GENETICS AND GENODERMATOSES

MUCO-CUTANEOUS FINDINGS IN HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT): GENOTYPE-PHENOTYPE CORRELATION OF 62 ADULT ITALIAN PATIENTS.

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Introduction: HHT is an autosomal dominant disorder characterized by epistaxis, telangiectases in muco-cutaneous and gastrointestinal sites, arteriovenous malformations (AVMs) and familial inheritance.

Objective: To correlate the muco-cutaneous findings with other manifestations of the disease, such as severity of epistaxis, visceral involvement, genetic mutations.

Materials and Methods: We made a prospective study on 62 patients visited from March 2014 to March 2018. We introduced a new cutaneous score (HHT Skin Score) that includes localization, number and morphology of skin and mucosal assessable telangiectases. All patients were tested for genetic mutation and were checked for visceral AVMs.

Results: Telangiectases were located on nose, ears, lips, fingertips, but also on feet and toes. Pin-sized vessels resulted the most common subtype, papulous telangiectases were present at lips and tongue, while arborized ones were prevalent on ears, nose and temples. We founded a direct correlation between cutaneous involvement (HHT Skin Score) and epistaxis scoring system (ESS). Oral cavity involvement resulted associated with abundant epistaxis, and with and increased risk of hepatic AVMs. Numerous papulous labial or lingual telangiectases resulted associated with pulmonary AVMs. HHT type 1 (26% of our patients) was related to pulmonary AVMs and mucosal telangiectases, while pulmonary AVMs and feet and ears lesions resulted associated to HHT type 2 (64% of our patients).

Conclusions: Our study demonstrates a very good genotype-phenotype correlation between genetic mutation and specific clinical aspects.