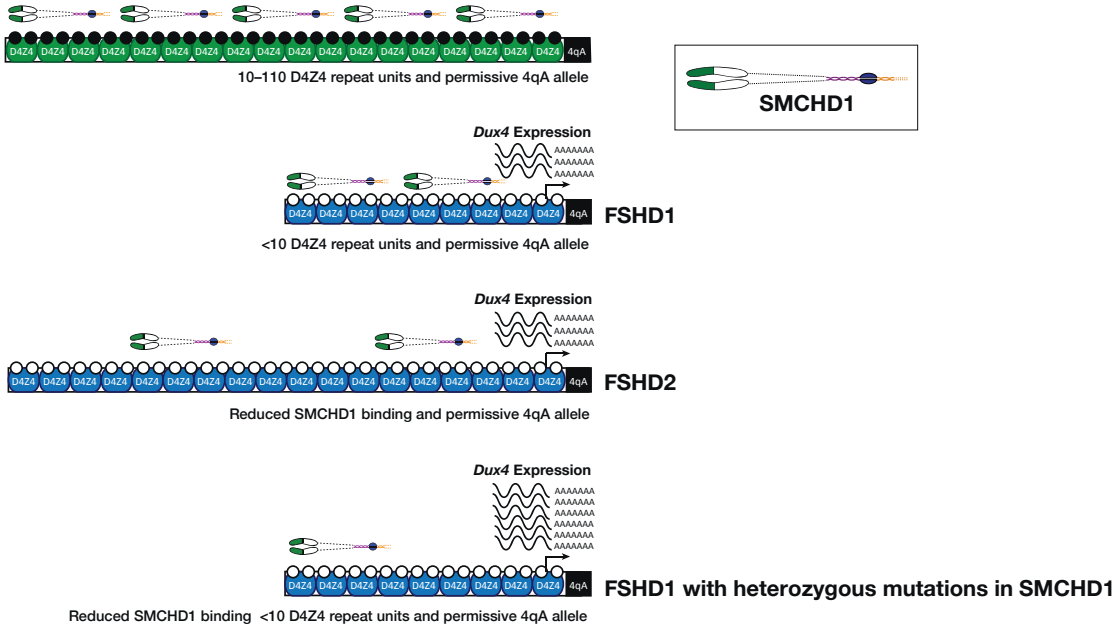
Walter+Eliza Hall Institute of Medical Research
DISCOVERIES FOR HUMANITY

Activating SMCHD1 to treat Facioscapulohumeral muscular dystrophy (FSHD)

- There is currently no specific treatment available for FSHD
- SMCHD1 possesses enzymatic (ATPase) activity; altered ATPase activity has been shown in disease
- SMCHD1 activation is a potential FSHD therapy

The opportunity

FSHD is a genetic muscle disorder mostly affecting muscles of the face, shoulder blades and upper arms. It is estimated to affect about 1 in 7500-20,000 individuals worldwide. FSHD is caused by DUX4 expression, which is normally silenced in adult cells by SMCHD1 and other epigenetic repressors.



The technology

We have shown that SMCHD1 contains a GHKL-ATPase domain and confirmed that it possesses enzymatic activity. Decreased ATPase activity has been found in FSHD patients suggesting that activation of SMCHD1 activity could be a potential FSHD therapy.

Opportunities for partnership

We are seeking partners to co-develop SMCHD1 activators for FSHD therapy:

We have:

- Foremost experts in SMCHD1, a world class structural biology program and an understanding of SMCHD1 structure
- Good quality recombinant protein and validated screening assays

We are seeking partners:

- With culture systems of human muscle cell systems to validate activator hits
- To invest in our medicinal chemistry program to follow up on an upcoming fragment-based drug discovery campaign

