Protein folding results for VKGL missense variants in the PTCH1 gene Based on VKGL public consensus release April 2023, FoldX 5.0, and AlphaFold2 human proteome v4 At a $\Delta\Delta G$ threshold of 4.75 the PPV is 100%, the sensitivity is 57%, the NPV is 92% and the specificity is 100% GA484R (DQQ) and variant protein energy change SA1132F **VKGL** classification reference LB/B Difference in Gibbs free between GRCh37 refere LP/P VUS GA91C GA509D LA175P SA1132P GA37R PA725S GA866E SA1425L PA568L AA167D SA827GRA893H RA1442Q VA1034L DA513Y TA1052M 500 1,000 0 1,500 Amino acid position within the protein alpha chain of PTCH1