

Gene discovery offers new hope for people living with chronic skin disease

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Dr. Chelisa Cardinez (pictured) says the discovery finally provides an explanation for why patients with psoriasis may go on to develop psoriatic arthritis, leaving them with joint pain, stiffness and swelling. Credit: Jamie Kidston/ANU

Scientists from The Australian National University (ANU) have discovered a gene mutation is responsible for causing psoriasis—a

chronic inflammatory skin disease that causes patients to develop red, scaly and itchy patches across their body. The research is published in *Nature Communications*.

According to ANU researcher Dr. Chelisa Cardinez, if two copies of this mutated gene (known as IKBKB) are present, patients with psoriasis may go on to develop psoriatic arthritis, leaving them with [joint pain](#), stiffness and swelling. Thanks to the world-first discovery from ANU, scientists now know what causes the progression from a skin-only disease to a skin and joint disease.

It's hoped the findings will lead to improved diagnosis and treatment for patients with psoriasis and psoriatic arthritis—conditions that patients say carry stigma in the community.

"Using a [mouse model](#), we identified that this mutation led to an abnormal function in a group of immune cells known as regulatory T cells," Dr. Cardinez, from the ANU John Curtin School of Medical Research (JCSMR), said.

"These cells are normally considered gatekeepers of the immune system. However, we found that this mutation alters the function of these cells, causing them to contribute to inflammation and promote the onset of disease."

Rebecca Davey is one of at least 500,000 Australians that live with psoriasis. She also happens to have psoriatic arthritis and says the stiffness and pain she feels when she gets out of bed in the morning can be extreme.

"People don't understand the debilitating effects these conditions can have on the individual and in fact a whole family when someone is in constant pain, has [poor sleep](#) from pain, and feels constantly fatigued,"

Ms Davey said.

"My psoriatic arthritis drugs have largely reduced the larger outbreaks on my skin, but you do have to consider everything you put on your skin and the fabrics you wear. As a former nurse, even the constant hand washing that was required for work would cause my skin to flare up. It's one of the reasons why I no longer work in the hospital system."

Psoriasis and psoriatic arthritis are forms of autoimmune disease. These types of diseases occur when the immune system attacks [healthy cells](#) after wrongly perceiving them as a threat. According to Arthritis Australia, three out of every 10 Australians with psoriasis develop psoriatic arthritis.

Although there is no cure for psoriasis, there are treatments that can help manage the condition. In October 2023, the Pharmaceutical Benefits Scheme (PBS) listed a new, subsidized drug for Australians living with severe psoriasis.

Ms Davey, who is also CEO of Arthritis ACT, says it's important to break down the stigma associated with these conditions. She says psoriasis is very misunderstood in the community.

"So many people are accused of having poor hygiene due to the plaques or even just minor skin lesions as they erupt. It's not the individual's fault that their skin is in the condition it's in; [psoriasis](#) is a painful, debilitating condition," she said.

"I had no idea what was causing my hands to flare up all the time. Our poor GPs often don't recognize these conditions early.

"In regional and [rural areas](#) there is a drastic shortage of specialists both in dermatology and rheumatology to diagnose and treat these conditions,

and people can wait over a year for an appointment if their symptoms are less dramatic.

"We must raise greater awareness of invisible disabilities such as those created by these conditions. A person might look ok from the outside, but in reality they are struggling on a daily basis."

Dr. Cardinez said, "Studies have shown that delays in [psoriatic arthritis](#) diagnosis is linked to worse clinical outcomes for patients. Therefore, earlier detection and treatment of these immune diseases is key to improving health outcomes.

"By developing a better understanding of the IKBKB gene and the role it plays in promoting the onset of these diseases, it could bring us a step closer to one day finding a cure, which would offer new hope for hundreds of thousands of Australians."

More information: IKK2 controls the inflammatory potential of tissue-resident regulatory T cells in a murine gain of function model, *Nature Communications* (2024). [DOI: 10.1038/s41467-024-45870-3](https://doi.org/10.1038/s41467-024-45870-3)

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