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Phenotypical and Genotypical Diagnosis of Cystic Fibrosis in 26 Cases from East and South Algeria

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Abstract : Cystic fibrosis (CF), the most common lethal genetic disease in the Europe population, is caused by mutations in the transmembrane conductance regulator gene (CFTR). It affects most organs including an epithelial tissue, base of hydroelectrolytic transepithelial transport, notably that aerial ways, the pancreas, the biliary ways, the intestine, sweat glands and the genital tractus. The gene whose anomalies are responsible of the cystic fibrosis codes for a protein Cl channel named CFTR (cystic fibrosis transmembrane conductance regulator) that exercises multiple functions in the cell, one of the most important in control of sodium and chlorine through epithelia. The deficient function translates itself notably by an abnormal production of viscous secretion that obstructs the execrator channels of this target organ: one observes then a dilatation, an inflammation and an atrophy of these organs. It also translates itself by an increase of the concentration in sodium and in chloride in sweat, to the basis of the sweat test. In order to do a phenotypical and genotypical diagnosis at a part of the Algerian population, our survey has been carried on 16 patients with evocative symptoms of the cystic fibrosis at that the clinical context has been confirmed by a sweat test. However, anomalies of the CFTR gene have been determined by electrophoresis in gel of polyacrylamide of the PCR products (polymerase chain reaction), after enzymatic digestion, then visualized to the ultraviolet (UV) after action of the ethidium bromide. All mutations detected at the time of our survey have already been identified at patients attained by this pathology in other populations of the world. However, the important number of found mutation with regard to the one of the studied patients testifies that the origin of this big clinical variability that characterizes the illness in the consequences of an enormous diversity of molecular defects of the CFTR gene.

Keywords : cystic fibrosis, CFTR gene, polymorphism, algerian population, sweat test, genotypical diagnosis
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