Hereditary Haemorrhagic Telangiectasia (HHT) Paediatric and Adult Care Pathway

HSE National Rare Diseases Office

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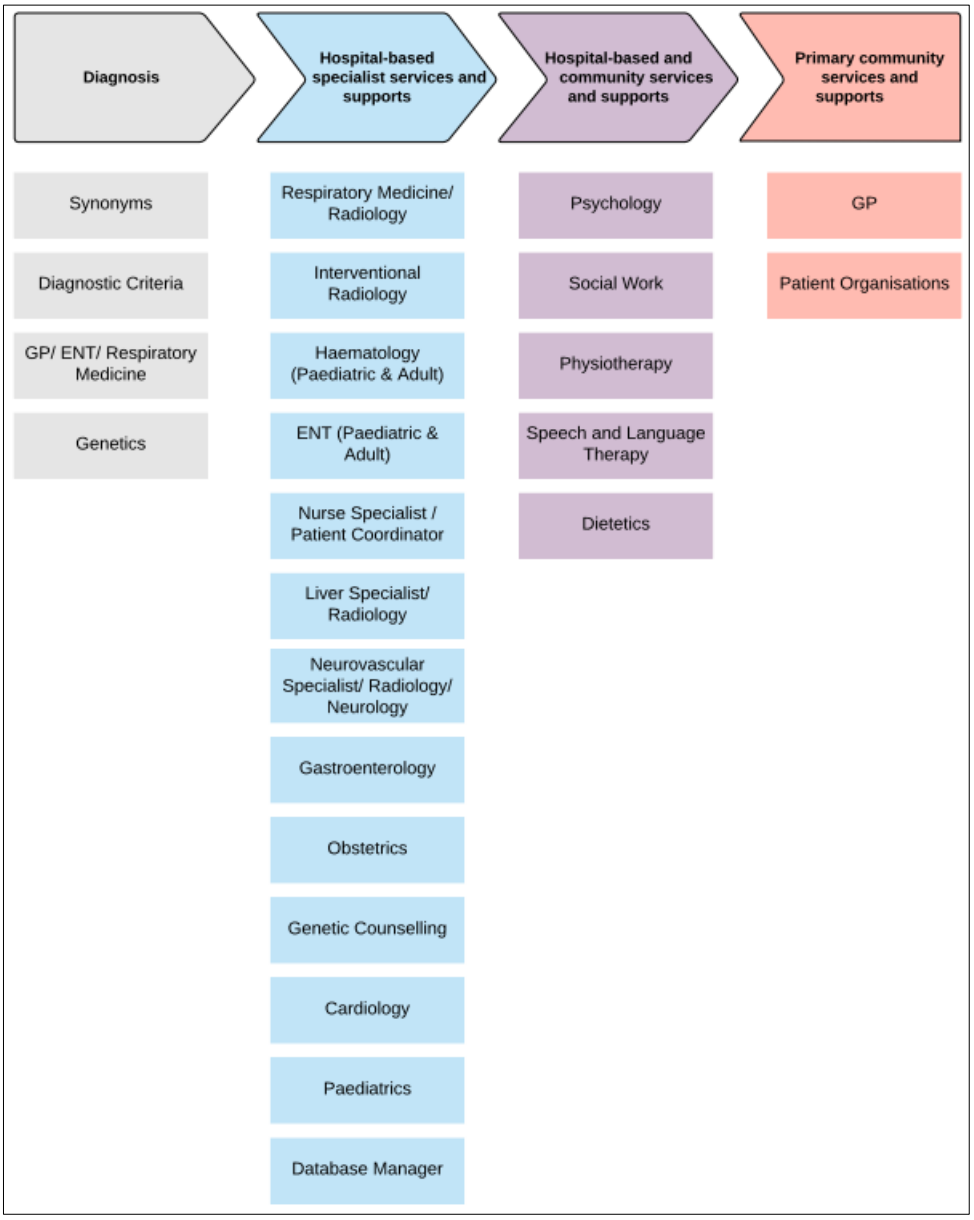

Clinical Characteristics:

Orphacode: 774

Disease Definition: An inherited disorder of angiogenesis characterized by mucocutaneous telangiectases and visceral arteriovenous malformations

