

Supplementary file 1

To detect potential pathways and target genes in infantile Pompe patients using computational analysis

Aynur Karadağ Gürel^{1*}, Selçuk Gürel²

¹Department of Medical Biology, School of Medicine, Usak University, Usak, Turkey

²Department of Pediatrics, School of Medicine, Bahcesehir University, İstanbul, Turkey

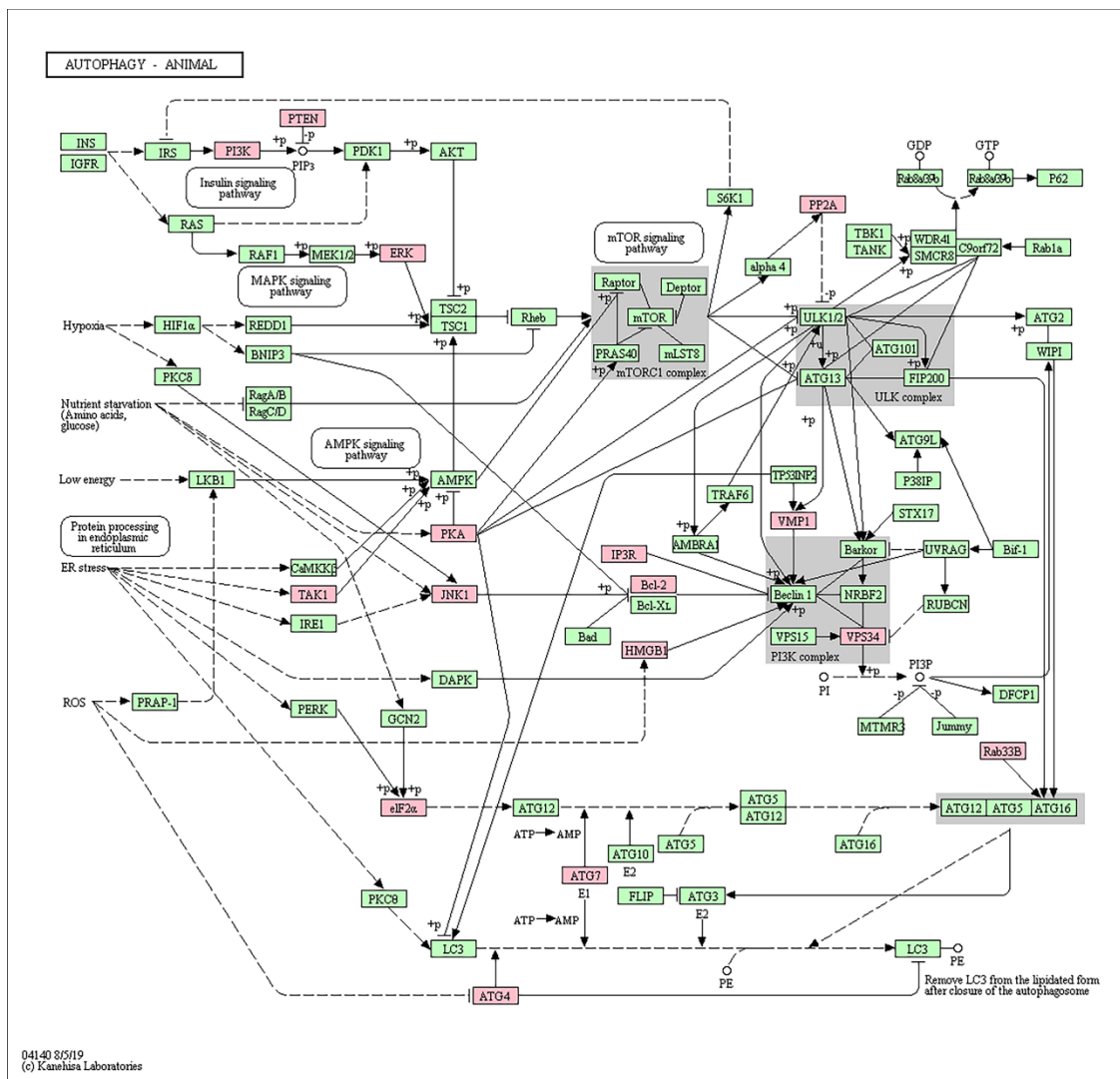


Fig.S1. KEGG pathway functional analysis of differentially expressed genes on Autophagy pathway.

