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ReachMD
www.reachmd.com
info@reachmd.com
(866) 423-7849

Recognizing Early Signs of LGMD2I/R9

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Dr. Vissing:
This is CME on ReachMD, and my name is John Vissing. And in this episode, we will review the clinical presentation of limb-girdle muscular dystrophy type 2I/R9.

So as for other limb-girdles, the clinical presentation can be very variable. It can have a childhood onset. This relates to more severe mutations in the FKRP gene. It can be reminiscent, almost, of the phenotype we see in Duchenne or very severe Becker muscular dystrophy patients. These patients typically lose ambulation in their late teens and are even in need of assisted ventilation when they reach age 30. This is about one-third of the patients.

Two-thirds of the patients have a milder phenotype. They are typically diagnosed later on in their 20s. Some, even later than that. And they have those symptoms early on, not being able to run so fast in school and so on, but this goes unnoticed very often. And on clinical presentation, maybe you don't see the weakness at this point, but you can see calf hypertrophy in most of the patients, which is very characteristic. Another characteristic feature is rhabdomyolysis in about a third to more 40% of the patients, actually, have a debut with rhabdomyolysis. So this should also be a red lamp, if you see a patient like this. Later on, of course, the patients do develop proximal weakness. This starts in the lower limbs first, with atrophy and weakness in the hamstring muscles, the adductor muscles, mostly. Patients start having a waddling, hyperlordotic gait. And later on, they also get weakness in the other extremities where you can find scapular winging and also weakness of the upper arm muscles.

Patients also tend to develop some cardiac affection. Not all the patients, but typically half of them do develop some kind of cardiomyopathy. This usually comes with time.There is a weak relationship with the severity of the skeletal muscle phenotype, so that patients who have a childhood onset that have a more severe cardiac phenotype. But it's not a completely straightforward relationship, so you should be really aware of this, even in the mildly affected patients.

Patients typically do not develop any affection of external eye muscles. The cognition is typically normal, also, in these patients. And the facial muscles are not affected. In patients with the childhood onset, you can see a remarkable hypertrophy of the tongue, which is something we don’t really understand so well why this occurs. And it can be bothersome now that we are providing assisted ventilation to the patients. It grows and grows with age and can be grossly enlarged when you reach the age of 60 or 70.And there, you typically have to do something about it, and mostly by reducing the tongue by surgical operations.

The CK is usually elevated always in these patients, but with time CK will drop as you get less and less muscle. And in the more severe cases, in the wheelchair-bound patients, the CK can actually be normal. But you will otherwise always see an elevated CK level in these patients.

So with this, I will wrap up. And thank you for listening.

Announcer:
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