Diagnosis	Synonyms	Hereditary Haemorrhagic Telangiectasia (HHT); Osler Weber Rendu Syndrome
Diagnosis	Diagnostic Criteria	Curaçao diagnostic criteria for Hereditary Haemorrhagic Telangiectasia (1998): Diagnosis of HHT: • Definite - 3 criteria present • Possible/ suspected - 2 criteria present • Unlikely - <2 criteria present 1. Epistaxis (nose bleeds): spontaneous and recurrent 2. Telangiectasia: multiple, at characteristic sites (lips, oral cavity, nose, face, fingers) 3. Visceral arterio-venous malformations (AVMs) – pulmonary, cerebral, hepatic, spinal, or gastrointestinal telangiectasia (with or without bleeding) 4. Family history: a first degree relative with HHT according to these criteria
Diagnosis	GP/ ENT/ Respiratory Medicine	Recurrent epistaxis, multiple mucocutaneous telangiectasia, pulmonary arteriovenous malformations (PAVMs), other visceral AVMs, stroke, brain abscesses, iron-deficiency anaemia (otherwise unexplained, esp. after 4-5th. decade of life), family history of same
Diagnosis	Genetics	Pathogenic variants in *ENG*, *ACVRL1* or *SMAD4* genes confirms diagnosis (pathogenic variants in other genes possible; no typical pathogenic variant detected in <10%)
Hospital-based specialist services and supports	Respiratory Medicine/ Radiology	Pulmonary Arteriovenous Malformation (PAVM) screening - Clinical evaluation in childhood (for cyanosis, dyspnoea, clubbing): supine and upright pulse oximetry, chest radiography and/or trans-thoracic echocardiography (TTCE) with agitated saline (bubble echo); Bubble echo post-puberty, pre-pregnancy, post-pregnancy and follow up as indicated. If echo positive, CT thorax (unenhanced usually sufficient). Repeat CTs at intervals according to current guidelines. Antibiotic prophylaxis prior to dental and surgical procedures if Pulmonary Arteriovenous Malformation (PAVM) not definitely excluded. SCUBA-diving must be avoided if Pulmonary Arteriovenous Malformation (PAVM) not definitely excluded
Hospital-based specialist services and supports	Interventional Radiology	Pulmonary Arteriovenous Malformation (PAVM) treatment - transcatheter embolotherapy
Hospital-based specialist services and supports	Haematology (Paediatric & Adult)	Manage iron deficiency anaemia - Oral and/ or intravenous iron supplementation, transfusion, consider risk benefit before using anticoagulant or anti-platelet therapy for other indications, Clinical Trials of anti-angiogenic agents, anti-VEGF (Vascular Endothelial Growth Factor)

Hospital-based specialist services and supports	ENT (Paediatric & Adult)	Assess and treat epistaxis - humidification, topical lubricants, lubricated low-pressure pneumatic packing, endonasal laser, electrical or chemical coagulation (only of benefit in short-term to stop acute bleeding, may exacerbate bleeding long-term), Clinicians and patients may consider a specific ablative therapy based on local expertise, understanding that ablative therapy is a temporizing treatment for epistaxis and perforation of the nasal septum is a known complication of all techniques (Faughan et al, 2020 Second International HHT Guidelines) Septal dermoplasty or nostril closure (only in extreme cases), Nasal artery embolization (only in emergency situation)
Hospital-based specialist services and supports	Nurse Specialist	Patient and family education and support Transition planning Liaison with other health care professionals Link families to patient organisations
Hospital-based specialist services and supports	Liver Specialist/ Radiology	Doppler ultrasound or CT screen for Hepatic Arteriovenous Malformations (HAVMs) if symptom suggest complicated HAVMs - high output cardiac failure, portal hypertension, biliary disease, portosystemic encephalopathy. Screening for asymptomatic hepatic shunts may be offered, but is not standard management, as no treatment is recommended in these circumstances. Treatment – treat as for other causes of high-output cardiac failure, cirrhosis/portal hypertension, biliary ischaemia. Hepatic artery embolization – should be avoided, as it is only a temporizing procedure associated with significant morbidity and mortality. Liver biopsy should be avoided in any patient with proven or suspected HHT. Liver transplantation in highly selected cases
Hospital-based specialist services and supports	Neurovascular Specialist/ Radiology/ Neurology	Asymptomatic screening for Cerebral Arteriovenous Malformations (CAVMs) – contrast-enhanced MRI brain in all patients (once only). If Cerebral Arteriovenous Malformation (CAVM) detected: consider endovascular embolization, neurosurgical resection, radiosurgery. Investigate neurological symptoms (e.g. unexplained headaches, epilepsy or focal neurological deficit) – could be due to complications of Pulmonary Arteriovenous Malformation (PAVM) (paradoxical embolization, stroke, brain abscess). Migraine incidence increased in patients with Pulmonary Arteriovenous Malformations (PAVMs)

