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STUDIES ON UNUSUAL EXPRESSION OF NEUROFILAMENT-L MRNA  
IN A MUTANT STRAIN OF THE JAPANESE QUAIL (QUV)

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A mutant strain of the Japanese quail (QUV) that develops a neurological disorder has been established in the Nippon Institute for Biological Science. This QUV disorder is characterized by generalized quivering that occurs immediately after hatching. Histological analysis has shown that the size of the axonal calibers of Quv is smaller than that of normal controls, and that neurofilaments (NFs) are not found in Quv neurons. NFs consist of three subunit proteins, NF-L, NF-M, and NF-H. None of these NF-L proteins could be detected in Quv neuronal tissues, and a non- or hypophosphorylated form of NF-M protein existed only in a very small population of the neurons. The axonal hypotrophy, primarily caused by NF deficiency in axons, might be responsible for development of the disorder. In order to investigate the mechanism of the deficiency of NF-L protein in Quv, the expression of mRNA and the gene structure of NF-L protein were analyzed using Northern and Southern blot hybridization. When RNAs extracted from Quv brains and those of apparently healthy control quails were analyzed by Northern blot hybridization using mouse NF-L cDNA as a probe, 2.5Kb mRNA was observed in the control but not in Quv. This result showed that the deficiency of NF-L protein was responsible for the lack of NF-L mRNA in Quv. NF-M mRNA with a size of 3.5Kb was observed in both Quv and control brains. Therefore, the lack of the expression of NF-L mRNA might not affect the expression of NF-M mRNA. A restriction map of the NF-L gene was constructed from the results of the Southern blot analysis using several restriction enzymes. The map and the analysis of the NF-L gene by the PCR method showed that there was not a remarkable rearrangement of large portion of exons and introns in the Quv NF-L gene. However, some rearrangement of the NF-L gene occurred at the 5' upstream region, which should contain the regulatory sequence for the expression of NF-L mRNA. These results suggested that such a change in DNA structure might be responsible for the deficiency of the expression of NF-L mRNA in Quv.