

UBERON:0004129 growth plate cartilage

(Unique DEGs among top-100 found by the distance-based method, 47 genes)
5 genes are specifically related to growth plate cartilage

gm2016: predicted gene 2016.

gm2022: predicted pseudogene 2022.

gfy: olgi-associated olfactory signaling regulator.

ly6c2: lymphocyte antigen 6 complex, locus C2.

gm11127: predicted gene 11127.

cmtm7: CKLF-like MARVEL transmembrane domain containing 7:

mapkapk3: mitogen-activated protein kinase-activated protein kinase 3:

Stress-activated serine/threonine-protein kinase **involved in cytokines production, endocytosis, cell migration, chromatin remodeling and transcriptional regulation**. Following stress, it is phosphorylated and activated by MAP kinase p38- alpha/MAPK14, leading to phosphorylation of substrates. Phosphorylates serine in the peptide sequence, Hyd-X-R-X(2)-S, where Hyd is a large hydrophobic residue. MAPKAPK2 and MAPKAPK3, share the same function and substrate specificity, but MAPKAPK3 kinase activity and level in protein expression...

malrd1: MAM and LDL receptor class A domain containing 1.

gldc: glycine decarboxylase:

The glycine cleavage system **catalyzes the degradation of glycine**. The P protein (GLDC) binds the alpha-amino group of glycine through its pyridoxal phosphate cofactor; CO(2) is released and the remaining methylamine moiety is then transferred to the lipamide cofactor of the H protein (GCSH).

tecpr2: tectonin beta-propeller repeat containing 2.

kdelr3: KDEL (Lys-Asp-Glu-Leu) endoplasmic reticulum protein retention receptor 3:

Required for the **retention of luminal endoplasmic reticulum proteins**. Determines the specificity of the luminal ER protein retention system. Also **required for normal vesicular traffic through the Golgi**. This receptor recognizes K-D-E-L.

nckap1l: NCK associated protein 1 like.

trpt1: tRNA phosphotransferase 1:

Catalyzes the last step of tRNA splicing, the transfer of the splice junction 2'-phosphate from ligated tRNA to NAD to produce ADP-ribose 1''-2'' cyclic phosphate.

exoc6: exocyst complex component 6:

Component of the **exocyst complex involved in the docking of exocytic vesicles with fusion sites on the plasma membrane**. Together with RAB11A, RAB3IP, RAB8A, PARD3, PRKCI, ANXA2, CDC42 and DNMBP promotes transcytosis of PODXL to the apical membrane initiation sites (AMIS), apical surface formation and lumenogenesis.

eprs: glutamyl-prolyl-tRNA synthetase:

Catalyzes the attachment of the cognate amino acid to the corresponding tRNA in a two-step reaction- the amino acid is first activated by ATP to form a covalent intermediate with AMP and is then transferred to the acceptor end of the cognate tRNA. **Component of the GAIT (gamma interferon-activated inhibitor of translation) complex which mediates interferon-gamma- induced transcript-selective translation inhibition in inflammation processes**. Upon interferon-gamma activation and subsequent phosphorylation dissociates from the multisynthetas...

nod1: nucleotide-binding oligomerization domain containing 1:

Enhances caspase-9-mediated apoptosis. Induces NF-kappa- B activity via RIPK2 and IKK-gamma. Confers responsiveness to intracellular bacterial lipopolysaccharides (LPS). **Forms an intracellular sensing system** along with ARHGEF2 for the **detection of microbial effectors during cell invasion by pathogens**. Recruits NLRP10 to the cell membrane following bacterial infection.

tm6sf1: transmembrane 6 superfamily member 1:

May function as sterol isomerase.

mgat5: mannoside acetylglucosaminyltransferase 5:

Catalyzes the addition of N-acetylglucosamine in beta 1- 6 linkage to the alpha-linked mannose of biantennary N-linked oligosaccharides. It is one of the most **important enzymes involved in the regulation of the biosynthesis of glycoprotein oligosaccharides**.

fermt3: fermitin family member 3:

Plays a central role in cell adhesion in hematopoietic cells. Acts by activating the integrin beta-1-3 (ITGB1, ITGB2 and ITGB3). Required for integrin- mediated platelet adhesion and leukocyte adhesion to endothelial cells. Required for activation of integrin beta- 2 (ITGB2) in polymorphonuclear granulocytes (PMNs).

slco3a1: solute carrier organic anion transporter family, member 3a1:

