

Figure S2. Multiple alignment of the equine, human and murine KIT proteins. The sequences were derived from Genbank accessions AM420315 (horse), NP_000213 (human), and NP_066922 (mouse). Individual protein domains are indicated by colored shading. The position of selected known human and murine KIT mutations is shown. The four newly described equine KIT mutations are highlighted in grey.

HORSE	MRGARGAWDFLCVLLLLLFRVQTGSSQPSVSPGELSPSPSIHPAKSELIVSVGDEIRLLCAD	60
HUMAN	MRGARGAWDFLCVLLLLLFRVQTGSSQPSVSPGEPSPSPSIHPKGSDDLIVRVGDEIRLLCTD	60
MOUSE	MRGARGAWDLLCVLLLLLRGQTATSQPSASPGEPSPSPSIHPAQSELIVEAGDTLSLTCID	60
	<1-22 signal peptide >	
	↓ K86Rfs17X (human piebaldism)	
HORSE	PGFVKWTFET-WGQLSENTHKEWVTEKAEATNTGYSYCTNEGGLSSSIYVFRDPAKLF	119
HUMAN	PGFVKWTFEI-LDETENKQNEWITEKAEATNTGKYCTCNKHGLSNSIYVFRDPAKLF	119
MOUSE	PDFVVRTFKTYFNEMVYENKNEWIQEKAERTRGTYSNSNGLTSSSIYVFRDPAKLF	120
HORSE	FDPSLYGKEGSDTLVRCPLTDPEVTNYSLMACEGKSLPKDLTFVADPKAGITIRNVKREY	179
HUMAN	VDRSLYKGEDNDTLVRCPLTDPEVTNYSKGCQKPLPKDLRFIPDPKAGIMIKSVKRAY	179
MOUSE	VGLPLFGKEDSDALVRCPLTDQPVSNYSLIECDGKSLPTDLTFVNPVKAGITIKNVKRAY	180
	↓ K236X (dominant white, Arabian Horses)	
HORSE	HRLCLRCSADKDGKSVLSNFKFTLVRAAIRAVPVVSVKASYLLREGEEFVSTCLIKDVS	239
HUMAN	HRLCLHCSVDQEGKSVLSEKFIKVRPAFKAVPVVSVKASYLLREGEEFVSTCLIKDVS	239
MOUSE	HRLCVRCAQRDGTWLSHDKFTLVKREAIKAIPIVVPVETSHLLKKGDTFTVCTIKDVS	240
HORSE	SSVDSMWIRENSR--TKEQVKSSSSHQGDFNFVRQERLTISPARVNDSGVFMCIYANNTFG	297
HUMAN	SSVYSTWKRENSQ--TKLQEKYNSWHHGDFNYERQATLTISSARVNDSGVFMCIYANNTFG	297
MOUSE	TSVNSMWLKMNPQPHIAQVKNHSHRQDFNYERQETLTISSARVDDSGVFMCIYANNTFG	300
HORSE	SANVTTLLEVVDKGFINVPMMNTTVFVNDGENVDLIVEYESYKPEHQQWIYMNRTSTD	357
HUMAN	SANVTTLLEVVDKGFINIPMINTTVFVNDGENVDLIVEYEAQPKPEHQQWIYMNRTSTD	357
MOUSE	SANVTTLKVVKEGFINISFVKNTTVFVTDGENVDLIVEYEAQPKPEHQQWIYMNRTSAN	360
	↓ V410F (mouse, Kit ^{W-2Ba0} , semidominant white)	
HORSE	KWEDYPKSENESENIRYVSELHLTRLKGTGEGTYTFLVNSDSDVSSVTFNVVNTKPEILT	417
HUMAN	KWEDYPKSENESENIRYVSELHLTRLKGTGEGTYTFLVNSDVAIAFNVVNTKPEILT	417
MOUSE	KGKDYVKSNDKSNIRYVNLRLTRLKGTGEGTYTFLVNSDASASVTFNVVNTKPEILT	420
HORSE	RDRLMNGMLQCVAAGFPEPTIDWYFCPGTEQRCSIPVGPVDVKIQNSSVSPFGKLVVQSS	477
HUMAN	YDRLVNGMLQCVAAGFPEPTIDWYFCPGTEQRCSASVLPVDVQTLNSSGPPFGKLVVQSS	477
MOUSE	YDRLINGMLQCVAAGFPEPTIDWYFCTGAEQRCTTPVSPVDVQVNVSPFGKLVVQSS	480
HORSE	IDYSAFKHNGTVECRAYNDVGKSSAFFNFPAFK---EQIHPHTLFTPLLIGFVVAAGMMC	533
HUMAN	IDSSAFKHNGTVECKAYNDVGKTSAYFNFAFKGNNKEQIHPHTLFTPLLIGFVIVAGMMC	537
MOUSE	IDSSVFRHNGTVECKASNDVGKSSAFFNFPAFK---EQIQAHTLFTPLLIGFVVAAGMAM	536
	<521-543 transmembrane domain	
	↓ G595R (mouse Kit ^{W-71c} , semidominant white spotting)	
	↓ E582K (mouse Kit ^{W-37J} , semidominant white spotting)	
	↓ Y569F (mouse Kit ^{tm2Ber} , dominant extreme white spotting)	
	↓ Y567F (mouse Kit ^{tm1Ber} , dominant extreme white spotting)	
