# Medical Imaging AI Advances Muscular Dystrophy Diagnosis

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Muscular dystrophy, an inherited disease with several variants that can appear anywhere from early childhood to middle age, is exceptionally tricky to diagnose. First, the patient's genetic profile must be plotted out in great detail. Then it is examined and compared with large sets of genomic data stored at research centers and hospitals. Analysis is painstaking, requiring physicians to do a great deal of manual work. The entire process can take 24 weeks to deliver results.

A new method uses high-performance computing and AI inferencing to do much of the heavy lifting, relieving doctors of tedious manual labor. By pointing them in the right direction, it can shorten the time to diagnosis to 16 weeks or shorter, depending on patients' condition. That's also good news for patients and their families, who can start treatments sooner, and for researchers, who can advance their knowledge of the disease.

## Collaboration Leads to IoT Healthcare Solutions

This new diagnosis method came about through the close collaboration of two Taiwanese companies. Avalue Technology Inc., a provider of IoT computing equipment, has deep experience with hospitals and labs. Biomdcare Corporation specializes in medical software,

imaging, and screening tools. Together, the companies developed a muscular dystrophy screening kit that can analyze massive genomic data quickly and deliver results that are 97% accurate.

To create the Genomics Analysis Platform – Muscular Dystrophy Screening Kit, both companies had to overcome the vexing challenges that make working with medical data so time-consuming and difficult. "We worked together to develop hardware and software with the latest technology," says Rus Lu, Senior Product Manager at Avalue.

Avalue was tasked with finding a way to efficiently transport and process the enormous sets of data to be analyzed.

"We knew the solution required a very high-performance CPU, so we applied the latest Intel$^®$ Processors," Lu says. The company also built an extra graphics card slot into its server to accommodate the dense medical images and used a 10-gigabit Intel Ethernet chipset to avoid common bottlenecks slowing data transmission.

The Biomdcare software had to identify the patient's disease variant and classify any correlations among the vast stores of genomic data it was able to use for comparison. Working with data sets this large is beyond the processing capability of most medical laboratories, and is usually confined to research centers and universities.

Biomdcare used the Intel$^®$ OpenVINO$^{™}$ Toolkit to develop an AI-assisted software program that combs through all the data, filters out irrelevant results, and homes in on promising correlations. It then fully analyzes these correlations before handing them over to doctors.

#AI algorithms have an insatiable appetite for #data, since the more they crunch, the more accurate their results will be. Avalue Technology Inc. and Biomdcare Corporation via @insightdottech

## Analyzing Medical Images with AI

Ironically, given the enormous amount of data it must sort through, one of the main problems Biomdcare encountered was having a small amount of relevant data to work with.

There are several reasons for this. The first is that AI algorithms have an insatiable appetite for data, since the more they crunch, the more accurate their results will be. And unlike the products and machinery AI systems were originally designed to analyze, humans are unique, their genetic data imbued with exponentially more permutations.

"In a factory, a lot of data and photos are generated about defective items and problems on the production line every day. But in healthcare, we don't have as much data compared to the overall size of the patient population. Low data counts are always a problem for AI solutions in healthcare," explains Richard Lin, Marketing Director of Biomdcare.

Diseases involving genetic mutations are especially complex, and analyzing them is never a straightforward task. The problem is compounded for muscular dystrophy, a rare disease roughly estimated to affect fewer than four per 100,000 people globally, according to the National Institutes of Health. In addition, its variants are so different from one another that medical organizations often refer to the condition as a *group* of diseases.

These issues would normally make it difficult to find enough specific correlations to confidently point toward a diagnosis. But Biomdcare developed a proprietary process to make it work. "We use a smaller database to generate a more accurate AI model. It's a key value of our solution," Lin says.

After the data has been analyzed, a report is prepared for physicians and stored—along with all the genomic data and the patient's records—on Avalue servers. Medical professionals can access it on a software platform containing simple annotation and workflow tools, which they use to arrive at a final diagnosis.

## Smart Healthcare Solutions Help Patients and Researchers

In addition to making doctors' work easier, the muscular dystrophy screening kit provides a faster way for patients to learn if they have the disease. "In Taiwan, about 1 in 40 patients may have the recessive gene but are without symptoms," says Olivia Wang, Product Manager at Biomdcare.

People who have a family member with the disease or couples starting a family can use the screening kit to learn if they have it or are carriers. Though there is currently no cure, treatments have been developed to improve muscle strength, and in some cases, slow disease progression. A faster diagnosis enables patients and their families to prepare for the future and seek treatment options sooner.

As more people use the screening kit, the data it produces will expand medical repositories, furthering research and helping AI systems achieve even better accuracy. "We hope our solution will help the research community build more reference data to help other patients who need these tests," Wang says.

The companies are also extending their technology to screen for other diseases, including breast cancer and human papillomavirus. "We believe AI model-training solutions can help in many situations," Lin says. "We anticipate developing more disease screening solutions in the future."

*Edited by [Georganne Benesch, Associate Editorial Director for insight.tech](#).*