A case of Hereditary hemorrhagic telangiectasia incidentally founded

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Introduction: Hereditary Hemorrhagic Telangiectasia (HHT), also known as Osler-Weber-Rendu disease, is a rare autosomal dominant disorder characterized as mucocutaneous telangiectasia, recurrent hemorrhage and familial history. The prevalence of HHT is around 1/5,000~8,000, and there are only few case reports are published previously. We report a case of a 19-year-old patient who was incidentally found to have HHT.

Case report: A 19-year-old male visited hospital with abnormal chest X ray. He took the image for military entrance as routine in Korean young man. In his early teens, he had suffered from frequent epistaxis, however, there was no other disease that bothered him and he denied any of familial histories. His chest X ray showed multifocal infiltrations of both lungs. Chest computered tomography revealed multiple pulmonary arteriovenous malformations in RUL, RML, and BLLs. On agitated saline transthoracic contrast echocardiography, the agitated saline bubbles appeared in the left side of the heart after 4 beats of the heart which meaned positive for shunting. For closure of the shunts, radiologic interventionist performed pulmonary angiography figuring out several arteriovenous malformations (AVMs). Embolizations were done with coils in multiple sites. Additionally, we tried to seartch AVMs in other organs, but there was no additional AVMs. The patient was evaluated the genetic test such as ENG, ACVRL1, GDF2 and SMAD4 genes. Conclusionly ENG gene (449C>A) was detected and the diagnosis of hereditary hemorrhagic telangiectasia (HHT), type II.

Conclusion: HHT is an autosomal dominant vascular disorder making multiple AVMs in multiple in various organs including the lungs, gastrointestinal tract, liver, and brain. In most cases of HHTs, patients usually have symptoms including recurrent epistaxis, cutaneous telangiectasia especially during young. However, even though there is no symptoms, we have to consider the hereditary problems and careful history taking is needed as well.,

Figure 1.

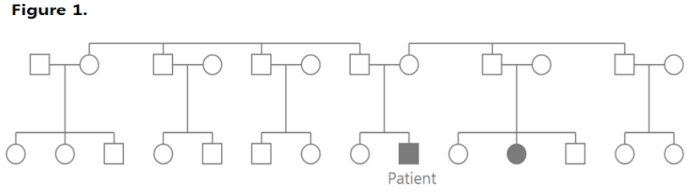

Figure 2.

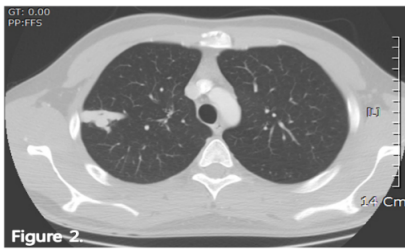

Figure 3.

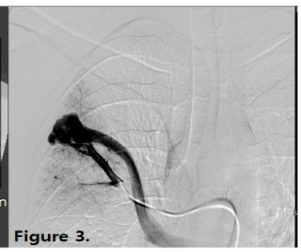

Figure 4.