ACP’s Repository Speeds Research, Cuts Costs, and Leads to Diagnostic Breakthrough

Tom Aune is a professor of medicine at Vanderbilt University in Nashville, as well as an MS researcher and entrepreneur. He is also an enthusiastic proponent of using samples and data from the ACP Repository to quickly advance MS research. Says Tom, “I have used the ACP Repository samples extensively and I calculate that it has saved me close to 5 years and over $1.5 million. Another way to think of it is that an experiment that has cost me $20,000 to do with samples from ACP, would have cost me $1 million with samples obtained from other sources. The value, and the positive impact the repository has had on our research and on the lives of some people with MS, really cannot be overstated!”

Since 2005, Tom and a team of colleagues have been searching for a means of diagnosing MS with a simple blood test. They hope to find an addition to the use of magnetic resonance images (MRIs), which are costly, invasive and slow, because patients typically undergo multiple MRIs of different parts of the brain over a period of months or even years to determine whether brain degeneration has occurred. It is not uncommon for the process of diagnosing MS to take five years from the date that symptoms were first reported.

But early intervention with one of the available disease modifying therapies may lead to better outcomes. In contrast, delayed or slow diagnosis can result in unnecessary damage to the brain, a threat that was underscored by the widely praised report: Brain Health: Time Matters in Multiple Sclerosis. Published in 2015, the report recommended specific actions to achieve the best possible outcome for every person with MS. It stated, “Early intervention is vital” but noted that instead, “Significant delays often occur before a person with symptoms suggestive of MS sees a neurologist for diagnosis and treatment. This is despite diagnosis being 10 times more rapid now than in the 1980s and substantial evidence that early treatment is more effective than later treatment.” The report urged healthcare professionals to “involve people with MS proactively in decision-making and in managing their disease,” and referred to the need for “specialized diagnostic procedures.”

Brain degeneration is not the only indicator of MS. Inflammation occurs when the body’s own immune cells attack the nervous system, as can happen in people with MS along any area of the brain, optic nerve, and spinal cord. Tom’s research led him and his colleague, Chase Spurlock, also a Vanderbilt
faculty member, to establish a private biotech company, IQuity, which is focused on this indicator and has developed a gene expression test that can measure the inflammatory component and reveal the presence of MS at a point before it would be obvious from an MRI scan.

IQuity’s new test analyzes ribonucleic acid (RNA) markers in a simple blood sample. Gene expression gives rise to a class of messenger molecule, called messenger RNA (mRNA) and changes in gene expression patterns are a “fingerprint” of disease. Measuring RNA molecules determines if the expression of the gene encoding that RNA is turned on or off. These measurements are captured through a simple blood test and provide a snapshot of information being processed in a living cell. Tom’s research revealed that people suffering from MS exhibit a distinct RNA expression pattern in their blood. This is critical to early disease detection since those unique patterns distinguish them from patients with other neurological diseases. The test he and his colleagues have developed can identify these patterns and confirm a suspected diagnosis with greater than 90% accuracy.

Having access to the ACP Repository has enabled Aune and his colleagues to examine samples from people before a diagnosis of MS was delivered, at the point of diagnosis and following diagnosis, and to spot variations in messenger RNA patterns in each group.