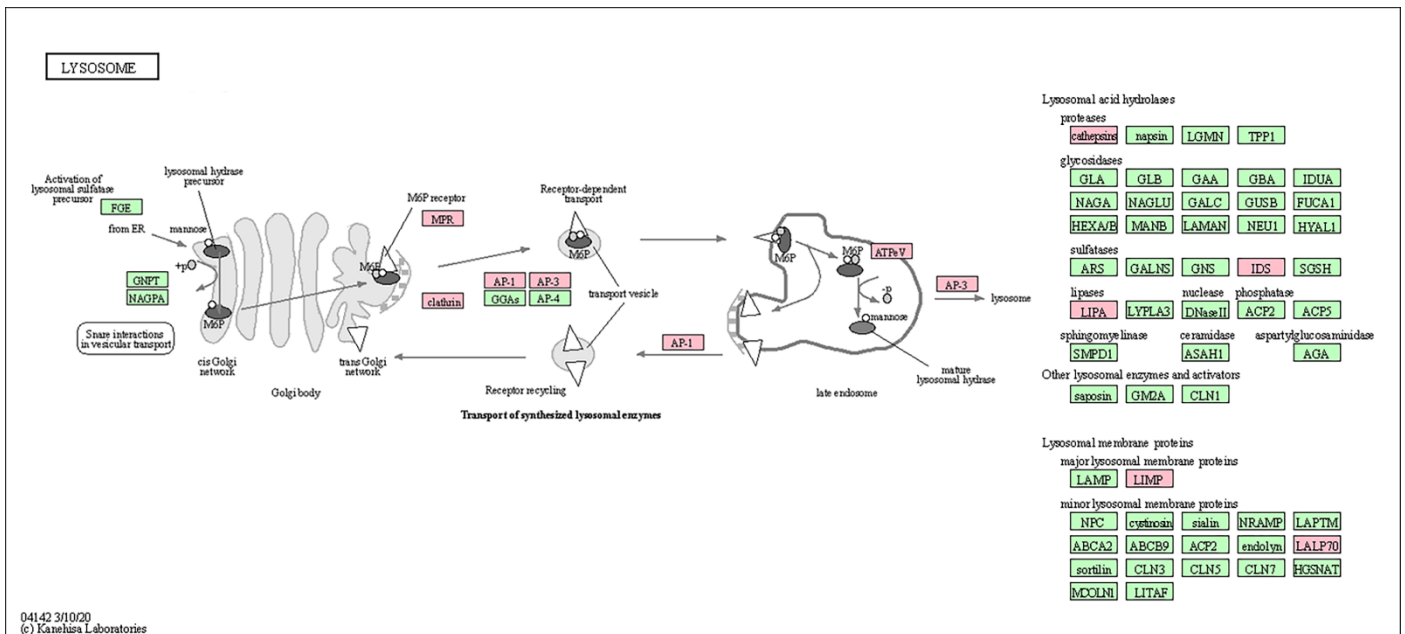


Fig.S2. KEGG pathway functional analysis of differentially expressed genes on Lysosomes pathway.