Hospital-based specialist services and supports	Gastroenterology	GI bleeding from telangiectasia occurs in 25-33% of HHT patients especially from 5th decade of life on. Screen by directed endoscopy if GI bleed suspected (e.g. anaemia disproportionate to epistaxis). Purpose of endoscopy is principally to exclude other significant cause of bleeding. Treatment – iron supplementation, hormone therapy, antifibrinolytics, endoscopic cauterisation of telangiectasias. Treatment largely ineffective long-term – iron supplementation is mainstay Screen for polyposis coli (SMAD4 gene only – associated with juvenile polyposis and early colorectal cancer) and GI cancer: Colonoscopy from age 15y every 3 years (if no polyps found), or every year, with upper GI endoscopy (if colonic polyps are found). Other patients with non-SMAD4 HHT should be screened for colorectal cancer following general population guidelines
Hospital-based specialist services and supports	Obstetrics	High-risk pregnancy management. Contrast-enhanced spinal MRI if epidural anaesthesia planned - used in some centres, but is not supported by expert guidelines, and epidural anaesthesia should not be withheld in women with HHT
Hospital-based specialist services and supports	Genetic Counselling	Review of genetic results (inheritance pattern is autosomal dominant), recurrence risk, identification of at-risk relatives, cascade testing (if familial pathogenic variant identified), reproductive options
Hospital-based specialist services and supports	Cardiology	Screen for aortic dilation/ aneurysm (SMAD4 mutation only)
Hospital-based specialist services and supports	Paediatrics	Screen for (and treat where necessary) Pulmonary Arteriovenous Malformations (PAVMs) and Cerebral Arteriovenous Malformations (CAVMs) (as outlined in other sections above). Manage epistaxis as above
Hospital-based specialist Services and supports	Database Manager & Patient Coordinator	Patient contact and advice Screening protocol management Registry Create and maintain database of patients attending service Record patient biographical, clinical and research data Audit and quality improvement Ensure minimum data set standards for ERN registry
Hospital-based and community	Psychology	Support for chronic condition

services and supports		
Hospital-based and community services and supports	Social Work	Psychosocial support: Assess social and family supports, safeguarding Link with community supports as required e.g., GP, Public Health Nurse, Primary Care Social Worker, Local authority Social Worker, Mental Health Social Worker, Disability Social Worker, TUSLA Offer 1-1 counselling or GP referral to Counselling in primary care (CIPC) www.hse.ie/eng/services/list/4/mental-health-services/counsellingpc/ Financial support (as required): Patient advocacy, support applications for Medical Card, Disability Allowance, Supplementary Welfare allowance, Exceptional Needs payment, Long-term illness card, direct to Citizens' information www.citizensinformation.ie/ Housing and/or mobility issues: Advocacy and support Home Care Packages: if issues with activities of daily living - arrange application for inpatients, liaise with Public Health Nurse to arrange for out-patients Respite Care: liaise with public health nurse or community disability services to arrange Employment issues: Link to Intreo public employment and EmployAbility services www.gov.ie/en/campaigns/fb84c0-intreo/ www.gov.ie/en/service/8578c4-access-the-employability-service/
Hospital-based and community services and supports	Physiotherapy	Stroke Rehab – Neurorehabilitation team Lung Transplantation Post-op management - Cardiothoracic / Lung Transplant team
Hospital-based and community services and supports	Speech and Language Therapy	Stroke or respiratory issues - Assessment, support and intervention for speech, language and communication needs (SLCN) and/or feeding, eating, drinking & swallowing (FEDS) skills based on individual needs and priorities Patient advocacy, patient / staff education, training and awareness
Hospital-based and community services and supports	Occupational Therapy	Stroke or Respiratory issues - Assessment and intervention for difficulties participating in activities of daily living.

Hospital-based and community services and supports	Dietetics	Intervention is focused on improving performance and reducing the risk of deterioration in these abilities. Areas of assessment and intervention include self-care, productivity, and leisure. Iron-deficiency anaemia – advise on healthy balanced diet with focus on increasing dietary iron intake Stroke - artificial nutrition support and/or educate on texture modified diets for dysphagia management and provide dietary counselling for secondary prevention of stroke Nutrition assessment where malnutrition risk identified with nutrition screening, to ensure adequate nutritional and hydration status
Primary / community services and supports	GP	Screen for iron deficiency anaemia (due to blood loss from GI telangiectasia) in asymptomatic individuals with HHT > 50 y.o. Younger patients with significant blood loss from epistaxis, or symptomatic patients at any age, should also be checked for anaemia. Refer to National HHT Centre for screening for Pulmonary Arteriovenous Malformations (PAVMs) and Cerebral Arteriovenous Malformations (CAVMs) Management of inter-current conditions Co-ordination of local services and supports Refer/communicate across services
Primary / community services and supports	Patient Organisations	Advocacy, support, information: HHT Ireland (www.hhtireland.org) Rare Ireland Family Support Network (www.rareireland.ie) Rare Diseases Ireland (www.rdi.ie)

Clinical Leads:

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