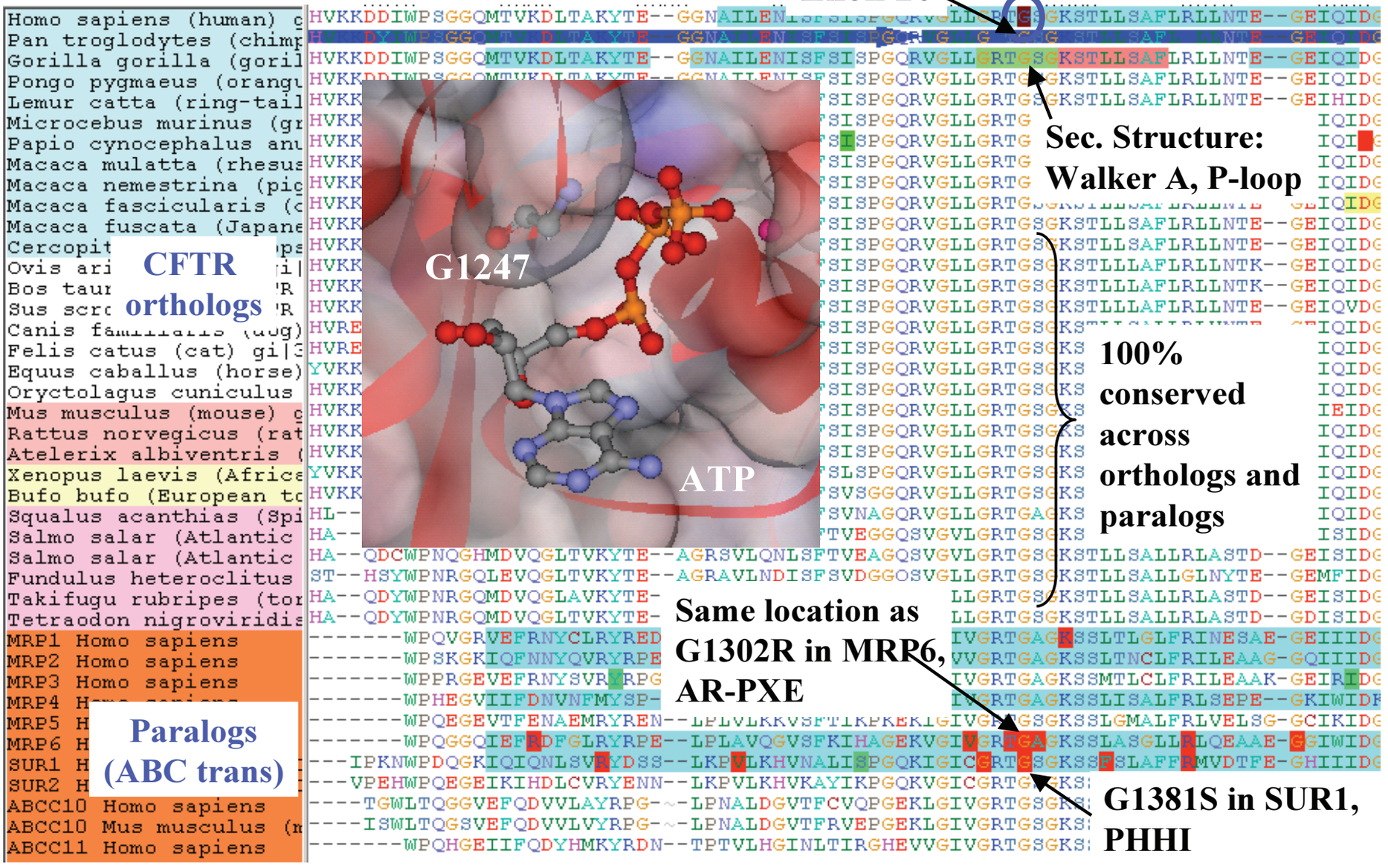
Annotated hand-curated alignments



Arlene Buller, PhD

The Mutation Inference Scoring Tool being developed for the Molecular Genetics Laboratory at Quest Diagnostics in San Juan Capistrano uses public databases to align the nucleotide sequence of a patient's CFTR gene with an uncertain variant with homologous or related genes. This alignment shows the degree of evolutionary conservation of the patient's mutation and helps to arrive at a quantitative estimate of how likely it is that the mutation is deleterious. In this case, the mutation in question, G1247R, corresponds to the site of known mutations in two other human genes that code for transport proteins. Structural information shows that the mutation is found in the P loop of the CFTR protein.