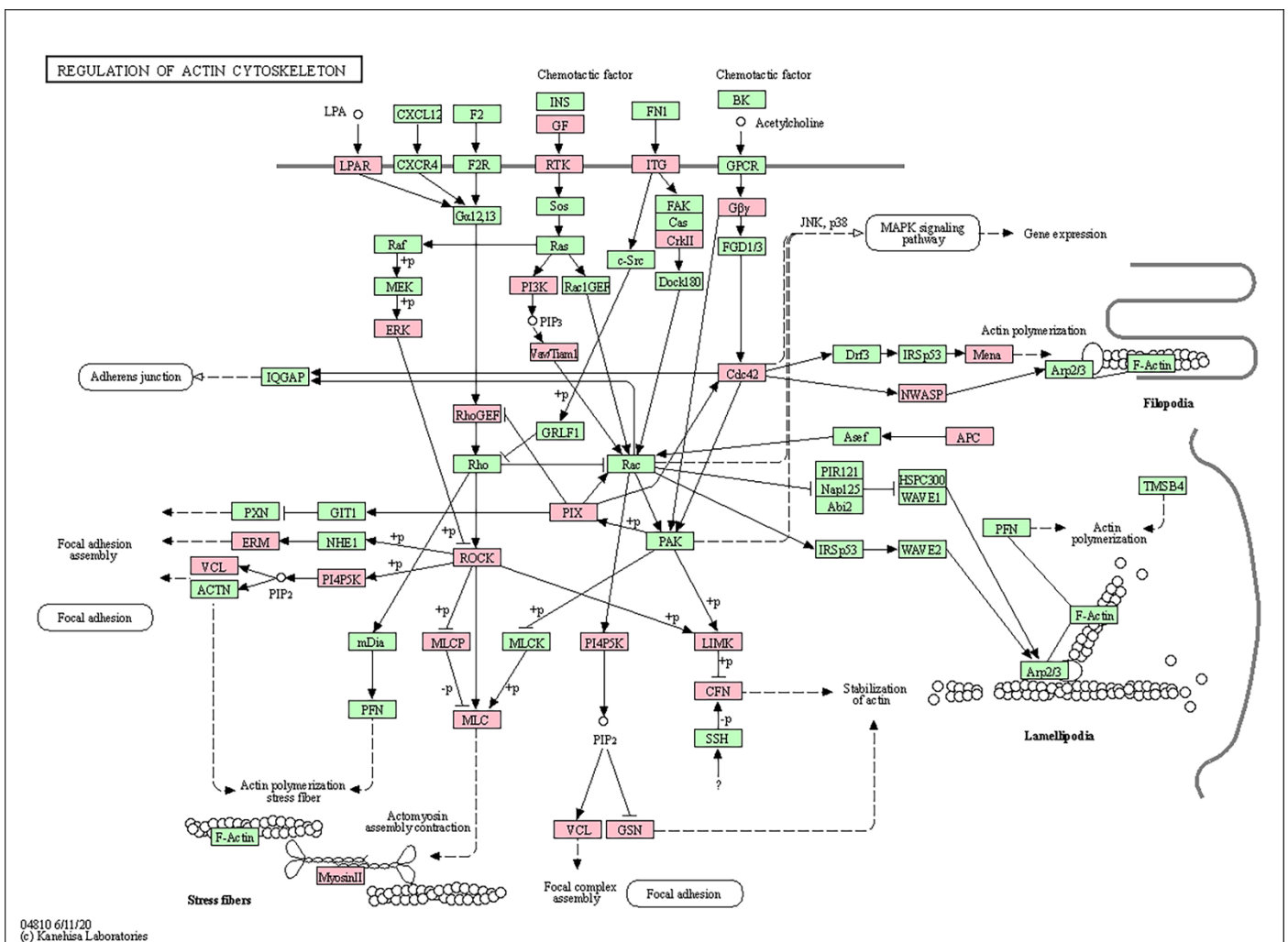


Fig.S3. KEGG pathway functional analysis of differentially expressed genes on Regulation of Actin Cytoskeleton Pathway.

