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Title: "Synonymous mutations are not always silent. The consequences of synonymous mutations in the CFTR Gene"

Synopsis: Synonymous mutations in the protein coding regions of genes result in alternative codon usage for amino acids, but do not change the amino acid sequence of the encoded proteins. With the growing number of GWAS studies, synonymous mutations (sSNPs) are linked to diseases. Studies analyzing the consequences of such mutations have determined multiple mechanisms by which they can alter the structure and function of the encoded proteins. We analyzed the consequences of the I507-ATC>ATT synonymous codon change in ΔF508 CFTR in detail and identified sSNPs in the CFTR gene which have consequences on protein structure, function and response to drugs. The seminar will summarize our present understanding of how synonymous mutations alter gene expression and provide experimental evidence from the analysis of the human CFTR gene. Because synonymous mutations can alter the severity of numerous human diseases or their response to treatments, understanding the molecular mechanisms by which they alter protein structure and function should be of interest to a wide audience.