

GeDiPNet Shared-Gene Disease Pairs

Similarity: Jaccard index on shared curated genes (disease\_gdp). Significance: upper-tail hypergeometric test ( $P(X \geq k)$ ), Benjamini-Hochberg FDR-corrected across all tested pairs. Corroborated Shared Genes: count of shared genes backed by  $\geq 2$  distinct disease\_gdp.main\_source values combined across both diseases (e.g. GWAS-backed for one, ClinVar-backed for the other still counts); Shared Gene Names lists each gene's own source count in parentheses. Overlap Coefficient: shared genes / MIN(each disease's own total gene count) -- complements the Jaccard-based Similarity Score by showing how much of the SMALLER disease's entire known gene-set is covered by this pair, which Jaccard (dividing by the union) can understate. Shared Cluster: cluster id if both diseases fall in the same label-propagation cluster (scripts/build\_disease\_clusters.php), blank otherwise. See scripts/build\_related.php on gedipnet.bicnirrh.res.in. Data as of: 2026-10-20 03:00:20. Filters applied: disease name contains "Cerebello-oculo-facio-genital syndrome" (5 matching pairs). · Rows: 5 · Generated: 2026-10-20 07:17:35

| Disease A                                              | Disease B                                                                                              | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|--------------------------------------------------------|--------------------------------------------------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Cerebellar hypoplasia                                  | Cerebello-oculo-facio-genital syndrome                                                                 | 1            | 0                         | MAB21L1 (1)       | 0.14286          | 1.00000             | 3.874e-4 | 7.329e-4    |                |
| Abnormality of the eye                                 | Cerebello-oculo-facio-genital syndrome                                                                 | 1            | 0                         | MAB21L1 (1)       | 0.25000          | 1.00000             | 1.937e-4 | 4.414e-4    |                |
| Abnormality of the genital system                      | Cerebello-oculo-facio-genital syndrome                                                                 | 1            | 0                         | MAB21L1 (1)       | 0.33333          | 1.00000             | 1.291e-4 | 3.269e-4    |                |
| Cerebellar, ocular, craniofacial, and genital syndrome | Cerebello-oculo-facio-genital syndrome                                                                 | 1            | 1                         | MAB21L1 (6)       | 0.33333          | 1.00000             | 1.291e-4 | 3.269e-4    |                |
| Cerebello-oculo-facio-genital syndrome                 | Congenital labioscrotal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome | 1            | 1                         | MAB21L1 (3)       | 0.50000          | 1.00000             | 6.457e-5 | 1.985e-4    |                |