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LIST OF ENTRIES

A

22q13 deletion syndrome
Aarskog syndrome
Aase syndrome
Abetalipoproteinemia
Acardia
Accutane embryopathy
Achondrogenesis
Achondroplasia
Achoo syndrome
Acrocallosal syndrome
Acromegaly
Adams-Oliver syndrome
Adrenoleukodystrophy
Aicardi syndrome
Alagille syndrome
Albinism
Alcoholism
Alkaptonuria
Alpha-1 antitrypsin
Alpha-thalassemia X-linked mental retardation syndrome
Alzheimer disease
Amelia
Amniocentesis
Amyoplasia
Amyotrophic lateral sclerosis
Androgen insensitivity syndrome
Anemia, sideroblastic X-linked
Anencephaly
Angelman syndrome
Ankylosing spondylitis
Apert syndrome
Arginase deficiency
Arnold-Chiari malformation
Arthrogyposis multiplex congenita
Arthropathy-camptodactyly syndrome

Asperger syndrome
Asplenia
Asthma
Ataxia-telangiectasia
Attention deficit hyperactivity disorder
Autism
Azorean disease

B

Bardet-Biedl syndrome
Batten disease
Beals syndrome
Beare-Stevenson cutis gyrata syndrome
Beckwith-Wiedemann syndrome
Beta thalassemia
Bicuspid aortic valve
Biotinidase deficiency
Bipolar disorder
Bloom syndrome
Blue rubber bleb nevus syndrome
Brachydactyly
Branchiootorenal syndrome
Breast cancer
Bruton agammaglobulinemia

C

Cadasil
Campomelic dysplasia
Canavan disease
Cancer
Cardiofaciocutaneous syndrome
Carnitine palmitoyltransferase deficiency
Carpenter syndrome

Caudal dysplasia
Celiac disease
Central core disease
Cerebral palsy
Charcot-Marie-Tooth disease
Charge syndrome
Chediak-Higashi syndrome
Chondrodysplasia punctata
Chondrosarcoma
Choroideremia
Chromosomal abnormalities
Chromosome
Cleft lip and palate
Cleidocranial dysplasia
Clubfoot
Cockayne syndrome
Coffin-Lowry syndrome
Coffin-Siris syndrome
Cohen syndrome
Coloboma
Color blindness
Cone-rod dystrophy
Congenital adrenal hyperplasia
Congenital heart disease
Congenital hypothyroid syndrome
Conjoined twins
Corneal dystrophy
Cornelia de Lange syndrome
Corpus callosum agenesis
Costello syndrome
Crane-Heise syndrome
Craniosynostosis
Cri du chat syndrome
Crouzon syndrome
Cystic fibrosis
Cystinosis
Cystinuria

D

Dandy-Walker malformation
 Deletion 22q11 syndrome
 Dementia
 Dentatorubral-pallidolusian atrophy
 Depression
 Diabetes
 Diastrophic dysplasia
 Distal arthrogryposis syndrome
 DNA
 Donohue syndrome
 Down syndrome
 Duane retraction syndrome
 Dubowitz syndrome
 Duchenne muscular dystrophy
 Dyschondrosteosis
 Dysplasia
 Dystonia

E

Ectodermal dysplasia
 Ectrodactyly-ectodermal dysplasia-clefting
 Ehlers-danlos syndrome
 Ellis-van Creveld syndrome
 Emery-Dreifuss muscular dystrophy
 Encephalocele
 Engelmann disease
 Epidermolysis bullosa
 Epilepsy
 Essential hypertension
 Essential tremor

F

Fabry disease
 Facioscapulohumeral muscular dystrophy
 Factor V Leiden thrombophilia
 Fahr disease
 Familial adenomatous polyposis
 Familial dysautonomia
 Familial mediterranean fever
 Familial nephritis
 Fanconi-Bickel syndrome
 Fanconi anemia
 Fetal alcohol syndrome

FG syndrome
 Fibroblast growth factor receptor mutations
 Fluorescent in situ hybridization
 Fragile X syndrome
 Fraser syndrome
 Freeman-Sheldon syndrome
 Friedreich ataxia
 Frontonasal dysplasia
 Frontotemporal dementia
 Frys syndrome

