HEREDITARY HEMORRHAGIC TELANGIECTASIA

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Hereditary hemorrhagic telangiectasia, also known as Osler-Weber-Rendu syndrome, is a genetic condition of malformed blood vessels. These irregular collections of blood vessels are known as arteriovenous malformations (AVM) and can lead to recurrent and excessive bleeding. The most commonly affected areas of the body include the gastrointestinal tract, oral cavity, lungs, liver and brain. When these AVM's affect the skin, they are referred to as telangiectasias. Diagnostic criteria for hereditary hemorrhagic telangiectasia requires the presence of three of the following four features: (1) recurrent nosebleeds (2) AVM's on the bodies internal organs (3) telangiectasias in the gastrointestinal tract and (4) having a close relative with the disease.

This medical condition is inherited in an autosomal dominant fashion, meaning children of an affected parent have a 50% chance of acquiring the disease. This gives rise to the hereditary nature of the disease. Individuals may develop this condition in the absence of a family history and likely represents a spontaneous genetic mutation.

One of the most striking features of hereditary hemorrhagic telangiectasia is a strong family history of this bleeding disorder. The initial diagnosis may be suspected in children and adolescents with recurrent nosebleeds. Commonly, the presence of the syndrome is often not confirmed until later in the patient's life. Physical exam findings will likely reveal numerous small 'cherry red' non-blanching maculae's measuring 2-5mm in size, scattered about the mouth, lips, tongue, ears and other areas of the skin. Other manifestations suggestive of the condition include increasing number and size of telangiectasias, bloody stools, vomiting of blood or iron deficiency anemia.

The best therapy for hereditary hemorrhagic telangiectasia is prevention. Most clinicians recommend various imaging modalities of the body including an MRI of the brain in all newly diagnosed patients. This is done in an attempt to discover any life threatening AVM's. These identified lesions can be prophylactically embolized by intentionally occluding the blood vessels. Destruction of these lesions may be necessary in certain cases and is usually reserved for small-localized areas. Procedures such as **electrocautery**, **laser** and estrogen therapy may be employed. If bleeding persists, surgical intervention may be required.

This information has been provided to you compliments of the American Osteopathic College of Dermatology and your physician.