RESEARCH LETTER

Positional oximetry to screen for pulmonary arteriovenous malformations in HHT

ABSTRACT
Orthodeoxia has been described in patients with hereditary haemorrhagic telangiectasia (HHT) and pulmonary arteriovenous malformations (PAVM), but its usefulness in screening children for PAVMs is unknown. We show here that while oximetry is overall lower in children with significant PAVMs, supine and upright oximetry is of no additional diagnostic value indicating that positional oximetry is not a useful screening test for PAVMs.

To the editor:
Screening for pulmonary arteriovenous malformations (PAVM) is performed in patients with hereditary haemorrhagic telangiectasia (HHT) to avoid complications, such as brain abscesses and stroke. While the clinical usefulness of screening is more controversial in children, international guidelines do recommend screening regardless of age. Orthodeoxia refers to a decrease in oxygen saturation when changing from the supine to upright position. Individual reports have described orthodeoxia as a finding that led to the diagnosis of PAVMs in HHT, but the diagnostic yield of using positional oximetry as a screening test for PAVMs in children is unknown. Studies in adults have shown upright oximetry to be a better predictor of right to left shunting through pulmonary AVMs than supine oximetry. Here, we aimed to assess the utility of pulse oximetry in the upright and supine position as a screening tool for PAVMs in children with HHT.

We retrospectively extracted data from the paediatric HHT database at the Hospital for Sick Children for the time period of 1997–2012. Screening procedures for pulmonary AVMs included an agitated saline echocardiogram and a CT thorax until 2009, when based on the finding that the echocardiogram had 100% sensitivity for detection of PAVMs, CT scanning was limited to patients with a positive echocardiogram. Genetic testing for HHT mutations was performed by sequencing of the endoglin and ALK-1 gene as well as the SMAD-4 gene in patients with negative testing results. Oxygen saturation measurements in both supine and upright position were performed as part of the clinical assessment after a period of at least 5 min of rest for each position.

Data for 209 children were included. Based on the screening procedure results, patients were divided into four groups (table 1): 116 had neither a clinically nor genetically confirmed diagnosis of HHT and were considered controls. Of 33 patients with PAVMs, 14 were considered to be either clinically significant or associated with feeding vessel > 2 mm and resulted in an interventional procedure (embolisation); the remaining 19 patients had small PAVMs with feeding vessel < 2 mm on chest CT that were followed clinically, and 60 had a genetically confirmed diagnosis without any evidence of PAVMs. Patients in group 1 (PAVMs requiring embolisation) had significant lower oxygen saturations in both body positions compared to any of the other groups (p < 0.01; ANOVA). There were no significant differences in upright versus supine oxygen saturations in any of the groups (table 1). Oxygen saturation decreased in one patient in group 1 from 81% in the supine position to 70% upright; no individual in any other group had a positional change in oxygen saturation of more than 2%. These data indicate that oximetry differentiates children with larger PAVMs from those with smaller PAVMs as well as children without PAVMs. On the other hand, positional oximetry has a low diagnostic yield in paediatric HHT. Therefore oximetry, but not positional oximetry, is a suitable screening test to assess for PAVM in children with HHT.

Susan Carpenter, Janina Josepovic, Giuseppe Latino, Felix Ratjen

1Division of Respiratory Medicine, Department of Paediatrics, The Hospital for Sick Children, Toronto, Canada
2Physiology & Experimental Medicine, Research Institute, The Hospital for Sick Children, Toronto, Canada
3University of Toronto, Toronto, Canada

Correspondence to Professor Felix Ratjen, Division of Respiratory Medicine, Department of Paediatrics, Rm. 4534, The Hospital for Sick Children, 555 University Avenue, Toronto, Ontario, Canada MSG 1X8; felix.ratjen@sickkids.ca

Contributors SC, GAL, JJ, FR: were involved in data collection, data analysis and writing of the manuscript.

Competing interests None.

Ethics approval REB SickKids Hospital.

Provenance and peer review Not commissioned; externally peer reviewed.


Received 12 February 2014

Accepted 13 March 2014


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Thorax published online April 4, 2014

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