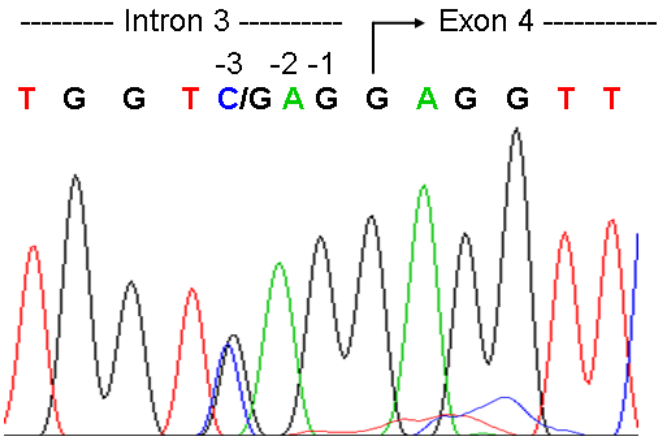
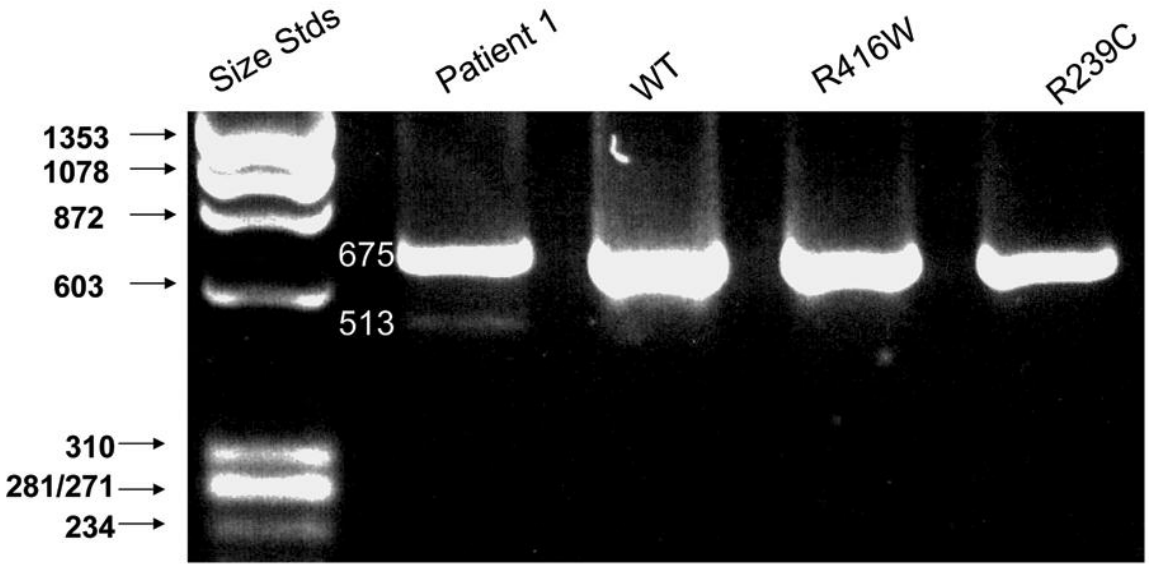
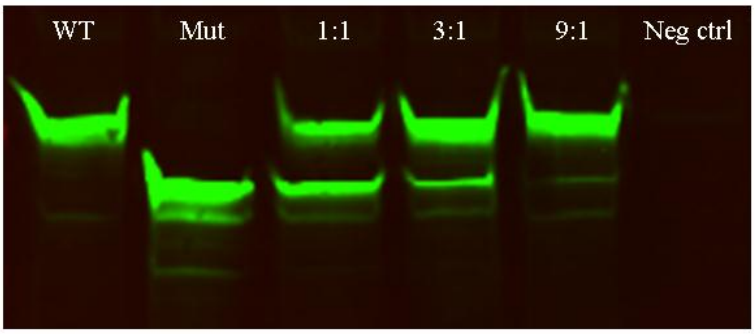


