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CASE REPORT ON OSLER WEBER RENDU SYNDROME

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ABSTRACT
Background: Osler-weber-rendu disease (OWRD) is a rare autosomal dominant disorder that affects blood vessels through out the body resulting in tendency for bleeding. Recurrent and severe epistaxis is the common clinical presentation which leading to severe anemia. **Case Report:** We present a case of Osler-weber-rendu formed by complication from brain abscess. 45-year-old male came to the hospital for evaluation of Anemia and recurrent Epistaxis and he had a history on the right temporal craniotomy and excision of an abscess. ENT examination was significant for the presence of telangiectasias on the tongue. **Conclusion:** The patient suffered from temporal lobe abscess for which he underwent right temporal craniotomy which is suggestive feature for brain abscess which is more common in patients with OWRD than in the healthy population.

KEYWORDS: OWRD, ENT.

INTRODUCTION
Osler-Weber-Rendu disease (OWRD) is a rare autosomal dominant disorder that affects blood vessels throughout the body (causing vascular dysplasia) and results in a tendency of bleeding. The condition is also known as hereditary hemorrhagic telangiectasia (HHT). HHT develops through mucocutaneous telangiectases and arteriovenous malformations (AVMs), a potential source of severe morbidity and mortality.[1] The most common presentation is recurrent and severe epistaxis, which often leads to severe transfusion-related anemia.[2] There is also a prevalence of GI bleeding.[3] The four criteria for clinical diagnosis are as follows: family history of HHT, Epistaxis, Telangiectasis, and visceral lesions.[4] Frequent nosebleeds and melena in the nose and GI tract may be the result of telangiectasia. Patients with severe HHT disease suffer from severe bleeding and iron deficiency anemia.[5]

CASE REPORT
A 45-year-old male presented to the hospital for evaluation of Anemia and recurrent Epistaxis. The patient had a history of Anemia secondary to GI bleeding. There was no associated history of hemoptysis or dyspnea for two months. He was already diagnosed with a similar condition to the hospital. Computed tomography (CT) of the brain shows a peripherally enhanced thin-walled hypo dense lesion with edema on the right temporal lobe He had history on the right temporal craniotomy and excision of an abscess. Physical examination reveals pallor and ENT examination was significant for the presence of telangiectasias on the tongue. There was active bleeding from the nose. Lab investigation revealed significant Anemia with a hemoglobin count of 2.6g/dl with the presence of microcytosis, anisopoikilocytosis, and hypothermia on peripheral smear. On further investigation reveals elevated neutrophil depleted lymphocytes and RBC level was significantly decreased with 1.84cells/cubic millimeter and ESR were significantly elevated with a value of 98mm/hr. A CT of the chest was negative for anterior venous malformations. The patient was managed with an anterior nasal package and 2 units of packed cell transfusions.

DISCUSSION
Hereditary hemorrhagic telangiectasia(HHT), also known as Osler-Weber-Rendu syndrome, is a rare autosomal dominant genetic disorder that leads to abnormal blood vessel formation in the skin, mucous membranes, and often in organs such as the lungs, liver, and brain.[6] Telangiectasia (small vascular malformations) can occur in the nose and gastrointestinal tracts skin and mucosal linings. The skin lesions characteristically occur on the lips, the nose, and the fingers, and the skin of the face in sun-exposed areas.[7]The variants of HHT are HHT type-1 and type-2 are due to defective endoglin (ENG) and activin like receptor kinase (ALK1) genes. The diagnosis of OWRD or HHT is based on epistaxis, telangiectasias, visceral lesions and family history with a first degree relative

with HHT. Patients with the HHT type-1 genotype have a higher prevalence of pulmonary and cerebral AVMs, and more severe GI bleeding than in HHT type-2.[8] The diagnosis is considered definite if any three of the above-mentioned criteria are present. Our patient had the three criteria present. In the absence of pulmonary AVMs, his symptomatology is attributed to anemia resulting from frequent bleeding from the nose.[9] Neurological manifestation mechanisms other than bleeding include paradoxical emboli with brain abscess, ischemic stroke, cerebral hypoxia caused by right-to-left shunting, air embolism, and secondary polycythemia.[10]