G

Galactokinase deficiency
 Galactosemia
 Gastric cancer
 Gastroschisis
 Gaucher disease
 Gene
 Gene mutations
 Gene pool
 Gene therapy
 Genetic counseling
 Genetic disorders
 Genetic mapping
 Genetic testing
 Genetics and congenital anomalies
 Genitalia ambiguous
 Genotype and phenotype
 Glaucoma
 Glycogen storage diseases
 Gm1-gangliosidosis
 Goltz syndrome
 Greig cephalopolysyndactyly
 Griscelli syndrome

H

Haim-Munk syndrome
 Hair loss syndromes
 Hallermann-Streiff syndrome
 Hand-foot-uterus syndrome
 Harlequin fetus
 Hemifacial microsomia
 Hemihypertrophy hemihyperplasia
 Hemochromatosis
 Hemolytic-uremic syndrome
 Hemophilia
 Hepatocellular carcinoma

Hereditary angioneurotic edema
 Hereditary colorectal cancer
 Hereditary desmoid disease
 Hereditary hearing loss and deafness
 Hereditary multiple exostoses
 Hereditary pancreatitis
 Hereditary spastic paraplegia
 Hereditary spherocytosis
 Hermansky-Pudlak syndrome
 Hermaphroditism
 Hirschsprung disease
 Holoprosencephaly
 Holt-Oram syndrome
 Homocystinuria
 Human genome project
 Huntington disease
 Hydrocephalus
 Hydrolethalus syndrome
 Hydrops fetalis
 Hyperlipoproteinemia
 Hypochondrogenesis
 Hypochondroplasia
 Hypophosphatasia
 Hypophosphatemia
 Hypospadias and epispadias

I

Ichthyosis
 Imprinting
 Incontinentia pigmenti
 Infantile refsum disease
 Inheritance

J

Jackson-Weiss syndrome
 Jacobsen syndrome
 Jervell and Lange-Nielsen syndrome
 Joubert syndrome

K

Kabuki syndrome
 Kallmann syndrome
 Kartagener syndrome
 Karyotype
 Kennedy disease

Klinefelter syndrome
Klippel-Feil sequence
Klippel-Trenaunay-Weber syndrome
Krabbe disease

L

Larsen syndrome
Laterality sequence
Leber congenital amaurosis
Lebers hereditary optic atrophy
Leigh syndrome
Lesch-Nyhan syndrome
Leukodystrophy
Li-Fraumeni syndrome
Limb-girdle muscular dystrophy
Lissencephaly
Long QT syndrome
Lowe oculocerebrorenal syndrome

M

Macular degeneration—age-related
Major histocompatibility complex
Malignant hyperthermia
Mannosidosis
Marfan syndrome
Marshall-Smith syndrome
Marshall syndrome
MCAD deficiency
McCune-Albright syndrome
McKusick-Kaufman syndrome
Meckel-Gruber syndrome
Meckels diverticulum
Menkes syndrome
Metaphyseal dysplasia
Methylmalonic acidemia
Methylmalonicaciduria due to methylmalonic coa mutase deficiency
Microphthalmia with linear skin defects
Miller-Dieker syndrome
Moebius syndrome
Moyamoya
Mucopolipidosis
Mucopolysaccharidoses
Mucopolysaccharidosis type I
Mucopolysaccharidosis type II
Muir-Torre syndrome
Multifactorial inheritance

Multiple endocrine neoplasias
Multiple epiphyseal dysplasia
Multiple lentigenes syndrome
Multiple sclerosis
Multiplex ligation-dependent probe amplification
Muscular dystrophy
Myasthenia gravis
Myopia
Myotonic dystrophy
Myotubular myopathy

N

Nail-patella syndrome
Narcolepsy
Nephrogenic diabetes insipidus
Neu-Laxova syndrome
Neural tube defects
Neuraminidase deficiency
Neuraminidase deficiency with beta-galactosidase deficiency
Neurofibromatosis
Nevoid basal cell carcinoma
Niemann-Pick disease
Nijmegen breakage syndrome
Noonan syndrome
Norrie disease

O

Oculo-digito-esophago-duodenal syndrome
Oculodentodigital syndrome
Oligohydramnios sequence
Omphalocele
Oncogene
Opitz syndrome
Oral-facial-digital syndrome
Organic acidemias
Ornithine transcarbamylase deficiency
Osler-Weber-Rendu syndrome
Osteoarthritis
Osteogenesis imperfecta
Osteoporosis
Otopalatodigital syndrome
Ovarian cancer

