عنوان مقاله:

An Integrated Functional Analysis of Multiple RNA-Sequencing and Microarray Studies: Understanding of Comprehensive Molecular Changes in Muscular Dystrophieswith Limb-Girdle Pattern of Weakness

محل انتشار:

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خلاصه مقاله:

Muscular dystrophy (MD) is a group of genetic disorders characterized by progressive muscle wasting and weakness [1]. Molecular analysis of patient samples sheds light on understanding the pathogenesis of disease and leads to therapeutic strategies which are currently close to or in human clinical trials [2]. Duchenne (DMD) is the most common form of muscular dystrophies presenting in early childhood. It is an X-linked disorder which is caused by mutations in the largest known human gene, DMD which encodes dystrophin. Limb-girdle (LGMD) is another form of muscular dystrophies with a high prevalence. Among the various types of LGMDs, 2A is the most common form that is caused by mutations in CAPN3 gene which is normally responsible for encoding the enzyme calpain 3 [3]. LGMD2B is another common form of LGMD in European countriesdue to mutations in dysferlin gene (DYSF) [4]. Since both LGMD2A and LGMD2B cause limb girdle pattern of weakness, they can even be clinically indistinguishable from that of DMD. Because research studies on DMD are much more than other two types, understanding common pathogenetic mechanisms, between DMD and LGMDs may be useful for generalizing possible therapeutic strategies. Also, identifying type-specific pathogenetic mechanisms might suggest differential therapeutic approaches. To do this, gene set enrichment analysis of common and non-common differentially expressed genes can be useful. Herein, we performed an integrated functional analysis of 2 single cellRNA-sequencing and 3 microarray studies to obtain .subnetworks involved in common and differential pathogenesis of these three MD types

کلمات کلیدی: