radic patients. Pathologically, Goebel et al. [10] also reported that muscle fascicles with numerous inclusion bodies were adjacent to completely normal fascicles, and such focal degeneration seems to be a characteristic feature of this disorder [6]. It is still uncertain whether familial cases of RBM share a common pathogenetic mechanism with that in sporadic RBM.

Histopathologically there was fiber type grouping in our proband, suggesting a neurogenic process as well. Although needle electromyogram of the right tibialis anterior muscle showed a few giant spikes, careful clinical examination and peripheral nerve conduction studies showed no neurogenic changes. A prominent finding in RBM is that the atrophic fibers with reducing bodies are frequently aggregated in some fascicles, sparing the rest of the fascicle [6,10]. With disease progression, the changes extend diffusely and consequently fibrotic tissue proliferation ensues [6]. This selectivity of fascicular involvement may differ from muscle to muscle, reflecting different degrees and clinical variability.

The origin and significance of the reducing bodies remain unknown. Since these inclusions are usually present around and in the vicinity of myonuclei, association with nuclear changes appears possible, more so since these bodies have the same electron density as that of chromatin granules. By immunohistochemical staining, however, these bodies have no nuclear component.

Acknowledgements

We thank Ms Fumie Uematsu for taking electron micrographs on muscle biopsies. The authors thank Dr. May Christine V. Malicdan for her critical comments on the manuscript. This work is supported in part by the "Research on Health Sciences focusing on Drug Innovation" and the "Research on Psychiatric and Neurological Diseases and Mental Health" from the Japanese Health Sciences Foundation; in part by the "Grant-in-Aid for Scientific Research" from the Japan Society for the Promotion of Science; and in part by the "Research Grant (17A-10) for Nervous and Mental Disorders" from the Ministry of Health, Labour and Welfare.

References

[1] Brooke MH, Neville HE. Reducing body myopathy. Neurology 1972;22:829–40.
[2] Hübner G, Pongratz D. Reducing body myopathy – ultrastructure and classification (author's transl). Virchows Arch A Pathol Anat Histol 1981;392:97–104.
[3] Oh SJ, Meyers GJ, Wilson Jr ER, Alexander CB. A benign form of reducing body myopathy. Muscle Nerve 1983;6:278–82.
[4] Carpenter S, Karpati G, Holland P. New observations in reducing body myopathy. Neurology 1985;35:818–27.
[5] Bertini E, Salviati G, Apollo F, Ricci E, Servidei S, Broccolini A, et al. Reducing body myopathy and desmin storage in skeletal muscle: morphological and biochemical findings. Acta Neuropathol (Berl) 1994;87:106–12.
[6] Kiyomoto BH, Murakami N, Kobayashi Y, Nihei K, Tanaka T, Takeshita K, et al. Fatal reducing body myopathy. Ultrastructural and immunohistochemical observations. J Neurol Sci 1995;128:58–65.
[7] Reichmann H, Goebel HH, Schneider C, Toyka KV. Familial mixed congenital myopathy with rigid spine phenotype. Muscle Nerve 1997;20:411–7.
[8] Goebel HH. Congenital myopathies with inclusion bodies : a brief review. Neuromuscul Disord 1998;8:162–8.
[9] Figarella-Branger D, Putzu GA, Bouvier-Labit C, Pouget J, Chateau D, Fardeau M, et al. Adult onset reducing body myopathy. Neuromuscul Disord 1999;9:580–6.
[10] Goebel HH, Halbig LE, Goldfarb L, Schober R, Albani M, Neuen-Jacob E, et al. Reducing body myopathy with cytoplasmic bodies and rigid spine syndrome : a mixed congenital myopathy. Neuropediatrics 2001;32:196–205.