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MR-Neurography of the sural nerve in patients with hereditary amyloidosis

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Background

Sural nerve biopsies are often performed in order to detect the underlying disease in patients suffering from unclear polyneuropathic symptoms. In transthyretin familial amyloid-polyneuropathy (TTR-FAP) the diagnostic value of invasive sural nerve biopsies is controversially discussed as it often lacks to detect amyloid deposits [Simmons et al, J Neurol Sci 1993]. As we recently reported, amyloid related nerve-injury in TTR-FAP can be unambiguously determined in large-caliber nerves (sciatic, tibial and common peroneal nerve) by applying high-resolution MR-Neurography (MRN) [Kollmer et al, Brain 2015]. However, the diagnostic yield of MRN of the small-caliber sural nerve, representing the target nerve specimen for biopsies, is still unclear and was subject to this investigation.

Methods

We prospectively enrolled 25 patients with manifest TTR-FAP, 10 asymptomatic gene-carriers with confirmed mutations in the TTR-gene, and 40 age/gender-matched healthy volunteers. Besides detailed neurological and electrophysiological examinations in all patients, a sural nerve biopsy was obtained in 12/25 manifest TTR-FAP patients. All participants underwent the following high-resolution MRN protocol (3Tesla/Magnetom/TIM-TRIO/Siemens):1) axial 2D-T2-TSE-fs (TR/TE 5970/55ms, voxel-size 0.4x0.3x3.5mm^3); 2) axial 2D-dual-echo-TSE-fs (TR 5210ms, TE1/TE2 12/73ms, voxel-size 0.4x0.3x3.5 mm^3).

On each axial imaging slice the sural nerve was identified and manually segmented. After signal-normalization (histogram-based, comparison with control population), nerve-voxels were statistically classified as nerve-lesion-voxels by operator-independent, threshold-based segmentation. The apparent-T2-relaxation-time and proton-spin-density were calculated for all nerve-lesion-voxels.

Results

Sural nerve lesion-voxels were found to be significantly higher in manifest TTR-FAP vs. controls ($p<0.0001$), in asymptomatic gene-carriers vs. controls ($p<0.0001$) and in manifest TTR-FAP vs. asymptomatic carriers ($p=0.0035$). Wilcoxon rank-sum-test revealed with high statistical significance that proton-spin-density was higher in severely affected TTR-FAP patients ($p<0.0001$), in moderate TTR-FAP ($p<0.0001$) and also in asymptomatic gene-carriers ($p=0.0003$) compared to healthy controls. The apparent-T2-relaxation-time was significantly increased in symptomatic TTR-FAP ($p<0.05$) but not in asymptomatic gene-carriers ($p=0.4286$) compared to controls.

Conclusion

MRN of the sural nerve is a new, non-invasive and highly sensitive diagnostic tool, which can clearly differentiate between symptomatic TTR-FAP, asymptomatic gene-carrier status and healthy controls by evaluating nerve-lesion-voxels and proton-spin-density. Additional analyzes of the apparent-T2-relaxation-time can further confirm symptomatic disease. Results of this evaluation may have a strong impact for a better diagnostic interpretation of negative sural nerve biopsies.

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