

# Mental health and counseling intervention for hereditary hemorrhagic telangiectasia (HHT) during the COVID-19 pandemic: perspectives from Italy

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**Abstract. – OBJECTIVE:** Hereditary hemorrhagic telangiectasia (HHT) is an inherited disorder of fibrovascular tissue, transmitted as an autosomal dominant trait. This rare disease can involve one or more organs and clinical manifestations interest several medical specialties.

**MATERIALS AND METHODS:** A review of recent literature and our clinical experience shows that COVID-19 pandemic greatly influences the autonomy and psychic sphere of patients with HHT, causing them further distress.

**RESULTS:** Often patients affected by HHT experience a sense of loneliness due to the rarity of this pathology and COVID-19 pandemic adds a burden for them and their caregivers who have to face emotional experiences that interfere with personal, social and working functioning.

**CONCLUSIONS:** Multidisciplinary approach and web-mediated counseling intervention could offer a valid and personalized support for patients affected by HHT and their caregivers during quarantine due to COVID-19 pandemic.

## Key Words:

Hereditary hemorrhagic telangiectasia, COVID-19, Counseling, Personalized Medicine, Multidisciplinary Approach

## Introduction

The COVID-19 epidemic started in Wuhan, the most populated city in Eastern China, where an increasing number of pneumonia cases of un-

known aetiology began. The causative agent of these lung diseases is the new Coronavirus Sars-CoV-2, from the same family as the coronaviruses responsible for SARS and MERS. Rapidly the virus arrived in the rest of the world and, particularly, in Italy, which was the first Western country to be severely affected. At the end of January 2020 the World Health Organization declared a global state of emergency. The most common symptomatic picture is represented by fever, dry cough, fatigue, dyspnoea, sputum production, sore throat, headache and myalgia. The most critical forms of the disease are characterized by respiratory failure, septic shock, multi-organ dysfunction. The range of symptoms with which COVID-19 infection can occur varies from asymptomatic to severe pneumonia and presentation with serious illnesses can have a fatal outcome<sup>1</sup>. There are still no vaccines available and there is little evidence to support the efficacy of potential therapeutic agents. In addition, presumably the population does not have any prior immunity against the new Coronavirus, therefore everybody is considered susceptible. The most serious forms of disease are more common among the elderly, immunosuppressed subjects or patients with other concomitant chronic pathologies. To date, these risk groups represent the majority of cases of serious illness and death. In order to mitigate the impact of the epidemic, urgent measures like isolation in quarantine have been put in place for the management of the emergency.

Hereditary hemorrhagic telangiectasia (HHT), also known as Rendu-Osler-Weber disease, is an inherited autosomal dominant genetic disorder characterized by vascular anomalies. HHT has always been diagnosed on the basis of clinical characteristics, with the presence of at least three of the Curaçao criteria: epistaxis, multiple telangiectasis, visceral venous arterial malformations, a first-degree relative suffering from HHT. Currently, it can also be diagnosed with genetic tests, through a single DNA pathogenic sequence variant, generally in *ENG*, *ACVRL1* or *SMAD4*<sup>2</sup>.

HHT affects one in 5000 people regardless of gender and it occurs in all ethnic groups and continents. The most frequently encountered signs are spontaneous and recurrent nosebleeds, which can cause chronic iron deficiency anaemia and poor quality of life<sup>3</sup>. As a consequence, subjects with HHT result to be more vulnerable to depression and other psychiatric disorders<sup>4</sup> and suffer higher levels of psychological distress than the general population<sup>5</sup>.

The disease also causes skin and mucous membrane manifestations or telangiectasias affecting the fingertips of the fingers and lips, oral cavity, nasal mucosa and conjunctiva. The most serious manifestations are hemorrhages and linked to the presence of arteriovenous malformations, are mainly located in cerebral, pulmonary and gastrointestinal districts. There is no specific treatment that can definitively cure this pathology; nevertheless, it is necessary to adopt therapeutic interventions in order to limit the symptoms of the various involved districts<sup>6</sup>.

### ***Web-Mediated Counseling for Patients With HHT***

The patient suffering from a rare disease frequently experiences his pathology in a condition of loneliness, now amplified by the isolation prescribed by the quarantine. Sense of impotence linked to the awareness that there are no specific therapies increases significantly along with fear of being infected with COVID-19. This context pervasively affects the lives of people with HHT and their families resulting in an increased risk of developing aggressive behaviors<sup>7</sup>, increasing use of alcohol and psychoactive drugs, evolving or worsening psychiatric or neurological disorders<sup>8</sup> and cardiological or multiple comorbidities<sup>9-11</sup>.

Social distancing and quarantine are the main measures taken to combat the pandemic and lead to a life reduced to its essence with a greater impact in patients suffering from HHT<sup>12</sup>. Uncer-

tainty and fear are the most experienced affective states towards an invisible enemy. The current suspension condition induced by the COVID-19 emergency deprives the human being of essential interhuman contact producing an emotional desertification. For this reason, in our clinical activity it results important to convert usual counseling interventions to distance, web-mediated counseling, aimed to offer a helping professional relationship for patients affected by HHT and their caregivers.

Rare events can represent a resource, help to grow and understand complicated realities and can open the way for the discovery of unknown mechanisms. This concept has prompted us to form a group with a multidisciplinary approach according to a model that also focuses on the emotional experiences of patients. It is known that the lack of attention to psychological distress can negatively alter the response to therapies, with possible negative influence on the prognosis: in patients suffering from HHT the care of their emotional difficulties can positively influence their quality of life and improve compliance with treatments.

Our model is based on the client-centred counseling intervention and aims to promote an effective functioning of the individual. The goal is to reach the well-being of the person by avoiding that negative psychological experiences pervade patients or their caregivers. Drawing on the personal resources already present in each individual it is possible to help to restore personal skills, to activate coping processes and to develop resilient skills (Table I).

### **Conclusions**

Multidisciplinary approach and web-mediated counseling intervention could offer a valid and personalized support for patients affected by HHT and their caregivers during quarantine due to COVID-19 pandemic. Since rare diseases are psychologically highly stressful for both patients and their families, in our perspective it is essential to build a bridge between the patient and the social and emotional network in which he lives and is inserted, through individual or group counseling interventions.

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### **Conflict of Interest**

The Authors declare that they have no conflict of interests.

**Table I.** Web-mediated counseling intervention for Hereditary Haemorrhagic Telangiectasia (HHT) during the COVID-19 pandemic.

Experienced affective states	Counseling intervention
Uncertainty and fear towards an invisible enemy	Exercise the capacity to remain in oscillation between continuity and discontinuity, to stay in uncertainty, in indefiniteness, having faith in the creative response of one's unconscious
Emotional desertification due to the deprivation of interhuman contacts	Help to create an open mental space indispensable for the birth of affects and thoughts, through the discovery of own individuality in relation with the fruitful otherness
Anguish thoughts of never being able to recover, oscillation between denial and moments of aggressive revolt	Suggest that rare events can represent a resource, a possibility to grow and understand complicated realities through the discovery and elaboration of unknown mechanisms
Fears for the future, hopelessness	Contain distressing thoughts and promote the building of one's own sense of experience, develop resilient skills, redesign a new approach with feelings and habits

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