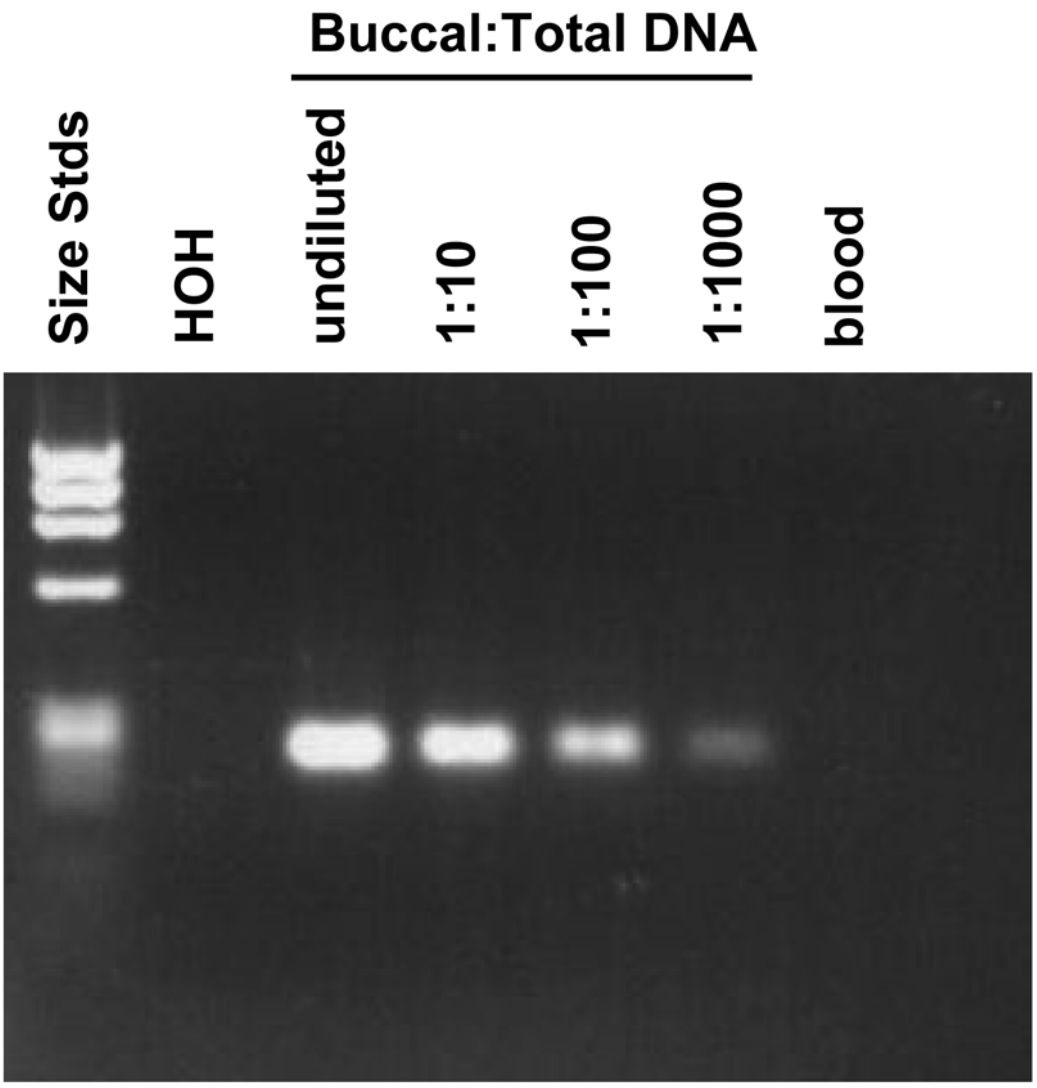
Supp. Figure S1. Detection of the heterozygous c.(619-3C>G) mutation in Patient 1. Sequencing data are shown for the area of the intron 3/exon 4 boundary.



