Human Genome Landmarks

Selected Genes, Traits, and Disorders

www.ornl.gov/hgmis/posters/chromosome

genomics.energy.gov

1
246 million base pairs

Cataracts
Malignant transformation suppression
Ehlers-Danlos syndrome, type VI
Glaucoma, primary infantile
Hirschsprung disease, cardiac defects
Schwartz-Jampel syndrome
Hypophosphatasia, infantile, childhood
Breast cancer, ductal
Cutaneous malignant melanoma/dysplastic nevus
p53-related protein
Serotonin receptors
Schnyder crystalline corneal dystrophy
Kostmann neutropenia
Oncogene MYC, lung carcinoma-derived
Deafness, autosomal dominant
Porphyria
Epiphyseal dysplasia, multiple, type 2
Intervertebral disc disease
Lymphoma, non-Hodgkin
Breast cancer, invasive intraductal
Colon adenocarcinoma
Maple syrup urine disease, type II
Atrioventricular canal defect
Fluorouracil toxicity, sensitivity to
Zellweger syndrome
Stickler syndrome, type III
Marshall syndrome
Stargardt disease
Retinitis pigmentosa
Cone-rod dystrophy
Macular dystrophy, age-related
Fundus flavimaculatus
Hypothyroidism, nongoitrous
Exostoses, multiple
Pheochromocytoma
Psoriasis susceptibility
Limb-girdle muscular dystrophy, autosomal dominant
Pycnodysostosis
Vohwinkel syndrome with ichthyosis
Erythrokeratoderma, progressive symmetric
Anemia, hemolytic
Elliptocytosis
Pyropoikilocytosis
Spherocytosis, recessive
Schizophrenia
Lupus nephritis, susceptibility to
Migraine, familial hemiplegic
Emery-Dreifuss muscular dystrophy
Cardiomyopathy, dilated
Lipodystrophy, familial partial
Dejerine-Sottas disease, myelin P-related
Hypomyelination, congenital
Nemaline myopathy, autosomal dominant
Lupus erythematosus, systemic, susceptibility
Neutropenia, alloimmune neonatal
Viral infections, recurrent
Antithrombin III deficiency
Atherosclerosis, susceptibility to
Glaucoma
Tumor potentiating region
Nephrotic syndrome
Sjogren syndrome
Coagulation factor deficiency
Alzheimer disease
Cardiomyopathy
Factor H deficiency
Membroproliferative glomerulonephritis
Hemolytic-uremic syndrome
Nephropathy, chronic hypocomplementemic
Epidermolysis bullosa
Popliteala pterygium syndrome
Ectodermal dysplasia/skin fragility syndrome
Usher syndrome, type 2A
Kenny-Caffey syndrome
Diphenylhydantoin toxicity

Homocystinuria
Neuroblastoma (neuroblastoma suppressor)
Rhabdomyosarcoma, alveolar
Neuroblastoma, aberrant in some
Exostoses, multiple-like
Opioid receptor
Hyperprolinemia, type II
Bartter syndrome, type 3
Prostate cancer
Brain cancer
Charcot-Marie-Tooth neuropathy
Muscular dystrophy, congenital
Erythrokeratodermia variabilis
Deafness, autosomal dominant and recessive
Glucose transport defect, blood-brain barrier
Hypercholesterolemia, familial
Neuropathy, paraneoplastic sensory
Muscle-eye-brain disease
Medulloblastoma
Basal cell carcinoma
Corneal dystrophy, gelatinous drop-like
Leber congenital amaurosis
Retinal dystrophy
B-cell leukemia/lymphoma
Lymphoma, MALT and follicular
Mesothelioma
Germ cell tumor
Sezary syndrome
Colon cancer
Neuroblastoma
Glycogen storage disease
Osteopetrosis, autosomal dominant, type II
Waardenburg syndrome, type 2B
Vesicoureteral reflux
Choreoathetosis/spasticity, episodic (paroxysmal)
Hemochromatosis, type 2
Leukemia, acute
Gaucher disease
Medullary cystic kidney disease, autosomal dominant
Renal cell carcinoma, papillary
Insensitivity to pain, congenital, with anhidrosis
Medullary thyroid carcinoma
Hyperlipidemia, familial combined
Hyperparathyroidism
Lymphoma, progression of
Porphyria variegata
Hemorrhagic diathesis
Thromboembolism susceptibility
Systemic lupus erythematosus, susceptibility
Fish-odor syndrome
Prostate cancer, hereditary
Chronic granulomatous disease
Macular degeneration, age-related
Epidermolysis bullosa
Chitotriosidase deficiency
Pseudohypoaldosteronism, type II
Hypokalemic periodic paralysis
Malignant hyperthermia susceptibility
Glomerulopathy with fibronectin deposits
Metastasis suppressor
Measles, susceptibility to
van der Woude syndrome (lip pit syndrome)
Rippling muscle disease
Hypoparathyroidism-retardation-dysmorphism syndrome
Ventricular tachycardia, stress-induced polymorphic
Fumarase deficiency
Chediak-Higashi syndrome
Muckle-Wells syndrome
Zellweger syndrome
Adrenoleukodystrophy, neonatal
Endometrial bleeding-associated factor
Left-right axis malformation
Prostate cancer, hereditary
Chondrodysplasia punctata, rhizomelic, type 2

2
243 million base pairs

Melanoma-associated gene
Thyroid iodine peroxidase deficiency
Goiter, congenital
Lipoproteinemia, hypobeta, abeta-, hyperbeta-, and apo-
ACTH deficiency
Obesity, adrenal insufficiency, and red hair
LCHAD deficiency
Trifunctional protein deficiency, type 1
HELLP syndrome, maternal, of pregnancy
Fatty liver, acute, of pregnancy
Deafness, autosomal recessive
Glaucoma, primary infantile
Gingival fibromatosis, hereditary
Holoprosencephaly
Ovarian dysgenesis
Carney complexes
Endometrial carcinoma
Zellweger syndrome
Adrenoleukodystrophy, neonatal
Alstrom syndrome
Preeclampsia/eclampsia
Welander distal myopathy
Kappa light chain deficiency
Pancreatic stone protein
Lissencephaly
Renal tubular acidosis with deafness
BRCA1-associated RING domain (breast cancer)
Achromatopsia
Rhabdomyosarcoma, down-regulated in
Diazepam-binding inhibitor
Thrombophilia due to protein C deficiency
Purpura fulminans, neonatal
Liver cancer oncogene
Xeroderma pigmentosum, group B
Trichothiodystrophy
Nemaline myopathy, autosomal recessive
Convulsions, familial febrile
Progressive intrahepatic cholestasis
Edstrom myopathy
Mesomelic dysplasia, Kantaputra type
Cardiomyopathy, familial hypertrophic
Bardet-Biedl syndrome
Ehlers-Danlos syndromes
Aneurysm, familial arterial
Diabetes mellitus, insulin-dependent
Primary pulmonary hypertension (familial primary)
Cleft palate, isolated
Wrinkly skin syndrome
Amyotrophic lateral sclerosis, juvenile recessive
Lactic acidosis due to defect in iron-sulfur cluster of complex I
Ichthyosis
Finnish lethal neonatal metabolic syndrome
T-cell leukemia or lymphoma
Bjornstad syndrome (pili torti and deafness)
Myopathy, desmin-related, cardioskeletal
Cardiomyopathy, dilated
Natural resistance-associated macrophage protein
Hyperoxaluria, primary, type 1
Alport syndrome, autosomal recessive
Hematuria, familial benign
Brachydactyly-mental retardation syndrome
Oguchi disease
Epidermolysis bullosa

Tremor, familial essential
Oculodigitoesophagoduodenal syndrome
Anaplastic lymphoma kinase (Ki-1)
Pseudovaginal perineoscrotal hypospadias
Xanthinuria, type I
Colorectal cancer, hereditary, nonpolyposis, type 1
Ovarian cancer
Muir-Torre syndrome
Human T-cell leukemia virus enhancer factor
Precocious puberty, male
Pseudohermaphroditism, male, with Leydig cell hypoplasia
Hypogonadotropic hypogonadism
Micropenis
Leydig cell adenoma, with precocious puberty
Sitosterolemia
Cystinuria
Doyne honeycomb degeneration of retina
Dyslexia, specific
Muscular dystrophy
Miyoshi myopathy
Myopathy, distal, with anterior tibial onset
Orofacial cleft
Parkinson disease, type 3
Vitamin K-dependent coagulation defect
Pancreatitis-associated protein
Pulmonary alveolar proteinosis, congenital
Glaucoma, open angle, B (adult-onset)
Diabetes mellitus, non-insulin-dependent
Ectodermal dysplasia, autosomal dominant and recessive
Hypothyroidism, congenital
Nephronophthisis
Colorectal cancer
Cardiomyopathy, dilated
Spastic cerebral palsy, symmetric, autosomal recessive
Epilepsy
Ataxia, episodic
Deafness, autosomal dominant
Myasthenic syndrome, slow-channel congenital
Rhizomelic chondrodysplasia punctata, type 3
Cardiomyopathy, dilated
Duane retraction syndrome
Synpolydactyly, type II
Colorectal cancer, hereditary nonpolyposis, type 3
Neurogenic differentiation
Arrhythmogenic right ventricular dysplasia
Myasthenia gravis, neonatal transient
Cataracts
Paroxysmal nonkinesiogenic dyskinesia
Choreoathetosis, familial paroxysmal
Cerebrotendinous xanthomatosis
Acyl-Coenzyme A dehydrogenase
Carbamoylphosphate synthetase I
Waardenburg syndrome, types I and III
Rhabdomyosarcoma, alveolar
Craniofacial-deafness-hand syndrome
Brachydactyly, type A1
Goodpasture antigen
Serotonin receptor
Bethlem myopathy
Programmed cell death
Leigh syndrome, French-Canadian type
Ultraviolet damage, repair of
Crigler-Najjar syndrome, type I

