Functional Annotation of Gene Lists

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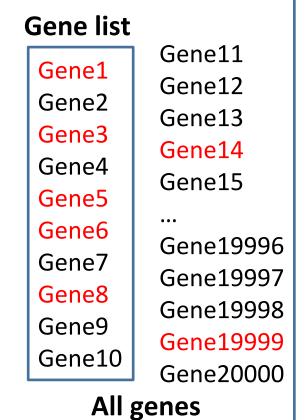
Functional Annotation

- After a bioinformatics analysis identifies a list of relevant probes, the follow-up questions are
 - What genes do the probes correspond to?
 - Look at GPL (GEO platform) data
 - What are the functions of the specific genes?
 - Look at, e.g., http://www.genecards.org
 - What biological processes or pathways are associated with these genes?
 - Use, e.g., DAVID (https://david.ncifcrf.gov) to perform a gene set enrichment analysis

Gene set enrichment analysis

In a list of interest, 50% of genes are related to a functional process, such as *cell cycle* (Gene Ontology, KEGG databases)

Out of all possible genes (background), 20% are related to the same functional process



The gene list is 2.5 times as likely to contain a cell cycle related gene as is the background

P-values (and adjusted p-values) determine whether the gene list is significantly enriched in cell cycle related genes?

Gene Ontology (GO)

- A controlled vocabularly for
 - Biological Processes (BP)
 - Molecular Functions (MF)
 - Cellular Components (CC)
- Relationships between terms leads to a "tree" structure
- http://geneontology.org
- Let's search TP53, look at Ontology, click on GO Term, and look at Graph Views

KEGG Pathways

- Molecular interactions and reactions related to metabolism, cellular processes, diseases, and others
- http://www.genome.jp/kegg/pathway.html
- Let's look at "pathways in cancer"