Title of Abstract: Multiple peripheral pulmonary artery stenoses and moyamoya disease: a possible novel pulmonary and systemic arteriopathy mimicking pulmonary arterial hypertension


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BACKGROUND: Moyamoya disease is a chronic, occlusive, cerebrovascular disorder of unknown pathogenesis. Recently, a few cases of the disease have been reported with extracranial arterial lesions, suggesting diffuse arteriopathy such as fibromuscular dysplasia as the underlying cause. Extracranial arterial lesions include pulmonary hypertension: idiopathic pulmonary arterial hypertension (IPAH), chronic thromboembolic pulmonary hypertension (CTEPH) and multiple peripheral pulmonary artery stenoses (PPS). We present two cases with moyamoya disease and multiple peripheral PPS.

CASE PRESENTATIONS: First case, a 19-year-old Japanese man, was diagnosed with moyamoya disease at the age of 9 years. On a routine follow-up visit, he was found to have systemic hypertension caused by renal artery stenosis and pulmonary hypertension at the age of 13. Pulmonary angiogram revealed multiple peripheral PPS resulting in high central pulmonary artery pressure. Second case, a 14-year-old Japanese boy, was referred to our hospital for investigation of pulmonary hypertension, initially to be considered idiopathic. We suspected PPS from systolic murmur all over the lung fields and CT supported the presence of PPS. Right heart catheterization using micro pressure wire revealed significant pressure gradients in several pulmonary arteries and angiogram confirmed multiple peripheral PPS causing central pulmonary hypertension. From experience of the first case, we took the coexistence of moyamoya disease into consideration although he did not have past history of neurological symptom. Brain MRI revealed occlusion of bilateral internal carotid arteries and moyamoya vessels, resulting in the diagnosis of moyamoya disease. The second case did not have systemic hypertension or renal artery stenosis.

CONCLUSIONS: Although multiple peripheral PPS and moyamoya disease is a very rare combination, it is clinically important as a possible novel pulmonary and systemic arteriopathy and may have been misdiagnosed as IPAH or CTEPH. Systolic murmur on lung field can be a clue to recognition of PPS and pulmonary angiogram is useful to confirm it. We plan to clarify the pathophysiology of this unique combination using genomic technology.

TYPE: Case Study