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New technology could treat rare disorder
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Researchers will use 3D facial analysis technology to help understand and eventually guide the treatment of a rare genetic disorder that affects one in 50,000 people as the result of a new agreement.

The agreement, between Takeda Global, SingHealth in Singapore, WA's King Edward Memorial Hospital, Curtin University and FrontierSI, will advance diagnosis and treatment monitoring for rare diseases, which globally impact more than 300 million people.

The study, funded by Takeda Pharmaceutical Asia-Pacific Medical Affairs in Rare Diseases, will enable researchers to analyse the facial features of Hereditary Angioedema (HAE), a rare genetic disorder characterised by severe and sometimes life-threatening episodic swelling mainly of the face and upper torso, across Singapore and Western Australia.

It will ensure clinicians at SingHealth, Singapore's largest group of healthcare institutions, are trained to capture and analyse 3D facial images using Curtin University's Cliniface software platform.

Perth nurse Emily Wheeler, aged 23, is living with HAE and taking part in the study.

As a result of HAE, Emily experiences swelling mainly in her stomach, adding up to 4kg of fluid to her abdomen during painful attacks that usually last a week.

Before her diagnosis, she faced losing part of her bowel and being forced to go through menopause at 21 years before doctors thankfully discovered the reason for her unexplained swelling was actually HAE -through a simple blood test.

While it took Emily two years and countless surgeries to eventually be diagnosed with HAE, she hopes this research project will ensure a quicker diagnosis and brighter future for others.

"If I can help one person by telling my story, I think that's an achievement – if someone hears this and thinks 'I have those symptoms or a doctor ticks a box of symptoms and it points to HAE, I just think that would be amazing," she said.

Currently, only subjective clinical descriptions of the nature of facial swelling in people with Hereditary Angioedema exist.

Giving clinicians an objective understanding of the facial swelling will help assess the onset of an attack, its severity, recovery and treatment, potentially assisting with the initial diagnoses.

Computer scientist Dr Richard Palmer, from Curtin University's School of Earth and Planetary Sciences, who developed the Cliniface technology, said the 3D facial images of 900 people would be processed through the Cliniface platform, extracting more than 50 facial measurements from each image.

"The resulting de-identified data will be transmitted from SingHealth to the Cliniface team at Curtin University for analysis, allowing us to generate modelling that outlines facial features specific to the Singapore population," Dr Palmer said.

Emily Wheeler is hoping the facial analysis will fast-track the diagnosis of HAE.