

A 16-Year-Old Girl with Hereditary Hemorrhagic Telangiectasia

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1. Clinical Images

A 16-year-old girl was admitted to the hospital with severe abdominal pain that could not be relieved. She was initially misdiagnosed as enterocolitis and did not improve after symptomatic treatment, the rupture of the spleen and massive blood accumulation in the abdominal cavity were detected by ultrasonography and CT (Figure 1). Splenectomy surgery was performed immediately and the pathological diagnosis of splenic sinus shore cell hemangioma was made after operation. Chest CT and enhanced scan revealed multiple pulmonary arteriovenous malformations in both lungs, the largest of which was located in the left lower lobe (Figure 2). Laboratory tests showed a low red blood cell count (2.86×10^{12}) and hemoglobin (98G/L) and low Oxygenation (84). The patient was found to have recurrent epistaxis since childhood and had been admitted to the hospital recently for failure to stop bleeding. The left nasal mucosal telangiectasia and pulsatile bleeding point were found by nasal endoscopy, electrocoagulation was performed under nasal endoscope to stop bleeding, and without obvious cause to faint, accompanied by nausea, vomiting the next day. The patients were re-admitted to the hospital and underwent interventional embolization for the large pulmonary arteriovenous malformations in the lower lobe of the left lung (Figure 3), post-operative Oxygenation (96) were significantly increased and patients' quality of life was improved. His father died of a ruptured arteriovenous malformation and his grandmother had a history of recurrent nosebleeds. The patient was highly suspected to have hereditary hemorrhagic Telangiectasia. A single-gene genetic disease gene test—whole-exome analysis, showed that the ENG: c.360+1

G>A variant was a pathogenic variant, associated with hereditary hemorrhagic Telangiectasia, the variant is a classical shear site mutation. Our diagnosis was confirmed and the patient is currently being followed up.

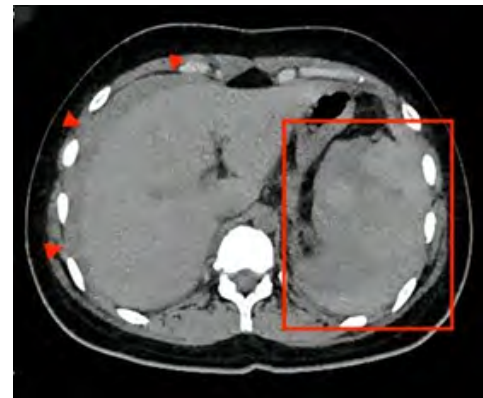


Figure 1: The rupture of the spleen and massive blood accumulation in the abdominal cavity



Figure 2: Multiple pulmonary arteriovenous malformations in both lungs



Figure 3: Before and after interventional embolization