3
199 million base pairs

von Hippel-Lindau syndrome
Renal cell carcinoma
Fanconi anemia, complementation group D
Biotinidase deficiency
Xeroderma pigmentosum, complementation group C
Cardiomyopathy, dilated, autosomal dominant
Endplate acetylcholinesterase deficiency
Arrhythmogenic right ventricular dysplasia
Teratocarcinoma-derived growth factor
Hepatoblastoma
Pilomatricoma
Ovarian carcinoma, endometrioid type
Hypobetalipoproteinemia, familial
GM1-gangliosidosis
Mucopolysaccharidosis
BRCA1 associated protein (breast cancer)
Hemolytic anemia
Septooptic dysplasia
Progressive external ophthalmoplegia, type 2
Larsen syndrome, autosomal dominant
HIV infection, susceptibility/resistance to
Ichthyosiform erythroderma, congenital
Long QT syndrome
Brugada syndrome
Heart block, progressive and nonprogressive
Deafness, autosomal recessive
Waardenburg syndrome
Tietz syndrome
Glycogen storage disease
Dementia, familial, nonspecific
Pituitary hormone deficiency, combined
Thyrotropin-releasing hormone deficiency
Deafness, autosomal recessive
Hypomagnesemia, primary
Tremor, familial essential
Charcot-Marie-Tooth neuropathy
Malignant hyperthermia susceptibility
Hypocalciuric hypercalcemia, type I
Neonatal hyperparathyroidism
Hypocalcemia, autosomal dominant
Atransferrinemia
Propionicacidemia, type II or pccB
Hailey-Hailey disease
Retinitis pigmentosa, autosomal dominant and recessive
Night blindness, congenital stationery, rhodopsin-related
Cataracts, juvenile-onset and congenital
Common acute lymphocytic leukemia antigen
Blepharophimosis, epicanthus inversus and ptosis type 1
Hemosiderosis, systemic
Sucrose intolerance
Cerebral cavernous malformations
Myelodysplasia syndrome
Apnea, postanesthetic
Ovarian cancer
Megakaryocyte growth and development factor
Thrombocythemia, essential
Peroxisomal bifunctional enzyme deficiency
Thrombophilia due to HRG deficiency
Leukoencephalopathy with vanishing white matter
Lipoma-preferred-partner gene fused with HMGIC

Moyamoya disease
Muscular dystrophy, limb-girdle, type IC
Obesity, severe
Diabetes mellitus, insulin-resistant
Marfan-like connective tissue disorder
Thyroid hormone resistance
Usher syndrome, type IIB
Pseudo-Zellweger syndrome
Lung cancer, small-cell
Colon cancer
Deleted in lung and esophageal cancer
Metaphyseal chondrodysplasia, Murk Jansen type
Carnitine-acylcarnitine translocase (deficiency)
Epidermolysis bullosa
Colorectal cancer, hereditary nonpolyposis, type 2
Turcot syndrome with glioblastoma
Muir-Torre family cancer syndrome
Hyperglycinemia, nonketotic
Pancreatic cancer
Spinocerebellar ataxia
Pituitary ACTH-secreting adenoma
Ventricular tachycardia, idiopathic
Night blindness, congenital stationary
T-cell leukemia translocation altered gene
Wernicke-Korsakoff syndrome, susceptibility to
Bardet-Biedl syndrome
Nonpapillary renal carcinoma
Protein S deficiency
Ventricular, skeletal, slow
Cardiomyopathy, hypertrophic
Myotonic dystrophy
Coproporphyria
Harderoporphyrinuria
Oroticaciduria
Neuropathy, hereditary motor and sensory, Okinawa type
Dopamine receptor
Psoriasis susceptibility
Moebius syndrome
Alkaptonuria
Glaucoma, primary open angle
Hypertension, essential
Usher syndrome (Finland)
Nephronophthisis, adolescent
Ataxia telangiectasia
Short stature
Myeloid leukemia factor, acute
Ectropic viral integration site (oncogene EVI1)
3q21q26 syndrome
Encephalopathy, familial, with neuroserpin inclusion bodies
Diabetes mellitus, noninsulin-dependent
Fanconi-Bickel syndrome
Lymphomas
Eukaryotic translation initiation factor (squamous cell lung cancer)
Limb-mammary syndrome
Tumor protein p63
Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome
Optic atrophy
Lipoma
Bernard-Soulier syndrome, type C melanoma-associated

4
191 million base pairs

Cherubism (familial benign giant-cell tumor of the jaw)
Dopamine receptor
Huntington disease
Night blindness, congenital stationary, type 3
Retinitis pigmentosa, autosomal recessive
Retinal degeneration, autosomal recessive
Wolfram syndrome
Craniosynostosis, Adelaide type
Phenylketonuria
Parkinson disease, familial
Pituitary tumor-transforming gene
Stargardt disease
Dentin dysplasia, Shields type II
Leukemia, acute myeloid
Periodontitis, juvenile
Muscular dystrophy, limb-girdle, type 2E
Melanoma growth-stimulating activity
Hyper-IgE syndrome
Renal tubular acidosis
Mucolipidosis
Lymphocytic leukemia, acute T-cell
Alcoholism, susceptibility to
Wolfram syndrome
Sclerotylosis
Huriez syndrome
Rieger syndrome
Iridogoniodysgenesis syndrome
Severe combined immunodeficiency
Afibrinogenemia
Anterior segment mesenchymal dysgenesis
Tryptophan oxygenase
Aspartylglucosaminuria
Hepatitis B virus integration site
Hepatocellular carcinoma
Progressive external ophthalmoplegia, type 3
Coagulation factor XI
Facioscapulohumeral muscular dystrophy
Eutropenia, neonatal alloimmune
Fletcher factor

Deafness, autosomal dominant
Achondroplasia
Hypochondroplasia
Thanatophoric dysplasia, types I and II
Crouzon syndrome with acanthosis nigricans
Muencke syndrome
Mucopolysaccharidosis
Wolf-Hirschhorn syndrome
Hypodontia
Dopamine receptor
Ellis-van Creveld syndrome
Weyers acrodental dysostosis
Huntington-like neurodegenerative disorder
Retinitis pigmentosa, autosomal recessive
Psoriasis susceptibility
Analbuminemia
Amelogenesis imperfecta
Piebaldism
Mast cell leukemia
Mastocytosis with associated hematologic disorder
Germ cell tumors
Dentinogenesis imperfecta
Myeloid/lymphoid or mixed-lineage leukemia
Parkinson disease, type 1
Polycystic kidney disease, adult, type II
Hypogonadotropic hypogonadism
Abetalipoproteinemia
Mannosidosis, beta
C3b inactivator deficiency
Long QT syndrome with sinus bradycardia
Fibrodysplasia ossificans progressiva
Fibrinogenemia
Amyloidosis, hereditary renal
Hair color, red
Pseudohypoaldosteronism type I, autosomal dominant
Glutaricaciduria, type IIC
Hypercalciuria
Beukes familial hip dysplasia
Facioscapulohumeral muscular dystrophy region

5
181 million base pairs

Dopamine transporter
Attention-deficit hyperactivity disorder, susceptibility to
Cri-du-chat syndrome, mental retardation in
Chondrocalcinosis
Taste receptor
Alpha-methylacyl-CoA racemase deficiency
Differentially expressed in ovarian cancer
Ketoacidosis
Leukemia inhibitory factor receptor
Myopathy, distal, with vocal cord and pharyngeal weakness
Molybdenum cofactor deficiency, type B
Endometrial carcinoma
Klippel-Feil syndrome
Anemia, megaloblastic
Sandhoff disease
Spinal muscular atrophy, juvenile
X-ray repair
Convulsions, familial febrile
Adenomatous polyposis coli
Gardner syndrome
Colorectal cancer
Desmoid disease
Turcot syndrome
Ehlers-Danlos syndromes
Neonatal alloimmune thrombocytopenia
Myelodysplastic syndrome
Limb-girdle muscular dystrophy, autosomal dominant
Deafness
Bronchial hyperresponsiveness (bronchial asthma)
Hemangioma, capillary infantile
Spinocerebellar ataxia
Macrocytic anemia
Gastric cancer
Non small-cell lung cancer
Retinitis pigmentosa, autosomal recessive
Charcot-Marie-Tooth neuropathy
Netherton syndrome
Treacher Collins-Franceschetti syndrome
Pituitary tumor-transforming gene
Coagulation factor XII (Hageman factor)
Myeloid malignancy, predisposition to
Craniosynostosis, type 2
Parietal foramina
Leukotriene C4 synthase deficiency
Dopamine receptor
Hermansky-Pudlak syndrome

Homocystinuria-megaloblastic anemia, cbl E type
Craniometaphyseal dysplasia
Leigh syndrome
Polycystic ovary syndrome
Hirschsprung disease
Severe combined immunodeficiency
Dwarfism
Malignant hyperthermia susceptibility
Pituitary hormone deficiency
Cytotoxic T-lymphocyte-associated serine esterase
Hanukah factor serine protease
Maroteaux-Lamy syndrome
Serotonin receptor
Schizophrenia susceptibility locus
Wagner syndrome
Erosive vitreoretinopathy
Basal cell carcinoma
Obesity with impaired prohormone processing
Diphtheria toxin receptor
Contractural arachnodactyly, congenital
Cutis laxa, recessive, type I
Deafness
Cortisol resistance
Corneal dystrophy
Eosinophilia, familial
Serotonin receptor
Schistosoma mansoni infection, susceptibility/resistance to
Natural killer cell stimulatory factor-2
GM2-gangliosidosis, AB variant
Startle disease, autosomal dominant and recessive
Diastrophic dysplasia
Atelosteogenesis
Achondrogenesis
Epiphyseal dysplasia, multiple
Asthma, nocturnal, susceptibility to
Obesity, susceptibility to
Muscular dystrophy, limb-girdle, type 2F
Carnitine deficiency, systemic primary
Atrial septal defect with atrioventricular conduction defects
Arthrogryposis multiplex congenital, neurogenic
Leukemia, acute promyelocytic, NPM/RARA type
Vascular endothelial growth factor receptor
Lymphedema, hereditary
Cockayne syndrome
Pancreatitis, hereditary

