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| --- | --- |
| **Row Labels** | **Sum of count** |
| 12q14 microdeletion syndrome | 1 |
| 1p36 deletion syndrome | 4 |
| 9q deletion syndrome | 1 |
| Abnormal corpus callosum | 1 |
| abnormal development | 13 |
| Acute coronary syndrome | 1 |
| Acute lymphoblastic leukemia | 915 |
| Acute monocytic leukemia | 3 |
| Acute myeloid leukemia | 102 |
| Acute promyelocytic leukemia | 7 |
| Addison''s disease | 1 |
| Adenocarcinoma | 7 |
| Adenoid cystic carcinoma | 71 |
| Adrenocortical carcinoma | 3 |
| Aetanephric adenomas | 1 |
| Alcoholism | 15 |
| Alveolar rhabdomyosarcoma | 2 |
| Alzheimer''s disease | 7 |
| Amyotrophic lateral sclerosis | 63 |
| Anaplastic large cell lymphoma | 1 |
| Anaplastic thyroid cancer | 3 |
| Angio-oedema | 7 |
| Aniridia syndrome | 3 |
| Aortic dissecting aneurysms | 36 |
| Apert syndrome | 1 |
| Arrhythmogenic right ventricular cardiomyopathy | 1 |
| Asthma | 11 |
| Astrocytoma | 32 |
| Atopic | 1 |
| Attention deficit hyperactivity disorder | 21 |
| Atypical hemolytic uremic syndrome | 3 |
| Autism | 407 |
| Autoimmune disease | 3 |
| Autosomal dominant nocturnal frontal lobe epilepsy | 1 |
| Autosomal dominant partial epilepsy | 1 |
| Bannayan-Riley-Ruvalcaba syndrome | 1 |
| Barakat syndrome | 1 |
| Barrett''s adenocarcinoma | 10 |
| Barrett''s syndrome | 21 |
| Basal cell lymphoma | 41 |
| Basal-like breast cancer | 1 |
| Beckwith-Wiedemann syndrome | 12 |
| Behavioral abnormalities | 1 |
| Berardinelli-seip congenital generalized lipodystrophy | 2 |
| Bethlem myopathy | 1 |
| Biliary cancer | 2 |
| Bipolar disorder | 88 |
| Bladder cancer | 9 |
| body mass index | 1 |
| Bone cancer | 3 |
| Brachydactyly type E | 1 |
| Breast cancer | 13862 |
| Burkitt''s lymphoma | 53 |
| Cancer | 2614 |
| Carcinoma ex pleomorphic adenoma | 5 |
| Cardiomyopathy | 2 |
| Carotid artery disease | 2 |
| Central neurocytomas | 13 |
| Centronuclear myopathy | 1 |
| Cervical cancer | 18 |
| Charcot-marie-tooth disease | 7 |
| Chondrodysplasia | 1 |
| Chordoma | 40 |
| Chronic lymphocytic leukemia | 519 |
| Chronic motor tic disorder | 4 |
| Chronic myelogenous leukemia | 1 |
| Chronic myeloid leukemia | 4 |
| cleft palate | 1 |
| CNS Malformation Syndrome | 1 |
| Cockayne syndrome | 1 |
| Cognitive impairment | 6 |
| Colon cancer | 3 |
| Colorectal cancer | 151 |
| Cone-rod dystrophy | 1 |
| Congenital abnormalities | 2 |
| Congenital anomalies of the kidney and urinary tract | 65 |
| Congenital diaphragmatic hernia | 27 |
| Congenital heart defect | 3 |
| Congenital long qt syndrome | 1 |
| Cornelia de Lange syndrome | 1 |
| Coronary artery disease | 2 |
| Coronary Disease | 1 |
| Costello syndrome | 1 |
| craniofacial dysmorphism | 3 |
| Crohn''s disease | 3 |
| Cryptorchidism | 2 |
| Cutaneous malignant melanoma | 3 |
