

Transcript Details

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Friedreich's Ataxia: Impact of Frataxin Loss

Announcer:

You're listening to *NeuroFrontiers* on ReachMD. Today, we'll hear from Dr. Arnie Koeppen, who will be discussing the impacts of frataxin deficiency in patients with Friedreich's ataxia. Dr. Koeppen is a neurologist at the Albany Stratton VA Medical Center and Professor Emeritus at Albany Medical College in New York.

Let's hear from him now.

Dr. Koeppen:

Frataxin deficiency is systemic, and in theory, other organs requiring high energy should be affected also. But they are not. The impact of frataxin deficiency is on the heart—the most common cause of death in Friedreich's ataxia—and on certain areas of the brain and the peripheral nervous system, including the so-called dorsal root ganglion.

Oxidative stress has been speculated to be critical in the pathogenesis of Friedreich's ataxia in the heart and in the brain. And some of the more recent drugs in the pipeline are devoted to hindering or to stopping oxidative stress, but nothing has proved to be entirely effective.

Being a neuropathologist, I've looked at 67 cases of Friedreich's ataxia—autopsy specimens—and the target is on the cerebral cortex, the so-called dentate nucleus of the cerebellum, and the dorsal root ganglion.

The emphasis on the spinal cord goes back all the way to the 19th century and Friedreich himself. It's probably discredited, largely through my own work. Medical students still think it's a disease of the spinal cord, but we have to abandon that idea.

Others research cardiomyopathy. I did too. I got several articles that I wrote about the heart in Friedreich's. But the idea is that it's a cardiac hypertrophy. The cardiac muscles of the ventricles swell and they become fibrotic, hindering their contraction. But also, the supraventricular heart—which is the right atrium and the left atrium—is involved, and patients with Friedreich's tend to get cardiac arrhythmia in addition to hypertrophic cardiomyopathy.

Announcer:

That was Dr. Arnie Koeppen talking about how frataxin deficiency impacts the heart and the brain. To access this and other episodes in our series, visit *NeuroFrontiers* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!