

**FATAL LUNG HEMORRHAGE ASSOCIATED WITH PULMONARY
ARTERIOVENOUS MALFORMATIONS IN CHILDREN**

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Purpose: To report the investigation of the cause of massive of hemoptysis that led to death of a 9-year-old girl with HHT. To report the screening results of her family, mutation analysis, and to review the world literature on this topic.

Methods: The postmortem examination of the 9-year-old girl was reviewed. Family members were screened by pulse oximetry, contrast echocardiography (CE), and brain MRI. Patients with positive CE underwent thin section (5mm) unenhanced spiral CT of the lung. Mutation analysis was performed by multiplex PCR. Symptoms of PAVM from 130 pediatric patients reported in the world literature were tabulated.

Results: At postmortem examination, the girl had a large right upper lobe PAVM and grade IV and V Heath-Edwards changes of pulmonary hypertension. 17/19 family members with HHT were screened and 9/17 (53%) had positive CE. 8/9 with positive CE had normal pulse oximetry and 6/9 had an abnormal unenhanced spiral CT. 2/9 had large PAVM (artery>3mm) and were treated by embolotherapy. These individuals had normal pulmonary artery pressures (PAP). Mutation analysis is pending.

7 children (ages 1-18), from a database of 130 with PAVM reported, developed hemoptysis and in 4/7 it was lethal. 3/7 had normal PAP and PAP was not reported in the remainder.

Conclusions: While pulmonary hypertension aggravated the hemoptysis of the patient reported, our review indicates that, in children, who do not have concomitant pulmonary hypertension, large PAVM can cause morbidity and mortality. The high prevalence of PAVM in this family suggests that they may be the first family reported with an endoglin mutation and pulmonary hypertension.