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**Background:** Distal-leg skin biopsies are an endorsed objective test for confirming SFPN. ENF densities ≤5th centile of the population distribution are considered diagnostic of SFPN. Most laboratories use a single threshold (e.g., 3.8 ENF/linear mm) to determine normality of ENF. The value of factoring demographics into diagnostic thresholds is untested.

**Objective:** To develop and test a model of normal ENF density that incorporates demographics.

**Materials and Methods:** With IRB permission, we obtained distal-leg skin biopsies from 373 normal volunteers (8-92 years) including 42 children. PGP9.5-immunolabeled ENF were measured using standard clinical methods.

**Results:** Young people aged 8-23 had far more ENF than older adults (426 vs. 227/mm²; p < 0.001). Females had more ENF than males (314 vs. 247/mm²; p < 0.001) and Asians had more thanagematched Whites, Blacks, and Hispanics (336 vs. 237/mm²; p < 0.001). 13 subjects ≤23 years with repeat biopsies at different ages lost 46 ENF/mm² on average per year, whereas older subjects (n = 9) lost 13 ENF/mm² per year. We developed and compared a multivariate model of ENF density incorporating age, gender, and race to the single diagnostic threshold. Had we applied the single threshold to all 105 biopsies from patients ≤40 years that our lab interpreted as having SFPN in 2012-2013, using the multivariate model, 75% would have received false negative (normal) diagnoses.

**Conclusions:** Different models yield contradictory interpretations of the same biopsies. Incorporating demographic covariates improves diagnostic sensitivity, especially for young patients. Repeat biopsies document rapid reduction in epidermal innervation during young adulthood.

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**Neuromuscular Disorders**

**Serum and muscular kl-6/muc1 are useful biomarker for gne-myopathy**


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**Introduction:** GNE-myopathy is an autosomal recessive myopathy with distal muscle weakness and the formation of rimmed vacuoles (RVs). Mutations in GNE result in the decrease of sialic acid that is necessary for glycosylation of MUC1. The extracellular domain of MUC1 has the highly glycosylated tandem repeat domain containing precursor of sialylated carbohydrate antigen KL-6. In this study, we examined KL-6/MUC1 in muscle, serum, and fibroblasts.

**Materials and methods:** The muscle biopsy specimens and laboratory data of GNE-myopathy (n = 6), sporadic inclusion body myositis (sIBM; n = 12), polymyositis (PM; n = 8), and normal control (NC; n = 5) were examined. These specimens were subjected to immunohistochemistry, immunofluorescent technique, and western blot analysis. Fibroblasts from two GNE-myopathy patients cultured with or without ManNAc were also examined by western blot analysis.

**Results:** In GNE-myopathy, inclusions and RVs were immunopositive for pTDP-43, MUC1 and KL-6. Western blot analysis showed the increase of MUC1-C and KL-6 in GNE-myopathy patients cultured with or without ManNAc. Serum and muscular KL-6/MUC1 might be a useful biomarker for gne-myopathy.

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