

Common miRNA signatures in a group of rare neuromuscular disorders

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Neuromuscular disorders (NMD) are heterogeneous group of genetic diseases that encompasses many different syndromes and diseases that either directly or indirectly impairs the function of skeletal muscle. However, there are currently no effective and common therapeutic approaches to prevent or delay the progression of these diseases. Recent studies revealed important regulatory roles for small noncoding RNAs, called microRNAs (miRNAs), in skeletal muscle function under physiological and pathological conditions. In this study, we aim to identify common miRNA signatures associated with etiopathogenesis of different neuromuscular diseases (Duchenne Muscular Dystrophy, Megaconial Congenital Muscular Dystrophy (CMD), Ullrich CMD and alpha-dystroglycanopathy), each caused by mutations in different nuclear genes encoding proteins with distinct roles. For this purpose, skeletal muscle biopsies from selected NMDs presenting mitochondrial damage (n=12, 3 from each group) and control individuals (n=3) were analyzed by using Affymetrix GeneChip miRNA 4.0 Array. To identify differentially expressed miRNAs in patients, raw data was analyzed by two different programs, MeV-SAM and Affymetrix TAC. Differentially expressed miRNAs whose expression were found to be statistically significant by both programs (miRNAs that showed an increase/decrease by 2 fold in patient samples compared to the control group) were identified as candidates. We then identified potential target genes of these candidate miRNAs by using miR-Walk and classified them by using GENE ONTOLOGY-PANTHER databases. Our results revealed that 17 miRNAs were differentially expressed in patients and 5 of these miRNAs are likely involved in skeletal muscle differentiation. Our commonality approach will provide contribution to the literature by identifying common potential therapeutic targets and/or biomarkers related to different rare NMDs