6
170 million base pairs

Iridogoniodysgenesis
Anterior segment mesenchymal dysgenesis
Rieger anomaly
Axenfeld anomaly
Coagulation factor XIII
Keratosis palmoplantaris striata
Spinocerebellar ataxia
Schizophrenia susceptibility locus
Maple syrup urine disease, type Ib
Bare lymphocyte syndrome, type I
Atrial septal defect, secundum type
Adrenal hyperplasia, congenital
Renal glucosuria
Beryllium disease, chronic, susceptibility to
Leukemia, pre-B-cell transcription factor
Tumor necrosis factor (cachectin)
Malaria, cerebral, susceptibility to
Retinitis pigmentosa
Platelet-activating factor
Asthma and atopy, susceptibility to
Peroxisomal biogenesis disorder
Anemia, hemolytic, Rh-null, suppressor type
Methylmalonicaciduria, mutase deficiency type
Hemolytic anemia
Char syndrome
Gluten-sensitive enteropathy (celiac disease)
Cone-rod dystrophy
Inflammatory bowel disease
Mixed polyposis syndrome, hereditary
Leber congenital amaurosis, type V
Serotonin receptors
Macular dystrophy, retinal, North Carolina type
Obesity, severe
Diabetes mellitus, insulin-dependent
Muscular dystrophy, congenital merosin-deficient
Arthropathy, progressive pseudorheumatoid, of childhood
Rhizomelic chondrodysplasia punctata, type 1
Deafness
Cardiomyopathy, dilated, autosomal dominant
Human immunodeficiency virus type I susceptibility
Epilepsy, myoclonic, Lafora type
Opioid receptor
Estrogen receptor
Breast cancer
Estrogen resistance
Insulin-like growth factor-2 receptor
Hepatocellular carcinoma
Tumorigenicity, suppression of
Loss of heterozygosity, ovarian
Ovarian cancer, serous
Myeloid/lymphoid or mixed-lineage leukemia
Pancreatic beta cell, agenesis of
uniparental disomy
Conjunctivitis, ligneous
Coronary artery disease, susceptibility to
Complex neurologic disorder
Xeroderma pigmentosum, variant type

Multiple myeloma oncogene
Orofacial cleft
Leukemia, acute nonlymphocytic
Fanconi anemia, complementation group E
Ankylosing spondylitis
Stickler syndrome, type II
OSMED syndrome
Weissenbacher-Zweymuller syndrome
Deafness, nonsyndromic sensorineural
Dyslexia
Hemochromatosis
Porphyria variegata
Pemphigoid, susceptibility to
Immune suppression to streptococcal antigen
Sialidosis, types I and II
Panbronchiolitis, diffuse
Psoriasis susceptibility
Ehlers-Danlos-like syndrome
Cone dystrophy
Polycystic kidney and hepatic disease, autosomal recessive
Retinal degeneration, slow (peripherin)
Retinitis pigmentosa, peripherin-related and punctata albescens
Macular dystrophy
Butterfly dystrophy, retinal
Cleidocranial dysplasia
Dental anomalies, isolated
Nystagmus, autosomal dominant
Bullous pemphigoid antigen 1
Pelviureteric junction obstruction
Stargardt disease, autosomal dominant
Epilepsy, juvenile myoclonic
Brain-specific angiogenesis inhibitor
Diazepam-binding inhibitor
Schizophrenia susceptibility locus
Salla disease
Sialic acid storage disorder, infantile
Chorioretinal atrophy, progressive bifocal
Melanoma, absent in
Metaphyseal chondrodysplasia, Schmid type
Spondylometaphyseal dysplasia, Japanese type
Hepatic fibrosis susceptibility
Oculodentodigital dysplasia (Syndactyly type III)
Hereditary persistence of fetal hemoglobin, heterocellular
Argininemia
Leukemia
Immune interferon, receptor for
Mycobacterial infection, atypical, familial disseminated
BCG infection, generalized familial
Tuberculosis, susceptibility to
Diabetes mellitus, transient neonatal
Pleomorphic adenoma (ZAC tumor supressor)
Parkinson disease, juvenile, type 2
Plasminogen Tochigi disease
Thrombophilia, dysplasminogenemic
Plasminogen deficiency, types I and II

7
158 million base pairs

Ewing sarcoma
Turcot syndrome with glioblastoma
Colorectal cancer, hereditary nonpolyposis, type 4
Osteopenia/osteoporosis
Macular dystrophy, dominant cystoid
Retinitis pigmentosa
Growth hormone deficient dwarfism
Hand-foot-uterus syndrome
Hyperinsulinism, familial
Charcot-Marie-Tooth neuropathy, neuronal type D
Alpha-ketoglutarate dehydrogenase deficiency
Myopathy
T-cell tumor invasion and metastasis
Argininosuccinicaciduria
Hyperreflexia
Clostridium perfringens enterotoxin receptor
Supravalvar aortic stenosis
Williams-Beuren syndrome
Cutis laxa
Cytoplasmic linker
Williams-Beuren syndrome chromosome region 4
Chronic granulomatous disease
Malignant hyperthermia susceptibility
P-glycoprotein/multiple drug resistance
Colchicine resistance
Cholestasis
Split hand/foot malformation (ectrodactyly) type 1
Paraoxonase
Coronary artery disease, susceptibility to
Plasminogen activator inhibitor, type I
Thrombophilia
Hemorrhagic diathesis
Hemochromotosis
Osteogenesis imperfecta
Ehlers-Danlos syndrome, type VIIA2
Osteoporosis, idiopathic
Marfan syndrome, atypical
Deafness, autosomal recessive
Pendred syndrome
Deafness, autosomal recessive
Enlarged vestibular aqueduct
Lipoamide dehydrogenase deficiency
Hemolytic anemia
Suppression of tumorigenicity (breast)
Obesity
Taste receptors
Renal tubular acidosis, distal, autosomal recessive
Deafness, autosomal recessive
Trypsinogen deficiency
Pancreatitis, hereditary
Glaucoma-related pigment dispersion syndrome

Lunatic fringe
Craniosynostosis, type 1
Saethre-Chotzen syndrome
Blepharophimosis, epicanthus inversus, and ptosis
Deafness, autosomal dominant
Myeloid leukemia
Cerebral cavernous malformations
Wilms tumor suppressor locus
Amphiphysin (Stiff-Man syndrome)
Greig cephalopolysyndactyly syndrome
Pallister-Hall syndrome
Polydactyly
Glioblastoma amplified sequence
Spinal muscular atrophy, distal
Autism, susceptibility to
Limb-girdle muscular dystrophy, autosomal dominant
Platelet glycoprotein IV deficiency
Cerebral cavernous malformations
Colon cancer
Zellweger syndrome
Adrenoleukodystrophy, neonatal
Refsum disease, infantile
Mucopolysaccharidosis
Osteoporosis, postmenopausal, susceptibility
Citrullinemia, adult-onset type II
Ulcerative colitis, susceptibility to
Adenoma, down-regulated in
Chloride diarrhea, congenital, Finnish type
Cardiomyopathy, familial hypertrophic
Renal cell carcinoma, papillary, familial and sporadic
Hepatocellular carcinoma, childhood type
Speech-language disorder
Basal cell carcinoma, sporadic
Retinitis pigmentosa, autosomal dominant
Cystic fibrosis
Congenital bilateral absence of vas deferens
Sweat chloride elevation without CF
Colorblindness, blue cone pigment
Myotonia
Glaucoma, open angle
Human ether-a-go-go-related gene
Long QT syndrome
Preeclampsia, susceptibility to
Coronary spasm, susceptibility to
Holoprosencephaly
Serotonin receptor
Growth rate controlling factor
Currarino syndrome
Sacral agenesis
Triphalangeal thumb-polysyndactyly syndrome
X-ray repair

8
146 million base pairs

Epilepsy, progressive, with mental retardation
Keratolytic winter erythema
Prostate cancer tumor suppressor, putative
Liver cancer, deleted in
Alopecia universalis
Atrichia with papular lesions
Scurvy
Schizophrenia susceptibility locus
Plasminogen activator deficiency
Spastic paraplegia, autosomal recessive
Lipoid adrenal hyperplasia
Monocytic leukemia
Retinitis pigmentosa
Pleomorphic adenoma
ACTH deficiency
Convulsions, familial febrile
Ataxia with isolated vitamin E deficiency
Achromatopsia
CMO II deficiency
Zellweger syndrome
Refsum disease, infantile form
Lymphoma, non-Hodgkin
Colon adenocarcinoma
Dihydropyrimidinuria
Cohen syndrome
Glaucoma, open angle
Epidermolysis bullosa simplex, Ogna type
Neuropathy, hereditary motor and sensory
Epilepsy
Oncogene PVT (MYC activator)
Nephroblastoma overexpressed gene
Exostoses, multiple, type 1
Chondrosarcoma
Trichorhinophalangeal syndrome type I
Prostate stem cell antigen
Rothmund-Thomson syndrome
Meleda disease

Microcephaly, primary autosomal recessive
Hyperlipoproteinemia
Chylomicronemia syndrome, familial
Combined hyperlipemia, familial
Farber lipogranulomatosis
Hepatocellular cancer
Colorectal cancer
Hemolytic anemia
Hypotrichosis, Marie Unna type
Torsion dystonia, adult onset, of mixed type
Werner syndrome
Spherocytosis
Pfeiffer syndrome
Chondrocalcinosis, with early-onset osteoarthritis
Opiate receptor, kappa
Salivary gland pleomorphic adenoma
Duane retraction syndrome
Charcot-Marie-Tooth neuropathy, autosomal recessive
Branchiootorenal syndromes
Branchiootic syndrome
Adrenal hyperplasia, congenital
Aldosteronism
Nijmegen breakage syndrome
Giant cell hepatitis, neonatal
Renal tubular acidosis-osteopetrosis syndrome
Segmentation syndrome
Spastic paraplegia
Brain-specific angiogenesis inhibitor
Papillomavirus type 18 integration site
Muscular dystrophy with epidermolysis bullosa
Macular dystrophy, atypical vitelliform
Renal cell carcinoma
Langer-Giedion syndrome
Burkitt lymphoma
Hypothyroidism, hereditary congenital
Goiter, adolescent multinodular and nonendemic

