The human disease network

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Disorder Class

- Bone
- Cancer
- Cardiovascular
- Connective tissue disorder
- Dermatological
- Developmental
- Ear, Nose, Throat
- Endocrine
- Gastrointestinal
- Hematological
- Immunological
- Metabolic
- Muscular
- Neurological
- Nutritional
- Ophthalmological
- Psychiatric
- Renal
- Respiratory
- Skeletal
- multiple
- Unclassified

Disorder Name

18 Acampomelic campodelic dysplasia
26 Achondrogenesis-hypochondrogenesis, type II
53 Adrenal hyperplasia, congenital
77 Aldosterone to renin ratio raised
87 Alpha-1-antichymotrypsin deficiency
92 Alpha-thalassemia/mental retardation syndrome
96 Alternating hemiplegia of childhood
107 Analgesia from kappa-opioid receptor agonist, female-specific
117 Angiotensin I-converting enzyme
126 Anterior segment anomalies and cataract
129 Anxiety-related personality traits
137 Apparent mineralocorticoid excess, hypertension due to
144 Arrhythmogenic right ventricular dysplasia
162 Athabaskan brainstem dysgenesis syndrome
171 Attention-deficit hyperactivity disorder
182 Bannayan-Riley-Ruvalcaba syndrome
192 Beare-Stevenson cutis gyrata syndrome
198 Beta-2-adrenoreceptor agonist, reduced response to
210 Blepharophimosis, epicanthus inversus, and ptosis
217 Bone mineral density variability
275 Carpal tunnel syndrome, familial
279 Cavernous malformations of CNS and retina
287 Central hypoventilation syndrome
292 Cerebrooculofacioskeletal syndrome
294 Cerebrovascular disease, occlusive
313 Cholesteryl ester storage disease
320 Chorioretinosis, hypochyroidism, and respiratory distress
329 Chylomicronemia syndrome, familial
344 Cold-induced autoinflammatory syndrome
347 Colonic aganglionosis, total, with small bowel involvement
357 Conotruncal anomaly face syndrome
377 Craniofacial anomalies, empty sella turcica, corneal endothelial changes
378 Craniofacial-deafness-hand syndrome
379 Craniofacial-skeletal-dermatologic dysplasia
396 Cyclic ichthyosis with epidermolytic hyperkeratosis
418 Dentinogenesis imperfecta, Shields type
422 Dermatofibrosarcoma protuberans
434 Dilated cardiomyopathy with woolly hair and keratoderma
438 Disordered steroidogenesis, isolated
439 Dissection of cervical arteries
441 Dopamine beta-hydroxylase deficiency
452 Dyggve-Melchior-Clausen disease
453 Dysalbuminemic hyperthyroxinemia
461 Dyssegmental dysplasia, Silverman-Handmaker type
463 Dystransthyretinemic hyperthyroxinemia
471 Elite sprint athletic performance
474 Emery-Dreifuss muscular dystrophy
527 Fatty liver, acute, of pregnancy
535 Fibrocalculous pancreatic diabetes
539 Fibular hypoplasia and complex brachydactyly
544 Fluorouracil toxicity, sensitivity to
545 Focal cortical dysplasia, Taylor balloon cell type
549 Foveomacular dystrophy, adult-onset, with choroidal neovascularization
558 Fuchs endothelial corneal dystrophy
584 Giant platelet disorder, isolated
594 Glomerulocystic kidney disease, hypoplastic
604 Glutathione synthetase deficiency
626 Greig cephalopolysyndactyly syndrome
646 Hearing loss, low-frequency sensorineural
665 Hemosiderosis, systemic, due to aceruloplasminemia
679 High-molecular-weight kininogen deficiency
699 Homocystinuria-megaloblastic anemia, cbl E type
701 Homozygous 2p16 deletion syndrome
727 Hyperferritinemia-cataract syndrome
733 Hyperkalemic periodic paralysis
734 Hyperkeratotic cutaneous capillary-venous malformations
780 Hypoparathyroidism-retardation-dysmorphism syndrome
785 Hypoplastic enamel pitting, localized
792 Hystrix-like ichthyosis with deafness
803 Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
809 Infundibular hypoplasia and hypopituitarism
830 Jervell and Lange-Nielsen syndrome
833 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
843 Keratitis-ichthyosis-deafness syndrome
845 Keratoderma, palmoplantar, with deafness
847 Keratosis palmoplantaris striata
868 Laryngonychocutaneous syndrome
891 Leukoencephalopathy with vanishing white matter
