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## ABERRANT AND ALTERNATIVE SPLICING OF ABCC7 (CFTR)

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Cystic fibrosis (CF) is the most common lethal genetic disease in European origin populations. The severity of the pathology depends on the mutations present on CFTR (cystic fibrosis transmembrane regulator) gene coding CFTR protein involved in the active transport of ions through the apical membrane of epithelial cells. Understanding how each mutation affects CFTR function is crucial for the clinical approach and therapeutic strategies. Therefore, mutations are classified into six established classes, according to their characteristics. Our research on CFTR mutations directly results from the activity of the molecular genetic laboratory at Poitiers University Hospital therapeutic strategies. Here we present mutations impact on alternative or aberrant splicings. For example, the polymorphism TG (11) T (5) aggravates the effects of the mutation c.1392G>T, inducing exon 9 complete deletion and also encoding a frame shift transcript (with a difference of 5 nucleotides), on aberrant splicing. Analysis of mRNA obtained from parental airway epithelial cells confirmed these *in cellulo* results (Farhat et al., 2015). In conclusion, the classification of a single mutation in one or more of the six described classes is important but not sufficient in a clinical approach, as the possible presence of a complex allele in a CF patient may alter the specific effect of this mutation. Therefore, an individual approach is required to perform a precise diagnostic and propose a specific treatment when available.