Volume 9, Issue 10, October – 2024 International Journal of Innovative Science and Research Technology
ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/IJISRT24OCT1822

Osler-Weber-Rendu Disease: A Rare Contributor to Recurrent Iron Deficiency Anemia

Kalva Suchitra Reddy[1]
Internal Medicine Department,
Mamata Medical College, India

Mekala Siddhanth Kumar[2]
Internal Medicine Department,
Bhaskar Medical College, India

Usha Topalkatti[3]
Internal medicine Department
Spartan Health Science University,
School of Medicine, Saint Lucia

Param Kadam[4]
Internal Medicine Department
Maharashtra Institute of
Medical Education and Research, Pune

Abstract:- Hereditary hemorrhagic telangiectasia (HHT), also called Osler-Weber-Rendu syndrome, is a rare genetic disorder inherited in an autosomal dominant pattern. It is marked by recurrent nosebleeds, arteriovenous malformations (AVMs), mucocutaneous telangiectasias, and often a family history of the condition. This case involves a 55-year-old woman with a prolonged history of spontaneous bleeding, particularly from lesions on her tongue, leading to chronic iron deficiency anemia. The case emphasizes the importance of considering HHT in patients with anemia and unexplained recurrent bleeding. It also underscores the value of targeted therapies and genetic counseling in managing the disorder.

I. INTRODUCTION

Osler-Weber-Rendu syndrome, also known as Hereditary Hemorrhagic Telangiectasia (HHT), is a genetic disorder affecting approximately 1 in 6,000 people. It follows an autosomal dominant inheritance pattern and is typically identified by a triad of symptoms: telangiectasias, frequent nosebleeds, and a family history of the condition. Individuals with HHT often develop arteriovenous malformations (AVMs) in various organs, adding complexity to the disease. The condition is caused by mutations in the ENG or ACVRL1 genes, which result in abnormal and fragile blood vessel formation.

II. CASE REPORT

A 55-year-old post-menopausal woman, born to non-consanguineous parents, presented with red, raised lesions on her tongue and hands that had been present for 25 years. She also reported a 20-year history of exertional shortness of breath and fatigue. Her medical history included recurrent episodes of nosebleeds and spontaneous bleeding from tongue lesions, leading to numerous blood transfusions over two decades. Her family history was significant, as two of her sisters and one of her children had similar lesions.

On clinical examination, the patient was pale, and erythematous, blanchable lesions were noted on both palms, along with compressible, non-pulsatile papulonodular lesions on the tongue and petechiae on the palate. There was no organ enlargement. Laboratory tests indicated microcytic hypochromic anemia with a normal reticulocyte count and coagulation profile. Tests for antinuclear antibodies (ANA), hemoglobinuria, and sickling were negative. Imaging studies, including abdominal ultrasonography, revealed a liver cavernoma, and carotid angiography showed several telangiectatic lesions in the tongue supplied by both lingual arteries. Chest X-ray and fundoscopy were normal, and no arteriovenous malformations (AVMs) were found in the intestines, lungs, or kidneys.

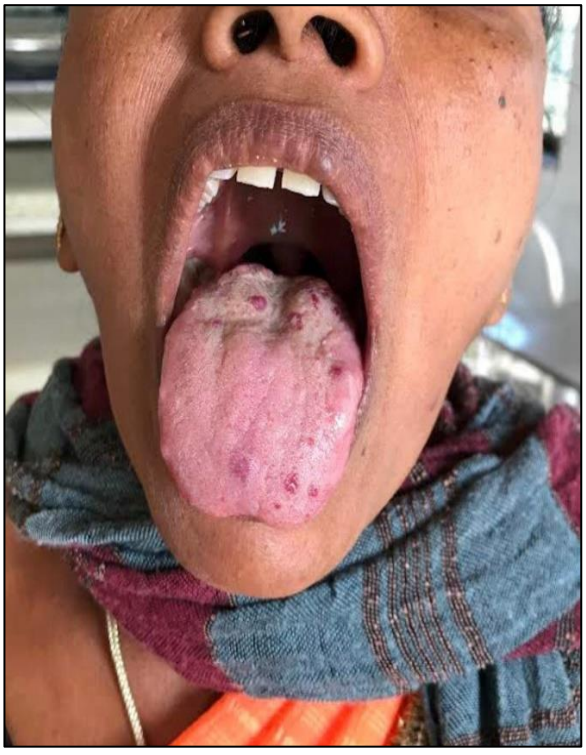

Fig 1: Erythematous Papulonodular Lesions on the Tongue

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/IJISRT24OCT1822

Fig 2: Erythematous Macular Lesions on Both Palm

HISTOPATHOLOGY REPORT (SMALL BIOPSY)

Biopsy No. 497 / H-19 / 7478

Clinical details : C/o Multiple erythematous papulonodular lesions.

Nature of specimen : Tongue Biopsy

Gross : Received single grey white soft tissue bit measuring 0.4 x 0.4 x 0.1 cm.

(Entire tissue submitted for histopathological examination)

Microscopic Examination :

Section shows stratified squamous epithelium keratinising type. No loss of polarity or hyperchromasia. Submucosa shows dilated lymphatics separated by spindle cells.

