

# Genetics and PAH Research findings



In February 2020, the Pulmonary Hypertension Association (PHA UK) conducted an online survey to ascertain what people with Pulmonary Arterial Hypertension (PAH) understand about the role of genetics in PAH, and to identify any information gaps. The survey was conducted in collaboration with *Professor Nick Morrell*, British Heart Foundation Professor of Cardiopulmonary Medicine at the University of Cambridge.

## HOW THE SURVEY WAS CARRIED OUT: METHOD AND RESPONSES

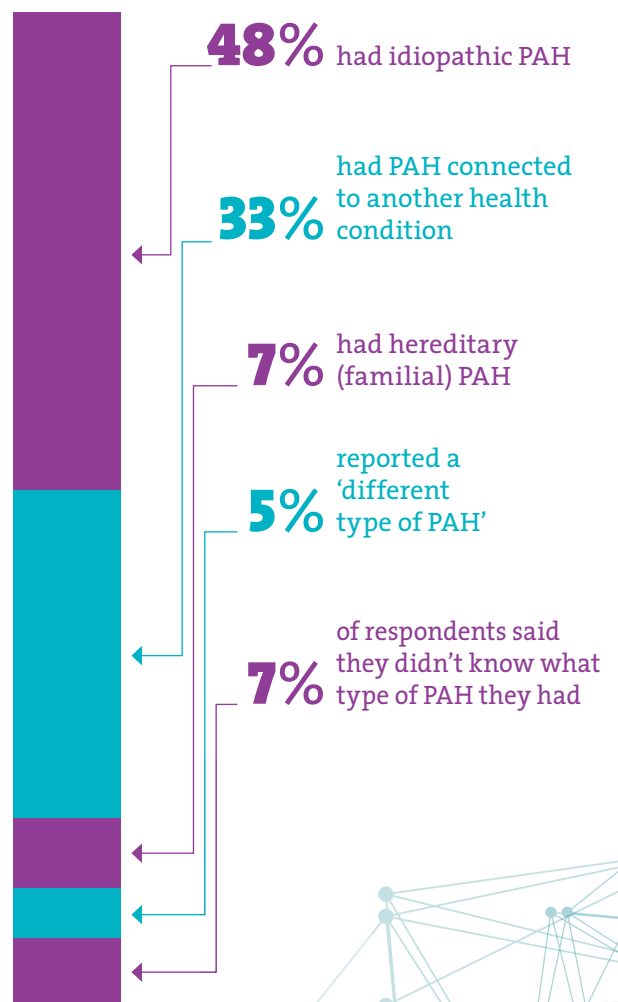
Online survey tool SurveyMonkey was used to carry out the research. Participants were recruited via the PHA UK's social media channels and via an e-newsletter.

The survey consisted of 11 questions, with space for additional comments at the end. It was open for two weeks and only those with a diagnosis of Pulmonary Arterial Hypertension (PAH) were asked to respond.



**75%**  
were female

**25%**  
were male



# The findings

## AWARENESS AND UNDERSTANDING

**50%** of respondents were not aware that PAH can be linked with a fault in genes.



**40%** of respondents have had a discussion with a healthcare professional about the links between genetics and PAH, and the research and studies around it.



*Of those people, just 39% said they understood what was being discussed.*

## INFORMATION NEEDS

**78%** feel there is not enough information available to them about the links between genetics and PAH, and the research and studies around it



**98%** of respondents said they would find clear and accessible information produced by the PHA UK vital, very useful, or useful. *(31% vital, 49% very useful, 18% useful)*



## GENETIC TESTING

**74%** respondents said they would want to be referred for genetic testing if they knew there was a chance of their PAH being caused by a faulty gene. *20% said they 'don't know'*



**80%** of respondents said that if they found out they had a faulty gene causing PAH, they would want their family members to be offered genetic testing. *19% said they 'don't know'*



## WHAT NEXT?

■ ■ *The results of this survey show a strong desire amongst people affected by PAH to understand more about this important subject. There is an obvious need for clear and accessible information. The PHA UK will now work with leading UK experts to produce resources that meet this need and that can be shared with family and friends. We would like to thank everyone who took part in our survey.* ■ ■

Iain Armstrong  
Chair, PHA UK



■ ■ *We have known for some time that there is often a genetic cause for PAH, especially if more than one individual is affected in a family. This study is the first of its kind in a large patient population, and I'm delighted that the PHA UK has carried it out. The results show just how important the issue is to people affected by the condition. Ongoing research by experts is showing the importance of genetics in pulmonary arterial hypertension. It's important that more work is carried out to engage the patient population and their families to ensure the best care and management - for both the present and future.* ■ ■

Professor Nick Morrell  
British Heart  
Foundation Professor  
of Cardiopulmonary  
Medicine



If you have any questions about this survey, please contact the PHA UK on 01709 761450 or [media@phauk.org](mailto:media@phauk.org)

