

#233650 COMBINED CELLULAR AND HUMORAL IMMUNE DEFECTS
WITH GRANULOMAS; CCHIDG
#601457 SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL
RECESSIVE, T CELL-NEGATIVE, B CELL-NEGATIVE, NK
CELL-POSITIVE

OMIM Gene ID	HGNC	UniProtAC
179615	RAG1	P15918
179616	RAG2	P55895

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:0002331	2	8	4.95332e-06	pre-B cell allelic exclusion
GO:0033151	2	30	7.69535e-05	V(D)J recombination
GO:0033077	2	60	0.000313121	T cell differentiation in thymus
GO:0002562	2	63	0.000345495	somatic diversification of immune receptors via germline recombination within a single locus
GO:0016444	2	63	0.000345495	somatic cell DNA recombination
GO:0002200	2	66	0.00037946	somatic diversification of immune receptors
GO:0030183	2	113	0.00111945	B cell differentiation
GO:0030217	2	154	0.00208412	T cell differentiation
GO:0042113	2	186	0.00304364	B cell activation
GO:0030098	2	263	0.0060949	lymphocyte differentiation
GO:0006310	2	294	0.00761946	DNA recombination
GO:0042110	2	319	0.00897278	T cell activation
GO:0002521	2	375	0.0124054	leukocyte differentiation
GO:0046649	2	484	0.0206777	lymphocyte activation
GO:0045321	2	579	0.0296017	leukocyte activation
GO:0016568	2	721	0.0459174	chromatin modification

Table 2: Overrepresented GO terms with the standard enrichment

2 Network-based enrichment

No novel enriched terms