| Cutaneous T-cell lymphoma | 10 |
| Darier disease | 1 |
| Denys-drash syndrome | 2 |
| Depressive disorder | 6 |
| Developmental delay | 80 |
| Diamond-Blackfan anemia | 2 |
| Diffuse large b-cell lymphoma | 18 |
| Digeorge syndrome | 3 |
| DiGeorge-Velo cardiofacial | 1 |
| Dihydropyrimidine dehydrogenase deficiency | 1 |
| Disease | 6 |
| Disorders of sex development | 5 |
| DNA damage stimulus | 2 |
| DNA repair | 2 |
| Ductal carcinoma | 8 |
| Dwarfism | 2 |
| Dyslexia | 1 |
| dysmorphic | 9 |
| Dysmorphic features | 7 |
| early-passage human iPS cells | 1211 |
| Ectopia Lentis | 1 |
| Effusion lymphoma | 6 |
| Emanuel syndrome | 2 |
| Emphysema | 4 |
| Encephalopathy | 1 |
| Endometrial cancer | 16 |
| Endometrioid adenocarcinoma | 9 |
| Ependymoma | 214 |
| Epilepsy | 28 |
| Epithelial cancer | 1 |
| Esophageal adenocarcinoma | 12 |
| Esophageal cancer | 17 |
| Esophageal squamous carcinoma | 33 |
| Ewing''s sarcoma | 52 |
| Familial wilms tumor | 1 |
| Fever | 3 |
| Fibroblasts | 2 |
| Follicular lymphoma | 37 |
| Fragile x | 1 |
| Frasier syndrome | 1 |
| Fumarase deficiency | 1 |
| Gastric adenocarcinoma | 10 |
| Gastric cancer | 63 |
| Gastrointestinal cancer | 1 |
| Gastrointestinal stromal cancer | 28 |
| genitourinary abnormalities | 1 |
| Glaucoma | 7 |
| Glioblastoma | 9 |
| Glioblastoma multiforme | 234 |
| Glioma | 218 |
| Glomerulonephritis | 3 |
| GLUT1 deficiency syndrome | 5 |
| GLUT2 deficiency syndrome | 1 |
| GLUT3 deficiency syndrome | 2 |
| Glycogen storage disease | 1 |
| growth disorder | 2 |
| head and neck squamous cell carcinoma | 2 |
| Heart disease | 8 |
| Heart failure | 2 |
| Hematopoietic stem cell | 2 |
| Hepatoblastoma | 2 |
| Hepatocellular carcinoma | 70 |
| Hereditary coagulopathy | 2 |
| Hereditary prostate cancer | 1 |
| High-grade prostatic intraepithelial neoplasia | 31 |
| High-proliferative meningiomas | 1 |
| Hirschsprung''s Disease | 21 |
| HIV/AIDS | 2 |
| Hodgkin''s lymphoma | 45 |
| Holoprosencephaly | 10 |
| Honadal dysgenesis | 4 |
| Human papillomavirus | 1 |
| Human rectal carcinomas | 1 |
| Hydrolethalus syndrome | 1 |
| Hyperglycemia | 1 |
| Hypertension | 1 |
| Hypoplastic left heart syndrome | 8 |
| Hypotonia | 1 |
| idiopathic generalized epilepsy | 1 |
| Idiopathic thrombocytopenic purpura | 2 |
| Immune disease | 3 |
| Incontinentia Pigmenti | 12 |
| infertile | 2 |
| Intellectual disability | 53 |
| Intestinal disease | 2 |
| Intracranial aneurysm | 12 |
| Intracranial arachnoid cysts | 13 |
| Intracranial ependymoma | 10 |
| Intracranial tumor | 3 |
| Invasive pancreatic ductal carcinoma | 9 |
| Ischemic vascular disease | 1 |
| Kartagener syndrome | 1 |
| laryngomalacia | 2 |
| Leukemia | 10 |
| LiNormalfraumeni syndrome | 1 |
| Liposarcoma | 18 |
| liposarcomas | 3 |
| Lissencephaly | 1 |
| Liver carcinoma | 11 |
