

Mutation makes some people more vulnerable to sleeping sickness

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Researchers of the Institute of Tropical Medicine in Antwerp (ITM) and the University of Antwerp (UA), collaborating with their colleagues from the Université Libre de Bruxelles (ULB), discovered a mutation in the genetic composition of the human protein apolipoprotein L1. This

mutation is common in certain African tribes, and increases their risk of contracting the deadly sleeping sickness disease.

Trypanosomes are unicellular parasites causing serious diseases, including sleeping sickness in Africa. Each year about 4000 new cases are reported. In 2009, due to the success of sustained control programmes, the number of new cases fell below 10.000 for the first time in 50 years. Last year, about 4000 new cases were reported.

Apolipoprotein L1 protects the human body from all African trypanosomes, except *gambiense* in West and Central Africa and *rhodesiense* in East and South Africa. These two subspecies have developed defense mechanisms against the human apolipoprotein L1.

In 2013, the researchers examined a blood sample of an infected patient from Ghana. They were surprised to find that the blood contained an ancestor of *gambiense* that had developed only one of the two defense mechanisms against apolipoprotein L1. Theoretically, this variant could not infect humans. The sample proved otherwise.

ITM's Bart Cuypers conducted the genetic analysis of the sample. "The complete study was based on the DNA in one dried blood spot on filter paper," Cuypers explains, "By combining advanced molecular techniques and bio-informatics we could unravel the genetic code of both human and parasite."

Additional research on the patient DNA revealed that the APOL1 gene - the gene containing the code for apolipoprotein L1 - had a mutation that blocks apolipoprotein L1 from destroying certain trypanosomes. This mutation in the human genome is rare, but relatively prevalent in certain African tribes. These tribes are thus more at risk of contracting sleeping sickness.

Fully understanding the biological interaction between humans and parasites is crucial to develop new medicines in the long term. "Trypanosomes have been around for a long time and have evolved alongside humans on the African continent. This fascinating study is an example of the delicate interaction between human and parasite, causing both to develop defense mechanisms to outwit each other," says ITM's Dr. Stijn Deborggraeve.

Link

- The paper: [Apolipoprotein L1 Variant Associated with Increased Susceptibility to Trypanosome Infection](#)