

Gendoo: Functional profiling of gene and disease features using MeSH vocabulary

Supplementary Figure S2 - Snapshot of Gendoo system

Gendoo provides keywords for diseases, drugs, biological phenomena and anatomy associated with OMIM entries of interest. It generates two views: high-scoring lists (a) and hierarchical tree view (b). The high-scoring list (a) shows relevant MeSH keywords sorted by score of OMIM-MeSH association. The background color of each association reflects the significance of the relationship (p -value). The hierarchical-tree view (b) illustrates keywords by using the MeSH ontology. Users can grasp the associated keywords at an appropriate level by collapsing or expanding “branch” terms (detailed information).

(a)



The screenshot shows the Gendoo interface for OMIM entry 104300 (ALZHEIMER DISEASE). It displays a table of related MeSH terms with their links, information gain scores, and significance levels. The table is as follows:

MeSH term	Link	Information Gain	01	02
<input type="checkbox"/> Alzheimer Disease	[NCBI]	0.144344	■	■
<input type="checkbox"/> Dementia	[NCBI]	0.005968654	■	■
<input type="checkbox"/> Dementia, Vascular	[NCBI]	0.00425402	■	■
<input type="checkbox"/> Memory Disorders	[NCBI]	0.0027187833	■	■
<input type="checkbox"/> Syndrome	[NCBI]	0.00244078	■	■
<input type="checkbox"/> Lewy Body Disease	[NCBI]	0.00208072	■	■
<input type="checkbox"/> Down Syndrome	[NCBI]	0.002035617	■	■
<input type="checkbox"/> Disease Progression	[NCBI]	0.001721918538	■	■
<input type="checkbox"/> Atrophy	[NCBI]	0.00133269	■	■
<input type="checkbox"/> Nerve Degeneration	[NCBI]	0.0010829017	■	■
<input type="checkbox"/> Mental Retardation	[NCBI]	0.000855815	■	■
<input type="checkbox"/> Cerebral Amyloid Angiopathy	[NCBI]	0.000778517	■	■

(b)

