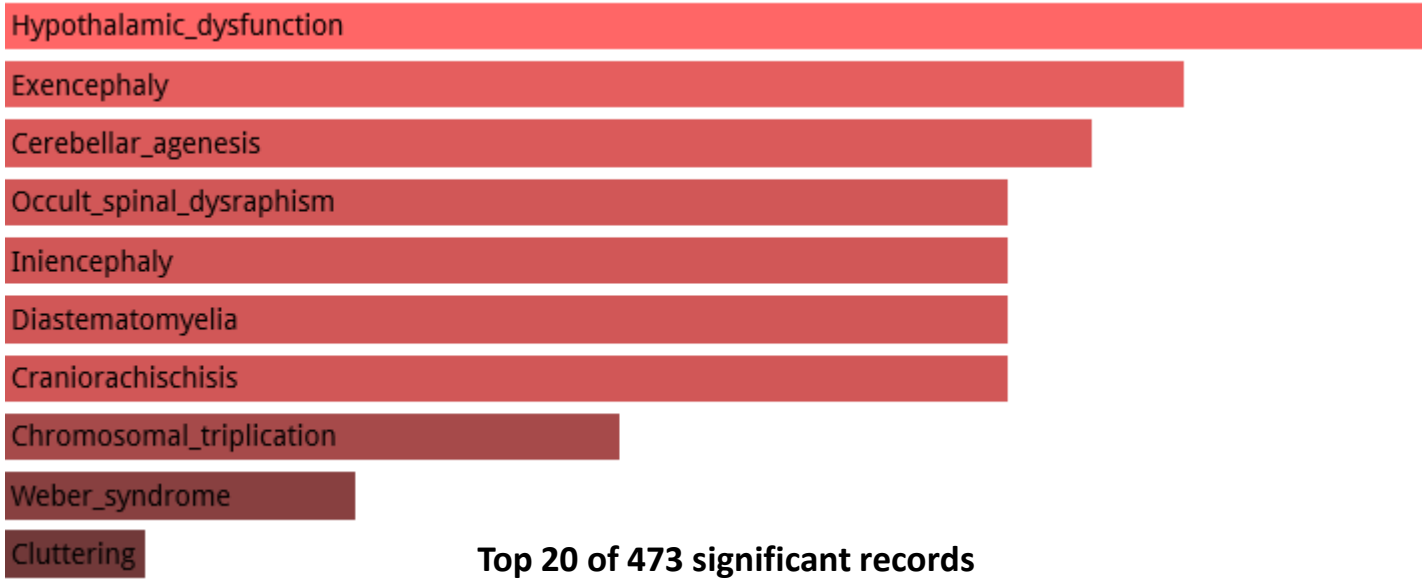
Supplemental Figure S5. Identification of genes implicated in more than 1,000 records classified as human rare diseases using the Enrichr bioinformatics platform (see Methods for details).