Mediates the Na(+)-independent transport of organic anions. Mediates transport of prostaglandins (PG) E1 and E2, thyroxine (T4), deltorphin II, BQ-123 and vasopressin.

adamts6: a disintegrin-like and metallopeptidase (reprolysin type) with thrombospondin type 1 motif, 6:

foxp1: forkhead box P1:

Transcriptional repressor. Can act with CTBP1 to synergistically repress transcription but CTBP1 is not essential. **Plays an important role in the specification and differentiation of lung epithelium.** Acts cooperatively with FOXP4 to **regulate lung secretory epithelial cell fate** and regeneration by restricting the goblet cell lineage program; the function may involve regulation of AGR2. Essential transcriptional regulator of B-cell development. **Involved in regulation of cardiac muscle.**

rnf187: ring finger protein 187:

E3 ubiquitin-protein ligase that acts as **a coactivator of JUN-mediated gene activation in response to growth factor signaling** via the MAP3K1 pathway, independently from MAPK8.

ccdc88a: coiled coil domain containing 88A:

Essential for the integrity of the actin cytoskeleton and for cell migration. **Required for formation of actin stress fibers and lamellipodia.** May be involved in membrane sorting in the early endosome. Plays a role as a key modulator of the AKT-mTOR signaling pathway controlling the tempo of the process of newborn neurons integration during adult neurogenesis, including correct neuron positioning, dendritic development and synapse formation. Enhances phosphoinositide 3- kinase (PI3K)-dependent phosphorylation and kinase activity of AKT1...

sp110: Sp110 nuclear body protein:

May act as a transcription factor. **Plays a role in the innate immunity against intracellular pathogens.** Required for resistance to M.tuberculosis and L.monocytogenes. Promotes apoptosis of infected cells.

tpgs1: tubulin polyglutamylase complex subunit 1

May act in the **targeting of the tubulin polyglutamylase complex.** Required for the **development of the spermatid flagellum.**

h2-q6: histocompatibility 2, Q region locus 6:

Involved in the **presentation of foreign antigens to the immune system.**

gabra5: gamma-aminobutyric acid (GABA) A receptor, subunit alpha 5:

GABA, the major inhibitory neurotransmitter in the vertebrate brain, mediates neuronal inhibition by binding to the GABA/benzodiazepine receptor and opening an integral chloride channel.

abcb7: ATP-binding cassette, sub-family B (MDR/TAP), member 7:

Could be **involved in the transport of heme from the mitochondria to the cytosol.** Plays a central role in the maturation of cytosolic iron-sulfur (Fe/S) cluster-containing proteins.

acaal1a: acetyl-Coenzyme A acyltransferase 1A:

adk: adenosine kinase:

ATP dependent phosphorylation of adenosine and other related nucleoside analogs to monophosphate derivatives. Serves as **a potential regulator of concentrations of extracellular adenosine and intracellular adenine nucleotides.**

alas1: aminolevulinic acid synthase 1.

aldh2: aldehyde dehydrogenase 2, mitochondrial:

Is capable of **converting retinaldehyde to retinoic acid.**

alox5ap: arachidonate 5-lipoxygenase activating protein:

Required for leukotriene biosynthesis by ALOX5 (5- lipoxygenase). Anchors ALOX5 to the membrane. Binds arachidonic acid, and could **play an essential role in the transfer of arachidonic acid to ALOX5. Binds to MK-886, a compound that blocks the biosynthesis of leukotrienes.**

ap3s1: adaptor-related protein complex 3, sigma 1 subunit:

This gene encodes the sigma subunit of the heterotetrameric adaptor protein complex AP-3 which is **involved in the formation of specialized lysosome-related compartments such as melanosomes.** Pseudogenes of this gene are found on chromosomes 1, 8, 16, 17 and X. Part of the AP-3 complex, an adaptor-related complex which is not clathrin-associated. The complex is associated with the Golgi region as well as more peripheral structures. It facilitates the budding of vesicles from the Golgi membrane and may be directly involved in trafficking to lysosomes. In concert with the BLOC-1 complex, **AP-3 is required to target cargos into vesicles assembled at cell bodies for delivery into neurites and nerve terminals.**

rhoc: ras homolog family member C:

Regulates a signal transduction pathway linking plasma membrane receptors to the assembly of focal adhesions and actin stress fibers. Serves as a microtubule-dependent signal that is required for the myosin contractile ring formation during cell cycle cytokinesis. Regulates apical junction formation in bronchial epithelial cells.

arhgdib: Rho, GDP dissociation inhibitor (GDI) beta:

