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Case Study

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OSLER-WEBER-RENDU SYNDROME – A CASE PRESENTATION

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ABSTRACT

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant inherited vascular malformation syndrome characterized by telangiectases and arteriovenous malformations. The dermal telangiectases are generally pinpoint to pinhead sized, very specifically concentrated on the hands, face, and lips, and not diffuse. We hereby report a case of hereditary hemorrhagic telangiectasia diagnosed in a 62 years old patient.

KEYWORDS: Hereditary hemorrhagic telangiectasia, telangiectases.

INTRODUCTION

Hereditary hemorrhagic telangiectasia is also known as Rendu-Osler-Weber syndrome. It is an autosomal-dominantly inherited vascular malformation syndrome characterized by telangiectases and arteriovenous malformations (AVMs) that occurs in 1 in 10,000 individuals.[1] It is characterized by telangiectasias of the skin and mucous membranes as well as large AVMs mainly in pulmonary, hepatic, and cerebral circulation. Clinically it appears as punctuate or splinter-like telangiectasias located on the lips, oral mucosa, upper extremities, nail beds, and trunk.[2]

CASE REPORT

We present a case of a 62-year-old patient who presented with complaints of spontaneous bleeding from the tip of fingers. The clinical examination revealed presence of punctuate or splinter-like telangiectases also occur on the mucosal surface of the tongue, lips and tip of fingers. (Figure 1) On the basis of clinical examination, the diagnosis of Hereditary hemorrhagic telangiectasia was made.

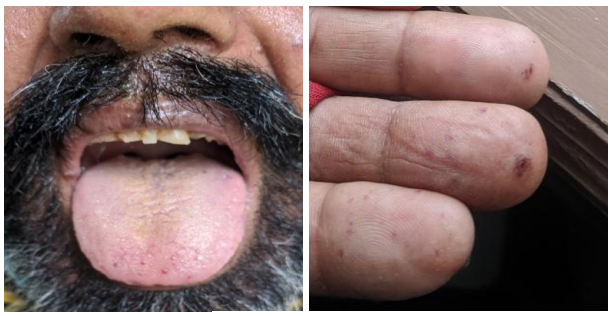

Fig 1&2: Shows telangiectases on the tongue and fingers.

DISCUSSION

Hereditary hemorrhagic telangiectasia (HHT) or Osler-Weber-Rendu syndrome is inherited as an autosomal dominant disorder. It is characterized by widespread telangiectases that can involve the skin, mucous membranes with prevalence of 1/10,000.[3]

It is thought that the abnormal vessels in HHT develop because of aberrant TGF signaling at some stage during vascular development and homeostasis due to mutations of HHT-associated genes (HHT1 and HHT2). It results in alteration in the elastic and muscle layers of vessel walls, making them more vulnerable to spontaneous rupture and injuries.[4]

The treatment options are primarily supportive and palliative and includes electrocauterization with diathermy, sclerotherapy, or laser therapy (Nd:YAG, IPL, argon).

Early diagnosis of these patients and their family members with timely referral, helps in reducing the complications.

Conflict of interest: nil.

REFERENCES

1. Marchuk, D. A., Guttmacher, A. E., Penner, J. A., and Ganguly, P. Report on the workshop on hereditary hemorrhagic telangiectasia July 10-11, 1997. *Am. J. Med. Genet*, 1998; 76: 269–273.
2. Chang DS, Kim MN, Hong CK, Ro BI. A case of hereditary hemorrhagic telangiectasia. *Korean J Dermatol*, 2002; 40: 1148–1150.

3. Begbie M, Wallace G, Shovlin C. Hereditary haemorrhagic telangiectasia (Osler-Weber-Rendu syndrome): a view from the 21st century. Postgrad Med J., 2003; 79(927): 18–24.
4. Juares AJ, Dell'Aringa AR, Nardi JC, Kobari K, Gradim Moron Rodrigues VL, Perches Filho RM. Rendu-Osler-Weber Syndrome: case report and literature review. *Braz J Otorhinolaryngol*, 2008; 74: 452–457.