Rare Diseases GeneRIF Gene Lists: 8,405 genes

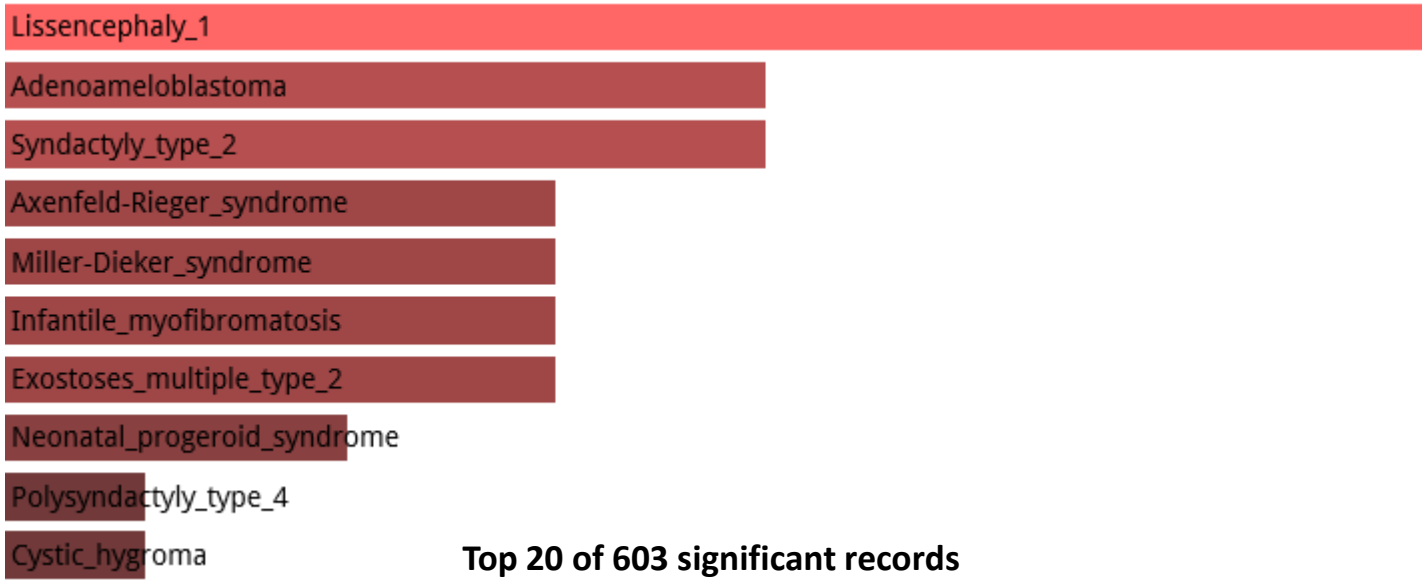


Top 20 of 473 significant records

Term	Overlap	P-value	Adjusted P-value
Hypothalamic_dysfunction	90/128	8.12E-11	6.55E-08
Exencephaly	158/256	1.38E-10	6.55E-08
Cerebellar_agenesis	198/335	1.7E-10	6.55E-08
Occult_spinal_dysraphism	157/255	2.05E-10	6.55E-08
Diastematomyelia	157/255	2.05E-10	6.55E-08
Craniorachischisis	157/255	2.05E-10	6.55E-08
Iniencephaly	157/255	2.05E-10	6.55E-08
Chromosomal_triplication	201/344	4.9E-10	1.37E-07
Weber_syndrome	94/139	8.85E-10	2.2E-07
Cluttering	106/162	1.41E-09	3.16E-07
Mental_retardation_epilepsy	190/326	1.92E-09	3.9E-07
Single_ventricular_heart	73/104	5.49E-09	1.02E-06
Sydenham's_chorea	209/369	8.42E-09	1.34E-06
Chorea_minor	209/369	8.42E-09	1.34E-06
Antisocial_personality_disorder	46/59	1.93E-08	2.87E-06
Sudden_infant_death_syndrome	103/162	2.37E-08	3.31E-06
Stress_cardiomyopathy	110/176	3.16E-08	4.16E-06
N_syndrome	222/401	3.89E-08	4.83E-06
Basilar_migraine	127/210	4.85E-08	5.7E-06
Corpus_callosum_agenesis	89/138	7.92E-08	8.85E-06

