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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Supplement table 6. The list of genes that are used to construct Myo7a gene network.** | | | | | | |  |  |  |  |
| **Gene ID** | **Gene symbol** | **Gene name** | **Location (Chr:Mb)** | **Mean expression** | **Correlation coefficient** | **N Cases** | **Correlation P value** | **Literature correlation score** | **Partial correlation P value** | **Gene function** |
| 17921 | Myo7a | myosin VIIA | Chr7: 105.199564 | 8.69 | 1.00 | 55 | 0.00 | 1.00 | #N/A | actin-binding molecular motor proteins commonly expressed in the inner ear and retina; mutations cause Usher syndrome |
| 16552 | Kif12 | kinesin family member 12 | Chr4: 62.826668 | 7.64 | 0.27 | 55 | 0.05 | 0.62 | 0.02 | molecular motor that functions with the microtubule cytoskeleton to play a role in intracellular transport and cell division; mutations cause beta cell oxidative stress |
| 208943 | Myo5c | myosin VC | Chr9: 75.079821 | 7.70 | 0.28 | 55 | 0.04 | 0.53 | 0.01 | actin binding protein expressed in epithelial and glandular tissues |
| 15371 | Hmx1 | H6 homeobox 1 | Chr5: 35.731507 | 8.33 | 0.28 | 55 | 0.04 | 0.52 | 0.01 | involved in the development of craniofacial structures; mutations cause oculoauricular syndrome |
| 74424 | Tmc5 | transmembrane channel-like gene family 5 | Chr7: 125.740811 | 7.32 | 0.39 | 55 | 0.00 | 0.49 | 0.04 | homologue of Tmc1, which is necessary for the normal function of cochlear hair cells |
| 244653 | Hydin | HYDIN, axonemal central pair apparatus protein | Chr8: 112.789148 | 7.32 | 0.30 | 55 | 0.03 | 0.45 | 0.01 | involved in cilia motility; mutations cause primary ciliary dyskinesia-5, characterized by accumulation of cerebrospinal fluid in the brain |
| 55992 | Trim3 | tripartite motif-containing protein 3 | Chr7: 112.752977 | 10.59 | 0.50 | 55 | 0.00 | 0.45 | 0.00 | localizes to cytoplasmic filaments; may play a role in myosin V-mediated cargo transport |
| 19156 | Psap | prosaposin | Chr10: 59.762218 | 13.49 | 0.36 | 55 | 0.01 | 0.44 | 0.00 | precursor for saposins A, B, C, and D, which facilitate the catabolism of glycosphingolipids with short oligosaccharide groups; mutations may cause hearing loss |
| 13628 | Eef1a2 | eukaryotic translation elongation factor 1 alpha 2 | Chr2: 180.882358 | 12.03 | 0.28 | 55 | 0.04 | 0.44 | 0.00 | delivers aminoacyl tRNA to the ribosome that is expressed in the brain, heart, and skeletal muscle; mutations result in epileptic encephalopathy and mental retardation |
| 218066 | Olfr11 | olfactory receptor 11 | Chr13: 21.730449 | 5.20 | 0.33 | 55 | 0.01 | 0.42 | 0.02 | part of a family of GPCRs that interact with odorant molecules to initiate a neuronal response that triggers the perception of smell |
| 14174 | Fgf3 | fibroblast growth factor 3 | Chr7: 152.023988 | 7.34 | 0.27 | 55 | 0.05 | 0.42 | 0.04 | amplification has been found in tumors and serves as a negative regulator of bone growth during ossification; may be involved in inner ear formation |
| 12305 | Ddr1 | discoidin domain receptor family, member 1 | Chr17: 35.818512 | 10.68 | 0.34 | 55 | 0.01 | 0.41 | 0.02 | a receptor tyrosine kinase that plays a role in communicating with cells in the microenvironment; expressed in epithelial cells and activated in collagen |
