

# Gene Ontology Signatures for Immune Cell-Types Inferred by Gene Expression Analysis

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**Abstract.** The Cell Ontology (CL) is a candidate OBO Foundry ontology for the representation of *in vivo* cell types. The CL is being revised to include logical definitions for cell types by using terms from other OBO ontologies such as the Gene Ontology (GO). For example, a “T-helper 17 cell (CL:0000899)” is *capable\_of* “interleukin-17 production (GO:0032620)”. Computational reasoners exploit these definitions to automate classification and ensure internal consistency in the ontology, and can sometimes expose unexpected but logically sound associations between cell types. These inter-ontology links are typically generated manually using biological knowledge and established literature. We are developing a method to go beyond these sources by integrating ontology and gene expression analysis in order to find “GO term signatures” of cell-types. Using data available from the Immunological Genome project ([www.immgen.org](http://www.immgen.org)), we have generated pair-wise gene expression comparisons between 88 immune cells types represented in the CL. By placing these datasets within an ontological context, GO term signatures are inferred not only for the sampled cell types but also for many ancestral cell types positioned higher in the hierarchy of the CL. Some of these signatures provide biological insights such as defining mature B cells as mature lymphocytes that are *capable\_of* MHC class II presentation. This work demonstrates the utility of the CL in gene expression analysis and can be used to semi-automate GO term signatures to cell types.

**Keywords:** Gene Ontology, Gene Expression Analysis, immune processes