Rare Diseases GeneRIF Gene Lists: 8,405 genes

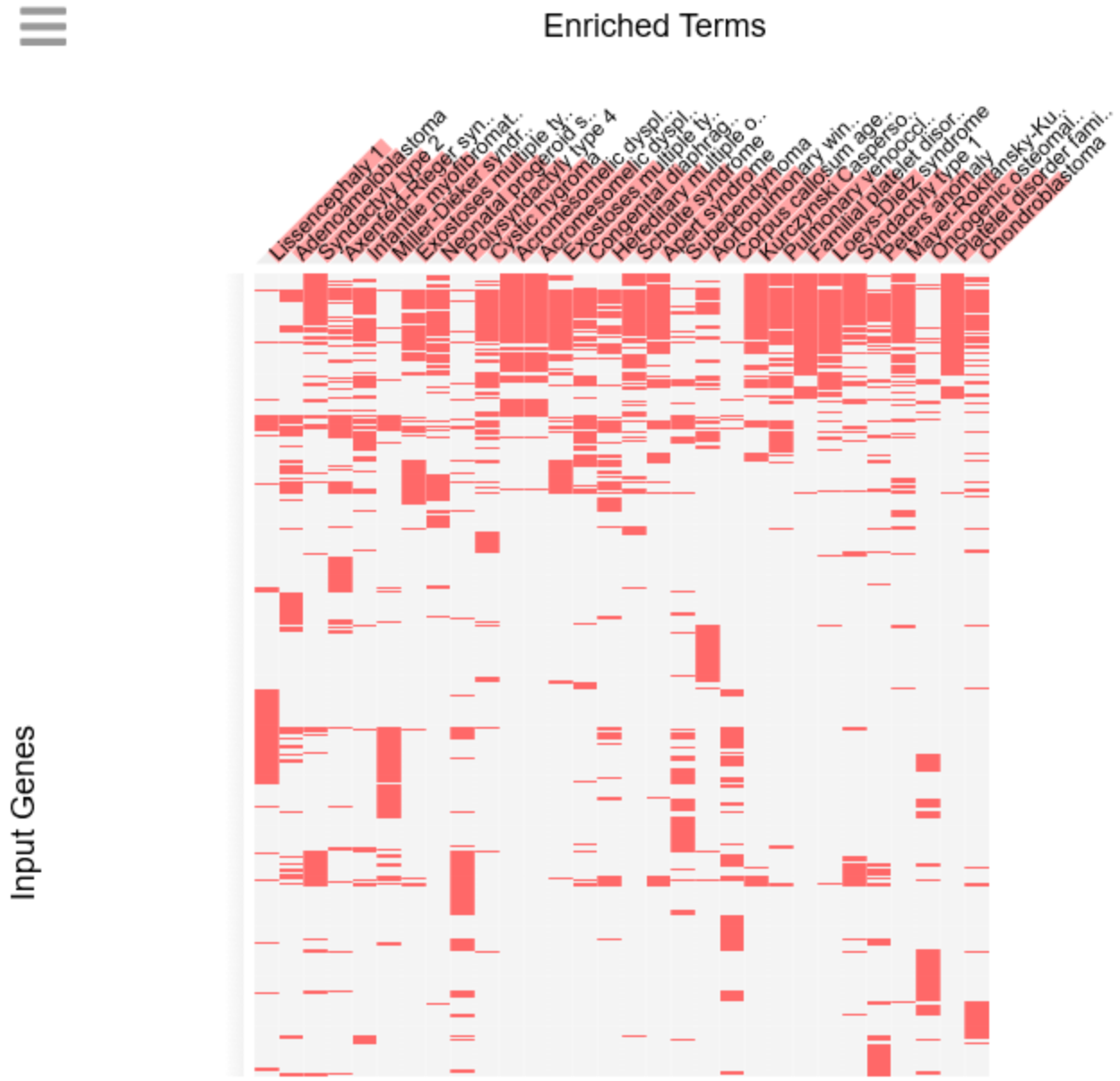
Rare Diseases GeneRIF ARCHS4 Predictions: 8,405 genes