9
136 million base pairs

Sex-reversal, autosomal
Hyperglycinemia, nonketotic
Suppression of tumorigenicity, pancreas
Diaphyseal medullary stenosis
Melanoma
Trichoepithelioma, multiple familial
Immotile cilia syndrome
Cartilage-hair hypoplasia
X-ray repair
Fanconi anemia, complementation group G
Sialuria
Hyperoxaluria, primary, type II
Cardiomyopathy
Deafness, autosomal recessive
Choreoacanthocytosis
Prostate-specific gene
Bamforth-Lazarus syndrome
Tyrosine kinase-like orphan receptor
Brachydactyly, type B1
Nephronophthisis (infantile)
Neuropathy, sensory and autonomic, type 1
Fructose intolerance
Basal cell carcinoma, sporadic
Muscular dystrophy, Fukuyama congenital
Basal cell nevus syndrome
Dysautonomia (Riley-Day syndrome)
Esophageal cancer
Endotoxin hyporesponsiveness
Amyotrophic lateral sclerosis, juvenile dominant
Berardinelli-Seip congenital lipodystrophy
Dystonia, torsion, autosomal dominant
Lethal congenital contracture syndrome
Leukemia, acute undifferentiated
Tuberous sclerosis
Hemolytic anemia
Telangiectasia, hereditary hemorrhagic
Ehlers-Danlos syndrome, types I and II
Joubert syndrome
Leukemia, T-cell acute lymphoblastic

Ovarian cancer
Albinism, brown and rufous
Interferon, alpha, deficiency
Leukemia
Cyclin-dependent kinase inhibitor
Venous malformations, multiple cutaneous and mucosal
Arthrogryposis multiplex congenita, distal, type 1
Galactosemia
Acromesomelic dysplasia, Maroteaux type
Myopathy, inclusion body, autosomal recessive
Hypomagnesemia with secondary hypocalcemia
Friedreich ataxia
Geniospasm
Bleeding diathesis
Hemophagocytic lymphohistiocytosis, familial
Chondrosarcoma, extraskeletal myxoid
Pseudohermaphroditism, male, with gynecomastia
Tangier disease
HDL deficiency, familial
Fanconi anemia, type C
Xeroderma pigmentosum
Epithelioma, self-healing, squamous
Leukemia, T-cell acute lymphoblastic
Muscular dystrophy, limb-girdle, type 2H
Bladder cancer
Sex reversal, XY, with adrenal failure
Leukemia transcription factor, pre-B-cell
Porphyria, acute hepatic
Lead poisoning, susceptibility to
Citrullinemia
Dopamine-beta-hydroxylase deficiency
Amyloidosis, Finnish type
Microcephaly, primary autosomal recessive
Leigh syndrome
Leukemia
Nail-patella syndrome
Prostaglandin D2 synthase (brain)
Pituitary hormone deficiency

10
135 million base pairs

Refsum disease, adult
Hypoparathyroidism, deafness, renal dysplasia
DiGeorge syndrome/velocardiofacial syndrome
Leukemia
Thrombocytopenia
Osaka thyroid oncogene
Ewing Sarcoma
Obesity, susceptibility to
Multiple endocrine neoplasia
Medullary thyroid carcinoma
Hirschsprung disease
Thyroid papillary carcinoma
Deafness, autosomal recessive
Serotonin receptor
Moebius syndrome
Hemolytic anemia
Hyperphenylalaninemia
Metachromatic leukodystrophy
Gaucher disease, variant form
SEMD, Pakistani type
Hermansky-Pudlak syndrome
Breast cancer
Multiple advanced cancers
Cowden disease
Lhermitte-Duclos syndrome
Bannayan-Zonana syndrome
Endometrial carcinoma
Polyposis, juvenile intestinal
Prostate cancer
Progressive external ophthalmoplegia
Corneal dystrophy, Thiel-Behnke type
Leukemia, T-cell acute lymphocytic
Spinocerebellar ataxia, infantile-onset
Split hand/foot malformation, type 3
Polycystic kidney disease
Meningioma-expressed antigen
Adrenal hyperplasia, congenital
Diabetes mellitus, insulin-dependent
Anterior segment mesenchymal dysgenesis
Cataract, congenital
Malignant brain tumors
Glioblastoma multiforme
Medulloblastoma
Crouzon syndrome
Jackson-Weiss syndrome
Beare-Stevenson cutis gyrata syndrome

Suppression of tumorigenicity, prostate
Prostate adenocarcinoma
Interleukin receptor, alpha chain, deficiency of
Arrhythmogenic right ventricular dysplasia
Myasthenic antigen B
Lambert-Eaton syndrome
Megaloblastic anemia
Diabetes mellitus, insulin-dependent
Severe combined immunodeficiency disease, Athabascan
Cockayne syndrome, type B
Cerebrooculofacioskeletal syndrome
Opsonic defect
Chronic infections
Retinal nonattachment, nonsyndromic congenital
Cardiomyopathy, dilated, autosomal dominant
Neuropathy, congenital hypomyelinating
Graves disease autoantigen
Hypermethioninemia, persistent, autosomal dominant
Hemophagocytic lymphohistiocytosis, familial
Retinitis pigmentosa, autosomal recessive and dominant
Urofacial syndrome (Ochoa syndrome)
Hypoglobulinemia and absent B cells
Hyperinsulinism-hyperammonemia syndrome
Spastic paraplegia
Dubin-Johnson syndrome
Warfarin sensitivity
Wolman disease
Cholesteryl ester storage disease
Tumor necrosis factor receptor superfamily, member 6
Autoimmune lymphoproliferative syndrome
Epidermolysis bullosa, generalized atrophic benign
Optic nerve coloboma with renal disease
Prostate cancer
Neurofibrosarcoma
Porphyria, congenital erythropoietic
Endometrial carcinoma
Gyrate atrophy of choroid and retina
Pancreatic lipase deficiency
Glaucoma
Pfeiffer syndrome
Apert syndrome
Saethre-Chotzen syndrome
Schizencephaly
Polykaryocytosis inducer (promoter)
Usher syndrome, autosomal recessive, severe

11
134 million base pairs

Beckwith-Wiedemann syndrome
Cyclin-dependent kinase inhibitor
Dopamine receptor
Autonomic nervous system dysfunction
Long QT syndrome
Jervell and Lange-Nielsen syndrome
Thalassemia
Diabetes mellitus, rare form
Hyperproinsulinemia, familial
Breast cancer
Rhabdomyosarcoma
Lung cancer
Segawa syndrome, recessive
Hypoparathyroidism, dominant and recessive
Tumor susceptibility gene
Breast cancer
Usher syndrome
Atrophia areata
Fanconi anemia, complementation group F
Leukemia, myeloid and lymphycytic
Acatalasemia
Aniridia
Peters anomaly
Cataract, congenital
Foveal hypoplasia, isolated
Keratitis
Severe combined immunodeficiency, B cell-negative
Reticulosis, familial histiocytic
Omenn syndrome
Wilms tumor, type 1
Denys-Drash syndrome
Frasier syndrome
Foramina parietalia permagna (Catlin marks)
Exostoses, multiple
Suppression of tumorigenicity, prostate
Prostate cancer
Spinocerebellar ataxia
Hyperlipidemia, combined
Osteoarthritis susceptibility, female-specific
Xeroderma pigmentosum, group E, subtype 2
High bone mass
Osteoporosis-pseudoglioma syndrome
Parathyroid adenomatosis
Centrocytic lymphoma
Multiple myeloma
Mammary tumor and squamous cell carcinoma
Anemia, pernicious, congenital
Multiple endocrine neoplasia
Hyperparathyroidism
Prolactinoma, carcinoid syndrome
Asthma, atopic, susceptibility to
Leukemia, acute promyelocytic
Retinitis pigmentosa, digenic
Cervical carcinoma
Macular dystrophy, vitelliform type (Best disease)
Spinal muscular atrophy with respiratory distress
Paraganglioma or familial glomus tumors
Folate receptor, adult
T-cell immune regulator
Osteopetrosis, recessive
Leukemia, acute myeloid and T-cell lymphoblastic
Ataxia-telangiectasia-like disorder
Apoptosis inhibitor
Deafness, autosomal dominant and recessive
Phenylketonuria
Hypertriglyceridemia
Immunodeficiency
Erythrocytosis, autosomal recessive benign
Glycogen storage disease
Jacobsen syndrome
Paragangliomas, familial nonchromaffin
Herpes virus entry mediator
Epstein-Barr virus modification site
Serotonin receptor
Porphyria, acute intermittent

Freeman-Sheldon syndrome variant
Jansky-Bielschowsky disease
Diabetes mellitus, insulin-dependent
Sickle cell anemia
Thalassemias, beta
Erythremias, beta
Heinz body anemias, beta
HPFH, deletion type
Bladder cancer
Wilms tumor, type 2
Adrenocortical carcinoma, hereditary
Sjogren syndrome antigen
Niemann-Pick disease, types A and B
Osteoporosis
Persistent hyperinsulinemic hypoglycemia of infancy
Deafness, autosomal recessive
Charcot-Marie-Tooth disease, type 4B
Leukemia, T-cell acute lymphoblastic
Hepatitis B virus integration site
Hepatocellular carcinoma
Lacticacidemia
T-cell leukemia/lymphoma
Diabetes mellitus, noninsulin-dependent
Xeroderma pigmentosum, group E
Cardiomyopathy, familial hypertrophic
Prostate cancer overexpressed gene
Coagulation factor II (thrombin)
Hypoprothrombinemia
Dysprothrombinemia
Complement component inhibitor
Angioedema, hereditary
Smith-Lemli-Opitz syndrome, types I and II
IgE responsiveness, atopic
Bardet-Biedl syndrome
Kaposi sarcoma
Diabetes mellitus, insulin-dependent
Meckel syndrome, type 2
Leigh syndrome
Alexander disease
McArdle disease
Somatotrophinoma
UV radiation resistance-associated gene
Vitreoretinopathy
Leukemia/lymphoma, B-cell
Pyruvate carboxylase deficiency
Usher syndrome, type 1B
Papillon-Lefevre syndrome
Albinism, oculocutaneous, type IA
Waardenburg syndrome
Glomerulosclerosis
Lung cancer
Ataxia-telangiectasia
T-cell prolymphocytic leukemia, sporadic
Lymphoma, B-cell non-Hodgkin
Breast cancer
Myopathy, desmin-related, cardioskeletal
ApoA-I and apoC-III deficiency
Hypertriglyceridemia
Hypoalphalipoproteinemia
Corneal clouding, autosomal recessive
Amyloidosis
Dopamine receptor
Dystonia, myoclonic
Ectodermal dysplasia, type 4 (Margarita type)
Hypomagnesemia, renal
Leukemia, myeloid/lymphoid or mixed-lineage
Lung cancer, non small-cell
Hydrolethalus syndrome
Porphyria, acute, Chester type
Megaloblastic anemia syndrome
Friend leukemia virus integration
Ewing sarcoma
Histiocytosis with joint contractures and deafness
Opioid-binding protein/cell adhesion molecule
Bartter syndrome, type 2

