Genetic key to lupus shows potential of personalised medicine

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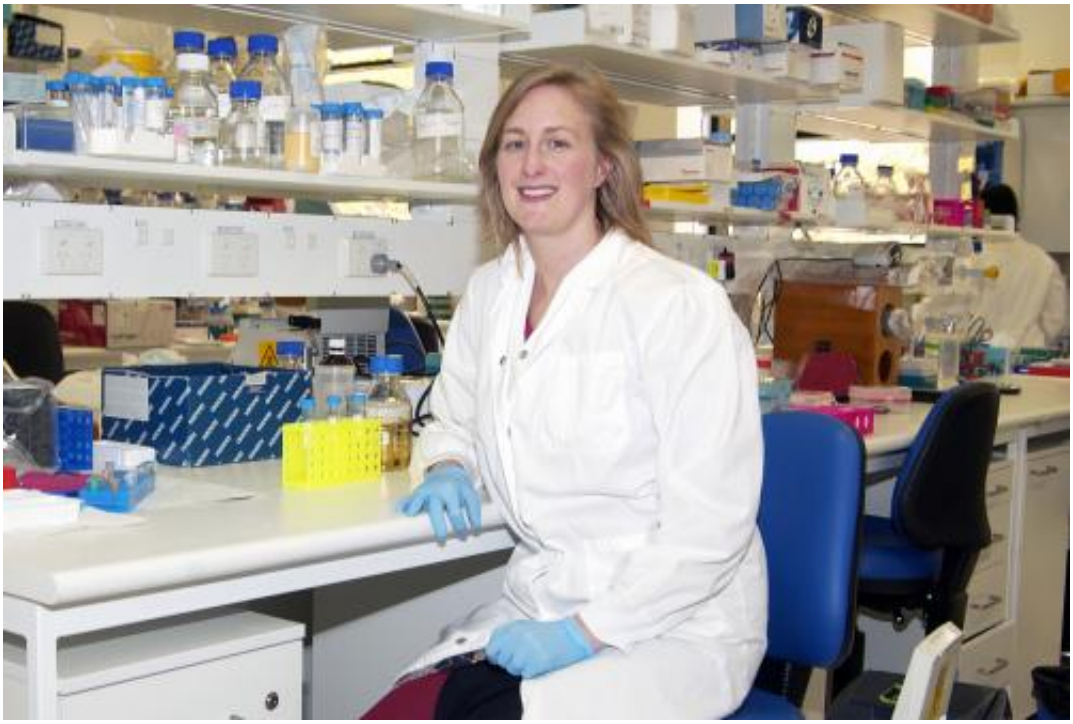

Dr Julia Ellyard

(Medical Xpress)—Medical researchers have used DNA sequencing to identify a gene variant responsible for causing lupus in a young patient.

The development shows that for the first time, it is feasible for researchers to identify the individual causes of lupus in patients by using DNA sequencing, allowing doctors to target specific treatments to

individual patients.

Lupus is a chronic autoimmune disease that affects one in 700 Australians, predominantly young and middle aged women.

Medical researchers at the Centre for Personalised Immunology, based at the John Curtin School of Medical Research (JCSMR), sequenced the genes of a young girl who suffered a stroke when she was four as a result of her lupus.

"We can now target her specific disease, and make treatments that will benefit her throughout her life," said lead researcher Dr Julia Ellyard, from the JCSMR.

Researchers identified a variant in the TREX1 gene. This mutation caused the patient's cells to produce a molecule called interferon-alpha. Clinical trials are already underway for drugs to target interferon-alpha in adults.

Dr Jeff Chaitow, head of rheumatology, a co-investigator and the patient's treating clinician at Sydney's The Children's Hospital at Westmead, said his young patient, now 10 years old, still needs regular steroids and immune suppressive drugs each day.

"New targeted therapy would be a major benefit in controlling her disease," he said.

Professor Carola Vinuesa, Co-director of the Centre for Personalised Immunology, said research was showing lupus was primarily caused by defects in only one or a few genes.

"This is the new age of personalised medicine," she said.

"This study shows that it is possible to unravel the detailed and individual genetic causes of lupus in individuals.

"Lupus is a heterogeneous disease and patients can experi.ence a number of different symptoms. We believe that there are different genetic causes of lupus. Understanding these defective genes and pathways in each individual will help tailor treatments."

Professor Matthew Cook, Co-director of the Centre for Personalised Immunology, said the results proved the potential benefits of personalised medicine, where doctors will be able to target treatments to individual patients.

"We are optimistic that this represents proof of principle for a new approach to diagnosis and treatment of a range of complex immunological disease," Professor Cook said.

More information: Results of the research are published in *Arthritis and Rheumatology*: onlinelibrary.wiley.com/doi/10 … 2/art.v66.8/issuetoc

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