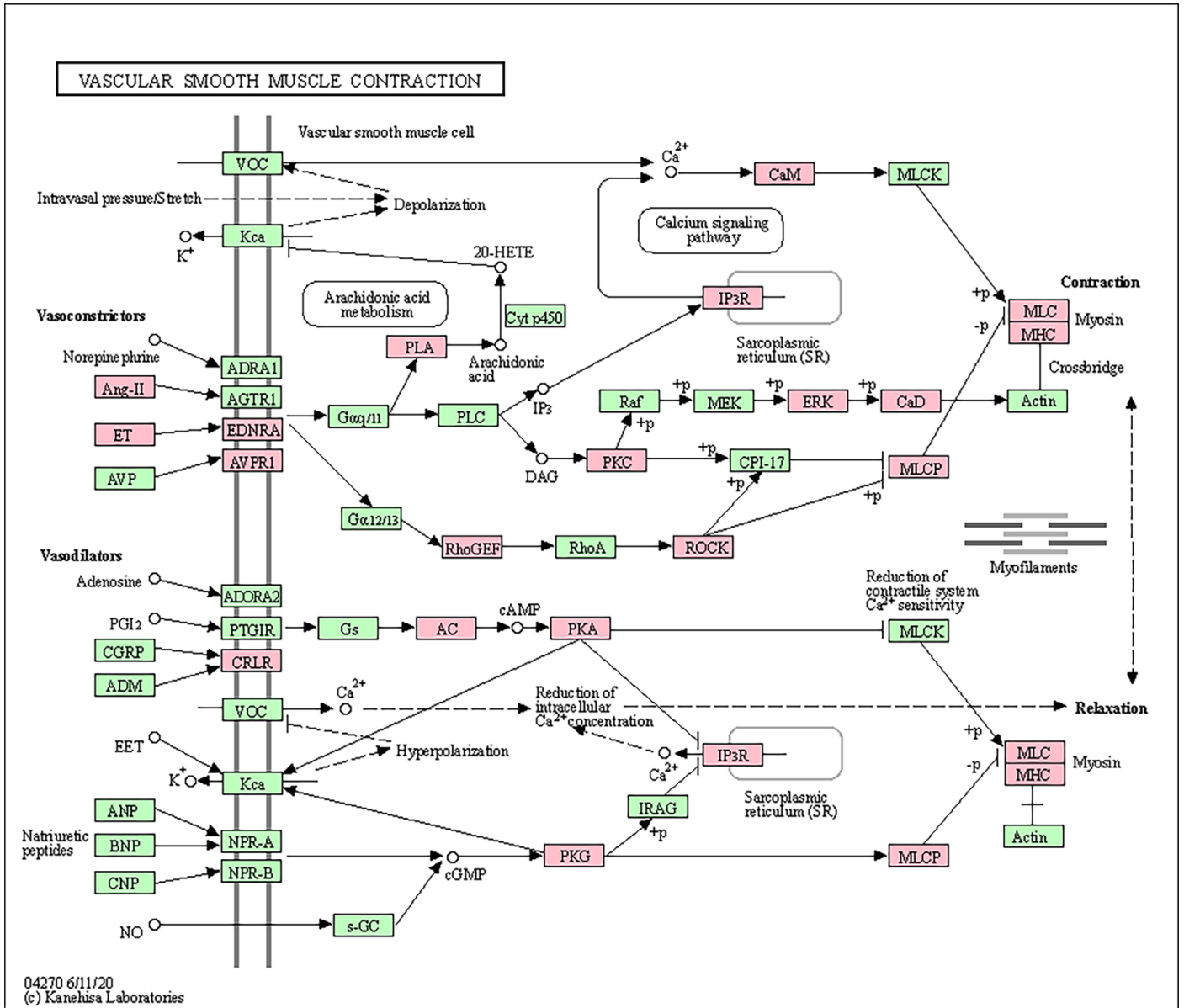


Fig.S4. KEGG pathway functional analysis of differentially expressed genes on Vascular smooth muscle contraction pathway.

