

#174900 JUVENILE POLYPOSIS SYNDROME; JPS

OMIM Gene ID	HGNC	UniProtAC
600993	SMAD4	Q13485
601299	BMPR1A	P36894

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:0060391	2	17	6.9504e-05	positive regulation of SMAD protein import into nucleus
GO:0060390	2	20	9.7101e-05	regulation of SMAD protein import into nucleus
GO:0010862	2	33	0.000269839	positive regulation of pathway-restricted SMAD protein phosphorylation
GO:0060393	2	45	0.000505945	regulation of pathway-restricted SMAD protein phosphorylation
GO:0007498	2	67	0.00112995	mesoderm development
GO:0030509	2	88	0.00195633	BMP signaling pathway
GO:0042307	2	102	0.00263246	positive regulation of protein import into nucleus
GO:0090100	2	103	0.00268459	positive regulation of transmembrane receptor protein serine/threonine kinase signaling pathway
GO:0046824	2	119	0.00358814	positive regulation of nucleocytoplasmic transport
GO:0060021	2	120	0.00364895	palate development
GO:0009798	2	132	0.0044186	axis specification
GO:0007179	2	189	0.00907947	transforming growth factor beta receptor signaling pathway
GO:0090316	2	193	0.00946888	positive regulation of intracellular protein transport
GO:0042306	2	208	0.011002	regulation of protein import into nucleus
GO:2000027	2	225	0.0128786	regulation of organ morphogenesis
GO:0071560	2	238	0.0144134	cellular response to transforming growth factor beta stimulus
GO:0090092	2	239	0.014535	regulation of transmembrane receptor protein serine/threonine kinase signaling pathway
GO:1900180	2	242	0.014903	regulation of protein localization to nucleus
GO:0071559	2	248	0.0156527	response to transforming growth factor beta
GO:0032388	2	259	0.0170749	positive regulation of intracellular transport
GO:0046822	2	262	0.0174736	regulation of nucleocytoplasmic transport
GO:0021998	1	1	0.0192884	neural plate mediolateral regionalization
GO:0048338	1	1	0.0192884	mesoderm structural organization
GO:0048352	1	1	0.0192884	paraxial mesoderm structural organization
GO:0007178	2	293	0.021862	transmembrane receptor protein serine/threonine kinase signaling pathway
GO:0048589	2	296	0.0223128	developmental growth
GO:0033157	2	322	0.026412	regulation of intracellular protein transport
GO:0051222	2	349	0.0310345	positive regulation of protein transport
GO:0003272	1	2	0.0385764	endocardial cushion formation
GO:0048382	1	2	0.0385764	mesendoderm development
GO:0060896	1	2	0.0385764	neural plate pattern specification
GO:0060897	1	2	0.0385764	neural plate regionalization
GO:0048732	2	391	0.0389655	gland development
GO:0001701	2	432	0.0475773	in utero embryonic development
GO:0043009	2	442	0.0498083	chordate embryonic development

Table 2: Overrepresented GO terms with the standard enrichment

2 Network-based enrichment

GO Term	N1	N2	P-value	Description
GO:0042659	2	37	0.0015134	regulation of cell fate specification
GO:0060033	2	39	0.00168383	anatomical structure regression
GO:0032924	2	43	0.00205195	activin receptor signaling pathway
GO:0048645	2	49	0.0026723	organ formation
GO:2000136	2	49	0.0026723	regulation of cell proliferation involved in heart morphogenesis
GO:2000826	2	68	0.00517646	regulation of heart morphogenesis
GO:0010453	2	89	0.00889859	regulation of cell fate commitment
GO:0030501	2	89	0.00889859	positive regulation of bone mineralization
GO:0070169	2	97	0.0105801	positive regulation of biomineral tissue development
GO:0010470	2	108	0.0131298	regulation of gastrulation
GO:0002053	2	113	0.0143796	positive regulation of mesenchymal cell proliferation
GO:0045778	2	124	0.017329	positive regulation of ossification
GO:0010464	2	132	0.0196469	regulation of mesenchymal cell proliferation
GO:0035137	2	137	0.0211694	hindlimb morphogenesis
GO:0071333	2	148	0.0247188	cellular response to glucose stimulus
GO:0071331	2	158	0.0281841	cellular response to hexose stimulus
GO:0071326	2	160	0.0289045	cellular response to monosaccharide stimulus
GO:0060485	2	161	0.0292681	mesenchyme development
GO:0007368	2	164	0.0303724	determination of left/right symmetry
GO:0009855	2	167	0.0314973	determination of bilateral symmetry
GO:0009799	2	168	0.0318768	specification of symmetry
GO:0001678	2	169	0.0322585	cellular glucose homeostasis
GO:0045669	2	184	0.0382576	positive regulation of osteoblast differentiation
GO:0030072	2	189	0.0403709	peptide hormone secretion
GO:0071322	2	192	0.0416661	cellular response to carbohydrate stimulus
GO:0002790	2	199	0.0447679	peptide secretion
GO:0030500	2	210	0.0498671	regulation of bone mineralization

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