

HHTIC London 2011 Research Newsletter

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HHTIC London, run by Dr Claire Shovlin, is an internationally-renowned centre for hereditary haemorrhagic telangiectasia (HHT) and pulmonary AVMs. These surprisingly common vascular disorders can cause significant ill health and lifestyle limitations. Our goal is to reduce death and disability in these under-recognised conditions, by asking the questions that matter most to patients.

1) PREVENTION OF COMPLICATIONS FROM PULMONARY AVMS (PAVMs)

PAVMs affect approximately 1 in 2 people with HHT, and also occur without HHT. A non surgical treatment by embolization was introduced to the UK by Professor David Allison at Hammersmith Hospital, and has been subsequently performed here by Dr James Jackson and Dr Ali Alsafi. Dr Jackson reported the safety and benefits of embolization in sequential series, and worked closely with the company manufacturers of embolization devices to improve device safety and develop new devices.

Examples of papers include:

JACKSON ET AL, COR ET VASA, 1990; 32: 191-196

DUTTON ET AL, AM J ROENT 1995; 165: 1119-1125

GUPTA ET AL, AM J ROENT. 2002; 179:347-55.

HART ET AL, EUR RADIOL. 2010:2663-70

Two papers specifically address pulmonary AVMs and pulmonary hypertension:

SHOVLIN ET AL, EUR RESP J 2008; 32:162-9

SHOVLIN, GIBBS AND JACKSON, EUR RESPIR REV 2009:18:111, 4-6

Most people with PAVMs do not know they have PAVMs, yet are still at risk of complications. From 1999, Dr Claire Shovlin analysed the differences between PAVM patients who had complications and those that did not. In the key paper of the 1995-2005/6 series, we were able to show that silent small PAVMs are an important under-recognised cause of stroke; that embolization treatment prevents strokes; but that most 'at-risk' individuals were unaware of their diagnosis of PAVMs and HHT when they had their strokes. The manuscript provided evidence enabling the Chairman of the NICE Dental Committee to work with us, issuing guidance specifically for PAVM patients.

SHOVLIN ET AL, THORAX 2008; 63; 259-66

SHOVLIN ET AL BRIT DENTAL J 2008 Nov 205(10):531-3

If any of you with PAVMs are finding doctors advising you that you do not need any treatment or do not need antibiotics before going to the dentist, please check that they have read these last 2 articles before finalising their advice to you.

2) CLINICAL DIAGNOSIS OF HHT

Dr Shovlin chaired the committee that developed the HHT diagnosis guidelines known as the Curaçao Criteria. Based on nosebleeds, blood spots in specific places, larger arteriovenous malformations (AVMs) and family history, these allow us to diagnose HHT and have been referenced by others on more than 500 occasions.

SHOVLIN ET AL, AM J HUM GENET 2000:91; 65-66

3) THROMBOSIS AND HHT

Given HHT is a bleeding disorder, it may surprise you to realise that people with HHT can still have blood clots (venous thromboemboli, VTE). In another key paper from the 1995-2005/6 series, we were able to show that people with HHT do have VTEs from time to time. This is important in the advice that doctors give you and we are following on with further studies which we hope to publish soon.

SHOVLIN ET AL, THROMB HAEMOST. 2007;98:1031-7

4) PREGNANCY AND HHT

For most women with HHT, pregnancy proceeds safely but there are occasional individuals where events go tragically wrong and mothers may die. We raised this in a publication in 1995, and in the 1999-2005 series, Dr Shovlin tried to ask why did this happen, and could we prevent any of these deaths? In the study of 484 HHT pregnancies published in 2008, and headlined by Reuters, we were able to show it was safer to know about HHT and PAVMs before pregnancy, and provided obstetricians with guidance for extra steps to make pregnancy safer. **This is important for all HHT families**

SHOVLIN ET AL, BJOG 2008 AUG;115(9):1108-15.

5) PUTTING IT ALL TOGETHER

To help doctors provide you with all the right information in areas outside of their own speciality, we summarise information in review articles. The two latest articles are:

GOVANI AND SHOVLIN, EUR J HUM GENET. 2009 JUL;17(7):860-71

SHOVLIN. BLOOD REV. 2010;24:203--19.

Imperial 2011

*2021 UPDATE: All information remains valid, including the Curaçao Criteria now cited > 1600 times. Please also see references in our 2020 Newsletter, especially the outputs from the British Thoracic Society Pulmonary AVM Clinical Statement Group (SHOVLIN ET AL THORAX 2017) and VASCERN HHT via <https://vascern.eu>, especially their explanation of WHY antibiotic prophylaxis is so important for people with PAVMs:

PREVENTION OF SERIOUS INFECTIONS IN HEREDITARY HEMORRHAGIC TELANGIECTASIA: ROLES FOR PROPHYLACTIC ANTIBIOTICS, THE PULMONARY CAPILLARIES-BUT NOT VACCINATION.

SHOVLIN C, BAMFORD K, SABBA C, MAGER HJ, KJELDSEN A, DROEGE F, BUSCARINI E, DUPUIS-GIROD S; VASCERN HHT. HAEMATOLOGICA. 2019 FEB;104(2):e85-e86.