Genetic Characterization of Pulmonary Fibrosis in Patients Who Suffer from Hermansky Pudlak Syndrome: An Autosomal Recessive Inherited Disease

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Hermansky Pudlak Syndrome is an autosomal recessive disease: a type of albinism correlated to the mutation of an HPS gene and the pulmonary function of patients. Puerto Rico has the highest incidence and prevalence of this condition. The objective is the characterization of the genotype and phenotype of Pulmonary Fibrosis in Puerto Rican albinos. Twenty-two Puerto Rican residents with HPS between 7 to 65 years of age were selected. The variables were based on the age of the individual in the following stages: when the symptoms started, HPS diagnosis and Pulmonary Fibrosis diagnosis. The correlation of phenotype and genotype of HPS was also studied. The genotype was analyzed by extracting DNA from the blood samples with the Archive-Pure DNA Purification Prime Kit. The phenotype was defined by lung tests done to the patients. Most of the patients showed pulmonary functions associated to the HPS1 mutation after amplifying the DNA and performing the PCR, which showed a deletion of 16 base pairs in the HPS1 gene. The mutation was found in 81.81% of the sample. Between 7 and 12 years of age no anomalies were found; between 13 and 21 no statistical significant anomalies were found, but the values in the “Total Vital Capacity” test changed. Between 22 and 65 years of age, a new medical percentage was found; this offers new data to the medical literature, changing the clinical data of this disease in Puerto Rico, since 53.3% of the patients had a significant decrease in their lungs’ capacity.

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