Researchers develop method to monitor patients with spinal muscular atrophy using sound waves

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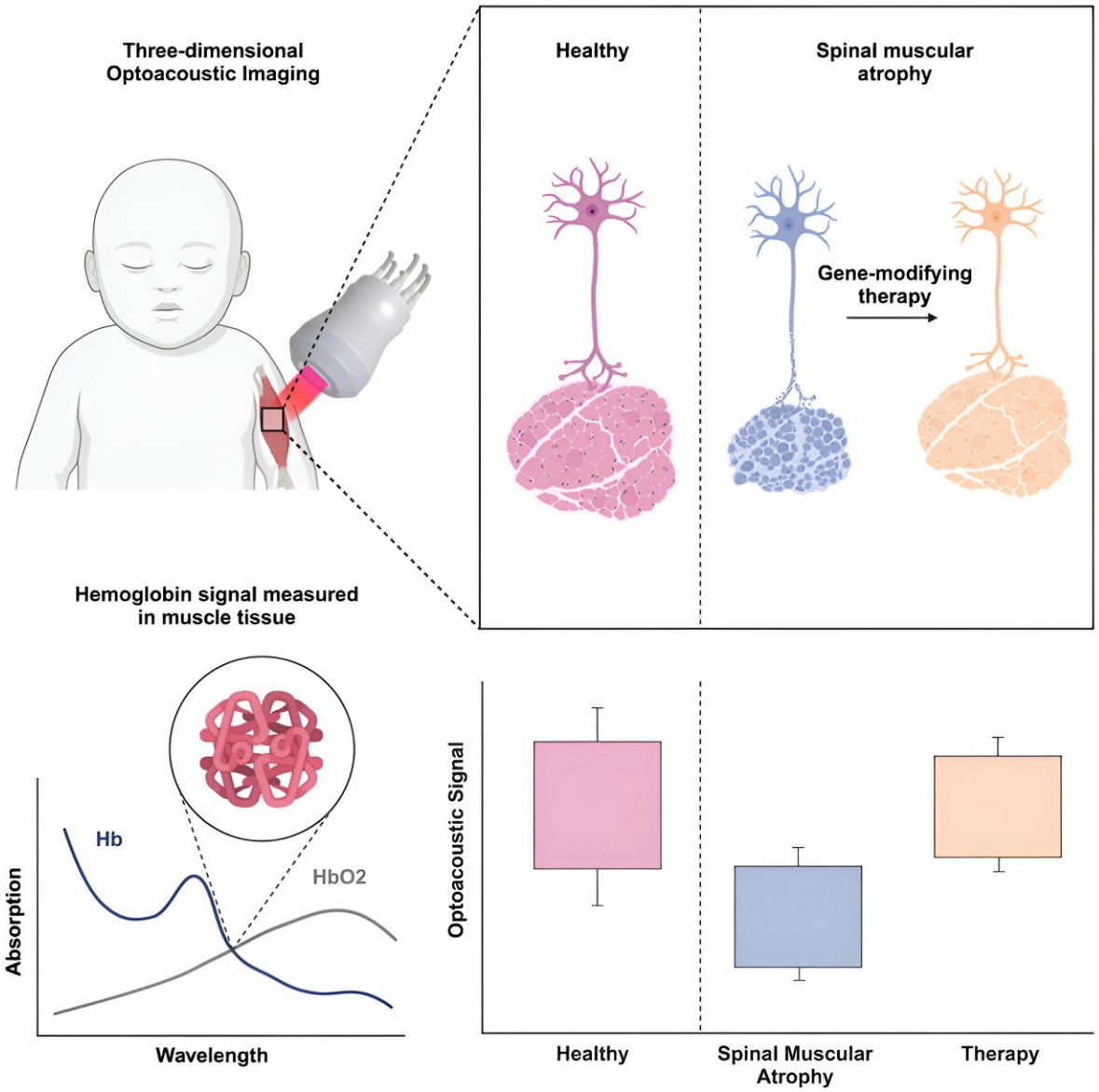

Credit: *Med* (2024). DOI: 10.1016/j.medj.2024.02.010

Spinal muscular atrophy (SMA) is a terrible disease in which a genetic mutation causes certain nerves responsible for sending signals to muscles to degenerate. This leads to muscles wasting away, and many patients have died a painful death due to this rare condition. Genetic treatments have only been available for a few years.

