Chest X-rays and computed tomography (CT) revealed right-sided pneumothorax and many diffuse cystic lesions in both lung fields. The X-rays were taken at admission (A). The CT was performed after improvement from pneumothorax (B). The histogram of electrophoresis of patient’s serum revealed flattening $\alpha_1$-globulin segment (arrow).

Pneumothorax Associated with $\alpha_1$-Antitrypsin Deficiency

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In Caucasians of northern European extraction, $\alpha$-AT deficiency is a relatively common hereditary disorder, while in Japanese, it has been reported with only 16 traits (1, 2). Individuals with severe $\alpha$-AT deficiency have at least a 20-fold increase in the risk of developing lung disease (3). The present report involves a patient with pneumothorax and an undetectable level of $\alpha$-AT in serum.

A 44-year-old Japanese woman was admitted to our hospital because of recurrent right-sided pneumothorax. One year earlier, she had suffered from right-sided pneumothorax for the first time and developed it 3 more times during the ensuing 3 months. Chest X-rays and computed tomography revealed right-sided pneumothorax and many diffuse cystic lesions in both lung fields in spite of her non-smoking status. Thoracic drainage was performed and pneumothorax improved after 2 weeks (Picture 1).

Picture 1. Chest X-rays and computed tomography (CT) revealed right-sided pneumothorax and many diffuse cystic lesions in both lung fields. The X-rays were taken at admission (A). The CT was performed after improvement from pneumothorax (B). The histogram of electrophoresis of patient’s serum revealed flattening $\alpha_1$-globulin segment (arrow).

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The \( \alpha_1 \)-globulin segment was 0.1%. Other laboratory findings pertaining to her liver function were normal (Picture 1). The \( \alpha_1 \)-AT content was below the measurable level (below 10 mg/dL). She was born in Tokushima Prefecture. Her father suffered from emphysema; and her mother and son, who were otherwise normal, had \( \alpha_1 \)-AT levels that were about one-half of normal. A reverse transcriptase polymerase chain reaction revealed the deletion of [268-270bp GGG\( \rightarrow \) GG] in exon II. Thus it was concluded that she represented a case of \( \alpha_1 \)-AT deficiency of null type: her phenotype was unknown.

We believe that careful follow-up of this patient is necessary considering that a possible lung transplantation may be necessary in the future, and that more detailed criteria are needed for such patients with complications of this disorder.

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