Spinal Muscular Atrophy Diagnosis

genetic blood tests, which can confirm the diagnosis of SMA.

an electromyography (EMG) test that measures the electrical activity of a muscle or a group of muscles (in some cases)

a creatine kinase (CPK) test (to distinguish from other types of neuromuscular diseases, if necessary).

Spinal Muscular Atrophy Magnetic Resonance Imaging

Magnetic resonance imaging (MRI) can provide potential biomarkers for SMA. It has been used to assess muscle volume and fat fraction of the upper and lower extremities [1) 2)]

Twenty-five patients with genetically confirmed SMA3b underwent MRI on a 1.5-Tesla MR scanner.

Results: MRI showed significantly more severe involvement of the iliopsoas than of the gluteus maximus muscles, and more severe involvement of the triceps brachii than of the biceps brachii muscles. The quadriceps femoris muscles were severely involved. The deltoid, adductor longus, portions of the hamstrings, gracilis, sartorius, and rectus abdominis muscles were well preserved. We found a significant positive correlation between MRI changes and disease duration for gluteus maximus and triceps brachii. Follow-up MRIs of 4 patients showed disease progression

Conclusions: This study confirms the pattern of selective muscle involvement suggested by previous studies and further refines muscle MRI changes in SMA3b. Progressive muscle involvement is implicated [3)].

Hooijmans et al. from The Netherlands evaluated 13 patients with SMA and 15 controls with a 3T MRI protocol consisting of DIXON method, DTI, and T2 sequences. qMRI measures were compared between groups and related to muscle force measured with quantitative myometry. The fat fraction was significantly increased in all upper arm muscles of the patients with SMA compared to controls and correlated negatively with muscle force. Additionally, the fat fraction was heterogeneously distributed within the Triceps Brachii (TB) and Brachialis (BR) muscle but not in the Biceps Brachii (BB) muscle. Diffusion indices and water T2 relaxation times were similar between patients with SMA and healthy controls but we did find a slightly reduced MD, λ1 and λ3 in the TB of patients with SMA. Furthermore, MD positively correlated with muscle force in the TB of patients with SMA. The variation in fat fraction further substantiates the selective vulnerability of muscles. The reduced DTI indices along with the positive correlation of MD with muscle force point to myofiber atrophy. The results showed the feasibility of qMRI to map disease state in the upper arm muscles of patients with SMA. Longitudinal data in a larger cohort is needed to further explore qMRI to map disease progression and to capture possible effects of therapeutic interventions [4)].

1)

Sproule DM, Montgomery MJ, Punyanitya M, Shen W, Dashnaw S, Montes J, Dunaway S, Finkel R,

Darras B, Vivo DC, Kaufmann P. Thigh muscle volume measured by magnetic resonance imaging is stable over a 6-month interval in spinal muscular atrophy. J Child Neurol. 2011 Oct;26(10):1252-9. doi: 10.1177/0883073811405053. Epub 2011 May 13. PMID: 21572051.

[2) 3)]

Durmus H, Yilmaz R, Gulsen-Parman Y, Oflazer-Serdaroglu P, Cuttini M, Dursun M, Deymeer F. Muscle magnetic resonance imaging in spinal muscular atrophy type 3: Selective and progressive involvement. Muscle Nerve. 2017 May;55(5):651-656. doi: 10.1002/mus.25385. Epub 2017 Jan 5. PMID: 27543937.

[4)]

Hooijmans MT, Habets LE, van den Berg S, Froeling M, Asselman FL, Strijkers GJ, Jeneson JAL, Bartels B, Nederveen AJ, van der Pol WL. Multi-modal MR imaging of the upper arm muscles of patients with Spinal Muscular Atrophy. NMR Biomed. 2022 Jan 20:e4696. doi: 10.1002/nbm.4696. Epub ahead of print. PMID: 35052014.