Determining Phrike (Phrke) Phenotype Causing Mutation through Micro-array Assay Analysis

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Spontaneous mutation in mice have been a driving force in the generation of model of human disease and in assigning genes to complex physiological processes. A prominent example of this is our understanding of the myelination of neurons. Mice with changes in myelination are easily detected because of distinctive motor/behavioral changes and comprise the largest subgroup of neurological mutant lines. We identified a mouse in our colony at UNC with a phenotype common to many mice which have been shown to have defective myelination, namely a "shivering" phenotype early in life. We have assigned this mutant mouse line the name "Phrike." The mice which developed this phenotype are inbred and the strain is designated 129S6 (129). As a first step to identification of the gene carrying the mutation, we backcrossed it onto a C57BL/6N (B6) genetic background. With each generation, the mutant mice are expected to carry less 129 DNA, and more B6 DNA. Furthermore, it is expected that DNA in linkage with Phrike will be of 129 origin. We now report the use of a micro-array platform designed to distinguish between the genome of inbred mouse lines, MiniMUGA, to localize the causative mutation in our Phrike mice. Specifically, we show that the Phrike phenotype maps to chromosome 17, and two possible regions are identified, which harbor genes known to play critical roles in myelination. We discuss the possibility that Phrike represents a novel allele of one of these genes or a new chromosome 17 gene involved in neuronal function.