Making a difference through personalized medicine

Researchers from WESTERN AUSTRALIA'S MURDOCH UNIVERSITY are transforming people's lives and having global impact through precision medicine programmes.

Murdoch University researchers' rise as world leaders in personalized medicine is dramatically illustrated by the story of Billy Ellsworth. Billy suffers from Duchenne muscular dystrophy (DMD), a fatal childhood condition afflicting boys, requiring them to use a wheelchair by age 12.

When he was 10, Billy was on that track — his breathing was erratic and he was unable to walk unaided on small inclines. But then Steve Wilton and Sue Fletcher, researchers who are now both at Murdoch University, designed a new drug aimed at helping people with DMD, called Exondys 51.

After taking Exondys 51 for two years, Billy's breathing had stabilized and he was able to walk up inclines independently, while whistling! Now 17 and still on the drug, he remains on his feet.

Exondys 51 delayed the loss of muscle function and reduced disease severity by restoring dystrophin, a protein that is missing in DMD sufferers. In late 2016, it was accelerated for approval by the US Food and Drug Administration.

"Despite being a first-generation drug, it has altered disease progression," says Wilton. "We're hearing some really positive stories of people responding to continued treatment with the drug."

Exondys 51 is just one example of the ground-breaking work being done by Murdoch University researchers on personalized medicine for rare and infectious diseases.

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Elizabeth Phillips is a professor at both Murdoch University and Vanderbilt University Medical Center. In global collaborations, she has identified biomarkers for adverse drug reactions that might occur when patients are being treated for high burden and once deadly diseases such as HIV. The work has lead to earlier diagnosis, treatment and prevention.

Between 2002 and 2008, Phillips, Simon Mallal and their team at Murdoch discovered a strong association between an adverse drug reaction to the HIV medication abacavir and the genetic marker HLA-B*57:01. Their championing of genetic screening in routine HIV clinical practice led to the first multicentre randomized clinical trial to show a specific genetic marker can be used to prevent abacavir hypersensitivity. This triumph in personalized medicine has now led to the prevention of drug hypersensitivity in thousands of patients, creating a roadmap from discovery to translation which Phillips and others are continuing to apply to make drugs safer globally.

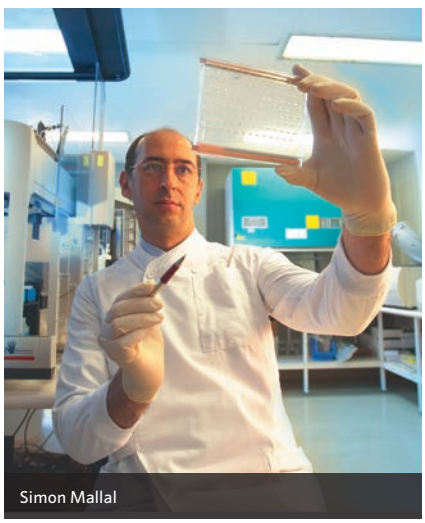
"It became very much a story of an enormous international collaborative effort," she says. "Not just between scientists and clinicians, but also with industry, to turn discoveries into tests used in clinical settings." ■

Murdoch UNIVERSITY

www.murdoch.edu.au

Stephen Wilton and Sue Fletcher

Professor Elizabeth Phillips



Simon Mallal