OBJECTIVE: To describe the clinical phenotype, long-term treatment outcome, and overall survival of sporadic late-onset nemaline myopathy (SLONM) with or without a monoclonal protein (MP).

METHODS: We conducted a retrospective chart review of patients seen between September 2000 and June 2017 and collected clinical, laboratory, and survival data. Treatment response was classified as mild, moderate, or marked as adjudged by predefined criteria.

RESULTS: We identified 28 patients with SLONM; 17 (61%) had an associated MP. Median age at symptom onset was 62 years. Diagnosis was often delayed by a median of 35 months from symptom onset. There was no difference in clinical or laboratory features between patients with or without MP. Although the majority of patients had proximal or axial weakness at onset, about 18% of patients had atypical presentations. A total of 7/9 (78%) patients receiving IV immunoglobulin (IVIg), 6/8 (75%) receiving hematologic therapy as either autologous stem cell transplant (ASCT) or chemotherapy, and 1/8 (13%) receiving immunosuppressive therapies responded to treatment ($p = 0.001$). All 3 patients with marked response were treated with IVIg; 2 of them had an MP. The 5-year and 10-year overall survival from symptom onset was 92% and 68%, respectively, with no difference between patients with or without MP.
CONCLUSION: SLONM has a wide spectrum of clinical presentations. In this contemporary case series, overall survival of patients did not seem to be affected by the presence of an MP. Initial treatment with IVIg is reasonable in all patients, followed by ASCT or chemotherapy as second-line therapy in patients with an associated MP.


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