

Changes in the myosin isoforms expressed during transformation of skeletal muscle phenotype

University of Birmingham - CFTR CF transmembrane conductance regulator [Homo sapiens (human)]



Description: -

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Notes: Thesis (Ph.D.) - University of Birmingham, Dept of Anatomy.
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Tags: #CFTR #CF #transmembrane #conductance #regulator #[Homo #sapiens #(human)]

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The most frequently occurring mutation in cystic fibrosis, DeltaF508, results in impaired folding and trafficking of the encoded protein. Mutations in this gene cause cystic fibrosis, the most common lethal genetic disorder in populations of Northern European descent. Am J Physiol Lung Cell Mol Physiol, 2020 Nov 1.

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. This section includes genomic Reference Sequences RefSeqs from all assemblies on which this gene is annotated, such as RefSeqs for chromosomes and scaffolds contigs from both reference and alternate assemblies. Nat Commun, 2020 Aug 26.

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These reference sequences are curated independently of the genome annotation cycle, so their versions may not match the RefSeq versions in the current genome build.

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Identify version mismatches by comparing the version of the RefSeq in this section to the one reported in above.

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The encoded protein functions as a chloride channel, making it unique among members of this protein family, and controls ion and water secretion and absorption in epithelial tissues. Model RNAs and proteins are also reported here.

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