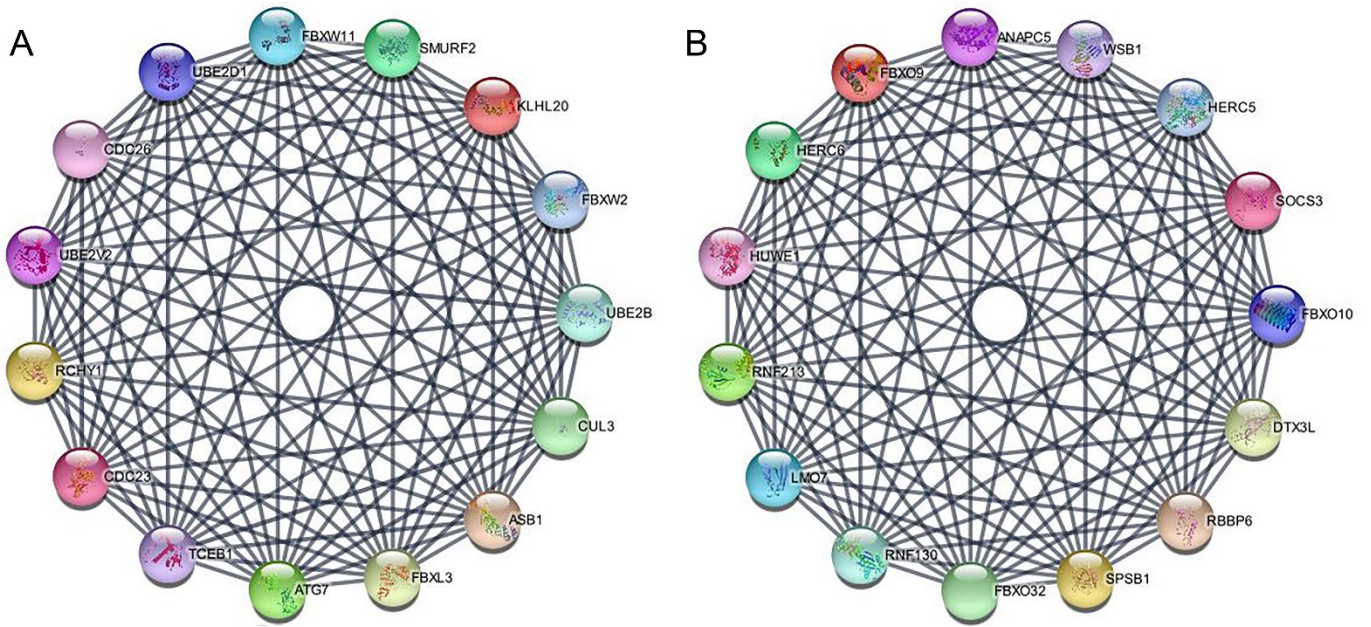


Fig.S5. Protein-protein interaction (PPI) network of 52wk poor vs pos downregulated (A) and upregulated (B) 15 hub genes by Cytoscape.