12
132 million base pairs

Lupus erythematosus
Hypophosphatemic rickets, autosomal dominant
Coagulation factor VIII (von Willebrand factor)
Tumor necrosis factor receptor superfamily
Periodic fever, familial
Keutel syndrome
Periodic fever, familial (Hibernian fever)
Episodic ataxia/myokymia syndrome
Pseudohypoaldosteronism, type I
Hemolytic anemia
Diabetes-associated peptide (amylin)
Lactate dehydrogenase-B deficiency
Colorectal cancer
Fibrosis of extraocular muscles, autosomal dominant
Adrenoleukodystrophy
Palmoplantar keratoderma, Bothnia type
Melanoma
Rickets, vitamin D-resistant
Anti-Mullerian hormone receptor, type II
Persistent Mullerian duct syndrome, type II
Activating transcription factor 1
Soft tissue clear cell sarcoma
Myopathy, congenital
Meesmann corneal dystrophy
Epidermolysis bullosa simplex
Cataract, polymorphic and lamellar
Sarcoma amplified sequence
Enuresis, nocturnal
Achondrogenesis-hypochondrogenesis, type II
Osteoarthrosis, precocious
Wagner syndrome, type II
SMED, Strudwick type
Scapuloperoneal syndrome
Sanfilippo syndrome, type D
Lipoma
Salivary adenoma
Uterine leiomyoma
Myopia, high grade, autosomal dominant
Darier disease
Spinocerebellar ataxia
Mevalonicaciduria
Hyperimmunoglobulinemia D and periodic fever
Spinal muscular atrophy
Phenylketonuria
Ulnar-mammary syndrome
Diabetes mellitus
Maturity-Onset Diabetes of the Young
Oral cancer

Dentatorubro-pallidoluysian atrophy
Emphysema
Alzheimer disease, susceptibility to
Inflammatory bowel disease
Leukemia, acute lymphoblastic
Hypertension, essential, susceptibility to
Leukemia factor, myeloid
Spastic paraplegia, autosomal dominant
Taste receptors
Glycogen storage disease, type 0
Hypertension with brachydactyly
Alzheimer disease, familial
Retinoblastoma-binding protein
Ichthyosis bullosa of Siemens
Telangiectasia, hereditary hemorrhagic
Leukemia: myeloid, lymphoid, or mixed-lineage
Allgrove syndrome
Diabetes insipidus, nephrogenic, dominant and recessive
Human papillomavirus type 18 integration site
Epidermolytic hyperkeratosis
Keratoderma, palmoplantar, nonepidermolytic
Cyclic ichthyosis with epidermolytic hyperkeratosis
White sponge nevus
Pachyonychia congenita
Fundus albipunctatus
Glioma
Myxoid liposarcoma
Stickler syndrome, type I
SED congenita
Kniest dysplasia
Glycogen storage disease
Rickets, pseudovitamin D deficiency
Interferon, immune, deficiency
Cornea plana congenita, recessive
Growth retardation with deafness and mental retardation
Spinal muscular atrophy, congenital nonprogressive
Cardiomyopathy, hypertrophic
Brachydactyly, type C
Noonan syndrome
Cardiofaciocutaneous syndrome
Tyrosinemia, type III
Lymphoma, B-cell non-Hodgkin, high-grade
Holt-Oram syndrome
Alcohol intolerance, acute
Tumor rejection antigen
Human immunodeficiency virus-1 expression
Amyloidosis, renal

13
113 million base pairs

Cholesterol-lowering factor
Deafness, autosomal dominant and recessive
Vohwinkel syndrome
Ectodermal dysplasia
Muscular dystrophy, limb-girdle, type 2C
Breast cancer, early onset
Pancreatic cancer
Disrupted in B-cell neoplasia
Leukemia, chronic lymphocytic, B-cell
MHC class II deficiency, group B
Hyperornithinemia, hyperammonemia, homocitrullinemia
Serotonin receptor
Retinoblastoma
Osteosarcoma
Bladder cancer
Pinealoma with bilateral retinoblastoma
Wilson disease
Postaxial polydactyly, type A2
Hirschsprung disease
Propionicacidemia, types I or pccA
Holoprosencephaly
Bile acid malabsorption, primary

Cataract, zonular pulverulent
Stem-cell leukemia/lymphoma syndrome
Spastic ataxia, Charlevoix-Saguenay type
Pancreatic agenesis
Maturity Onset Diabetes of the Young, type IV
Enuresis, nocturnal
Dementia, familial British
Rieger syndrome, type 2
X-ray sensitivity
Rhabdomyosarcoma, alveolar
Lung cancer, non small-cell
Spinocerebellar ataxia
Ceroid-lipofuscinosis, neuronal
Microcoria, congenital
Schizophrenia susceptibility
Xeroderma pigmentosum, group G
Coagulation Factor VII deficiency
Oguchi disease
Stargardt disease, autosomal dominant
Coagulation Factor X deficiency
Breast cancer, ductal

14
105 million base pairs

Chorea, hereditary benign
Meningioma-expressed antigen
Myopathy, distal
Defender against cell death
Temperature-sensitive apoptosis
Lysinuric protein intolerance
Ichthyosis, lamellar, autosomal recessive
Ichthyosiform erythroderma, congenital
Spastic paraplegia
Deafness, autosomal recessive
Deafness, autosomal dominant
Arrhythmogenic right ventricular dysplasia
Immunodeficiency
Glycogen storage disease
Phenylketonuria, atypical
Dystonia, DOPA-responsive
Leber congenital amaurosis, type III
Tyrosinemia, type Ib
Alzheimer disease
Machado-Joseph disease
Ovarian cancer
Microphthalmia, autosomal recessive
Cerebrovascular disease, occlusive
Leukemia/lymphoma, T-cell
Agammaglobulinemia
Achromatopsia

Basal ganglia calcification (Fahr disease)
Multinodular goiter
Retinitis pigmentosa, autosomal dominant
Leukemia/lymphoma, T-cell
Oculopharyngeal muscular dystrophy, autosomal recessive
APEX nuclease (multifunctional DNA repair enzyme)
Cardiomyopathy, familial hypertrophic
Oligodontia
Goiter, familial
Carbohydrate-deficient glycoprotein syndrome, type II
Elliptocytosis
Spherocytosis
Anemia, neonatal hemolytic, fatal and near-fatal
Arrhythmogenic right ventricular dysplasia
Marfan syndrome, atypical
DNA mismatch repair gene MLH3
Diabetes mellitus, insulin-dependent
Krabbe disease
Hypothyroidism, congenital
Thyroid adenoma, hyperfunctioning
Graves disease
Hyperthroidism, congenital
Usher syndrome, autosomal recessive
Emphysema-cirrhosis
Hemorrhagic diathesis
X-ray repair

15
100 million base pairs

Hypertension, essential, susceptibility to
CLL/lymphoma, B-cell
Lymphoma, diffuse large cell
Necdin
Prader-Willi syndrome
Angelman syndrome
Hair color, brown
Spastic paraplegia
Limb deformity
Schizophrenia, neurophysiologic defect in
Isovalericacidemia
Spherocytosis, hereditary, Japanese type
Bartter syndrome
Amytrophic lateral sclerosis, juvenile recessive
Dyserythropoietic anemia, congenital, type III
Griscelli syndrome
Deafness, autosomal recessive
Hepatic lipase deficiency
Marfan syndrome
Shprintzen-Goldberg syndrome
Ectopia lentis, familial
Leukemia, acute promyelocytic, PML/RARA type
Cardiomyopathy, familial hypertrophic
Enhanced S-cone syndrome
Glutaricaciduria, type IIA
Epilepsy, nocturnal frontal lobe, type 2
PAPA syndrome
Diabetes mellitus, insulin-dependent

Prader-Willi/Angelman syndrome (paternally imprinted)
Eye color, brown
Human coronavirus sensitivity
Albinism, oculocutaneous, type II and ocular
Andermann syndrome
Cardiomyopathy, dilated and familial hypertrophic
Epilepsy, juvenile myoclonic
Spinocerebellar ataxia
Microcephaly, primary autosomal recessive
Dyserythropoietic anemia, congenital, type I
Muscular dystrophy, limb-girdle, type 2A
Dyslexia
Amyloidosis, hemodialysis-related
Ceroid-lipofuscinosis, neuronal, late infantile
Gynecomastia, familial
Virilization, maternal and fetal
Colorectal cancer
Carbohydrate-deficient glycoprotein syndrome, type Ib
Bardet-Biedl syndrome
Tay-Sachs disease
GM2-gangliosidosis
Tyrosinemia, type I
Mental retardation, severe
Hypercholesterolemia, familial, autosomal recessive
Retinitis pigmentosa, autosomal recessive
Otosclerosis
Bloom syndrome

16
90 million base pairs

Methemoglobinemias, alpha
Erythremias, alpha
Heinz body anemias, alpha
Alpha-thalassemia/mental retardation
Axis inhibitor
Hepatocellular carcinoma
Rubenstein-Taybi syndrome
Tuberous sclerosis
Polycystic kidney disease, adult type I
Leukemia, acute myelomonocytic
Pseudoxanthoma elasticum
Epilepsy, myoclonic, infantile
MHC class II deficiency
Retinitis pigmentosa
Atopy, susceptibility to
Glycogenosis, hepatic, autosomal
Medullary cystic kidney disease, autosomal dominant
Convulsions, infantile and paroxysmal choreoathetosis
Arthrocutaneouveal granulomatosis (Blau syndrome)
Paroxysmal kinesigenic choreoathetosis
Wilms tumor
Hypodontia, autosomal recessive
Cocaine- and antidepressant-sensitive
Orthostatic intolerance
Leukemia, acute myelogenous
Macular dystrophy, corneal
Cataract, Marner type
Norum disease
Fish-eye disease
Tyrosinemia, type II
Breast cancer antiestrogen resistance
Fibrosis of extraocular muscles, congenital
Fanconi anemia, complementation group A
Lymphedema with distichiasis
Spastic paraplegia
Chronic granulomatous disease, autosomal
Giant axonal neuropathy
Urolithiasis, 2,8-dihydroxyadenine
Mucopolysaccharidosis
UV-induced skin damage, vulnerability to