| 26927 | Foxl2 | forkhead box L2 | Chr9: 98.856026 | 7.68 | 0.27 | 55 | 0.04 | 0.41 | 0.04 | transcription factor active in eyelids, ovaries, and pituitary; mutations cause blepharophimosis, ptosis, and epicanthus inversus syndrome and ovarian cancer |
| 14366 | Fzd4 | frizzled class receptor 4 | Chr7: 96.552876 | 9.93 | -0.40 | 55 | 0.00 | 0.41 | 0.04 | receptor for the Wingless type MMTV integration site family of signaling proteins and Norrin; signaling induced by Norrin regulates vascular development of vertebrate retina and controls blood vessels in the ear |
| 59056 | Evc | Ellis van Creveld gene homolog (human) | Chr5: 37.680337 | 8.36 | 0.29 | 55 | 0.03 | 0.40 | 0.02 | may be important in normal growth and development and found in primary cilia; mutations cause Ellis-van Creveld syndrome |
| 11567 | Avil | advillin | Chr10: 126.437765 | 7.68 | 0.28 | 55 | 0.04 | 0.40 | 0.04 | actin regulatory protein that may play a role in development of neuronal cells that form ganglia |
| 11881 | Arsb | arylsulfatase B | Chr13: 94.541634 | 10.37 | 0.29 | 55 | 0.03 | 0.40 | 0.03 | hydrolyzes sulfates by breaking down glycosaminoglycans; differential expression may result in cystic fibrosis and mucopolysaccharidosis type VI |
| 228770 | Rspo4 | R-spondin family, member 4 | Chr2: 151.668663 | 7.91 | 0.28 | 55 | 0.04 | 0.40 | 0.04 | increases Wnt signaling; mutations cause anonychia congenita |
| 18641 | Pfkl | phosphofructokinase, liver, B-type | Chr10: 77.449692 | 11.93 | 0.36 | 55 | 0.01 | 0.39 | 0.00 | subunit of liver enzyme that catalyzes the conversion of D-fructose 6-phosphate to D-fructose 1,6-biphosphate in glycolysis |
| 58911 | Sumf1 | sulfatase modifying factor 1 | Chr6: 108.057015 | 8.59 | -0.32 | 55 | 0.02 | 0.39 | 0.02 | catalyzes the hydrolysis of sulfate esters and activates arylsulfatase I; mutations cause multiple sulfatase deficiency |
| 17118 | Marcks | myristoylated alanine rich protein kinase C substrate | Chr10: 36.853049 | 10.59 | 0.30 | 55 | 0.03 | 0.39 | 0.03 | substrate for protein kinase C that localizes to plasma membrane; involved in cell motility, phagocytosis, membrane trafficking, and mitogenesis |
| 69807 | Trim32 | tripartite motif-containing 32 | Chr4: 65.266020 | 10.54 | 0.33 | 55 | 0.01 | 0.39 | 0.01 | localizes to cytoplasmic bodies and nucleus, where it interacts with HIV-1 Tat protein; ubiquitinates DTNBP1 (dysbindin) and promotes its degredation |
| 15394 | Hoxa1 | homeobox A1 | Chr6: 52.105366 | 6.40 | 0.27 | 55 | 0.04 | 0.39 | 0.02 | may be involved in placement of hindbrain during development; mutations cause Bosley-Salih-Alorainy syndrome and Athabaskan brainstem dysgenesis syndrome |
| 18223 | Numbl | numb-like | Chr7: 28.043452 | 9.46 | 0.26 | 55 | 0.05 | 0.39 | 0.01 | plays a role in neurogenesis and is expressed in the rat auditory epithelium |
| 17885 | Myh8 | myosin, heavy polypeptide 8, skeletal muscle, perinatal | Chr11: 67.090626 | 6.58 | 0.31 | 55 | 0.02 | 0.38 | 0.04 | motor protein that functions in skeletal muscle contraction, predominately in fetal skeletal muscle; mutations cause Trismus-Pseudocamptodactyly Syndrome |
| 246707 | Emilin2 | elastin microfibril interfacer 2 | Chr17: 71.601514 | 7.87 | 0.34 | 55 | 0.01 | 0.38 | 0.04 | extracellular matrix glycoprotein highly homologous to Emilin1, which is involved in the elasticity of tissues |