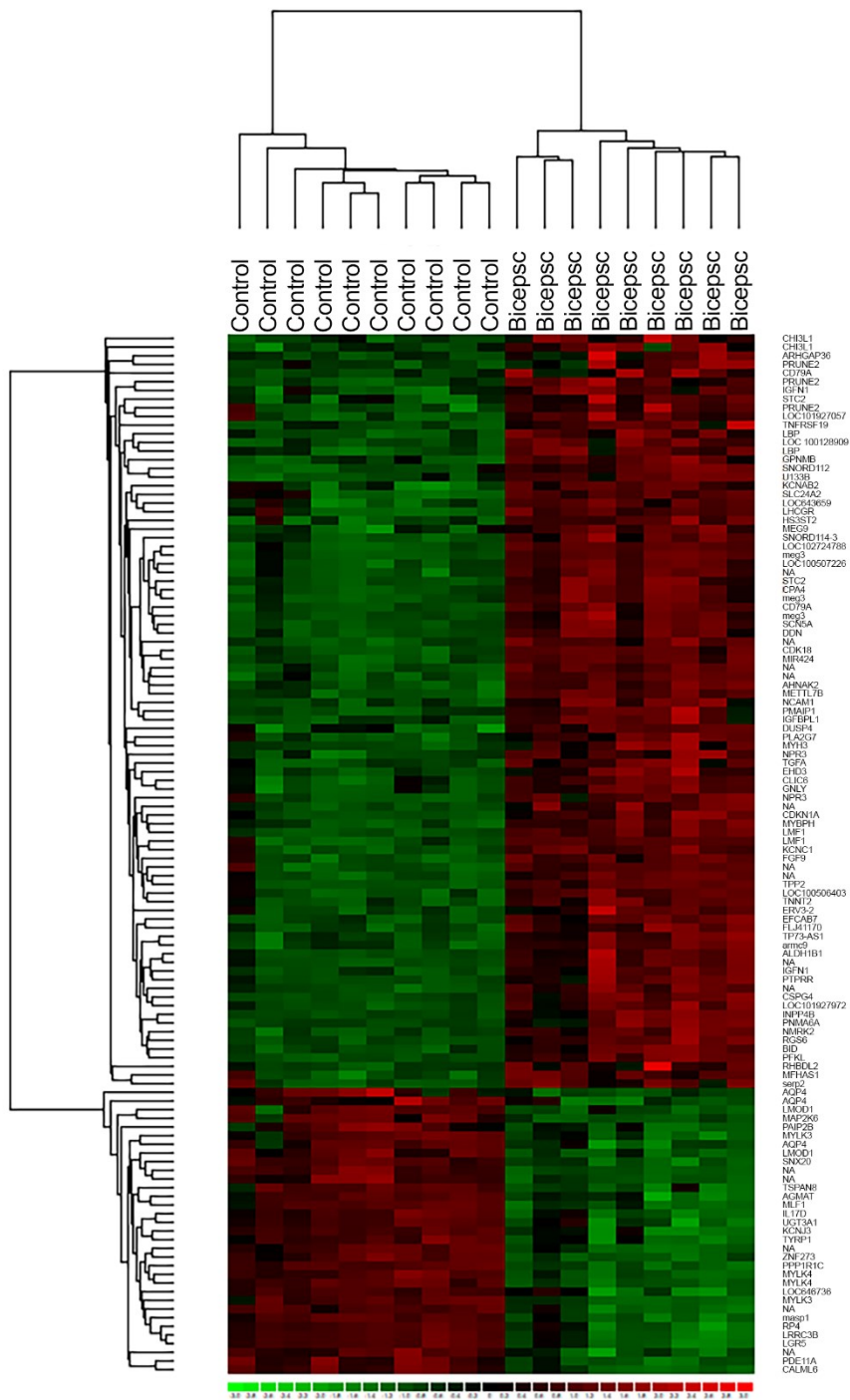


Fig. S6. A hierarchical clustering heat-map of the significant genes from biceps and control groups (p value <0.001 and $|\text{Log}_2\text{Foldchange}| \geq 1.5$). Red shows that the expression of genes is upregulated; green shows that the expression of genes is downregulated, and black shows no significant changes in gene expression).