The protein encoded by this gene is a member of the Rho guanine nucleotide dissociation inhibitor (GDI) family. This gene is expressed at high levels in hematopoietic cells. This protein is cytosolic, and dissociation of Rho from this protein is required for membrane association and activation of Rho by Guanine Nucleotide Exchange Factors (GEFs). C-terminal truncations of this gene product have been reported to promote metastasis. Regulates the GDP/GTP exchange reaction of the Rho proteins by inhibiting the dissociation of GDP from them, and the subsequent binding of GTP to them. **Regulates reorganization of the actin cytoskeleton mediated by Rho family members.**

arpc1b: actin related protein 2/3 complex, subunit 1B:

Functions as component of the Arp2/3 complex which is involved in **regulation of actin polymerization** and together with an activating nucleation-promoting factor (NPF) **mediates the formation of branched actin networks.**

slc7a3: solute carrier family 7 (cationic amino acid transporter, y⁺ system), member 3:
The protein encoded by this gene is a member of the system y⁺ cationic amino acid transporter family. Proteins of this family allow uptake of arginine from extracellular media. **Mediates the uptake of the cationic amino acids arginine, lysine and ornithine** in a sodium-independent manner.

b2m: beta-2 microglobulin:
Component of the class I major histocompatibility complex (MHC). Involved in the **presentation of peptide antigens to the immune system.**

bckdha: branched chain ketoacid dehydrogenase E1, alpha polypeptide:
The branched-chain alpha-keto dehydrogenase complex **catalyzes the overall conversion of alpha-keto acids to acyl-CoA and CO(2)**. It contains multiple copies of three enzymatic components- branched-chain alpha-keto acid decarboxylase (E1), lipoamide acyltransferase (E2) and lipoamide dehydrogenase (E3).

bgn: biglycan:
This gene encodes a small, leucine-rich repeat proteoglycan that **plays important roles in bone mineralization and connective tissue metabolism**. The encoded preproprotein undergoes post-translational processing during which chondroitin sulfate or dermatan sulfate chains are attached before incorporation into the extracellular matrix. May be **involved in collagen fiber assembly**.

btg3: B cell translocation gene 3:
This gene encodes B cell translocation gene 3, a member of the BTG gene family. This family is defined by a conserved N-terminal domain, known to bind transcription factors, and a less conserved C-terminal domain. This protein is thought to have anti-proliferative properties, and may be **involved in regulating the G1-S transition to suppress cell cycle progression**.

btik: Bruton agammaglobulinemia tyrosine kinase:
Non-receptor tyrosine kinase indispensable **for B lymphocyte development, differentiation and signaling**. Binding of antigen to the B-cell antigen receptor (BCR) triggers signaling that ultimately leads to B-cell activation. After BCR engagement and activation at the plasma membrane, phosphorylates PLCG2 at several sites, igniting the downstream signaling pathway through calcium mobilization, followed by activation of the protein kinase C (PKC) family members. PLCG2 phosphorylation is performed in close cooperation with the adapter protein B-cell...

btrc: beta-transducin repeat containing protein:
Substrate recognition component of a SCF (SKP1-CUL1-F-box protein) E3 ubiquitin-protein ligase complex which **mediates the ubiquitination and subsequent proteasomal degradation of target proteins**. Recognizes and binds to phosphorylated target proteins. SCF(BTRC) mediates the ubiquitination of CTNNB1 and participates in Wnt signaling. SCF(BTRC) mediates the ubiquitination of NFKBIA, NFKBIB and NFKBIE; the

degradation frees the associated NFκB1 to translocate into the nucleus and to activate transcription.

tspo: translocator protein:

Can bind protoporphyrin IX and may play a role in the transport of porphyrins and heme. Was initially identified as peripheral-type benzodiazepine receptor; can also bind isoquinoline carboxamides. **Promotes the transport of cholesterol across mitochondrial membranes** and may **play a role in lipid metabolism**, but its precise physiological role is controversial. According to some reports, it is not required for steroid hormone biosynthesis.

canx: calnexin:

Calcium-binding protein that interacts with newly synthesized glycoproteins in the endoplasmic reticulum. It may **act in assisting protein assembly and/or in the retention within the ER of unassembled protein subunits**. It seems to play a major role in the quality control apparatus of the ER by the retention of incorrectly folded proteins. Associated with partial T-cell antigen receptor complexes that escape the ER of immature thymocytes, it may function as a signaling complex regulating thymocyte maturation.