| Lobular carcinoma | 1 |
| Long-qt syndrome | 1 |
| low-grade B-cell lymphoma tumor | 32 |
| Lung adenocarcinoma | 185 |
| Lung cancer | 613 |
| Lung squamous cell carcinoma | 9 |
| lymphocytic leukemia | 37 |
| Lymphoma | 4 |
| Macular degeneration | 6 |
| Maculopathy | 35 |
| Malaria | 6 |
| Malignant fibrous histiocytomas | 12 |
| Malignant germ cell tumour | 1 |
| Malignant glioma | 4 |
| Malignant melanoma | 7 |
| Malignant mesothelioma | 14 |
| Malignant peripheral nerve sheath tumor | 18 |
| Man infertility | 1 |
| Mantle cell lymphoma | 25 |
| Meckel-Gruber syndrome | 1 |
| Medullary thyroid carcinoma | 7 |
| Medulloblastoma | 94 |
| Melanoma | 460 |
| Mental retardation | 40 |
| Merkel cell carcinoma | 2 |
| Metanephric adenoma | 9 |
| microdeletion syndrome | 2 |
| micrognathia | 1 |
| Migraine | 1 |
| Miller-Dieker syndrome | 1 |
| Monoclonal gammopathy of undetermined significance | 4 |
| Mortal | 2 |
| Mucolipidosis types II | 1 |
| Multiple endocrine neoplasia type 1 | 1 |
| Multiple Epiphyseal Dysplasia | 3 |
| Multiple myeloma | 86 |
| Multiple sclerosis | 1 |
| Muscle-eye-brain disease | 1 |
| Muscular dystrophy | 1 |
| Myelodysplastic syndrome | 11 |
| Myelofibrosis | 477 |
| Myeloma | 1 |
| Myeloproliferative neoplasm | 7 |
| Myoclonic Astatic Epilepsy | 1 |
| Myoclonic epilepsy | 1 |
| Myoepithelioma | 2 |
| Myofibroblastic sarcoma | 2 |
| Myxofibrosarcoma | 14 |
| Nasopharyngeal cancer | 5 |
| Neonatal seizures | 2 |
| Nephroblastoma | 1 |
| Neuroblastic tumor | 1 |
| Neuroblastoma | 66 |
| Neurocytoma | 12 |
| Neurodevelopmental disorder | 9 |
| Neuroendocrine carcinoma | 3 |
| Neuroticism | 6 |
| Non-muscle invasive bladder neoplasm | 5 |
| Non-small cell lung cancer | 50 |
| Non-syndromic sensorineural hearing loss | 11757 |
| Normal | 6371 |
| Obesity | 68 |
| Obsessive-compulsive disorder | 1 |
| Oculoskeletal dysplasia | 1 |
| Olfactory neuroblastoma | 8 |
| Oligodendroglial tumors | 2 |
| Oligozoospermia | 6 |
| Ollier disease | 10 |
| Oral cancer | 17 |
| Oral squamous cell carcinoma | 20 |
| Osteopoikilosis | 1 |
| Osteosarcoma | 265 |
| oto-dental syndrome | 5 |
| Ovarian cancer | 2911 |
| Ovarian clear cell carcinoma | 125 |
| ovarian endometriomas | 1 |
| Ovarian neoplasms | 13 |
| Ovine squamous-cell carcinoma | 15 |
| Palpebral fissures | 1 |
| Pancreas cancer | 372 |
| Pancreatic cancer | 4003 |
| Pancreatic ductal adenocarcinoma | 100 |
| Pancreatic endocrine tumor | 6 |
| Papillary thyroid carcinoma | 5 |
| Paraganglioma | 7 |
| Parathyroid adenoma | 4 |
| Parkinson disease | 3 |
| periventricular nodular heterotopia | 1 |
| Peters Plus syndrome | 7 |
| Pheochromocytoma | 8 |
| Phyllodes tumor | 4 |
| Pilocytic astrocytoma | 3 |
| Pilomyxoid astrocytoma | 1 |
| Pituitary adenoma | 3 |
| Plasma-cell dyscrasia | 1 |
| Platelet disorder | 1 |
