

Changes in a pseudogene mistakenly mapped as SNPs in the functional gene.

a, AGP1 aligned with AGP2. Boxed amino acids are the substitutions that were nsSNP entries for *AGP1* in dbSNP. These substitutions were derived from GenBank AC# AA700876, T27840, H67119, T73068, T70907, H82213, and AA550779, all of whose nucleotide sequences matched *AGP2*. Amino acids in lowercase bold italic are the substitutions predicted to be deleterious. *AGP2* was not included in the alignment used for prediction on *AGP1* because it was > 90% identical to *AGP1* and thus discarded (see Methods). b, FKSG25 aligned with the translated pseudogene and sequences from which nsSNPs were derived. The pseudogene has a deletion that introduces an early stop codon; the translated pseudogene can be aligned to FKSG25 after the deletion with a frameshift of two bases. AC067884, AL365277, AC024511 are the contigs from which nsSNPs (boxed) were interpreted and deposited into dbSNP and all contain the same deletion as the pseudogene. Substitutions predicted to be deleterious are in lowercase bold italic.

a

AGP1	MALSWVLTVL	SLLPPLLEAQI	PLCANLVPVP	ITNATLDQIT	GKWFYIASAF	RNEEYNKSVQ	EIQATFFFYFT
AGP2	.....	.....	.....	.....R..	.....	.....	.....
AGP1	PNKTEDTIFL	REYQTRQDQC	IYNTTYLNVQ	RENGTISRYV	GGQEHFAHLL	ILRDTKTYM	AFDVNDEKNW
AGP2	.....	.....N..	F..SS.....	.....V...E	..R..V....	<b>f</b> .....L..	<b>f</b> SYLD.....
AGP1	GLSVYADKPE	TTKEQLGEFY	EALDCLRIPK	SDVVYTDWKK	DKCEPLEKQH	EKERKQEEGE	S
AGP2	...F.....	.....	.....C..R	.....m.....	.....	.....	..

b

FKSG25	MAALRLLASV	LGRGVPAGGS	GLALSQGCAR	CFATSPRLRA	KFYADPVEMV	KDISDGATVM	IGGFGLCGIP
pseudogene	.....	.....	.....	.....P..	.....	.....	.....
AC067884	.....	.....	.....	.....	.....	.....	.....
AL365277	.....	.....	.....	.....P..	.....	.....	.....
AC024511	.....	.....	.....	.....P..	.....	.....	.....
FKSG25	ENLIAALLRT	RVKDLQVVSS	NVGVEDFGLG	LLLAARQVRR	IVCSYVGENT	LCESQYLAG	LELELTPQGT
pseudogene	.....	.....	.....	.....	.....	.....	.....
AC067884	.....	.....	.....	.....	.....	.....	.....
AL365277	.....	.....	.....	.....	.....	.....	.....
AC024511	.....	.....	.....	.....	.....	.....	.....
FKSG25	LAERIRAGGA	GVPAFYTPTG	YGTLVQEGGA	PIRYTPDGH	ALMSQPREVR	EFNGDHFLLE	RAIRADFAV
pseudogene	.....	.....	.....	.....	.....	.....	.....
AC067884	.....	.....	.....	.....	.....	.....	.....
AL365277	.....	.....	.....	.....	.....	.....	.....
AC024511	.....	.....	.....	.....	.....	.....	.....
***** ***							
FKSG25	KGWKADRAGN	VVFRRSARNF	NVPMCKAADV	TAVEVEEIVE	VGAFPPEDIH	VPNIYVDRVI	KGQKYEKRIE
pseudogene	.....	.....	.....	.....	.....	.....G...	.....
AC067884	.....	.....	.....	.....	.....	.....G...	.....
AL365277	.....	.....	.....	.....	.....	.....G...	.....
AC024511	.....	.....	.....	.....	.....	.....G...	.....
FKSG25	RLTIKKEEDG	DAGKEEDART	RIIRRAALEF	EDGMANLGI	GIPLLASNFI	SPSMTVHLHS	ENGILGLGPF
pseudogene	.....R.....	.....	.....	.....	.....	.....	.....
AC067884	.....R.....	.....	.....	.....	.....V.....	.....	.....
AL365277	.....R.....	.....	.....	.....	.....	.....	.....
AC024511	.....R.....	.....	.....	.....	.....	.....	.....
FKSG25	PTEDEVADL	INAGKQTVTV	LPGGCFFASD	DSFAMIRGGH	IQLTMLGAMQ	VSKYGDLANW	MIDCKKVKGM
pseudogene	.....	.....	.....	.....	.....	.....	.....s.....
AC067884	.....	.....	.....	.....	.....	.....	.....
AL365277	.....	.....	.....	.....	.....	.....	.....
AC024511	.....	.....	.....	.....	.....	.....	.....
FKSG25	GGAMDLVSSQ	KTRVVVTMQH	CTKDNTPKIM	EKCTMPLTGK	RCVDRIITEK	AVFDVHRKKH	LTLRELWEGL
pseudogene	.....	.....	.....	.....	.....	.....G.....	.....
AC067884	.....	.....	.....	.....	.....	.....	.....
AL365277	.....	.....	.....	.....	.....	.....R.....	.....
AC024511	.....	.....	.....	.....	.....	.....G.....	.....
FKSG25	TVDPIKKSTG	CAFAVSPNLR	PMQQVAP				
pseudogene	.....n.....	.....	.....				
AC067884	.....	.....	.....				
AL365277	.....	.....	.....V.....				
AC024511	.....n.....	.....	.....				