








Genetic counselling and testing in pulmonary arterial hypertension: a consensus statement on behalf of the International Consortium for Genetic Studies in PAH

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Shareable abstract (@ERSpublications)

Idiopathic, anorexigen-induced, congenital heart disease-associated and heritable PAH, and pulmonary veno-occlusive disease patients should be offered genetic counselling and testing with a gene panel including all disease genes for the condition <https://bit.ly/3ga3HEc>

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Abstract

Pulmonary arterial hypertension (PAH) is a rare disease that can be caused by (likely) pathogenic germline genomic variants. In addition to the most prevalent disease gene, *BMPR2* (bone morphogenetic protein receptor 2), several genes, some belonging to distinct functional classes, are also now known to predispose to the development of PAH. As a consequence, specialist and non-specialist clinicians and healthcare professionals are increasingly faced with a range of questions regarding the need for, approaches to and benefits/risks of genetic testing for PAH patients and/or related family members. We provide a consensus-based approach to recommendations for genetic counselling and assessment of current best practice for disease gene testing. We provide a framework and the type of information to be provided to patients and relatives through the process of genetic counselling, and describe the presently known disease causal genes to be analysed. Benefits of including molecular genetic testing within the management protocol of patients with PAH include the identification of individuals misclassified by other diagnostic approaches, the optimisation of phenotypic characterisation for aggregation of outcome data, including in clinical trials, and importantly through cascade screening, the detection of healthy causal variant carriers, to whom regular assessment should be offered.