The patient suffered from temporal lobe abscess for which he underwent right temporal craniotomy which is suggestive from McDonald MJ, Brophy BP et.al. who explained stroke and brain abscess is more common in patients with OWRD than in the healthy population. Also the diagnosis of this disease is made clinically on basis of the curacao criteria which shows recurrent epistaxis , visceral lessions on the toungue and telangietasias.

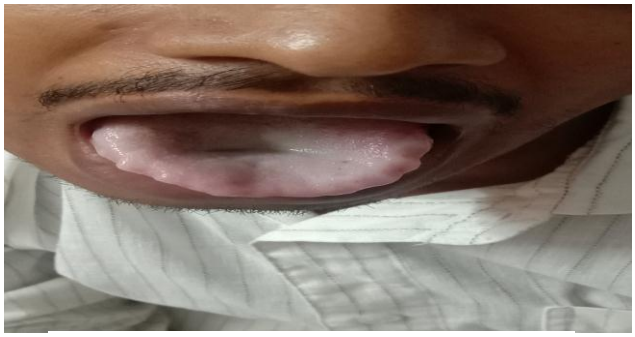

Figure 1.0: Its shows visceral lesion on tongue.

CONCLUSION

Brain abcess is the most potential complication of Osler-weber-rendu disease and the abcess is more likely to result from a bacterial seeding of an ischemic portion of the brain. The diagnosis of this disease is based on clinical manifestations suggestive of HHT.

REFERENCES

1. Giordano P, Lenato GM, Suppressa P, Lastella P, Dicuonzo F, Chiumarulo L, et al. Hereditary hemorrhagic telangiectasia: arteriovenous malformations in children. J Pediatr, 2013; 163(1): 179-86.
2. Franchini M, Frattini F, Crestani S, Bonfanti C. Novel treatments for epistaxis in hereditary hemorrhagic telangiectasia: a systematic review of the clinical experience with thalidomide. *J Thromb Thrombolysis*, 2013; 36(3): 355-7.
3. Nanda S, Bhatt SP. Hereditary hemorrhagic telangiectasia: epistaxis and hemoptysis. CMAJ, 2009; 180(8): 838.
4. Shovlin CL, Guttmacher AE, Buscarini E, et al. Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). *Am J Med Genet*, 2000; 91(1): 66-7.
5. Kjeldsen AD, Oxhøj H, Andersen PE, et al. Pulmonary arteriovenous malformations: screening procedures and pulmonary angiography in patients with hereditary hemorrhagic telangiectasia. Chest, 1999; 116(2): 432-9.
6. Govani FS, Shovlin CL "Hereditary hemorrhagic telangiectasia: a clinical and scientific review". *European Journal of Human Genetics*, 2009; **17**(7): 860–71.
7. Dupuis-Girod S, Bailly S, Plauchu H. "Hereditary hemorrhagic telangiectasia (HHT): from molecular biology to patient care". *J. Thromb. Haemost*, 2010; **8**(7): 1447–56.
8. Johnson DW, Berg JN, Baldwin MA, Gallione CJ, Marondel I, 7.Yoon SJ, et al. Mutations in the activin receptor-like kinase1 gene in hereditary hemorrhagic telangiectasia type 2. NatGenet, 1996; 13: 189-95.
9. McDonald MJ, Brophy BP, Kneebone C. Rendu-Osler-Weber syndrome: a current perspective on cerebral manifestations. *J Clin Neurosci*, 1998; 5(3): 345-50. .
10. Putman CM, Chaloupka JC, Fulbright RK et al. Exceptional multiplicity of cerebral arteriovenous malformations associated with hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu syndrome). AJNR Am J Neuroradiol, 1996; 17: 1733–42.