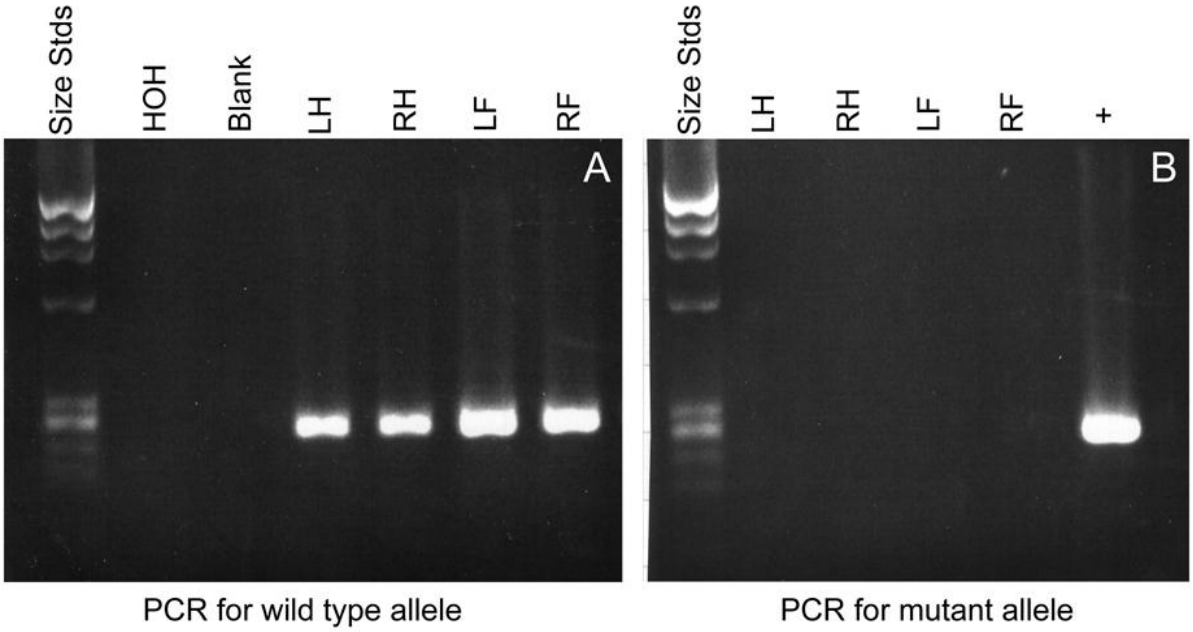
Supp. Figure S2. Detection of the *GFAP* splice variant. RT-PCR was performed as described in Materials and Methods using primers in exon 1 and exon 6 and RNA isolated from Patient 1, a wild type (WT) control, or two AxD controls known to have either an R416W or R239C mutation (Patients 10 and 5, respectively, of Brenner et al., 2001). Agarose gel electrophoresis of the amplified products shows that all RNAs yielded a fragment of the size expected from the wild type transcript (675 bp), but only the RNA from Patient 1 yielded a fragment of the size expected if exon 4 is skipped (513 bp).



Supp. Figure S3. Comparison of GFAP expression level to transfected plasmid level. SW13vim$^-$ cells were transfected with pcDNA3.1-hGF(WT), which expresses wild type GFAP (WT); pcDNA3.1-hGF(Δex4), which expresses GFAP lacking exon 4 (Mut); or the indicated ratios of WT:Mut plasmid. Neg ctrl was transfected with pUC19. In each case total plasmid transfected was 2 ug. Extracts from the transfected cells were immunoblotted for GFAP, and GFAP bands visualized and quantified using an Odyssey system. The calculated protein ratios for the plasmid transfection ratios were 1:1 = 0.94; 3:1 = 3.6; 9:1 = 11.2.



Supp. Figure S4. PCR assay for the I/D/F allele in Patient 2 blood DNA. Blood DNA from Patient 2 was subjected to PCR with a primer pair specific for the mutant I/D/F allele as described in Materials and Methods. To determine the sensitivity of the assay, various amounts of buccal DNA from Patient 2 were mixed with blood DNA from a wild type control as indicated. The same total amount of DNA was used as template for each reaction. The mutant allele was not detected in Patient 2 blood DNA (blood), but would have been detected if present at as little as 1 part in 1,000. HOH, no template added.



Supp. Figure S5. Allele-specific PCR analysis of nail DNA from Patient 2. PCR was performed for the wild type allele (A) or mutant allele (B) of the I/D/F mutation as described in Materials and Methods. Only the wild type allele was detected. HOH, no template; Blank, parallel DNA preparation procedure without nails; LH, left hand; RH, right hand; LF, left foot; RF, right foot; +, positive control (Patient 2 buccal DNA).