9.10 Hereditary Haemorrhagic Telangiectasia – an update on national numbers

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Since 2003, the Mercy University Hospital Cork has been the national centre for Hereditary Haemorrhagic Telangectasia (HHT) in Ireland. The centre's mission is to identify as many affected individuals and families as possible and offer screening for and management of the condition's many manifestations. Complications include vascular malformations (lung, brain, liver typically), epistaxis and telangectasia. Screening consists of clinical history, genetic testing, bubble study echocardiography, CT-Thorax and MRI Brain. The Mercy University Hospital has assessed 704 patients thus far through tracking index cases and their relatives, with 298 confirmed diagnoses of HHT.

It is estimated that only 10% of affected individuals with HHT have been formally diagnosed globally. We show that a large proportion of patient with HHT develop severe complications (epistaxis (360 patients) and pulmonary AVMs (300 positive bubble study echocardiograms) being the most common complications seen).

Conflict of Interest: *None to declare*