| 170731 | Mfn2 | mitofusin 2 | Chr4: 147.247704 | 11.38 | 0.28 | 55 | 0.04 | 0.38 | 0.00 | determines shape of mitochondria; mutations cause Charcot-Marie-Tooth disease type 2A |
| 224674 | Slc37a1 | solute carrier family 37 (glycerol-3-phosphate transporter), member 1 | Chr17: 31.432428 | 7.65 | 0.29 | 55 | 0.03 | 0.38 | 0.05 | translocates glucose-6-phosphate from cytoplasm to ER lumen |
| 66264 | Ccdc28b | coiled coil domain containing 28B | Chr4: 129.296518 | 8.71 | 0.28 | 55 | 0.04 | 0.38 | 0.01 | involved in ciliogenesis and localizes to centrosomes and basal bodies; associated with Bardet-Biedl syndrome |
| 16469 | Jrk | jerky | Chr15: 74.539591 | 7.81 | -0.32 | 55 | 0.02 | 0.38 | 0.01 | conserved protein smiliar to DNA-binding proteins; Childhood Absence Epilepsy (CAE) has been mapped to this chromosomal location |
| 15526 | Hspa9 | heat shock protein 9 | Chr18: 35.097068 | 11.42 | 0.28 | 55 | 0.04 | 0.38 | 0.02 | plays a role in cell proliferation, stress response, and maintenance of mitochondria; mutations cause Even-plus syndrome and skeletal dysplasia |
| 107656 | Krt9 | keratin 9 | Chr11: 100.048095 | 7.46 | 0.29 | 55 | 0.03 | 0.38 | 0.05 | type I cytokeratin found in the terminally differentiated epidermis of palms and soles; mutations cause epidermolytic palmoplantar keratoderma |
| 17996 | Neb | nebulin | Chr2: 51.992167 | 7.02 | 0.45 | 55 | 0.00 | 0.37 | 0.02 | actin-binding protein that localizes to the thin filament of sarcomeres in skeletal muscle; may regulate actin-myosin interactions |
| 20351 | Sema4a | semaphorin 4A | Chr3: 88.239881 | 8.96 | 0.29 | 55 | 0.03 | 0.37 | 0.05 | transmembrane protein expressed in the retina and brain; mutations cause retinal degenerative diseases, such as retinitis pigmentosa type 35 and cone-rod dystrophy type 10 |
| 110084 | Dnahc1 | dynein, axonemal, heavy chain 1 | Chr14: 32.073561 | 7.58 | 0.29 | 55 | 0.03 | 0.37 | 0.05 | motor protein complex that provides support between the radial spokes and outer doublet of a sperm tail; mutations are associated with primary ciliary dyskinesia and morphological anomalies of flagella |
| 12721 | Coro1a | coronin, actin binding protein 1A | Chr7: 133.843287 | 8.28 | 0.27 | 55 | 0.05 | 0.37 | 0.04 | involved in cell cycle progression, signal transduction, apoptosis, and gene regulation |
| 110821 | Pcca | propionyl-Coenzyme A carboxylase, alpha polypeptide | Chr14: 123.063473 | 10.35 | -0.33 | 55 | 0.01 | 0.37 | 0.03 | helps catalyze the carboxylation of propionyl CoA in the mitochondrial matrix to S-methylmalonyl CoA |
| 22040 | Trex1 | three prime repair exonuclease 1 | Chr9: 108.960446 | 7.80 | 0.32 | 55 | 0.02 | 0.37 | 0.03 | major 3'->5' DNA exonuclease in human cells that may serve as a proofreading function for human DNA polymerase |
| 11733 | Ank1 | ankyrin 1, erythroid | Chr8: 24.249266 | 8.28 | 0.33 | 55 | 0.01 | 0.37 | 0.00 | links integral memrane proteins to underlying spectrin-actin cytoskeleton and plays roles in cell motility, activation, proliferation, contact, and maintenance of specialized membrane domains; mutations may cause deafness |
| 11981 | Atp9a | ATPase, class II, type 9A | Chr2: 168.459938 | 10.34 | 0.28 | 55 | 0.04 | 0.37 | 0.01 | phospholipid flippase that localizes to endosomes and trans-Golgi networks |