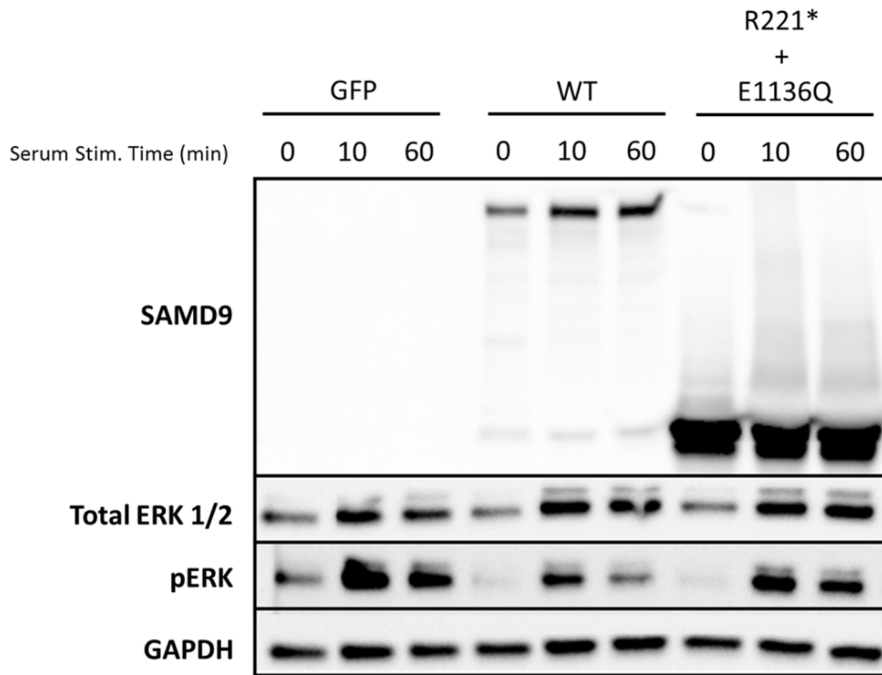


Supplemental Figure 4. Biologic characterization of the sibling specific *SAMD9* mutations in *cis* with p.E1136Q. A.) Western blot showing the p.R221* mutation rescues the restriction of ERK phosphorylation induction caused by the p.E1136Q. B.) Western blot showing the p.F583I mutation may slightly abrogate p.E1136Q restriction of ERK phosphorylation.

A



B

