SA's itch finally gets scratched

The genetic mutation responsible for a rare skin disease in Afrikaners has been found after a 40-year search at Wits University. Thandiswa Ngcungcu is the scientist responsible for the final breakthrough. She chose keratolytic winter erythema as a topic for her doctoral studies.

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The disease, colloquially known as Oudtshoorn skin, causes redness on the palms and soles, with cycles in which thick sections of skin peel, especially during winter.

Wits researchers started studying the disorder in the late 1980s, and in 1997 the trait was mapped to the short arm of chromosome 8. But the mutation remained elusive until Ngcungcu decided to try to track it down, said Wits spokesman Lisa Rautenbach.

International collaboration

Writing in this month's edition of the American Journal of Human Genetics, Ngcungcu and researchers say they discovered a mutation in a region between genes which was present in all Oudtshoorn skin sufferers they studied.

At the same time, a researcher from the University of Bergen discovered the cause of the disease in Norwegians. When they compared notes they found an overlap in a critical genomic region called an enhancer. After another year's work, they

demonstrated that the mutation causes a nearby gene to produce more protein than normal - the likely cause of skin peeling.

"Solving the mystery of [Oudtshoorn skin] was a journey of data analysis, ancestry mapping, genomic comparison and global collaboration," said Rautenbach. The discovery will allow dermatologists to make a definitive diagnosis of the disease in patients and is a starting point for researching possible treatments.

Rautenbach said Afrikaners were at high risk of inheriting several genetic disorders, the best known being familial hypercholesterolaemia - inherited high cholesterol leading to heart attacks early in life - and porphyria-sensitivity of the skin to ultraviolet exposure and adverse reactions to specific drugs. "These disorders are common because of founder mutations brought to South Africa by small groups of immigrants who settled in the Cape of Good Hope," she said.

Ngcungcu is now examining the genetics of albinism.

Source: The Times