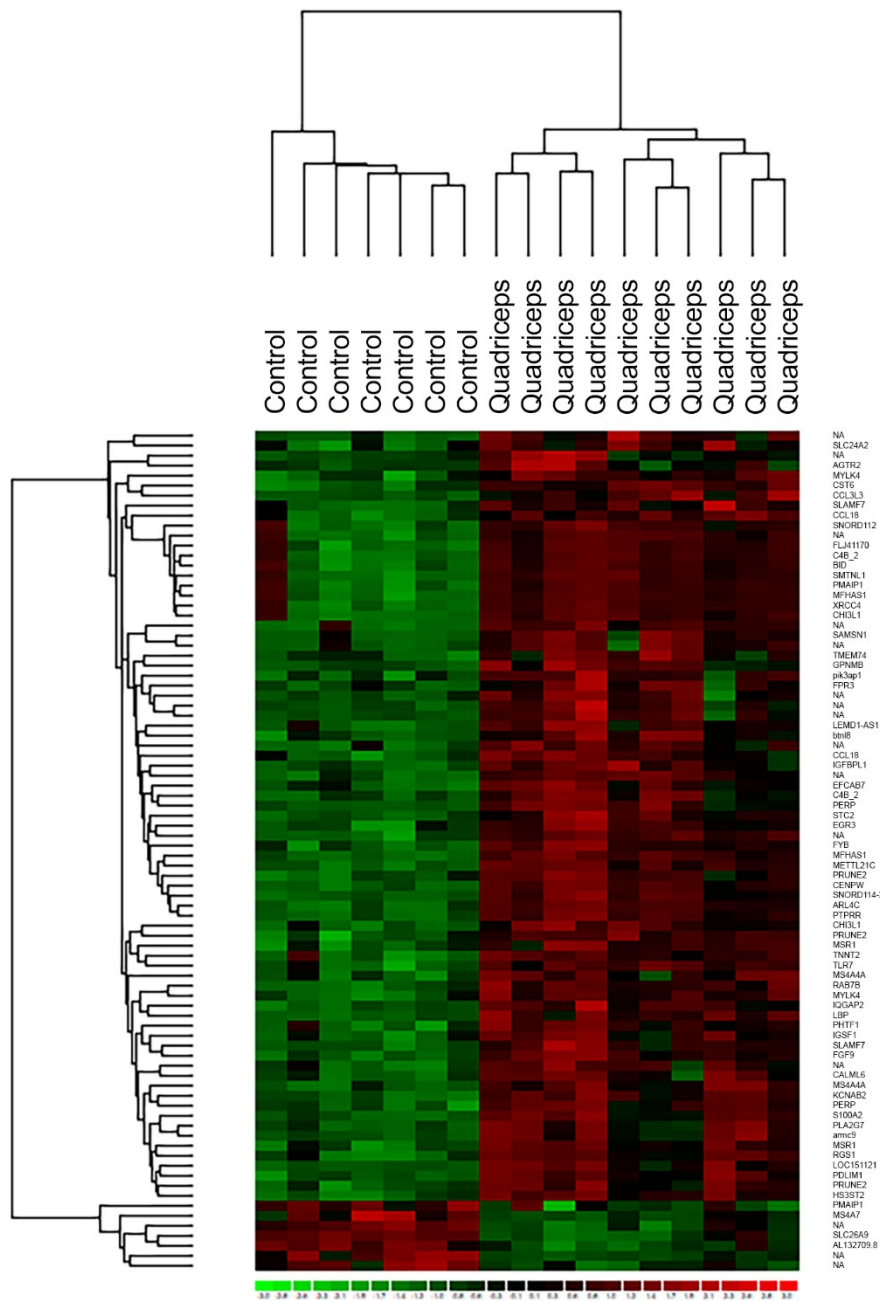


Fig. S7. A hierarchical clustering heat-map of the significant genes from quadriceps and control groups (p value < 0.001 and $|\text{Log}_2\text{Foldchange}| \geq 1.5$. Red shows that the expression of genes is upregulated; green shows that the expression of genes is downregulated, and black shows no significant changes in gene expression).

