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### **Artificial Intelligence Technology Provides New Evidence that Mutations in the Mitochondrial Genome Underlie Parkinson's Disease**

Rockville, MD -- April 05, 2005 – Pope John Paul II suffered from Parkinson's disease, and he was far from alone. Over a million Americans have Parkinson's, and many more suffer worldwide. Yet in spite of years of effort by medical researchers, tracking down the genetic roots of the disorder has proved devilishly difficult. This year, it seems, a combination of human and artificial intelligence may have taken a large step in the right direction. By applying advanced artificial intelligence technology to analyze data regarding mutations in mitochondrial DNA, scientists at Rockville bioinformatics firm Biomind and the University of Virginia have pinpointed a particular region of a particular gene on the mitochondrial genome that appears to be strongly associated with Parkinson's disease.

**[ For a slightly lengthier discussion of the history and science of this discovery, including an interesting story involving a bad batch of heroin, see:**

[http://www.biomind.com/pd\\_article.pdf](http://www.biomind.com/pd_article.pdf) ]

Parkinson's disease is a progressive disorder of the central nervous system, whose well-known victims – besides the late Pope -- include boxer Muhammad Ali and comedian Richard Pryor. Most common in people over age 45, its symptoms include rigidity, tremors, difficulty with balance and posture, and slowing of movements. Various treatments have been found that lessen the effects of the symptoms, but there is no known cure.

Parkinson's is rooted in nerve cells – and is particularly associated with degeneration of nerve cells in the area of the brain governing movement. In the late 1990's, Drs. Davis Parker and Russell Swerdlow at the UVA, together with scientists from San Diego firm MitoKor, did experiments showing that Parkinson's is associated with defects in the mitochondrial DNA of these nerve cells.

(The DNA one usually hears about lies in the nucleus of a cell, the cell's center. But mitochondria, the cell's energy-producing engines, also contain a small amount of DNA. The human mitochondrial genome only contains 37 genes,

whereas the nuclear genome contains around 25,000 at last count. But these 37 genes – inherited from the prehistorical time in which mitochondria were independent organisms -- carry out a lot of valuable functions. If they stop working properly, serious problems can ensue.)

What this 1999 work showed was that the problem lay somewhere in the mitochondrial genome. But it left the question: Which mutations caused the problem?

To answer this question, Dr. Parker, Dr. Rafal Smigrodzi and colleagues sequenced mitochondrial DNA drawn from the nerve cells of a number of Parkinson's patients, as well as a number of normal individuals, and looked for patterns. But to their surprise, when in 2003 they set about seriously analyzing this data, they found no simple, consistent pattern. There were no specific genetic mutations common to the Parkinson's patients and not the normal people.

Enter artificial intelligence. Biomind's AI software crunched the data and discovered that, while there are no specific mutations corresponding to Parkinson's disease, there are regions – and combinations of regions -- of the mitochondrial genome that tend to be mutated in Parkinson's patients. There are many different rules of the form "If there are mutations in this region of this mitochondrial gene and that region of that mitochondrial gene, then the person probably has Parkinson's disease."

The AI technology was written up in a technical journal article, which will appear later this year in the Journal of Artificial Intelligence in Medicine. And Dr. Parker and his colleague Dr. Janice Parks have already published a paper in Biochemical and Biophysical Research Communications, showing that mutations in a certain narrow region of ND5, a mitochondrial gene encoding a complex I subunit, correctly predict whether a person has Parkinson's disease with 94% accuracy. To discuss these results and others, this June Dr. Parker is convening the First International Brainstorming Conference on Parkinson's Disease, in Louisville, Kentucky.

The approach the Biomind/UVA team has taken for analyzing Parkinson's also has implications for other neurodegenerative diseases. According to Dr. Ben Goertzel, Biomind's Chief Science Officer and the primary creator of the algorithms used in the Parkinson's research, "it seems reasonably likely that applying the same techniques to data obtained from Alzheimer's patients will yield analogous results."

**For additional information, including preprints describing the detailed technical work, contact:** Dr. Ben Goertzel -- Phone: 240-505-6518 -- [info@biomind.com](mailto:info@biomind.com)