

PVRI International Consortium for Genetic Studies in PAH (PAH-ICON) Report 2021



Report by Nick Morrell

The International Consortium for Genetic Studies in PAH (PAH-ICON) continues to go from strength to strength with regular zoom meetings, and this year included a new series of Task Force webinars.

These webinars were initiated when the annual face-to-face symposium was suspended due to the ongoing COVID-19 pandemic and have proved to be a great success and will likely continue into the future.

Each webinar focused on a particular PAH-ICON Task Force with the first session, hosted by Professor Harm-Jan Bogaard, covering healthy carriers of BMPR2 mutations, including screening and research studies.

A further session focused on TBX4 mutations included a family member introducing a new patient organisation, **TBX4Life: developing an international family-driven initiative and research focus on TBX4.**

The sessions were organised in conjunction with the PVRI, were well attended and created many interesting discussions and leads for future genetic collaborations. It is only with the continued international collaborations and the set-up of PAH-ICON Task Forces, which are capable of generating thousands of samples and phenotype data from patients with idiopathic and heritable PAH, that we can learn more about the complicated genetic make-up of PAH.

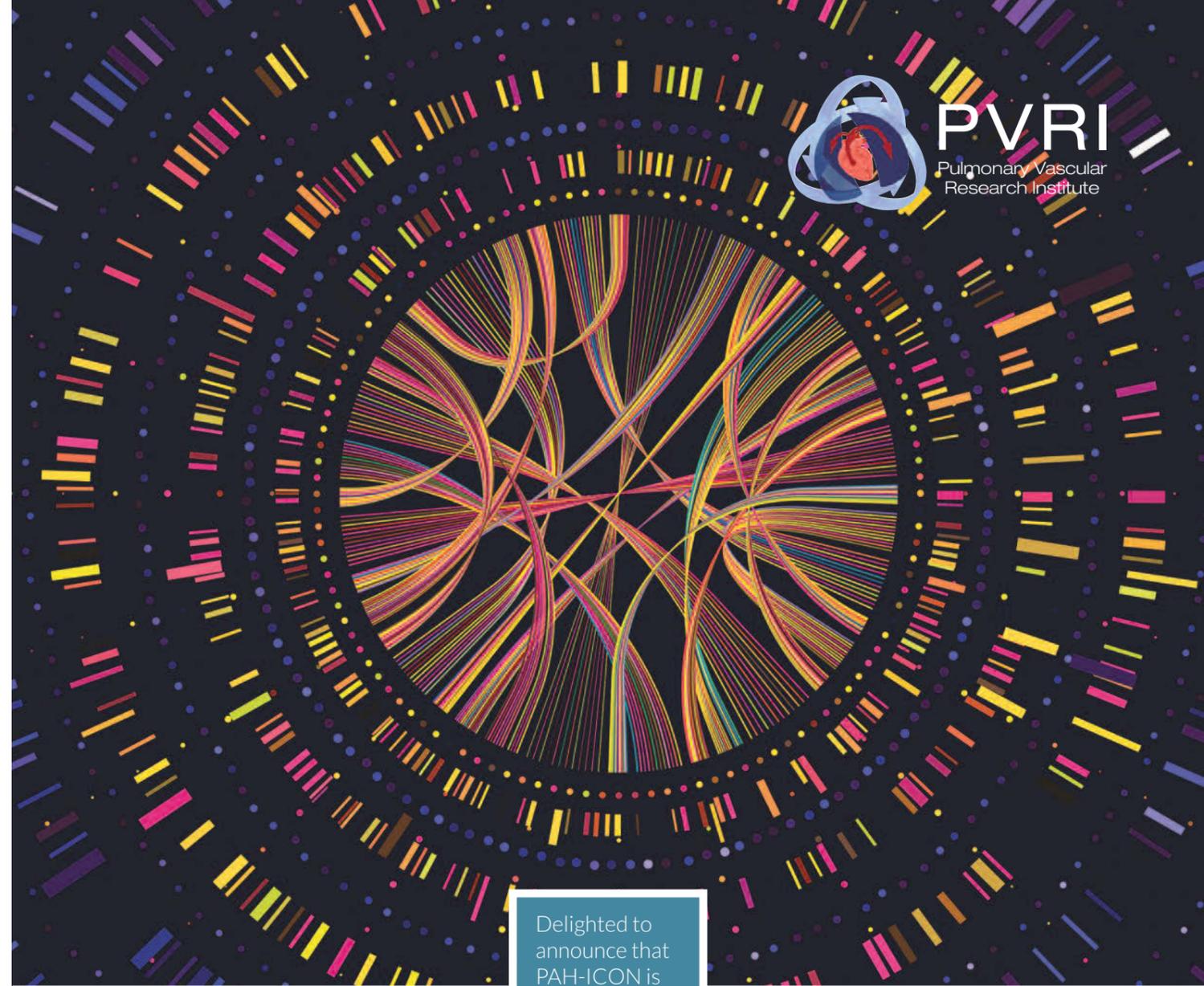
We aim to be able to receive samples to our PAH-ICON biorepository in the coming year to enable the collation of international samples to allow the identification of ultra-rare PAH variants.

PAH-ICON also goes beyond genetics, with Task Forces on metabolomics, proteomics and epigenetics.

We are delighted to announce that PAH-ICON is now formally affiliated with PVRI. A Memorandum of Understanding has been signed between both parties confirming that PAH-ICON operates under the umbrella of the PVRI as a fully established Task Force. This will ensure our goal of furthering knowledge of the genetic architecture underlying PAH.

In 2022, our international guidelines for genetic testing in PAH under the PAH-ICON/PVRI banner will be published and will provide recommendations on genetic testing for patients and their relatives.

We look forward to a productive 2022 and our **3rd PAH-ICON Symposium in Athens** following on from PVRI's Annual World Congress. ■



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