SHORT TELOMERE SYNDROME IS A RISK FACTOR FOR THE DEVELOPMENT OF IPF

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Objective:

Short telomere syndrome composes of a group of different affected genes that cause a decrease in telomere lengths and, which in turn, causes accelerated aging syndromes. Initially, it was associated with dyskeratosis congenita, but now it is seen that many other organs that have a high cell turnover, such as lungs, can be affected. Many gene mutations can be associated with short telomere syndrome, but not all will affect the lungs. Most commonly lungs are involved if one of the following gene mutations is found: TERT, TERC, RTEL1, PARN, DKC1, TINF2, and NAF1. On high-resolution CT scan the most prevalent pattern is UIP, but other patterns such as indeterminant UIP and rarely pleuroparenchymal fibroelastosis, chronic hypersensitivity pneumonitis, and unclassifiable fibrosis can be seen.

Here we will present a case of a patient with TERT mutation. The patient initially presented at the age of forty with a fever. CT scan that was done showed unspecific changes of the lung parenchyma. The next time the patient came to our Department was seven years later when a repeated HRCT showed

UIP pattern. After all other possible diseases were excluded, the diagnosis of IPF was established, and treatment with nintedanib was started. Initially, the disease was stable, but after six months a slow, but progressive decline in FVC was noted. After two years and eight months, a decline was more obvious. HRCT scan showed progression of the lung fibrosis and respiratory insufficiency which required oxygen therapy was noted. The patient was transferred to a referral center to be included in the lung transplant program. A reevaluation of the disease was done with whole-genome sequencing that showed the patient was a heterozygote for TERT mutation. Since nintedanib was ineffective, it was switched to pirfenidone; unfortunately, after seven months of treatment a progression of the disease was noted and the patient was hospitalized for developing pneumothorax, pneumomediastinum, and subcutaneous emphysema. Although a complete recovery of those was seen, the course of the hospitalization was complicated, with pneumonia which led to the patient's death.