Affects >85,000 European citizens

• Most cases undiagnosed

• Causes of preventable
  - Anaemia/blood transfusions
  - Brain abscess (1 in 16 cases at VASCERN centres)
  - Ischaemic strokes (>1 in 8 referrals)
  - Heart failure
  - Maternal death in pregnancy (1% of pregnancies)
  - Venous thromboemboli (VTE)

• Screen for
  - PAVMs, patient pathway
  - VASCERN/13
  - Prioritised antiangiogenic drugs & thromboemboli
  - Transfusions
  - Group
  - Buscarini
  - HHT Drug Registry
  - (1)
  - Intervention
  - Deficiency
  - Group on behalf of Knowledge (5 minute videos)
  - 100
  - Pregnancy - (>100 patient responses in review (VTE)
  - Separate questions for patients, to receive nosebleed advice in writing (VTE)
  - Failure (VASCERN and multisystemic pregnancy advice)
  - Cause of preventable
  - Vascular screening in March 2017

VASCERN-HHT Year 1 Impact (established 9th March 2017)

HHT Patient Pathway

Top 7 HHT Priorities

1. Anaemia
2. AVMs
3. Bleeding
4. Children
5. Hereditary
6. Medications
7. Informed care

HHT Drug Registry

• Separate questions for patients, scientists and doctors
• Prioritised antiangiogenic drugs
• Thalidomide and Bevacizumab
(BZB, Avastin)
>100 patient responses in review

HHT Drug Registry

• PAVM screen
• Antibiotics/other advice
• AI Rx + advice
• PA Rx + advice
• Pregnancy advice
• Other treatment
• Other advice
• Family elements
• Refer to (inter)national patient support groups

VASCERN-HHT Year 1 Outputs

• Patient pathway defined and revised
• Priorities defined by 61 patients/professionals
• Drug Registry designed and first data published
• 5 Outcome Measures defined and applied
• 11 cross-border clinical cases discussed
• 1 Guideline (Clinical Statement) produced*
• 13 Do’s and Don’ts for General Care produced
• 8 research projects with >1 VASCERN HHT Centre
• 2 YouTube Videos (published online 2018)
• 2 Educational workshops (published 2017)

in addition to VASCERN members’ existing activities, e.g.
• 3 HHT clinical trials completed/in progress (Dupuis Girod)
• 17 PubMed-publications; >40 research projects

HHT Do’s and Don’ts

1. Physical Activity
2. Breast feeding
3. Contraindicated medications
4. Antiplatelets/anticoagulants
5. Venous thromboemboli (VTE)
6. Haemorrhagic stroke
7. Brain abscesses:
8. Heart failure
9. Kidney failure
10. Multiple traumatic injuries
11. Bronchoscopies
12. Aortic dissection
13. Pneumothorax

For publication, March 2018
>15 more in preparation, 2018

Core Values:
• “Patient first, Safety first”
• “Combining evidence and experience to work together”

Other 2017-8 Formal HHT Education

YouTube Minipills of Knowledge (5 minute videos)
An overview of Hereditary Haemorrhagic Telangiectasia (SHOVLIN)
https://youtu.be/z2gALD8xSNE

What an ENT doctor needs to know about HHT and why
(KJELDSEN) https://youtu.be/k2V9q87NHe

12th HHT International Conference Dubrovnik, June 2017
Workshop on Management of Hepatic AVMs (Dupuis Girod & Buscarini),
Workshop on Inflammation, Immunity and Injury in HHT (SHOVLIN & BOTELLA)
VASCERN HHT PRIORITY EVALUATIONS 2016-2017
(SHOVLIN ET AL ON BEHALF OF THE VASCERN-HHT WORKING GROUP)
VASCERN HHT Survey 2: drug registry part 1
(BUSCARINI ET AL ON BEHALF OF THE VASCERN-HHT WORKING GROUP)

Pan-VASCERN-specific investment
(excludes local HCP activity eg patient reviews, ERN application)
>500 hours by Board (Shovlin, Dupuis Girod, Buscarini)
>150 hours by the Leads of the 5 other HHT WG Centres
(Note the still very small number of clinical experts)

For further information, please contact:
VASCERN HHT CHAIR
Claire L. Shovlin MB BChir, PhD FRCP (c.shovlin@imperial.ac.uk)
Professor of Practice (Clinical and Molecular Medicine), Imperial College London
https://vascern.eu/expertise/rare-diseases-wgs/hht-wg/