P

Paine syndrome
Pallister-Hall syndrome
Pallister-Killian syndrome
Pancreatic beta cell agenesis
Pancreatic cancer
Panic disorder
Parkinson disease
Paroxysmal nocturnal hemoglobinuria
Patent ductus arteriosus
Pedigree analysis
Pelizaeus-Merzbacher disease
Pendred syndrome
Pervasive developmental disorders
Peutz-Jeghers syndrome
Pfeiffer syndrome
Pharmacogenetics
Phenylketonuria
Pierre Robin sequence
Pituitary dwarfism
Poland anomaly
Polycystic kidney disease
Polycystic ovary syndrome
Polydactyly
Pompe disease
Porphyrias
Prader-Willi syndrome
Prenatal ultrasound
Prion diseases
Progeria syndrome
Propionic acidemia
Prostate cancer
Proteus syndrome
Prune-belly syndrome
Pseudoachondroplasia
Pseudoxanthoma elasticum
Pyloric stenosis
Pyruvate carboxylase deficiency
Pyruvate dehydrogenase complex deficiency
Pyruvate kinase deficiency

R

Raynauds disease
Refsum disease
Renal agenesis
Renal failure due to hypertension
Renpenning syndrome

Retinitis pigmentosa
 Retinoblastoma
 Rett syndrome
 Rheumatoid arthritis
 Rhizomelic chondrodysplasia punctata
 Rhodopsin
 Rieger syndrome
 RNA
 Roberts SC phocomelia
 Robinow syndrome
 Rothmund-Thomson syndrome
 Rubinstein-Taybi syndrome
 Russell-Silver syndrome

S

Saethre-Chotzen syndrome
 Schinzel-Giedion syndrome
 Schizophrenia
 Schwartz-Jampel syndrome
 Scleroderma
 Sclerosing bone dysplasias
 Scoliosis
 Sebastian syndrome
 Seckel syndrome
 Septo-optic dysplasia
 Severe combined immunodeficiency
 Short-rib polydactyly
 Shprintzen-goldberg craniosynostosis syndrome
 Sickle cell disease
 Simpson-Golabi-Behmel syndrome
 Sirenomelia
 Sjögren Larsson syndrome
 Skeletal dysplasia
 Smith-Fineman-Myers syndrome
 Smith-Lemli-Opitz syndrome
 Smith-Magenis syndrome
 Sotos syndrome
 Spastic cerebral palsy
 Spina bifida

Spinal muscular atrophy
 Spinocerebellar ataxia
 Spondyloepiphyseal dysplasia
 SRY (sex determining region y)
 Stargardt disease
 Stickler syndrome
 Sturge-Weber syndrome
 Sutherland-Haan syndrome
 Systemic elastorrhexis

T

Tangier disease
 TAR syndrome
 Tay-Sachs disease
 Teratogen
 Thalassemia
 Thalidomide embryopathy
 Thanatophoric dysplasia
 Thrombasthenia of Glanzmann and Naegeli
 Tourette syndrome
 Treacher Collins syndrome
 Trichorhinophalangeal syndrome
 Triose phosphate isomerase deficiency
 Triploidy
 Trismus pseudocamptodactyly syndrome
 Trisomy 8 mosaicism syndrome
 Trisomy 13
 Trisomy 18
 Tuberous sclerosis complex
 Turner syndrome

U

Urea cycle disorders
 Urogenital adysplasia syndrome
 Usher syndrome

V

Van der Woude syndrome
 Vater association
 Von Hippel-Lindau syndrome
 Von Willebrand disease

W

Waardenburg syndrome
 Walker-Warburg syndrome
 Weaver syndrome
 Weissenbacher-Zweymuller syndrome
 Werner syndrome
 Williams syndrome
 Wilson disease
 Wiskott-Aldrich syndrome
 Wolf-Hirschhorn syndrome
 Wolman disease

X

X-linked hydrocephaly
 X-linked mental retardation
 Xeroderma pigmentosum
 XX male syndrome
 XYY syndrome

Z

Zellweger syndrome
 Zygote