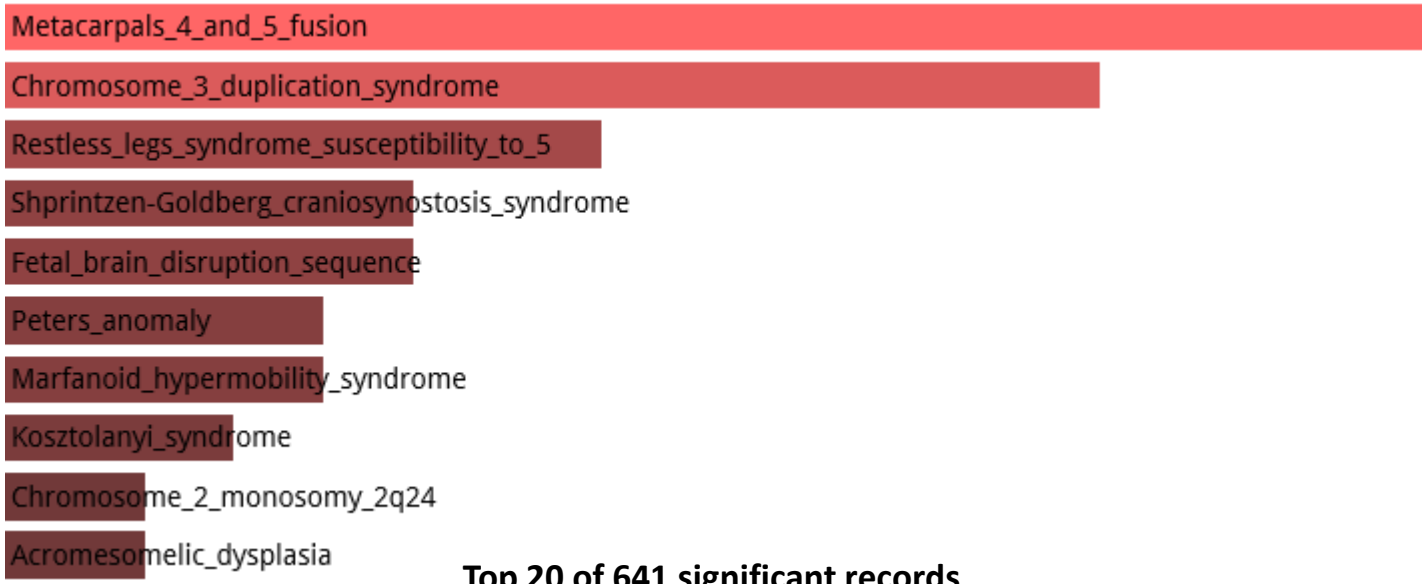
Top 20 of 603 significant records

Term	Overlap	P-value	Adjusted P-value
Lissencephaly_1	154/200	4.79E-24	1.08E-20
Adenoameloblastoma	151/200	4.35E-22	3.26E-19
Syndactyly_type_2	151/200	4.35E-22	3.26E-19
Axenfeld-Rieger_syndrome	150/200	1.85E-21	5.94E-19
Miller-Dieker_syndrome	150/200	1.85E-21	5.94E-19
Exostoses_multiple_type_2	150/200	1.85E-21	5.94E-19
Infantile_myofibromatosis	150/200	1.85E-21	5.94E-19
Neonatal_progeroid_syndrome	149/200	7.69E-21	2.16E-18
Polysyndactyly_type_4	148/200	3.11E-20	6.97E-18
Cystic_hygroma	148/200	3.11E-20	6.97E-18
Acromesomelic_dysplasia_Hunter_Thompson_type	147/200	1.22E-19	2.11E-17
Acromesomelic_dysplasia	147/200	1.22E-19	2.11E-17
Exostoses_multiple_type_1	147/200	1.22E-19	2.11E-17
Scholte_syndrome	146/200	4.69E-19	6.58E-17
Congenital_diaphragmatic_hernia	146/200	4.69E-19	6.58E-17
Hereditary_multiple_osteochondromas	146/200	4.69E-19	6.58E-17
Aortopulmonary_window	145/200	1.76E-18	1.88E-16
Corpus_callosum_agenesis	145/200	1.76E-18	1.88E-16
Apert_syndrome	145/200	1.76E-18	1.88E-16
Subependymoma	145/200	1.76E-18	1.88E-16

