Inherited antithrombin deficiency due to mutation SERPINC1 found after pulmonary thromboembolism

계명대학교 동산병원 내과[1]

연제혁[1], ***김태훈**[1]

We presented a case of a 46-year-old female diagnosed with pulmonary thromboembolism with a history of asthma, and recurrent cerebrovascular infarction. The patient had administrated anticoagulation for recurrent stroke with thrombotic thrombocytopenic purpura but stopped in 2016. The patient visited the emergency room for chest discomfort and right pleural. The patient was diagnosed with pulmonary thromboembolism. The patient was administered anticoagulation therapy immediately, but additional thrombotic events including stroke and occlusion of extremities vessels were developed. At the time, the patient experienced thrombocytopenia, and rivaroxaban was inevitably stopped but warfarin was started instead of rivaroxaban because we determined that the cause of thrombocytopenia was microangiopathy. The patient's coagulopathy was re-evaluated. Antiphospholipid syndrome was excluded, and it was confirmed that the antithrombin level was greatly reduced by less than 10%. After the next-generation sequencing test for mutation, inherited antithrombin deficiency was diagnosed due to a double mutation of the SERPINC1 gene (c.442T>C, and c.234C>T). This report is interesting because until now, a few cases of antithrombin deficiency was reported, but there was no report of a long-term clinical course of double mutation of SERPINC1. Currently, anticoagulation treatment with warfarin was maintained while the international normalized ratio is controlled within the therapeutic level and rehabilitation was being performed without additional thrombotic events.

Fig. 1