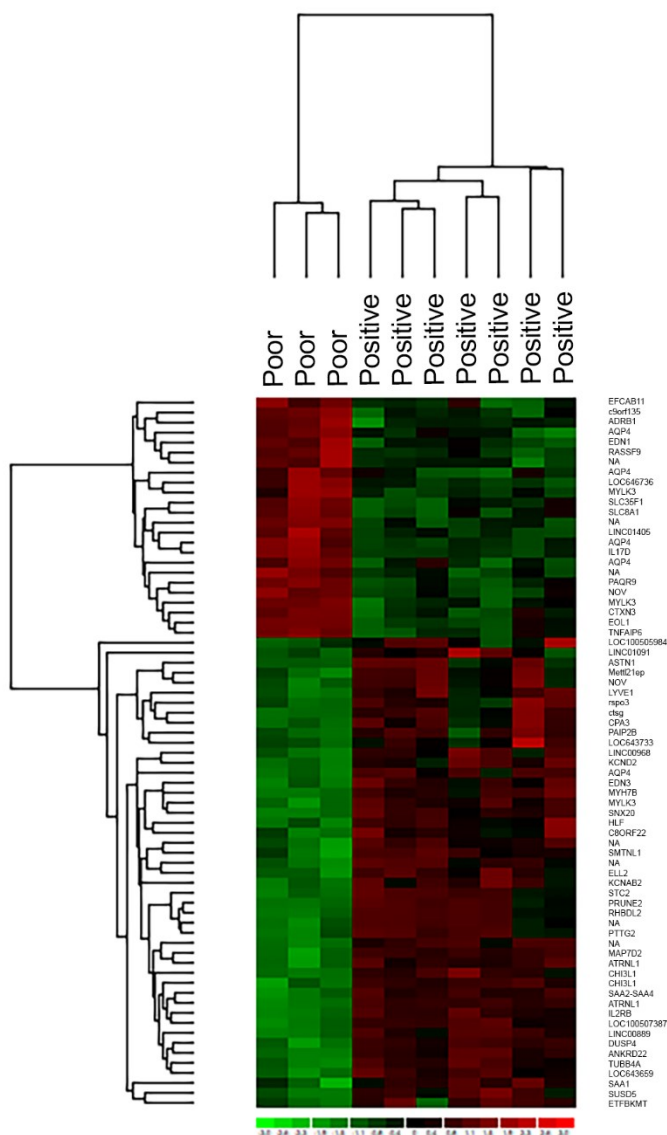


Fig. S8. A hierarchical clustering heat-map of the significant genes from Quadriceps 52wk poor and Positive groups (p value <0.001 and $|\text{Log}_2\text{Foldchange}| \geq 1.5$. Red shows that the expression of genes is upregulated; green shows that the expression of genes is downregulated, and black shows no significant changes in gene expression).

Table S1. KEGG pathway analysis results of upregulated and down regulated genes (top 10) in Biceps vs control groups.

Term	Upregulated genes	Gene Count	P _{Value}
hsa04064	NF-kappa B signaling pathway	15	0.000120029
hsa05200	Pathways in cancer	34	0.00326388
hsa04142	Lysosome	15	0.003453337
hsa04510	Focal adhesion	20	0.00919691

hsa04662	B cell receptor signaling pathway	9	0.024126821
hsa04514	Cell adhesion molecules (CAMs)	14	0.030007119
hsa04612	Antigen processing and presentation	9	0.040049277
hsa04390	Hippo signaling pathway	14	0.045888212
hsa04650	Natural killer cell mediated cytotoxicity	12	0.048026597
Term	Down-regulated genes	Gene Count	P_{Value}
hsa00020	Citrate cycle (TCA cycle)	9	4.47408E-05
hsa01200	Carbon metabolism	17	5.36166E-05
hsa04022	cGMP-PKG signaling pathway	19	0.000324151
hsa04310	Wnt signaling pathway	15	0.004367441
hsa04261	Adrenergic signaling in cardiomyocytes	15	0.004367441
hsa04350	TGF-beta signaling pathway	11	0.004988836
hsa04068	FoxO signaling pathway	14	0.008570011
hsa04120	Ubiquitin mediated proteolysis	14	0.01024335
hsa04390	Hippo signaling pathway	14	0.021649157
hsa04810	Regulation of actin cytoskeleton	17	0.032544255

Table S2. KEGG Pathway analysis results of upregulated and down regulated genes in quadriceps (0wk vs control) groups.

Up-regulated genes		Gene Count	P_{Value}
hsa04514	Cell adhesion molecules (CAMs)	20	0.0001
hsa04064	NF-kappa B signaling pathway	15	0.0001
hsa04115	p53 signaling pathway	13	0.0001
hsa04142	Lysosome	18	0.0001
hsa04612	Antigen processing and presentation	12	0.0017
hsa04620	Toll-like receptor signaling pathway	14	0.0031
hsa05200	Pathways in cancer	33	0.0071
hsa04668	TNF signaling pathway	13	0.0091
hsa04621	NOD-like receptor signaling pathway	8	0.0247
Down-regulated genes		Gene Count	P_{Value}
hsa04022	cGMP-PKG signaling pathway	11	0.0002
hsa04020	Calcium signaling pathway	11	0.0007
hsa04270	Vascular smooth muscle contraction	8	0.0033
hsa04510	Focal adhesion	10	0.0073
hsa05202	Transcriptional misregulation in cancer	8	0.0214
hsa04120	Ubiquitin mediated proteolysis	7	0.0268
hsa04261	Adrenergic signaling in cardiomyocytes	7	0.0276
hsa02010	ABC transporters	4	0.0367