Thalassemia, alpha
Erythrocytosis
Heinz body anemia
Hemoglobin H disease
Hypochromic microcytic anemia
GABA-transaminase deficiency
Cataract, congenital, with microphthalmia
Polycystic kidney disease, infantile severe
Ubiquitin-specific protease, herpes virus-associated
Xeroderma pigmentosum, group F
Microhydranencephaly
Tamm-Horsfall glycoprotein
Cerebellar degeneration-related antigen
Familial Mediterranean fever
Liddle syndrome
Pseudohypoaldosteronism, type I
Batten disease
Mitral valve prolapse, familial
Brody myopathy
Retinoblastoma-binding protein
Inflammatory bowel disease (Crohn disease)
Myxoid liposarcoma, fusion gene in
Cylindromatosis, familial
Spiegler-Brooke syndrome
Townes-Brocks syndrome
Retinoblastoma
Gitelman syndrome
Bardet-Biedl syndrome
Leukemia, acute myeloid, M4Eo subtype
Ras-related gene associated with diabetes
Endometrial carcinoma
Ovarian carcinoma
Breast cancer, lobular
Gastric cancer, familial
Benzene toxicity, susceptibility to
Leukemia, postchemotherapy, susceptibility to
Spinocerebellar ataxia
Stomatocytosis, dehydrated hereditary
Pseudohyperkalemia, familial

17
81 million base pairs

Canavan disease
Ovarian cancer
Miller-Dieker syndrome
Retinitis pigmentosa
Tumor protein p53
Colorectal cancer
Li-Fraumeni syndrome
Cystinosis, nephropathic
Diabetes mellitus, noninsulin-dependent
Cone dystrophy
Myasthenic syndrome
Deafness, autosomal recessive
Smith-Magenis syndrome
VLCAD deficiency
Maturity Onset Diabetes of the Young, type V
Hypertension, essential, susceptibility to
T-cell immunodeficiency, alopecia, and nail dystrophy
Chondrosarcoma, extraskeletal myxoid
Neurotransmitter transporter, serotonin (anxiety-related)
Neurofibromatosis, type 1
Watson syndrome
Leukemia, juvenile myelomonocytic
HIV-1 disease, delayed progression of
Meesmann corneal dystrophy
Muscular dystrophy, limb-girdle
Epidermolysis bullosa simplex, recessive
Pachyonychia congenita, Jackson-Lawler type
Steatocystoma multiplex
Wilms tumor, type 4
Glycogen storage disease (von Gierke disease)
Parkinsonism-dementia
Epidermolytic hyperkeratosis
Patella aplasia or hypoplasia
Osteogenesis imperfecta
Ehlers-Danlos syndrome, types I and VIIA
Osteoporosis, idiopathic
Ovarian carcinoma antigen
Neuroblastoma
Glanzmann thrombasthenia, type A
Thrombocytopenia, neonatal alloimmune
CLL/lymphoma, B-cell
Retinitis pigmentosa
Pituitary tumor, invasive
Myocardial infarction, susceptibility to
Alzheimer disease, susceptibility to
Myotonia congenita, atypical
Cramps, familial
Fetal Alzheimer antigen
Lung cancer, small-cell
Campomelic dysplasia with autosomal sex reversal
Apoptosis inhibitor
Diabetes mellitus, type II
Radical fringe

Bernard-Soulier syndrome
Breast cancer-related regulator of TP53
Hypermethylated in cancer
Lissencephaly
Subcortical laminar heterotopia
Leber congenital amaurosis, type I
Medulloblastoma
Cataract, anterior polar
Myasthenia gravis, familial infantile
Bruck syndrome
Sjogren-Larsson syndrome
Charcot-Marie-Tooth neuropathy
Dejerine-Sottas disease
Van der Woude syndrome modifier
Choroidal dystrophy, central areolar
Huntingtin-associated protein
Psoriasis susceptibility
Epidermolysis bullosa
Alzheimer disease, susceptibility to
Van Buchem disease
Malignant hyperthermia susceptibility
Leukemia, acute promyelocytic
Epidermolytic palmoplantar keratoderma
Pachyonychia congenita, Jadassohn-Lewandowsky type
Keratoderma, nonepidermolytic palmoplantar
Sclerosteosis
Muscular dystrophy, Duchenne-like, type 2
Adhalinopathy, primary
Breast cancer, early onset
Ovarian cancer
Leukemia, myeloid/lymphoid or mixed-lineage
Breast cancer, sporadic
Gliosis, familial progressive subcortical
Pseudohypoaldosteronism type II
Spherocytosis, hereditary
Hemolytic anemia
Renal tubular acidosis, distal
T-cell leukemia virus (I and II) receptor
Dementia, frontotemporal, with Parkinsonism
Trichodontoosseous syndrome
Glanzmann thrombasthenia, type B
Symphalangism, proximal
Synostoses syndrome, multiple
Mulibrey nanism
Growth hormone deficiency
Myeloperoxidase deficiency
Cataracts
Tylosis with esophageal cancer
Adrenoleukodystrophy, pseudoneonatal
Deafness, autosomal dominant
Leukemia, acute myeloid, therapy-related
Myasthenic syndrome, slow-channel congenital
Sanfilippo syndrome, types A and B

18
76 million base pairs

Myopia, high grade, autosomal dominant
Holoprosencephaly
Torsion dystonia, adult-onset, focal
Orthostatic hypotensive disorder of Streeten
Hepatitis B virus integration site
Retinoblastoma-binding protein
Amyloid neuropathy, familial
Amyloidosis, senile systemic
Carpal tunnel syndrome, familial
Pemphigus vulgaris antigen
Diabetes mellitus, insulin-dependent
Pancreatic cancer
Polyposis, juvenile intestinal
Leukemia/lymphoma, B-cell
Colorectal cancer
Lymphoma/leukemia, B-cell, variant
Combined factor V and VIII deficiency
Tumor necrosis factor receptor superfamily

Parkinson disease, susceptibility to
Glucocorticoid deficiency
Schizophrenia
Niemann-Pick disease, types C1 and D
Epidermolysis bullosa
Synovial sarcoma
Keratosis palmoplantaris striata
Cholestasis
Osteosarcoma
Cone dystrophy
Carnosinemia
Protoporphyria, erythropoietic
Squamous cell carcinoma
Osteolysis, familial expansile
Obesity, autosomal dominant
Paget disease of bone
Methemoglobinemia

19
63 million base pairs

Coxsackie virus sensitivity
Cyclic hematopoiesis
Fucosyltransferase-6 deficiency
Hypocalciuric hypercalcemia, type II
Leukemia, myeloid/lymphoid or mixed-lineage
Wegener granulomatosis autoantigen
Bleeding disorder
Persistent Mullerian duct syndrome, type I
Mucolipidosis
Glutaricaciduria, type I
Leprechaunism
Rabson-Mendenhall syndrome
Diabetes mellitus, insulin-resistant
Ichthyosis
Leukemia, T-cell acute lymphoblastoid
Liposarcoma
Mycobacterial and salmonella infections, susceptibility to
Eye color, green/blue
Hemiplegic migraine, familial
Episodic ataxia, type 2
Ataxia, spinocerebellar and cerebellar
Leukemia, acute myeloid
Mannosidosis, alpha, types I and II
Alzheimer disease, late onset
Glomerulosclerosis, focal segmental
Deafness, autosomal dominant
Hypercalcemia, familial benign, Oklahoma type, type III
Orofacial cleft
Charcot-Leyden crystal protein
Hemolytic anemia
Hydrops fetalis
Malignant hyperthermia susceptibility
Central core disease
Osteodysplasia, polycystic lipomembranous
Maple syrup urine disease, type Ia
Camurati-Engelmann disease
Myotonic dystrophy
Heart block, progressive familial, type I
Optic atrophy
3-methylglutaconicaciduria, type III
Cystic fibrosis modifier
Meconium ileus in cystic fibrosis, susceptibility to
Cone dystrophy
Leber congenital amaurosis
Retinitis pigmentosa, late-onset dominant
Diabetes mellitus, noninsulin-dependent
Hyperferritinemia-cataract syndrome
Hypogonadism, hypergonadotropic
Retinitis pigmentosa, autosomal dominant
Ectrodactyly, ectodermal dysplasia, cleft lip/palate

Ataxia, cerebellar, Cayman type
Convulsions, familial febrile
Guanidinoacetate methyltransferase deficiency
Muscular dystrophy
Hirschsprung disease
Peutz-Jeghers syndrome
Leukemia, acute lymphoblastic
Atherosclerosis, susceptibility to
Malaria, cerebral, susceptibility to
Sicca syndrome
Glioblastoma
Thyroid carcinoma, nonmedullary
Low density lipoprotein receptor
Hypercholesterolemia, familial
Arteriopathy, cerebral
Pseudoachondroplasia
Epiphyseal dysplasia, multiple
Severe combined immunodeficiency disease
Hair color, brown
Leigh syndrome
MHC class II deficiency
Exostoses, multiple, type 3
Benign familial infantile convulsions
Leukemia/lymphoma, B-cell
Spondylocostal dysostosis, autosomal recessive
Prostate-specific antigen
Spastic paraplegia , autosomal dominant
Cystinuria, types II and III
Nephrosis, congenital, Finnish type
Generalized epilepsy with febrile seizures plus
Ovarian carcinoma
Microcephaly, autosomal recessive
Hyperlipoproteinemia, types Ib and III
Myocardial infarction susceptibility
Cytochrome P450 (coumarin resistance)
Nicotine addiction, protection from
X-ray repair
Excision repair
Xeroderma pigmentosum, group D
Trichothiodystrophy
DNA ligase I deficiency
Polio virus receptor
Herpes virus entry mediator B
Glutaricaciduria, type IIB
Colorectal cancer
Leukemia, T-cell acute lymphoblastic
Shaw-related subfamily genes
Melanoma inhibitory activity
Cardiomyopathy, familial hypertrophic

