**OBJECTIVE:** To investigate the spectrum of undiagnosed congenital myopathies (CMs) in adults presenting to our neuromuscular clinic and to identify the pitfalls responsible for diagnostic delays.

**METHODS:** We conducted a retrospective review of patients diagnosed with CM in adulthood in our neuromuscular clinic between 2008 and 2018. Patients with an established diagnosis of CM before age 18 years were excluded.

**RESULTS:** We identified 26 patients with adult-onset CM and 18 patients with pediatric-onset CM who were only diagnosed in adulthood. Among patients with adult onset, the median age at onset was 47 years, and the causative genes were RYR1 (11 families), MYH7 (3 families) and ACTA1 (2 families), and SELENON, MYH2, DNM2, and CACNA1S (1 family each). Of 33 patients who underwent muscle biopsy, only 18 demonstrated histologic abnormalities characteristic of CM. Before their diagnosis of CM, 23 patients had received other diagnoses, most commonly non-neurologic disorders. The main causes of diagnostic delays were mildness of the symptoms delaying neurologic evaluation and attribution of the symptoms to coexisting comorbidities, particularly among pediatric-onset patients.

**CONCLUSIONS:** CMs in adulthood represent a diagnostic challenge, as they may lack the clinical and myopathologic features classically associated with CM. Our findings underscore the need for a revision of the terminology and current classification of these disorders.

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