

CDC's Advanced Molecular Detection (AMD) program fosters scientific innovation in genomic sequencing, epidemiology, and bioinformatics to transform public health and protect people from disease threats.

## Accurately Identifying Neglected Tropical Diseases

## Using AMD methods to improve diagnosis and clinical management of leishmaniasis

Leishmaniasis is a parasitic disease found in over 90 countries in the tropics, subtropics, and southern Europe. The disease is caused by more than 20 different species (types) of Leishmania parasites, which are spread by the bite of sand flies. Infection with different Leishmania species can cause different clinical forms and consequences of infection. The three main clinical forms are visceral leishmaniasis, which can affect internal organs of the body (such as the spleen, liver, and bone marrow) and may be life threatening; cutaneous leishmaniasis, which causes skin sores that may last for months to years; and mucosal leishmaniasis, a potentially disfiguring form caused by infection in the nose, mouth, or throat.



AMD is improving diagnosis for leishmaniasis, a parasitic disease spread by sand flies.

Treatment of leishmaniasis depends in part on the *Leishmania* species or strain and the geographic area in which infection was acquired. Identification of the Leishmania species with which a person is infected helps guide clinical decisions, such as which treatment to use and how to monitor for and prevent consequences of the infection.

In the U.S. civilian sector, CDC's reference diagnostic laboratory is the main source for identification of *Leishmania* species in human clinical specimens. Over the last several years, the number of specimens tested in CDC's laboratory has progressively increased; the lab confirmed upwards of 100 cases of *Leishmania* infection per year in 2013 and 2014. However, current laboratory methods have limitations; for example, they can be time-consuming to use (which can delay treatment decisions) and they do not always succeed in identifying some of the *Leishmania* species.





To increase the ability to provide accurate and timely lab results, CDC scientists are applying advanced molecular detection (AMD) techniques to their existing collection of *Leishmania* parasite strains and creating a database of the genetic diversity among *Leishmania* parasites; CDC scientists are sequencing and comparing the genomes of multiple species and strains. The goal is to develop laboratory methods that improve the diagnosis (including species identification) and clinical management of leishmaniasis and that thereby improve the patient outcomes.

For more information on Neglected Tropical Diseases, visit the CDC website at www.cdc.gov/globalhealth/ntd.

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## 2017 Update

Before this project began, no reliable genomic data existed for the 20 species of *Leishmania*. In the first year, project investigators began sequencing genomes of several *Leishmania* species to create a much needed genomic database. In addition, they used data collected through AMD to begin developing new methods to differentiate between *Leishmania* parasites. This work proved vital for one patient, whose *Leishmania* infection was initially attributed to the wrong species. Once CDC researchers identified the actual species involved, the patient received treatment that targeted the specific *Leishmania*. This reflects the importance of cutting-edge molecular methods in correctly identifying *Leishmania* species.

In the coming year, researchers will sequence genomes of seven additional Leishmania parasites to continue building the genomic reference database and developing species-specific molecular tests for *Leishmania*.