	↓ E584K (human piebaldism)	
	↓ F584L & F584C (4x human piebaldism)	
HORSE	VIVMVLTYKYLQKPMYEVQWVVEEINGNNYVYIDPTQLPYDHHKWEFPRNRLSFGKTLGA	593
HUMAN	IIVMILTYKYLQKPMYEVQWVVEEINGNNYVYIDPTQLPYDHHKWEFPRNRLSFGKTLGA	597
MOUSE	IIVMVLTYKYLQKPMYEVQWVVEEINGNNYVYIDPTQLPYDHHKWEFPRNRLSFGKTLGA	596
	transmembrane domain >> 544-581 juxtamembrane domain >> 582-937 protein kinase domain	
	↓ G597A (mouse Kit ^{W-e1} , dominant white)	
	↓ K641E (mouse Kit ^{tm1Bpr} , recessive white)	
	↓ A621S	
	↓ K642Sfs6X (2x human piebaldism)	
	↓ A602V (dominant white, Camarillo White Horses)	
HORSE	GAFGKVVVEATAYGLIKSDAAMTVAVKMLKPSAHLTEREALMSELKVLSYLGNHMNIENLL	653
HUMAN	GAFGKVVVEATAYGLIKSDAAMTVAVKMLKPSAHLTEREALMSELKVLSYLGNHMNIENLL	657
MOUSE	GAFGKVVVEATAYGLIKSDAAMTVAVKMLKPSAHLTEREALMSELKVLSYLGNHMNIENLL	656
	↓ T660M (mouse Kit ^{W-v} , semidominant white spotting)	
	↓ G664R (human piebaldism)	
	↓ G654R (dominant white, Thoroughbreds)	
HORSE	GACTVGGPTLVITEYCCYDGLLNFLRRKRHSFICSKQEDHAEAAALYKNLLHKSKESSCNS	713
HUMAN	GACTIGGPTLVITEYCCYDGLLNFLRRKRDSFICSKQEDHAEAAALYKNLLHKSKESSCSDS	717
MOUSE	GACTVGGPTLVITEYCCYDGLLNFLRRKRDSFIFSKQEEQAEAAALYKNLLHSTPEPSC-DS	715
	protein kinase domain >> 685-761 kinase insert domain	
	↓ Y719F (mouse Kit ^{tm1.1Bsm} , infertility, no pigmentation phenotype)	
	↓ Y717X (dominant white, Franches-Montagnes Horses)	
HORSE	TNEYMDMKPGVSYVVPVTKDKRRAARIGSYIERDVAPSIMEDDELALDLEDLLSFSYQVA	773
HUMAN	TNEYMDMKPGVSYVVPVTKDKRRSARIGSYIERDVPAIMEDDELALDLEDLLSFSYQVA	777
MOUSE	SNEYMDMKPGVSYVVPVTKDKRRSARIDSYIERDVPAIMEDDELALDLEDLLSFSYQVA	775
	685-761 kinase insert domain >> 582-937 protein kinase domain	
	↓ A835V (mouse Kit ^{W-n} , semidominant white spotting)	
	↓ R816W (mouse Kit ^{W-f} , semidominant white spotting)	
	↓ D790G (mouse Kit ^{W-42J} , dominant white)	
	↓ V831M (mouse Kit ^{W-41J} , semidominant white spotting)	
	↓ R796G (human piebaldism with sensineural deafness)	
HORSE	KGMAFLASKNCIHRDLAARNILLTHGRITKICDFGLARDIKNDSNYVVKGNARLPVKWMA	833
HUMAN	KGMAFLASKNCIHRDLAARNILLTHGRITKICDFGLARDIKNDSNYVVKGNARLPVKWMA	837
MOUSE	KAMAFLASKNCIHRDLAARNILLTHGRITKICDFGLARDIRNDSNYVVKGNARLPVKWMA	835
	↓ F856S (mouse Kit ^{Wads} , semidominant white spotting)	
	↓ T847P (human piebaldism)	
HORSE	PESIFNCVYTFESDVSYSYGFIFLWELFSLGSSPYGMPVDSKFKYKMIKEGFRMLSPEHAPA	893
HUMAN	PESIFNCVYTFESDVSYSYGFIFLWELFSLGSSPYGMPVDSKFKYKMIKEGFRMLSPEHAPA	897
MOUSE	PESIFNCVYTFESDVSYSYGFIFLWELFSLGSSPYGMPVDSKFKYKMIKEGFRMVSPEHAPA	895
HORSE	EMYDIMKTCWDADPLKRPTFKQIVQLIEKQISDSTNHIYSNLANCSPRQENS-AVDHSVR	952
HUMAN	EMYDIMKTCWDADPLKRPTFKQIVQLIEKQISESTNHIYSNLANCSPNRQKP-VVDHSVR	956
MOUSE	EMYDVMKTCWDADPLKRPTFKQIVQLIEKQISDSTKHIYSNLANCPNPNPENVVVDHSVR	955
HORSE	INSVGSASSTQPLLHVEDV	972
HUMAN	INSVGSSTASSQPLLHDDV	976
MOUSE	VNSVGSASSTQPLLHVEDA	975