

Access Guide to the HHT clinical pathway

Purpose of the guide:

In the course of life, a patient with a diagnosis or suspicion of HHT accesses his or her center of reference for various reasons and with different urgencies. Making the patient know how to access and the necessary documentation for reservations and access to the service means ensuring greater efficiency for the user.

Made by:

HHT Equipe Policlinico Gemelli

- Prof.ssa Eleonora Gaetani
- Prof.ssa Maria Elena Riccioni
- Prof. Giulio Cesare Passali

In collaboration with:

HHT Onlus – Italian Association of
Hemorrhagic Hereditary Teleangiectasia

Adult patient access guide

FIRST VISIT TO THE RARE DISEASES CLINIC

Service description and access mode

Visit necessary to confirm the clinical suspicion and to begin the diagnostic procedure.

Will have to make an initial outpatient visit:

- Those who have a chance of having inherited the pathology from their family;
- Those who have a suspicion of HHT.

How:

Contact via e-mail the HHT path (percorso.hht@policlinicogemelli.it) which will evaluate the request and fix the first outpatient appointment.

Ask your family doctor for a commitment with the following wording:

“[Internal visit for suspected HHT](#)”.

Exemption code: [R99](#) - which corresponds to the exemption for suspicion of rare pathology.

For patients who have already been diagnosed with the disease, also at another HHT Center, it will be sufficient to carry out binding for “[Internal visit for suspected HHT](#)” with the relative exemption code [RG0100](#).

The test will be delivered directly to the doctors of the HHT clinic, without the need to carry out any administrative practice.

Delivery times

Average response times to the first email: 3-7 days

Average times for the first outpatient visit: about 4 weeks

Average times for the execution of the complete genetic evaluation with research of known familiar variant and delivery of the definitive report: approximately 2 months;

The genetic outpatient assessment for new variant research is between 4-6 months.

FIRST SCREENING

Service description and access mode

The screening, foreseen by the international guidelines of the pathology, consists of a series of tests necessary to know the involvement of the disease in the various organs of the patient. It is an essential step for patient protection.

Screening is carried out on an outpatient basis, trying to concentrate more than one evaluation on the same day (minimum 2 visits: always internal evaluation and ENT on the first visit)

Blood tests, echocardiogram bubble, abdominal ultrasound, chest CT, brain MRI, possible endoscopic procedures and specialized consultancy, counseling and psychological support for patients and family.

Delivery times

4-6 months

Following the first screening and each visit, the patient will receive a written and updated report with indications for subsequent visits.

FOLLOW UP (*Control Visits*)

Service description and access mode

Following the first screening and each subsequent visit, the patient will receive a written report with the visits and procedures performed and the specific indications on the timing of the following checks.

These controls will be evaluated based on the individual screening result and will vary from patient to patient.

The expected timing in the absence of emergencies is as follows:

- **Control visit with execution of blood count and martial reserves:** once a year
 - **Abdomen ultrasound:**
 - Once every 3-5 years in the absence of liver involvement;
 - Once every 3 years in the presence of an isolated liver sign
 - **Echocardiogram with contrast:**
 - Once every 5 years in the absence of echocardiographic signs indicative of disease
 - **Chest CT:** once every 5 years in the presence of echocardiographic signs indicative of disease
 - **MRI brain:** systematic follow-up is not expected in the absence of neurological signs
 - **Endoscopic procedures:** systematic follow-up is not expected in the absence of signs of gastrointestinal bleeding or anemia.
- As with all chronic and rare diseases, the course is also equipped with a counseling service and psychological and psychiatric support.

Patients in charge of the HHT Center, where indicated, can take advantage of iron infusion therapy at the center.

When the patient goes to make the required exams, he brings with him the discharge sheet which includes the indications and above all the reasons for the examination.

This sheet should be presented to those who will take the exam so that they know the specific methods with which to perform it, but above all know what to go to investigate.

Once all the tests have been carried out, the patient returns to the family doctor to have a check-up done for the check-up visit to be done at the HHT Center of the Gemelli General Hospital.

The check must read: "[Internal medicine visit: Follow up for HHT](#)".

And here too the RG 0100 exemption applies.

With this commitment the patient contacts the center via email "First outpatient visit" (see above)

but the subject of the e-mail must be indicated: reservation follow up visit with exams already requested.

CONSULTATIONS ON PERIODICAL RESULTS OF THE BLOOD TESTS

Service description and access mode

Sending copy of the analysis exclusively via email to percorso.hht@policlinicogemelli.it specifying in the object: HHT patient analysis (name and surname)

Delivery times

The reply to the email received by the Center usually occurs within 3-7 working days.

EMERGENCIES

Service description and access mode

It may happen that a patient finds himself in an emergency situation in his own area of residence and needs advice from the Center. Urgent situations are considered:

- Serious manifestations of the disease and complications resulting in on-site hospitalization.
- Sudden and serious health complications, not directly related to HHT, in which the doctor managing this complication requires clarification and comparison with the patient's HHT center.

By email: it is advisable to write an email to percorso.hht@policlinicogemelli.it with the details of the emergency indicating in the subject of the email the same "URGENCY".

Outside the established times of the Center and in case of real emergency, the HHT Onlus, through the help line active every day, at any time, can be of support to patients and their families, but without being able to dispense any kind of clinical advice.

Delivery times

Short times

Do not contact the center by email for:

- Information on trials
- Information on research progress
- Information on meetings and conferences
- Contact requests with other patients

For these and other information the patient can:

- Attend the Annual Patient Doctors Conference
- Register with HHT Onlus to receive the quarterly newsletter with all the updates
- Join a regional group of HHT Onlus
- Contact the HHT Onlus Help Line at 333.6159012
- Write via email to HHT Onlus info@hhtonlus.org

Do you have suggestions to improve our service?

Write to sportellomalattierare@policlinicogemelli.it

Contacts

For the patient with HHT: percorso.hht@policlinicogemelli.it

HHT Onlus - Italian Association of Hemorrhagic Hereditary Teleangiectasia
Help Line 333 6159012 Email: info@hhtonlus.org

CEMAD

Centro Malattie Apparato Digerente
Digestive Disease Center