20
63 million base pairs

Creutzfeldt-Jakob disease
Gerstmann-Straussler disease
Insomnia, fatal familial
Pantothenate kinase associated neurodegeneration
Alagille syndrome
Corneal dystrophy
Inhibitor of DNA binding, dominant negative
Facial anomalies syndrome
Gigantism
Retinoblastoma
Rous sarcoma
Colon cancer
Galactosialidosis
Severe combined immunodeficiency
Hemolytic anemia
Obesity/hyperinsulinism
Pseudohypoparathyroidism, type Ia
McCune-Albright polyostotic fibrous dysplasia
Somatotrophinoma
Pituitary ACTH secreting adenoma
Shah-Waardenburg syndrome

Diabetes insipidus, neurohypophyseal
McKusick-Kaufman syndrome
Cerebral amyloid angiopathy
Thrombophilia
Myocardial infarction, susceptibility to
Huntington-like neurodegenerative disorder
Anemia, congenital dyserythropoietic
Acromesomelic dysplasia, Hunter-Thompson type
Brachydactyly, type C
Chondrodysplasia, Grebe type
Hemolytic anemia
Myeloid tumor suppressor
Breast cancer
Maturity Onset Diabetes of the Young, type 1
Diabetes mellitus, noninsulin-dependent
Graves disease, susceptibility to
Epilepsy, nocturnal frontal lobe and benign neonatal, type 1
Epiphyseal dysplasia, multiple
Electro-encephalographic variant pattern
Pseudohypoparathyroidism, type IB

21
46 million base pairs

Coxsackie and adenovirus receptor
Amyloidosis, cerebroarterial, Dutch type
Alzheimer disease, APP-related
Schizophrenia, chronic
Usher syndrome, autosomal recessive
Amytrophic lateral sclerosis
Oligomycin sensitivity
Jervell and Lange-Nielsen syndrome
Long QT syndrome
Down syndrome cell adhesion molecule
Homocystinuria
Cataract, congenital, autosomal dominant
Deafness, autosomal recessive
Myxovirus (influenza) resistance
Leukemia, acute myeloid

Myeloproliferative syndrome, transient
Leukemia, transient, of Down syndrome
Enterokinase deficiency
Multiple carboxylase deficiency
T-cell lymphoma invasion and metastasis
Mycobacterial infection, atypical
Down syndrome (critical region)
Autoimmune polyglandular disease, type I
Bethlem myopathy
Epilepsy, progressive myoclonic
Holoprosencephaly, alobar
Knobloch syndrome
Hemolytic anemia
Breast cancer
Platelet disorder, with myeloid malignancy

22
49 million base pairs

Cat eye syndrome
Thrombophilia
Rhabdoid predisposition syndrome, familial
Schizophrenia susceptibility locus
Bernard-Soulier syndrome, type B
Giant platelet disorder, isolated
Hyperprolinemia, type I
Cataract, cerulean, type 2
Leukemia, chronic myeloid
Ewing sarcoma
Neuroepithelioma
Li-Fraumeni syndrome
Fechtner syndrome
Amyotrophic lateral sclerosis
Pulmonary alveolar proteinosis
Meningioma, SIS-related
Dermatofibrosarcoma protuberans
Giant-cell fibroblastoma
Spinocerebellar ataxia
Waardenburg-Shah syndrome
Yemenite deaf-blind hypopigmentation syndrome
Debrisoquine sensitivity
Polycystic kidney disease
Leukodystrophy, metachromatic
Myoneurogastrointestinal encephalomyopathy
Leukoencephalopathy

DiGeorge syndrome
Velocardiofacial syndrome
Schindler disease
Kanzaki disease
NAGA deficiency, mild
Epilepsy, partial
Glutathioninuria
Opitz G syndrome, type II
Ubiquitin fusion degradation
Transcobalamin deficiency
Heme oxygenase deficiency
Manic Fringe
Leukemia inhibitory factor
Sorsby fundus dystrophy
Neurofibromatosis, type 2
Meningioma, NF2-related, sporadic
Schwannoma, sporadic
Neurolemmomatosis
Malignant mesothelioma, sporadic
Deafness, autosomal dominant
Colorectal cancer
Cardioencephalomyopathy, fatal infantile
Adenylosuccinase deficiency
Autism, succinylpurinemic
Glucose/galactose malabsorption
Benzodiazepine receptor, peripheral type
Methemoglobinemia, types I and II

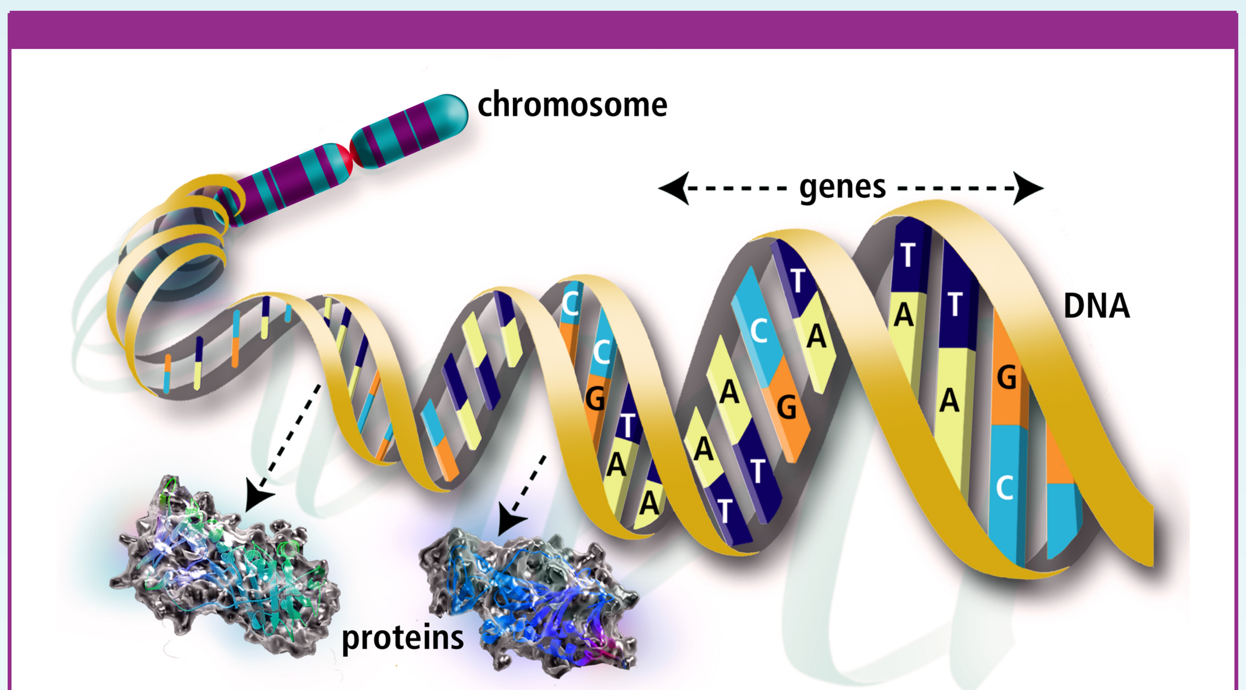
X
153 million base pairs

Short stature, idiopathic familial
Leri-Weill dyschondrosteosis
Langer mesomelic dysplasia
Leukemia, acute myeloid, M2 type
Chondrodysplasia punctata
Kallmann syndrome
Ocular albinism, Nettleship-Falls type
Oral-facial-digital syndrome
Nance-Horan cataract-dental syndrome
Heterocellular hereditary persistence of fetal hemoglobin
Pyruvate dehydrogenase deficiency
Glycogen storage disease
Coffin-Lowry syndrome
Mental retardation
Spondyloepiphyseal dysplasia tarda
Paroxysmal nocturnal hemoglobinuria
Infantile spasm syndrome
Aicardi syndrome
Deafness, sensorineural
Simpson-Golabi-Behmel syndrome, type 2
Adrenal hypoplasia, congenital
Dosage-sensitive sex reversal
Deafness, congenital sensorineural
Retinitis pigmentosa
Wilson-Turner syndrome
Cone dystrophy
Aland island eye disease (ocular albinism)
Optic atrophy
Night blindness, congenital stationary, type 1
Erythroid-potentiating activity
Arthrogryposis multiplex congenita
Night blindness, congenital stationary, type 2
Brunner syndrome
Wiskott-Aldrich syndrome
Thrombocytopenia
Dent disease
Nephrolithiasis, type I
Hypophosphatemia, type III
Proteinuria
Anemia, sideroblastic/hypochromic
Cerebellar ataxia
Renal cell carcinoma, papillary
Diabetes mellitus, insulin-dependent
Sutherland-Haan syndrome
Cognitive function, social
Mental retardation, nonspecific
Menkes disease
Occipital horn syndrome
Cutis laxa, neonatal
FG syndrome
Immunodeficiency, moderate and severe
Miles-Carpenter syndrome
Charcot-Marie-Tooth neuropathy, dominant
Mental retardation
X-inactivation center
Premature ovarian failure
Arts syndrome
Cleft palate and/or ankyloglossia
Megalocornea
Epilepsy (Juberg-Hellman syndrome)
Pelizaeus-Merzbacher disease
Spastic paraplegia
Alport syndrome
Cowchock syndrome
Hypertrichosis, congenital generalized
Ptosis, hereditary congenital
Apoptosis inhibitor
Panhypopituitarism
Thoracoabdominal syndrome
Simpson-Golabi-Behmel syndrome, type 1
Split hand/foot malformation, type 2
Hypoparathyroidism
Mental retardation, Shashi type
Lesch-Nyhan syndrome
HPRT-related gout
Lowe syndrome
Borjeson-Forssman-Lehmann syndrome
Testicular germ cell tumor
Hemophilia B
Warfarin sensitivity
Osseous dysplasia (male lethal), digital
Adrenoleukodystrophy
Adrenomyeloneuropathy
Colorblindness, blue monochromatic
Cardiac valvular dysplasia
Emery-Dreifuss muscular dystrophy
Heterotopia, periventricular
Favism
Hemolytic anemia
Colorblindness, green cone pigment
Incontinentia pigmenti, type II
Hydrocephalus
MASA syndrome
Spastic paraplegia
Rett syndrome
Mature T-cell proliferation
Myopia (Bornholm eye disease)
Mental retardation with psychosis
Endocardial fibroelastosis

