Pulmonary Hypertension in the setting of Hereditary Hemorrhagic Telangiectasia

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Background:
Hereditary Hemorrhagic Telangiectasia (HHT) is a heritable disease characterized by mucocutaneous telangiectasias, spontaneous or recurrent epistaxis, and arteriovenous malformations which can be associated with shunting or increased flow. A subset of patients develops pulmonary hypertension (PH). We aimed to determine prognostic factors of mortality in patients with HHT.

Methods:
Single center cohort study of all patients with confirmed HHT who underwent right heart catheterization and transthoracic 2D echocardiography for suspected PH between 1/1/1995-9/1/2013. All patients met at least 3 of 4 Curacao Criteria. Mean pulmonary artery pressure ≥ 25 mmHg defined PH. Data are reported as mean ± standard deviation.

Results:
Of 38 patients with HHT who underwent right heart catheterization and transthoracic echocardiography, 28 patients (74%) had PH. Mean age of these 28 patients was 56 ± 17 years. NT-BNP was 1173 ± 1901 pg/ml, and hemoglobin was 11.3 ± 2.6 g/dL. Thirteen patients (46%) were functional class III/IV. Pulmonary arteriovenous malformations, visceral arteriovenous malformations, and cerebral malformations were present in 11 (39%), 22 (79%), and 2 (7%) respectively. Right heart catheterization demonstrated a right atrial pressure (RAP) of 15 ± 10 mmHg, mean pulmonary artery pressure (PAP) of 41 ± 11 mmHg, and a pulmonary capillary wedge pressure of 17 ± 10 mmHg. Mean pulmonary vascular resistance was 4.5 ± 4.2 Wood Units. Echocardiography demonstrated an ejection fraction of 68 ± 9%, RVSP of 65 ± 23 mmHg, and RAP of 13 ± 5 mmHg. Moderate/severe right ventricular enlargement was present in 16 (57%) patients while 14 (50%) had abnormal right ventricular function including 9 (32%) patients with moderate to severe dysfunction. Adverse predictors of survival included: PH (p=0.06), functional class III/IV (p=0.003), increased RAP (p=0.006), moderate to severe RV enlargement (p=0.09), moderate to severe decrease in RV function (p=0.006).

Conclusion:
In patients with hereditary hemorrhagic telangiectasia, PH is common and associated with worse survival. Prognostic indicators include the presence of PH, functional class, increased RAP, right ventricular enlargement, and elevated RVSP.

Type: Clinical Science