

#215150 OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA; OSMED

OMIM Gene ID	HGNC	UniProtAC
120140	COL2A1	P02458
120290	COL11A2	P13942

Table 1: OMIM - UniProtAC mapping

Legend

- N1: #input proteins associated to the significant GO term
- N2: #proteins associated to the significant GO term
- P-value: Bonferroni-corrected p-value of Fisher's exact test
- *red*: go terms not related to the input proteins
- *blue*: go terms related to the input proteins (enriched uniquely by network-based method)
- *green*: go terms ancestors of terms enriched with the standard method (enriched uniquely by network-based method)

1 Standard enrichment

GO Term	N1	N2	P-value	Description
GO:0030199	2	76	0.000500176	collagen fibril organization
GO:0030574	2	78	0.000527029	collagen catabolic process
GO:0044243	2	84	0.000611795	multicellular organismal catabolic process
GO:0032963	2	96	0.000800282	collagen metabolic process
GO:0044259	2	105	0.000958234	multicellular organismal macromolecule metabolic process
GO:0044236	2	112	0.00109091	multicellular organismal metabolic process
GO:0022617	2	117	0.00119095	extracellular matrix disassembly
GO:0051216	2	118	0.00121148	cartilage development
GO:0060021	2	120	0.00125307	palate development
GO:0007605	2	175	0.002672	sensory perception of sound
GO:0050954	2	180	0.00282731	sensory perception of mechanical stimulus
GO:0001501	2	213	0.00396245	skeletal system development
GO:0022411	2	404	0.0142868	cellular component disassembly
GO:0030198	2	486	0.0206836	extracellular matrix organization
GO:0043062	2	487	0.0207689	extracellular structure organization
GO:0007600	2	586	0.0300816	sensory perception
GO:0071599	1	5	0.033117	otic vesicle development
GO:0060023	1	7	0.0463625	soft palate development

Table 2: Overrepresented GO terms with the standard enrichment

2 Network-based enrichment

GO Term	N1	N2	P-value	Description
GO:0030500	2	210	0.0190215	regulation of bone mineralization
GO:0070167	2	231	0.023026	regulation of biomineral tissue development

Table 3: Overrepresented terms with the network-based enrichment. Only terms not detected with the standard method.