAGATGTT  
GCTGGGATTGTGGAGCACTTGTAC A ACATGAACTGAAGAACAGAGATGTT

## Affected transcripts - 1

[XM\\_024446182.1\(SLC1A3\\_v001\):c.1361G>A](#)

## Affected proteins - 1

XM\_024446182.1 (SLC1A3\_i001):p.(Arg454Gln)

## Reference protein

1	MTKSNGEETPK	MGGRMERFQQ	GVRKRTLLAK	KKVQNITKED	VKSYLFRNAF	VLLTDTVAVIV
61	GTILGFTLRP	YRMSYREVKY	FSFPGELLMR	MLQMLVLP LI	ISSLV TGMAA	LD SKASGKMG
121	MRAVVYYMTT	TIIAVVIGII	IVII IH PGKG	TKENMHREGK	IVRVTAADAF	LDLIRNMFPP
181	NLVEACFKQF	KTNYEKRSFK	VPIQANETLV	GAVINNVSEA	METLTRITEE	LVPVPGSVNG
241	VNALGLVVFS	MCFGFVIGNM	KEQGQALREF	FDSLNEAIMR	LVAVIMWYAP	VGILFLIAGK
301	IVEMEDMGVI	GGQLAMYTVT	VIVGLLIHAV	IVLPLLYFLV	TRKNPWVFIG	GLLQALITAL
361	GTSSSSATLP	ITFKCLEENN	GVDKRVTRFV	LPVGATINMD	GTALYEALAA	IFIAQVNNFE
421	LNFGQIITIR	DRLRTTTNVL	GD SLGAGIVE	HLS <b>R</b> HELKNR	DVEMGNSVIE	ENEMKKPYQL
481	IAQDNETEKP	IDSETKM*				

## Protein predicted from variant coding sequence

1	MTKSNGEETPK	MGGRMERFQQ	GVRKRTLLAK	KKVQNITKED	VKSYLFRNAF	VLLTDTVAVIV
61	GTILGFTLRP	YRMSYREVKY	FSFPGELLMR	MLQMLVLP LI	ISSLV TGMAA	LD SKASGKMG
121	MRAVVYYMTT	TIIAVVIGII	IVII IH PGKG	TKENMHREGK	IVRVTAADAF	LDLIRNMFPP
181	NLVEACFKQF	KTNYEKRSFK	VPIQANETLV	GAVINNVSEA	METLTRITEE	LVPVPGSVNG
241	VNALGLVVFS	MCFGFVIGNM	KEQGQALREF	FDSLNEAIMR	LVAVIMWYAP	VGILFLIAGK
301	IVEMEDMGVI	GGQLAMYTVT	VIVGLLIHAV	IVLPLLYFLV	TRKNPWVFIG	GLLQALITAL
361	GTSSSSATLP	ITFKCLEENN	GVDKRVTRFV	LPVGATINMD	GTALYEALAA	IFIAQVNNFE
421	LNFGQIITIR	DRLRTTTNVL	GD SLGAGIVE	HLS <b>Q</b> HELKNR	DVEMGNSVIE	ENEMKKPYQL
481	IAQDNETEKP	IDSETKM*				

## Effects on Restriction sites

Raw variant	Created	Deleted
1		Hpy188III

## Experimental services

Genomic description: 1467G>A