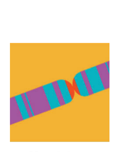
Hodgkin disease susceptibility, pseudoautosomal
Ichthyosis
Microphthalmia, dermal aplasia, and sclerocornea
Episodic muscle weakness
Mental retardation
Ocular albinism and sensorineural deafness
Amelogenesis imperfecta
Charcot-Marie-Tooth disease, recessive
Keratosis follicularis spinulosa decalvans
Hypophosphatemia, hereditary
Partington syndrome
Retinoschisis
Gonadal dysgenesis, XY female type
Mental retardation, non-dysmorphic
Agammaglobulinemia, type 2
Craniofrontonasal dysplasia
Opitz G syndrome, type I
Pigment disorder, reticulate
Melanoma
Duchenne muscular dystrophy
Becker muscular dystrophy
Cardiomyopathy, dilated
Chronic granulomatous disease
Snyder-Robinson mental retardation
Norrie disease
Exudative vitreoretinopathy
Coats disease
Renpenning syndrome
Retinitis pigmentosa, recessive
Mental retardation, nonspecific and syndromic
Dyserythropoietic anemia with thrombocytopenia
Chondrodysplasia punctata, dominant
Autoimmunity-immunodeficiency syndrome
Renal cell carcinoma, papillary
Faciogenital dysplasia (Aarskog-Scott syndrome)
Choroathetosis with mental retardation
Sarcoma, synovial
Prieto syndrome
Spinal muscular atrophy, lethal infantile
Migraine, familial typical
Androgen insensitivity
Spinal and bulbar muscular atrophy
Prostate cancer
Perineal hypospadias
Breast cancer, male, with Reifenstein syndrome
Ectodermal dysplasia, anhidrotic
Alpha-thalassemia/mental retardation
Juberg-Marsidi syndrome
Sutherland-Haan syndrome
Smith-Fineman-Myers syndrome
Hemolytic anemia
Myoglobinuria/hemolysis
Wieacker-Wolff syndrome
Torsion dystonia-parkinsonism, Filipino type
Leukemia, myeloid/lymphoid or mixed-lineage
Anemia, sideroblastic, with ataxia
Allan-Herndon syndrome
Deafness
Choroideremia
Agammaglobulinemia
Fabry disease
Mohr-Tranebjaerg syndrom
Jensen syndrome
Lissencephaly
Bazex syndrome
Mental retardation with growth hormone deficiency
Mental retardation, South African type
Lymphoproliferative syndrome
X inactivation, familial skewed
Pettigrew syndrome
Gustavson mental retardation syndrome
Immunodeficiency, with hyper-IgM
Retinitis pigmentosa
Wood neuroimmunologic syndrome
Heterotaxy, visceral
Albinism-deafness syndrome
Cone dystrophy, progressive
Prostate cancer susceptibility
Fragile X mental retardation
Epidermolysis bullosa, macular type
Diabetes insipidus, nephrogenic
Cancer/testis antigen
Dyskeratosis
Hemophilia A
Hunter syndrome
Mucopolysaccharidosis
Intestinal pseudoobstruction, neuronal
Melanoma antigens
Mental retardation-skeletal dysplasia
Myotubular myopathy
Otopalatodigital syndrome, type I
Colorblindness, red cone pigment
Goeminne TKCR syndrome
Waisman parkinsonism-mental retardation
Barth syndrome
Cardiomyopathy, dilated
Noncompaction of left ventricular myocardium
Von Hippel-Lindau binding protein

Y
50 million base pairs

Short stature homeo box, Y-linked
Short stature
Leri-weill dyschondrosteosis
Langer mesomelic dysplasia
Interleukin-3 receptor, Y chromosomal
Sex-determining region Y (testis-determining)
Gonadal dysgenesis, XY type
Protocadherin 11, Y-linked
Azoospermia factors
Male infertility due to spermatogenic failure
Growth control, Y-chromosome influenced
Chromodomain proteins
Retinitis pigmentosa, Y-linked

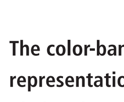


A Closer Look



Chromosomes are tightly coiled microscopic structures made up mainly of DNA, which consists of four different building blocks called bases (A, T, C, and G). The four bases are repeated millions of times to form each chromosome. The entire human genome consists of 23 pairs of chromosomes with one chromosome in every pair coming from each parent. With few exceptions (e.g., red blood cells), each of the trillions of cells in the human body contains a complete set of chromosomes—the genome.

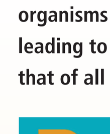
The color-banded figures in this poster are simplified representations (ideograms) of chromosomes, which can be distinguished by observing their features through a light microscope. Such features include length, position of a constricted region (centromere), and the particular patterns of light and dark bands that result from treatment with stains.



Genes are chromosome pieces whose base sequence (e.g., ATTCGGA) determines how, when, and where the body makes each of the many thousands of different proteins required for life. Humans have an estimated 25,000 genes, with an average length of about 3000 bases. Genes make up less than 2% of human DNA; the remaining DNA has important but still unknown functions that may include regulating genes and maintaining the chromosome structure.

Researchers hunt for disease-associated genes by looking for base changes found only in the DNA of affected individuals. Numerous disorders and traits mapped to particular chromosomes are displayed on this poster. Some disorders, such as cystic fibrosis (chr. 7) and sickle cell anemia (chr. 11), are caused by base sequence changes in a single gene. Many common diseases such as diabetes, hypertension, deafness, and cancers have more complex causes that may be a combination of sequence variations in several genes on different chromosomes, in addition to environmental factors.

In April 2003, scientists in the international **Human Genome Project (HGP)** completed a highly accurate, 3-billion base pair human genome reference DNA sequence. The completion of the HGP coincided with the 50th anniversary of the publication of the structure of the DNA molecule by James Watson and Francis Crick. This achievement earned them the Nobel Prize and spawned the new field of molecular biology that led to the inception of the HGP in 1990. DNA sequences are being obtained for the genomes of many other organisms as well and are critical for comparative studies leading to a greater understanding of human biology and that of all living organisms.



A New Era of Customized Medicine. Knowing the DNA sequence is important because it affects responses to particular medicines, resistance to infections and toxins, and it may even influence behavior. Sequence variations also can cause or contribute to such disorders as those named on this poster.

These new data and powerful DNA analysis tools will usher in a new era of medicine that could allow doctors to detect disease at earlier stages, make more accurate diagnoses, and customize drugs and other medical treatments to fit an individual's own DNA sequence. The eventual understanding of gene functions will lead to more focused and effective treatments with fewer side effects.

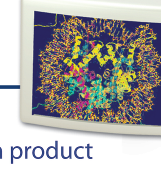
Explore the Human Genome Online!

Gene Gateway

www.ornl.gov/hgmis/posters/chromosome

Step-by-step instructions for using the Web to learn about:

Genetic Disorders
- Causes, inheritance, symptoms, diagnosis, treatments
- Associated genes
- Support groups and organizations
- Genetic health professionals
- Articles and other materials



Genes and Proteins
- Gene name, symbol, size, protein product
- Chromosome maps
- Gene and protein sequence data
- Similar sequences in other organisms
- Gene mutations associated with disorders
- Molecular structures of proteins

For More Information

- Human Genome Project Information: Comprehensive HGP information and a look at the "new biology" of the 21st century
  www.ornl.gov/hgmis/home.shtml
- Genomics and Its Impact on Science and Society: A Primer
  www.ornl.gov/hgmis/publicat/primer/
- Genome Education Resources
  www.ornl.gov/hgmis/education/education.shtml
- DOE Genomics:GTL: Explores how microbial and plant genomes function for bioenergy and other applications
  http://genomicsgtl.energy.gov
- Medicine and the New Genetics: How genetic technologies will revolutionize medicine
  www.ornl.gov/hgmis/medicine/medicine.shtml
- Nature Human Genome Collection: Detailed analyses of all the chromosomes
  www.nature.com/nature/supplements/collections/humangenome
- National Human Genome Research Institute: National Institutes of Health genome program
  www.nhgri.nih.gov
- Careers in Genetics and the Biosciences: Resources for students and teachers
  www.ornl.gov/hgmis/education/careers.shtml
- DOE Joint Genome Institute: Facility for integrated high-throughput sequencing and computational analysis
  http://jgi.doe.gov
- Ethical, Legal, and Social Issues: Implications surrounding use of genome data
  www.ornl.gov/hgmis/elsi/elsi.shtml

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www.ornl.gov/hgmis/posters/chromosome

Y-12 Graphic Design Services

Legend

Regions reflecting the unique patterns of light and dark bands seen on human chomosomes stained to allow viewing through a light microscope.

The centromere, or constricted portion, of each chromosome.

Chromosomal regions that vary in staining intensity and sometimes are called heterochromatin (meaning "different color").

Variable regions, called stalks, that connect a very small chromosome arm (a "satellite") to the chromosome.

Information Sources

Genes associated with the disorders and other traits listed on this poster were selected from *Online Mendelian Inheritance in Man (OMIM)*, which designated the status of each of these as confirmed or provisional as of July 2000. Listing of genes on Y chromosome updated November 2002. The number of base pairs reported for each chromosome is based on finished human genome sequence data from the National Center for Biotechnology Information Build 34, Version 2, accessed February 4, 2004 (www.ncbi.nlm.nih.gov/genome/guide/human/).

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genomics.energy.gov

DISCLAIMER
• For educational purposes only. This poster is not intended to provide medical advice. The extent of knowledge about any specific gene or disorder varies widely. For more detailed information, see the OMIM Web site (www.ncbi.nlm.nih.gov/Omim).

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