Molecular genetic analysis of 2 Chilean cystic fibrosis patients and their families

Análisis genético molecular de la fibrosis quística en dos pacientes chilenos y sus familias.

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As a contribution to establish the real incidence of cystic fibrosis (CF) and the prevalent mutations in the Chilean population a method for the detection of delta F-508 and R-553X, two of the most frequent mutations described worldwide, has been implemented. The method is based on the polymerase chain reaction (PCR) amplification of DNA followed by allele specific restriction enzymatic digestion. The application of this techniques allowed to confirm CF diagnosis in two patients and to detect asymptomatic carriers in both families. One of the patients showed normal sweat electrolyte concentration.