Now, a team led by Emmanuel Nedoschill, Ferdinand Knieling and Adrian Regensburger from the Translational Pediatrics working group at the Department of Pediatrics and Adolescent Medicine at Uniklinikum Erlangen have devised an elaborate procedure that shows promising results when used in combination with these treatments: short laser pulses create sound waves that then provide images of the muscle tissue.

The have published a paper on their findings in the journal *Med*.

"This method is similar to ultrasound scans that have been in use for a long time already," explains Nedoschill. "In just a few minutes, a scan taken outside the body can provide an image of the condition of the muscles inside the body."

One major advantage of this optical-acoustic imaging method is that even small children tend to cooperate without too much fuss as it is a non-surgical procedure that does not involve swallowing or injecting contrast agent or the like. That not only makes it easier for the medical team, it also improves the situation for the children and their parents during their stay at hospital.

The situation is usually incredibly stressful for those affected anyway. The disease is caused by only a slight change in the genome in the blueprint for a protein named "SNM," but the lack of this protein leads to the degeneration of certain nerves that are responsible for transferring signals to muscle cells. The affected muscles waste away. Lay persons can find it very upsetting to be faced with discussions on the consequences and the various ways in which the disease can progress.

One category is the "walkers," who are still capable of taking a few steps by themselves. The situation for the "sitters" is considerably worse. Without help, they are only capable of sitting, but cannot stand up by themselves. Worst of all, however, are the "non-sitters," who are not even able to sit anymore. If the muscles that are required for swallowing or breathing are affected, the disease can have fatal consequences.

Luckily, only 1 in approximately 10,000 newborns has the SNM genetic mutation. However, the suffering of those who are affected is so great that any improvement to the treatments available constitutes a major breakthrough, as is the case with the treatment known as "optoacoustic imaging" or "OAI" researched at the Department of Pediatrics and Adolescent Medicine at Uniklinikum Erlangen.

These treatments, which have only been available for a few years now, have led to significant breakthroughs in this disorder which was largely untreatable beforehand. Notable improvements have even been achieved in the most severe cases referred to as "non-sitters."

However, until now the only way to keep track of this success involves strenuous movement tests, that can last for several days. The very nature of the tests can also jeopardize their objectivity. Some people may make more effort than others, leading to better results in some children than in others. The mood of the children can also vary day to day, influencing the results of the tests.

The OAI procedure with its short laser pulses using near infrared light can significantly increase the objectivity of these observations. These pulses of light heat the affected tissue, which then emits sound waves that can give important information on the various structures within the patient's body. Tissue, for example, consists of collagen proteins that return a different spectrum of sound waves than muscles or fatty tissue.

"In muscles, we can identify the spectrum of hemoglobin in red blood cells that is responsible for transporting oxygen to the organism and removing carbon dioxide," explains Nedoschill. The more muscle cells there are and the more active they are, the more oxygen they require to do their job.

If the researcher from Uniklinikum Erlangen sees a lot of hemoglobin, he knows that this means that the muscle mass is considerable. On the other hand, if muscles are wasting away and becoming replaced with connective tissue, the three-dimensional images show how the disease is progressing and leading to greater quantities of collagen, thereby documenting the wasting of muscle mass.

This provides doctors such as Nedoschill with a tool that is as quick and easy to use as an ultrasound scan and delivers impressive images of how muscles and connective tissue come and go.

The research conducted in Erlangen based on tracking hemoglobin has indicated that children suffering from SMA have considerably less muscle tissue than the healthy control group. After receiving the life-saving genetic therapy, however, the concentration of hemoglobin increases, the muscles that had wasted away start to recover and the ultrasound signals soon start to be similar to those from healthy organisms.

Thanks to research at the Department of Pediatrics and Adolescent

Medicine in Erlangen, a relatively straightforward tool is now available for monitoring the progress of the muscle-wasting disease and the success of treatment.

More information: Emmanuel Nedoschill et al, Monitoring spinal muscular atrophy with three-dimensional optoacoustic imaging, *Med* (2024). DOI: 10.1016/j.medj.2024.02.010

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