913 Lower motor neuron disease, progressive, without sensory symptoms
930 Lynch cancer family syndrome II
942 Malignant hyperthermia susceptibility
945 Mandibuloacral dysplasia with type B lipodystrophy
959 Mastocytosis with associated hematologic disorder
969 Medullary cystic kidney disease
982 Melorheostosis with osteopoikilosis
1002 Methionine adenosyltransferase deficiency, autosomal recessive
1007 Methylcobalamin deficiency, cblG type
1016 Mitochondrial complex deficiency
1050 Myelomonocytic leukemia, chronic
1056 Myoglobinuria/hemolysis due to PGK deficiency
1057 Myokymia with neonatal epilepsy
1080 Nephrogenic syndrome of inappropriate antidiuresis
1090 Neural tube defects, maternal risk of
1096 Neurofibromatosis-Noonan syndrome
1104 Nevus, epidermal, epidermolytic hyperkeratotic type
1105 Newfoundland rod-cone dystrophy
1113 Noncompaction of left ventricular myocardium
1119 Norwalk virus infection, resistance to
1133 Oculodentoosseous dysplasia
1140 Oligodontia-colorectal cancer syndrome
1153 Ossification of the posterior longitudinal spinal ligaments
1164 Osteoporosis-pseudoglioma syndrome
1174 Palidoponotonigral degeneration
1183 Papillary serous carcinoma of the peritoneum
1227 Pigmentation of hair, skin, and eyes, variation in
1229 Pigmented paravenous chorioretinal atrophy
1232 Pituitary ACTH-secreting adenoma
1238 Pneumonitis, desquamative interstitial
1239 Pneumothorax, primary spontaneous
1263 Prion disease with protracted course
1265 Progressive external ophthalmoplegia with mitochondrial DNA deletions
1267 Prolactinoma, hyperparathyroidism, carcinoid syndrome
1297 Pyruvate dehydrogenase deficiency
1325 Rhizomelic chondrodysplasia punctata
1335 Robinow syndrome, autosomal recessive
1347 Sandhoff disease, infantile, juvenile, and adult forms
1361 Schwartz-Jampel syndrome, type 1
1376 Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis
1383 Severe combined immunodeficiency
1396 Silver spastic paraplegia syndrome
1401 Skin fragility-woolly hair syndrome
1414 Solitary median maxillary central incisor
1432 Spondylocarpotarsal synostosis syndrome
1438 Stapes ankylosis with broad thumb and toes
1446 Stevens-Johnson syndrome, carbamazepine-induced
1456 Subcortical laminar heterotopia
1466 Sweat chloride elevation without CF
1475 Tarsal-carpal coalition syndrome
1499 Thanatophoric dysplasia, type I and II
1518 Thanatophoric dysplasia, type II
1519 Transposition of great arteries, dextro-looped
1526 Trifunctional protein deficiency
1528 Trismus-pseudocamptodactyly syndrome
1542 Ullrich congenital muscular dystrophy
1545 Unna-Thost disease, nonepidermolytic
1555 VATER association with hydrocephalus
1565 Vitamin D-dependent rickets type I
1580 Warfarin resistance/sensitivity
1596 Weissenbacher-Zweymuller syndrome
1611 XLA and isolated growth hormone deficiency
1614 Yemenite deaf-blind hypoplastic thyroid syndrome
2327 Chronic infections, due to opsonin defect
2354 Congenital bilateral absence of vas deferens
2385 Creatine deficiency syndrome, X-linked
2785 Hypoplastic left heart syndrome
3037 Multiple cutaneous and uterine leiomyomata
3144 Optic nerve coloboma with renal disease
3212 Persistent hyperinsulinemic hypoglycemia of infancy
3229 Pigmented adrenocortical disease, primary isolated
3260 Premature chromosome condensation w/ microcephaly, mental retardation
3512 Total iodide organification defect
3558 Ventricular fibrillation, idiopathic
4291 Cerebral cavernous malformations
5170 Ovarian hyperstimulation syndrome
5233 Placental steroid sulfatase deficiency

Supporting Information Figure 13 | Bipartite-graph representation of the diseasome. A disorder (circle) and a gene (rectangle) are connected if the gene is implicated in the disorder. The size of the circle represents the number of distinct genes associated with the disorder. Isolated disorders (disorders having no links to other disorders) are not shown. Also, only genes connecting disorders are shown.