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Carriers of a single *CFTR* mutation are asymptomatic: an evolving dogma?

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CF carriers may be at higher risk of multiple *CFTR*-related diseases <https://bit.ly/3foJK7n>

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Cystic fibrosis (CF) is a genetic autosomal recessive disease due to mutations in the gene encoding for the cystic fibrosis transmembrane conductance regulator (CFTR) protein [1, 2]. Mutations in the *CFTR* gene may cause a reduction of CFTR protein function, leading to abnormal chloride and bicarbonate transport in epithelia, resulting in abnormal mucus properties and a multiorgan disease dominated by respiratory and gastro-intestinal abnormalities [3]. The level of CFTR protein function is an important determinant of disease in humans and CF patients carrying two disease-causing *CFTR* mutations usually have very low levels of functional CFTR protein. Carriers of a single *CFTR* mutation (also called CF carriers) express 50% CFTR protein function, a level that has been considered sufficient to stay healthy [3]. CF carriers represent approximately 1 in 35 Caucasian in the USA, accounting for more than 10 million individuals [4]. CF carriers are generally informed that they are at risk of transmitting the mutation to their children and/or to have a child with CF; they are also informed that having one *CFTR* gene mutation does not cause symptoms [4]. Having a single *CFTR* mutation has even been suspected to provide a selective advantage, as CF carriers may withstand secretory diarrhoea better than non-carrier individuals, leading to a possible protection against cholera [5]. This latter finding has been suggested to explain the high rate of CF carriers in Caucasian populations [1].