

## Ultra-fast, Meta-genomics Pathogen Detection Software for Diagnosing Infectious Diseases

SALT LAKE CITY, UT, May 26, 2016—Scientists at the University of Utah, ARUP Laboratories, and IDbyDNA, Inc., have developed ultra-fast, meta-genomics analysis software called Taxonomer that dramatically improves the accuracy and speed of pathogen detection. In a paper published today in [Genome Biology](#), the collaborators demonstrated the ability of Taxonomer to analyze the sequences of all nucleic acids in a clinical specimen (DNA and RNA) and to detect pathogens, as well as profile the patient's gene expression, in a matter of minutes.

Infectious diseases are one of the biggest killers in the world. Almost 5 million children under age 5 die each year from infectious diseases worldwide, yet many infections are treatable *if* the pathogen culprit can be quickly and accurately identified.

“In the realm of infectious diseases, this type of technology could be as significant as sequencing the human genome,” says co-author [Mark Yandell, PhD](#), professor of human genetics at the University of Utah (U of U), H.A. & Edna Benning Presidential Endowed Chair holder, co-director of the USTAR Center for Genetic Discovery, and co-founder of IDbyDNA. “Very few people have inherited genetic disease. But at some point, everyone gets sick from infections.”

It is difficult for infectious pathogens to hide when their genetic material is laid bare. Taxonomer opens up an entirely new approach for infectious disease diagnosis, driven by sophisticated genomic analysis and computational technologies. After a patient's sample is sequenced, the data are uploaded via the internet to Taxonomer. In less than one minute, the tool displays a thumbnail inventory of all pathogens in the sample, including viruses, bacteria, and fungi. The interactive, real-time user interface of Taxonomer is powered by the iobio system developed by the laboratory of Gabor Marth, DSc, professor of human genetics at the U of U and co-Director of the USTAR Center for Genetic Discovery.

“Our benchmark analyses show Taxonomer being ten to a hundred times faster than similar tools,” says co-author [Robert Schlberg, MD, Dr Med, MPH](#), a medical director at ARUP Laboratories and cofounder of IDbyDNA. Schlberg was awarded a \$100,000 grant from the Bill and Melinda Gates Foundation to apply Taxonomer toward decreasing high mortality rates of children with infectious diseases in resource-limited settings.

Schlberg points out that current diagnostic testing still relies heavily on growing cultures of suspected pathogens in the laboratory, which is often inconclusive and time consuming. Even with much faster tests like PCR, the number of pathogens that can be detected is limited.

Schlberg explains that Taxonomer can identify an infection without the physician having to decide what to test for, something a PCR-based test cannot do. In other words, a doctor doesn't have to suspect the cause of a patient's infection, but can instead simply ask, “What does my patient have?” and Taxonomer will identify the pathogens.

In the new study, Taxonomer was put to the test with real-world cases using data published by others and samples provided by ARUP Laboratories and the Centers for Disease Control and Prevention (CDC). Taxonomer determined that some patients who exhibited Ebola-like symptoms in the recent African outbreak did not have Ebola but severe bacterial infections that likely caused their symptoms. “This



technology can be applied whenever we don't know the cause of the disease, including the detection of sudden outbreaks of disease. It is very clear we urgently need more accurate diagnostics to greatly enhance the ability of public health response and clinical care," says Seema Jain, MD, medical epidemiologist at the CDC.

Another unique feature of Taxonomer is its ability to delve into human gene expression profiling, which provides information on how or if the patient's body is reacting to an infection. "As a clinician, this gives you a better idea, when we identify a pathogen whether it is *really* the cause of the disease," says Carrie L. Byington, MD, professor of pediatrics of the U of U and co-director of the Center for Clinical and Translational Science. "This tool will also allow us to determine if the patient is responding to a bacterial or viral infection when we don't find a pathogen or when we find multiple potential causes." She says that she sees the exceptional value of this tool for treating children, who experience more life-threatening infections early in life. "Seeing how a host [patient] reacts is extremely valuable; I believe this is a paradigm shift in how we diagnose people. It is why I wanted to be involved."

In a [previous paper published in the \*Journal of Clinical Microbiology\*](#), Schlberg and his collaborators demonstrated that high-throughput sequencing in combination with Taxonomer can reliably detect pathogens, and identify previously missed pathogens, in patient samples. "Taxonomer provides a critical step forward, as it is extremely fast, accurate, and easy enough to use for implementation in diagnostic laboratories," says Schlberg.

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#### **About ARUP Laboratories**

Founded in 1984, ARUP Laboratories is a leading national reference laboratory and a nonprofit enterprise of the University of Utah and its Department of Pathology. ARUP offers more than 3,000 tests and test combinations, ranging from routine screening tests to esoteric molecular and genetic assays. ARUP serves clients across the United States, including many of the nation's top university teaching hospitals and children's hospitals, as well as multihospital groups, major commercial laboratories, group purchasing organizations, military and other government facilities, and major clinics. In addition, ARUP is a worldwide leader in innovative laboratory research and development, led by the efforts of the ARUP Institute for Clinical and Experimental Pathology®.

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#### **About IDbyDNA Inc.**

Based in Sunnyvale, California, IDbyDNA is developing technologies to enable universal microorganism detection. IDbyDNA's mission is to help doctors and scientists to detect any pathogen in any sample. For more information about IDbyDNA, please visit [www.idbydna.com](http://www.idbydna.com). For Taxonomer, please visit [taxonomer.com](http://taxonomer.com).

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### **About University of Utah Health Sciences**

The University of Utah Health Sciences programs are internationally regarded for their research and clinical expertise in medicine, pharmacy, nursing, and health. Through the School of Medicine and Colleges of Pharmacy, Health, and Nursing, the University of Utah health sciences faculty conduct pioneering research in the genetics of disease, regenerative medicine, drug compounds, cancer, nutrition, and other areas. In addition, the Health Sciences programs also train many Utah physicians, pharmacists, nurses, therapists, and other health-care professionals.

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