Scientific team

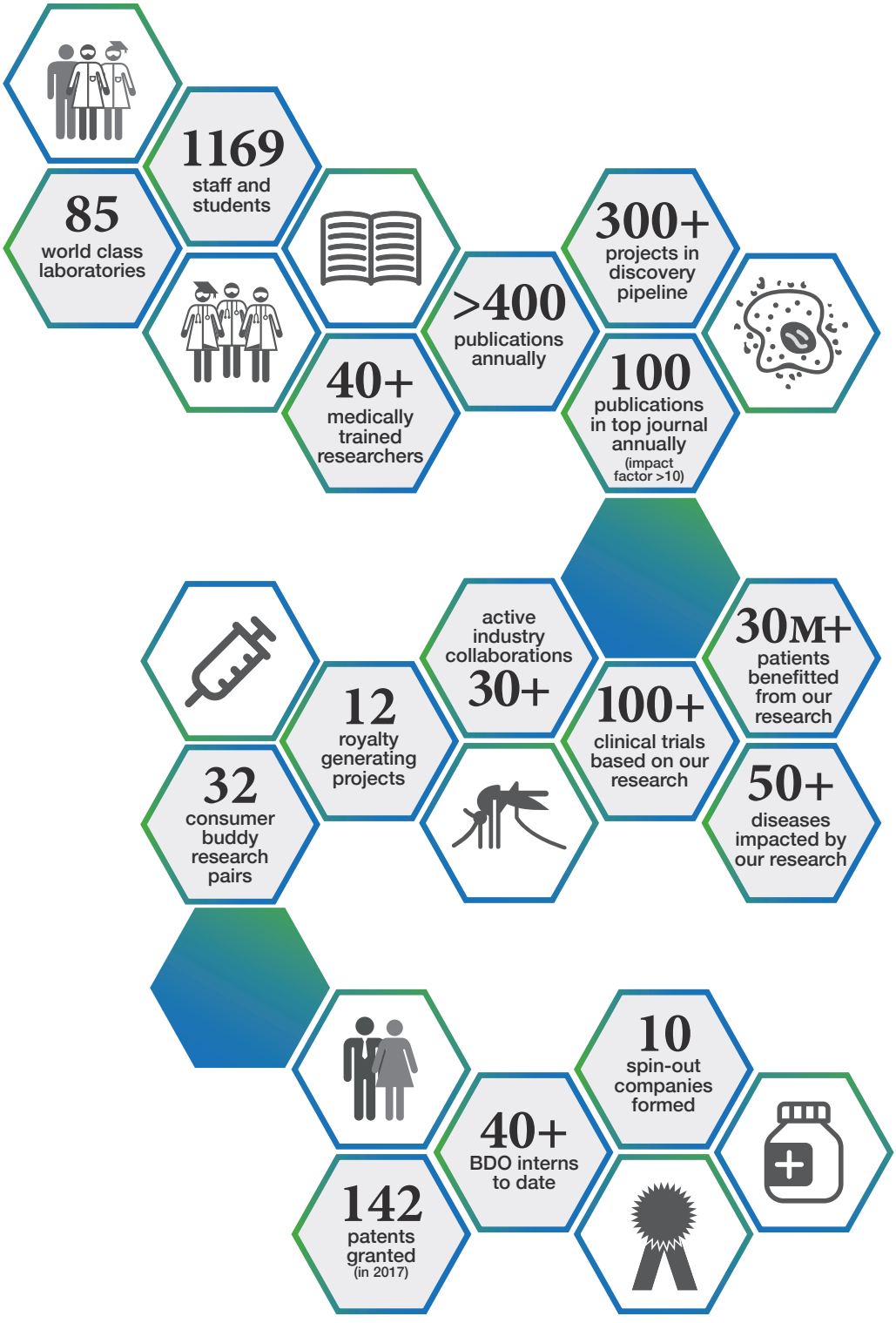
Associate Professor Marnie Blewitt
Division Head, Molecular Medicine Division

Associate Professor James Murphy
Laboratory Head, Cell Signalling and Cell Death Division

Walter+Eliza Hall
Institute of Medical Research
DISCOVERIES FOR HUMANITY

Walter and Eliza Hall Institute of Medical Research

At the Walter and Eliza Hall Institute our multidisciplinary research teams are focused on solving complex biological questions by integrating expertise in bioinformatics, clinical translation, computational biology, epidemiology, genomics, medicinal chemistry, proteomics, structural biology and systems biology. Our innovative science expands and improves the understanding of human biology and enables the translation of this new knowledge into novel therapies that benefit patients worldwide.



To discuss partnering opportunities, please contact **Dr Anne-Laure Puaux**, Head of Commercialisation, by email **puaux.a@wehi.edu.au** or phone +61 3 9345 2175.