| Pleomorphic liposarcoma | 18 |
| Pleomorphic xanthoastrocytoma | 1 |
| Pleural malignant mesothelioma | 14 |
| Poland''s syndrome | 1 |
| Polyposis syndrome | 1 |
| Potocki-Shaffer syndrome | 5 |
| Prader-willi syndrome | 1 |
| Predisposition to dermoid sinus | 4 |
| Preeclampsia | 1 |
| Primary central nervous system lymphoma | 5 |
| primary effusion lymphomas | 1 |
| Primary hepatocellular | 2 |
| Primary mediastinal b-cell lymphoma | 9 |
| Prostate cancer | 608 |
| Pulmonary chondroma | 3 |
| Pulmonary fibrosis | 1 |
| Quebec platelet disorder | 1 |
| Recurrent pregnancy loss | 25 |
| Renal cell carcinoma | 31 |
| renal disease | 3 |
| renal hypoplasia | 1 |
| Renal-coloboma syndrome | 1 |
| retinal degeneration | 1 |
| Retinitis pigmentosa | 1 |
| Retinoblastoma | 25 |
| retrognathia | 1 |
| Rett syndrome | 10 |
| Rhabdomyosarcoma | 6 |
| Rheumatoid arthritis | 3 |
| Rubinstein-Taybi syndrome | 1 |
| Salivary gland adenoid cystic carcinoma | 2 |
| Salivary gland tumor | 1 |
| Schizophrenia | 308 |
| Seminomas | 2 |
| Sertoli-cell only syndrome | 5 |
| Sezary syndrome | 92 |
| short stature | 1 |
| Silver-Russell syndrome | 3 |
| Sinonasal adenocarcinoma | 1 |
| skeletal anomalies | 1 |
| Small bowel adenocarcinoma | 9 |
| small cell lung cancer | 140 |
| Smith-Lemli-Opitz syndrome | 1 |
| Smith-Magenis syndrome | 1 |
| Soft tissue tumor | 2 |
| Spastic ataxia of charlevoix-saguenay | 1 |
| speech delay | 2 |
| Spinal muscular atrophy | 2 |
| Spitz naevi | 3 |
| Splenic marginal zone lymphoma | 4 |
| Squamous cell cancer | 21 |
| Stargardt''s disease | 1 |
| Stenocardia | 39 |
| Submicroscopic aberration syndrome | 1 |
| Sudden cardiac death | 9 |
| Systemic autoimmune disease | 1 |
| Systemic lupus erythematosus | 8 |
| TAR Syndrome | 2 |
| T-cell acute lymphoblastic leukemia | 27 |
| T-cell lymphomas | 90 |
| T-cell primary immunodeficiency | 1 |
| T-cell prolymphocytic leukemia | 136 |
| Testicular cancer | 1 |
| Testicular germ cell tumor | 3 |
| Tetralogy of Fallot | 21 |
| Thoracic aortic aneurysm | 44 |
| Thrombocytopenia-absent radius syndrome | 11 |
| Thymic lymphomas tumor | 1 |
| Thyroid cancer | 2 |
| Tourette syndrome | 1 |
| Trilogy of Fallot | 1 |
| Trisomy 5 syndrome | 2 |
| Type 1 diabetes | 2 |
| Type 2 diabetes | 8 |
| Urothelial carcinoma | 24 |
| Urothelial cell carcinoma | 43 |
| Urothelial tumor | 1 |
| Usher syndrome | 6 |
| Usher type I cohort | 8 |
| Uterine leiomyoma | 1 |
| Uterine serous papillary cancer | 1 |
| Uveal melanoma | 8 |
| van der Woude syndrome | 2 |
| Vasospasm | 39 |
| Velocardiofacial syndrome | 2 |
| WAGR syndrome | 10 |
| Wegener''s granulomatosis | 2 |
| Williams Syndrome | 32 |
| Williams-beuren syndrome | 12 |
| Wilms tumour | 53 |
| XY gonadal dysgenesis | 10 |
| XY sex reversal | 1 |
| **Grand Total** | **52746** |