Rare Diseases GeneRIF ARCHS4 Predictions: 8,405 genes



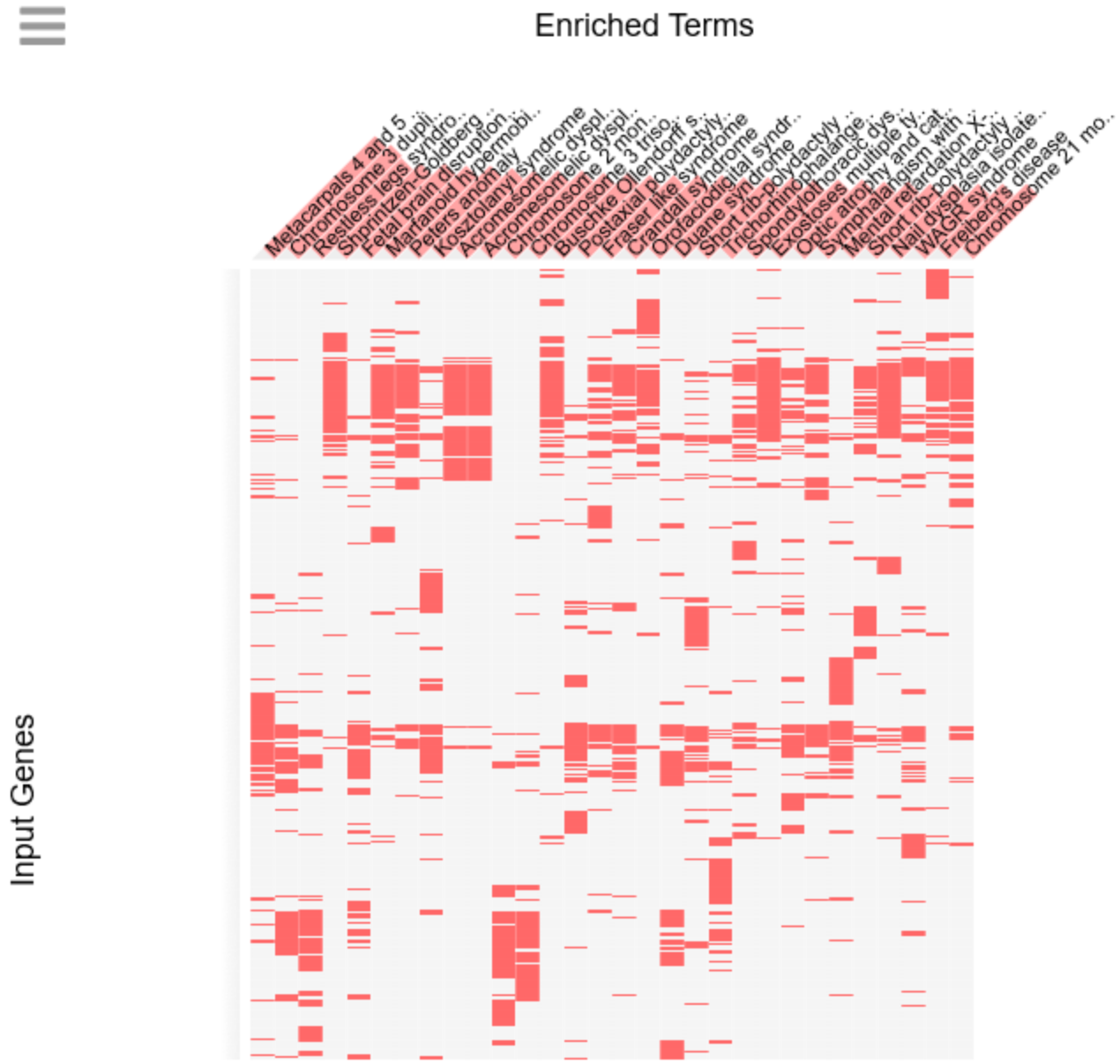
Rare Diseases AutoRIF ARCHS4 Predictions: 8,405 genes