Impression : Suggestive of **Lymphangioma**

Certificate # MC-2657

Dr. R S ASOK KUMAR
CONSULTANT PATHOLOGIST

Fig 4: Histopathological Report of Tongue Biopsy

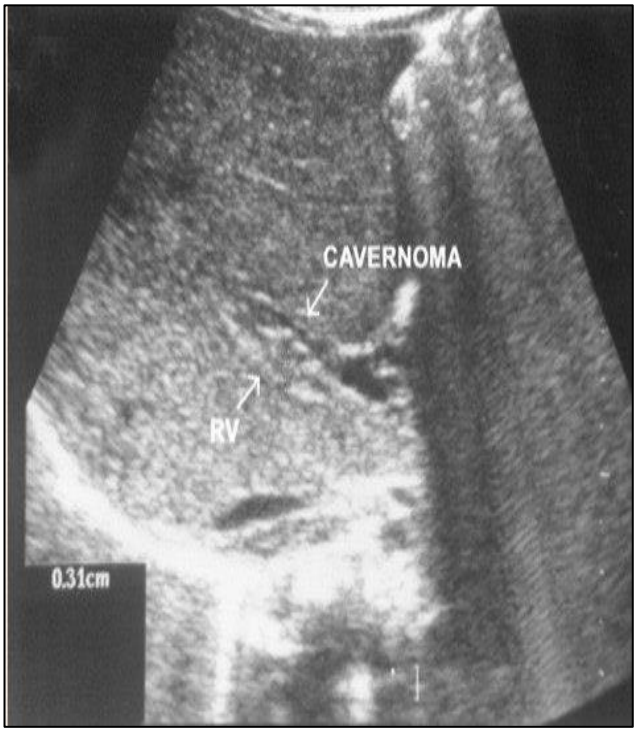

Fig 3: Liver Cavernoma on USG

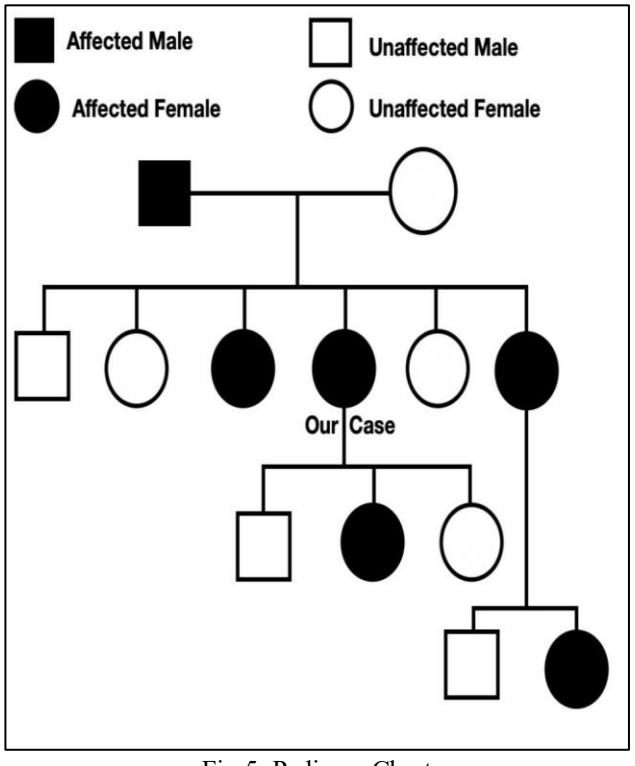

Fig 5: Pedigree Chart

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/IJISRT24OCT1822

III. DISCUSSION

Osler-Weber-Rendu syndrome, also known as Hereditary Hemorrhagic Telangiectasia (HHT), is a rare genetic disorder initially identified by Henri Rendu in 1896, with further descriptions by Sir William Osler in 1901 and Frederick Parks Weber in 1907. It is divided into two main types based on genetic mutations: HHT type 1, related to ENG gene mutations, and HHT type 2, linked to ACVRL1 gene mutations. Type 1 is more frequently associated with pulmonary and cerebral arteriovenous malformations (AVMs) and severe gastrointestinal bleeding, whereas type 2 often involves hepatic AVMs.

The disorder results from abnormal endothelial cell signalling, leading to fragile, dilated blood vessels that are prone to bleeding. Diagnosis is based on the Curacao criteria, which include recurrent spontaneous nosebleeds, multiple telangiectasias in locations such as the lips, oral cavity, fingers, and nose, AVMs in internal organs (e.g., lungs, liver, brain, spine, gastrointestinal tract), and a family history of the condition. A diagnosis is considered definitive when three or more of these criteria are met.

The patient in this case met all four Curacao criteria, confirming the diagnosis of HHT. Managing the condition often requires a multidisciplinary approach due to the complexity of the AVMs, with treatment options including cryotherapy, laser therapy, estrogen therapy, aminocaproic acid, cauterization, or surgical removal of AVMs. Given the genetic nature of HHT, genetic counselling and family screening are recommended for affected individuals and their relatives.

IV. CONCLUSION

HHT should be suspected in patients presenting with recurrent mucocutaneous bleeding, particularly frequent nosebleeds, chronic iron deficiency anemia, and a normal coagulation profile. Early diagnosis and a well-coordinated treatment plan, which includes family screening, are essential for minimizing complications and improving the quality of life for those impacted by the condition.

REFERENCES

[1]. Guttmacher AE, Marchuk DA, White RI Jr. Hereditary hemorrhagic telangiectasia. N Engl J Med. 1995;333:918-924.
[2]. Thappa DM, Laxmisha C. Recurrent bleeding from the tip of the index finger and mucocutaneous telangiectasia: Osler-Weber-Rendu syndrome. J Postgrad Med. 2002;48:274-277.
[3]. Shovlin CL, Hughes JM, Scott J, Seidman CE, Seidman JG. Characterization of endoglin and identification of novel mutations in hereditary hemorrhagic telangiectasia. Am J Hum Genet. 1997;61:68-79.