Headache and Epistaxis in family history associated with ischemic stroke

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A 34-year-old woman developed hemoptysis and headache with glittering visual complaint but checked into an emergency room with right arm weakness during 5 minutes. For a long time, she suffered from epistaxis. She had gotten electro-coagulation therapy for the control of nose bleeding in a military hospital. However, her epistaxis redeveloped even electro-coagulation therapy two times. We checked her chest and brain by a CAT and a MR scan. Thus, her chest CAT scan showed arteriovenous malformation (AVM) in the right apex and right lower lung and the MR brain scan indicated an AVM in her frontal head. Through the operation of her pulmonary AVMs, her epistaxis was dramatically controlled. It is known that pulmonary arteriovenous fistulas (PAVFs) was induced right to left shunt and became an embolic source to cause transient ischemic attack. However, it may cause critical neurological problems such as permanent hemiparesis, brain abscess and meningo-encephalitis when properly untreated. Furthermore, her pedigree declared it might be a genetic disease and we found the gene study proved the hereditary hemorrhagic telangiectasia (HHT) with ENG gene mutation from her and her mother. Finally, we have learned that adequate management like operation would be needed to control pulmonary AVMs to prevent critical neurologic complications and recurrent epistaxis or hemoptysis with family history may be inspired to test gene study for the hereditary hemorrhagic telangiectasia.