**P1-109**

## A case of pulmonary arteriovenous malformation as first sign of hereditary haemorragic teleangectasia.

*De Sando E. (1), Codazzi A.C.(2), Grassidonio S. (1), Danesino C. (3), Pagella F. (4), Spinozzi G. (4), Quaretti P. (5), Marseglia G.L. (1), Mannarino S. (2)*
*Department of Pediatrics, Foundation IRCCS Policlinico San Matteo, University of Pavia, Italy (1);*
*Unit of Pediatric Cardiology, Department of the Mother and Child Health, IRCCS Policlinico San Matteo Foundation, Pavia, Italy (2);*
*Department of Molecular Medicine, General Biology, and Medical Genetics Unit, University of Pavia, Pavia, Italy (3);*
*Department of Otorhinolaryngology, University of Pavia, IRCCS Policlinico San Matteo Foundation, Pavia, Italy (4);*
*Unit of Interventional Radiology, IRCCS Policlinico San Matteo Foundation Pavia, Pavia, Italy (5)*

We report the case of an 8-year-old girl who came to our Pediatric Department for urticaria and dermatitis. Blood exams indicated increase of haemoglobin and haematocrit (Hb 17.2g/dl, Hct 53.6%); physical examination showed perioral cyanosis, digital clubbing and reduced oxygen saturation (86-90%). Electrocardiogram and conventional echocardiogram were normal. Chest X-ray showed a parenchymal consolidation with vascular ectasias at right lung, evocative of pulmonary arteriovenous malformations (PAVM). Transthoracic contrast echocardiography showed an extensive opacification of the left ventricle without outlining the endocardium (grade 3). Chest CT demonstrated a large high-flow PAVM (3.5 x 3.7 x 5 cm) at the right lower lobe and three small PAVM at left lower lobe. Therefore the patient was submitted to embolization of the large PAVM with an improvement of saturation until 100%. An accurate physical examination showed mucocutaneous telangectasias. Because of the presence of 2/4 Curacao criteria, we suspected hereditary haemorragic teleangectasia (HHT), confirmed by the presence of ENG gene mutation. Two years after the embolization, the reopening of the large PAVM required a lobectomy of the right lower lobe. Actually she is in a state of general health with a good oxygen saturation.
HHT is a rare systemic fibrovascular autosomal dominant dysplasia. Its prevalence is currently estimated at one in 5,000 to 8,000. Mutations in ENG and ALK1 genes have been reported to cause up to 85% of HHT. The signs and symptoms of HHT are nonspecific and are extremely variable within families. HHT is often difficult to diagnose on the basis of history and physical examination alone, especially in children. In our case, the presence of PAVM led us to think about HTT, even without nosebleeds and family history that are usually the most common criteria. Mutations in ENG gene are more frequently associated with PAVM, so these patients need of a careful follow up for the risk of progression of vascular disease.