

Genetics discovery to help fight 'black fever'

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Scientists—including a geneticist at The University of Western Australia—are a step closer to developing a vaccine against a fatally infectious parasite carried in the bite of sandflies.

Visceral leishmaniasis, also known as black fever, is the second-largest parasitic killer in the world after malaria. The parasite migrates to organs such as liver, spleen and bone marrow and if left untreated will almost always be fatal. Symptoms include fever, weight loss, mucosal ulcers, fatigue, anemia and substantial swelling of the liver and spleen.

Leishmaniasis affects 12 million people and there are an estimated 1.5 million new cases annually mainly in India, Bangladesh, Nepal, Sudan, South Sudan, Ethiopia and Brazil.

UWA Winthrop Professor Jenefer Blackwell, Head, Genetics and Health, Telethon Institute for Child Health Research, was a senior author of a big international study which has uncovered a single major genetic risk factor for visceral leishmaniasis. She led the LeishGEN Consortium that carried out all of the field work and sample collection, first from the Cambridge Institute for Medical Research at the University of Cambridge and later from UWA and TICHR. She was also a member of the Wellcome Trust Case Control Consortium which carried out the [genetic analysis](#).

The study was published online this week in [Nature Genetics](#).

With scientists from India, Brazil, UK, Australia and the USA, Professor

Blackwell identified variation in a specific region of the major immune response locus, known to immunologists as the major histocompatibility complex (MHC), as the single most important [genetic risk](#) factor for disease.

The researchers compared genomes of 1346 people suffering from disease in India and Brazil against those of 2702 healthy controls. The findings were confirmed in an independent cohort of 941 cases and 990 healthy controls from India.

Teams in Australia, UK and the USA are using the results in [vaccine research](#) to study the way the immune system interacts with the disease in mice. "Earlier genetic studies of [visceral leishmaniasis](#) in inbred mice allowed us to clearly demonstrate the importance of the MHC in regulating this disease," Professor Blackwell said.

"Now, major advances in human genetics and the ability to compare the genomes of large numbers of people with and without the disease have allowed us to identify the precise molecular basis to this MHC control in humans. This will have a major impact on refining research towards the ultimate goal of a vaccine."

Provided by University of Western Australia

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