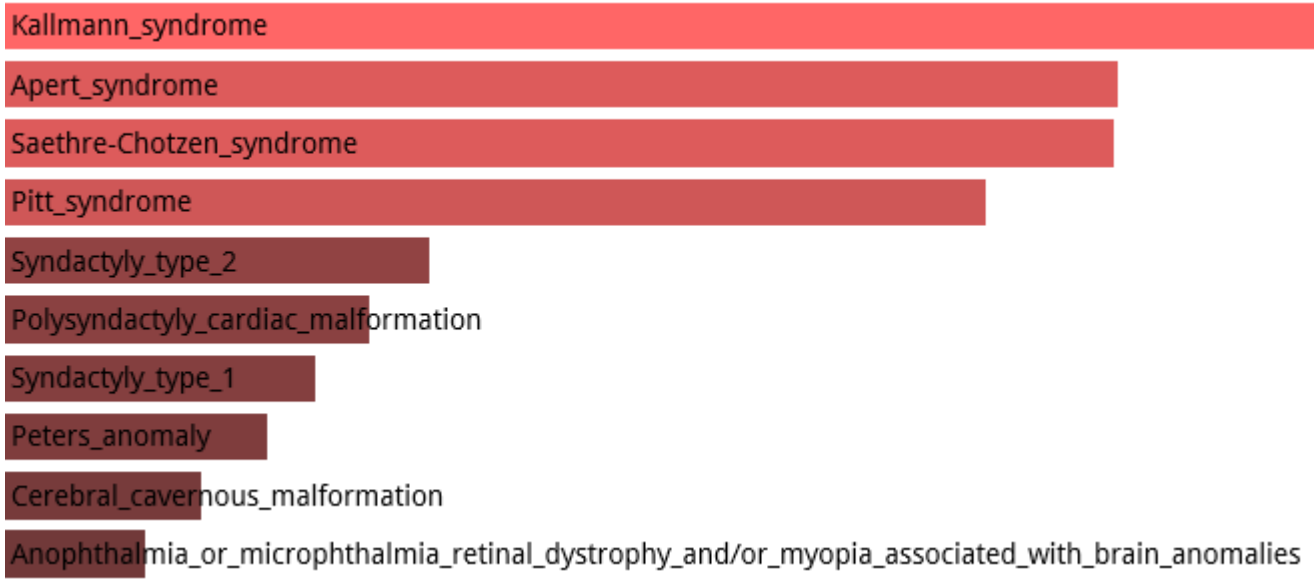
Top 20 of 641 significant records

Term	Overlap	P-value	Adjusted P-value
Metacarpals_4_and_5_fusion	163/200	1.33E-30	4.97E-27
Chromosome_3_duplication_syndrome	160/200	2.69E-28	5.01E-25
Restless_legs_syndrome_susceptibility_to_5	155/200	1.01E-24	1.25E-21
Shprintzen-Goldberg_craniosynostosis_syndrome	153/200	2.21E-23	1.65E-20
Fetal_brain_disruption_sequence	153/200	2.21E-23	1.65E-20
Peters_anomaly	152/200	9.95E-23	5.3E-20
Marfanoid_hypermobility_syndrome	152/200	9.95E-23	5.3E-20
Kosztolanyi_syndrome	151/200	4.35E-22	2.03E-19
Chromosome_2_monosomy_2q24	150/200	1.85E-21	6.28E-19
Acromesomelic_dysplasia_Hunter_Thompson_type	150/200	1.85E-21	6.28E-19
Acromesomelic_dysplasia	150/200	1.85E-21	6.28E-19
Chromosome_3_trisomy_3p	149/200	7.69E-21	2.2E-18
Buschke_Ollendorff_syndrome	149/200	7.69E-21	2.2E-18
Fraser_like_syndrome	148/200	3.11E-20	6.81E-18
Crandall_syndrome	148/200	3.11E-20	6.81E-18
Orofaciodigital_syndrome_11	148/200	3.11E-20	6.81E-18
Postaxial_polydactyly_mental_retardation	148/200	3.11E-20	6.81E-18
Short_rib-polydactyly_syndrome_type_4	147/200	1.22E-19	2.28E-17
Trichorhinophalangeal_syndrome_type_3	147/200	1.22E-19	2.28E-17
Duane_syndrome	147/200	1.22E-19	2.28E-17

Rare Diseases AutoRIF ARCHS4 Predictions: 8,405 genes



Rare Diseases AutoRIF Gene Lists: 8,405 genes



Top 20 of 1,116 significant records

Term	Overlap	P-value	Adjusted P-value
Kallmann_syndrome	159/247	9.25E-13	3.25E-09
Apert_syndrome	143/219	2.58E-12	3.25E-09
Saethre-Chotzen_syndrome	136/206	2.62E-12	3.25E-09
Pitt_syndrome	241/410	4.01E-12	3.73E-09
Syndactyly_type_2	124/188	2.62E-11	1.95E-08
Polysyndactyly_cardiac_malformation	77/104	3.2E-11	1.98E-08
Syndactyly_type_1	122/185	3.85E-11	2.04E-08
Peters_anomaly	75/101	4.53E-11	2.1E-08
Cerebral_cavernous_malformation	157/252	5.65E-11	2.33E-08
Anophthalmia_or_microphthalmia_retinal_dystrophy_and/or_myopia_associated_with_brain_anomalies	182/301	6.82E-11	2.36E-08
X-linked_periventricular_heterotopia	89/126	6.98E-11	2.36E-08
Aniridia	171/282	1.85E-10	5.73E-08
Dominant_cleft_palate	163/267	2.37E-10	6.76E-08
Craniofacial_and_skeletal_defects	94/137	2.64E-10	7.01E-08
Anodontia	140/223	2.84E-10	7.03E-08
Glaucoma_congenital	202/345	3.48E-10	8.08E-08
Childhood-Onset_Schizophrenia	168/278	3.87E-10	8.46E-08
Hennekam_syndrome	139/223	6.68E-10	1.38E-07
Kurczynski_Casperson_syndrome	114/176	8.57E-10	1.68E-07
Osteochondroma	177/299	1.31E-09	2.44E-07

Rare Diseases AutoRIF Gene Lists: 8,405 genes