

Subject Index for Volume 64

(E) = editorial; (L) = letter to the editor; (R) = review article

- ABC transporter, 739
ABCR, 1024
Acanthosis nigricans, 722
Achondroplasia, 722
Achromatopsia, 1679
ADCA, 1594
Adenomatous polyps, 378
ADMCKD2, 1655
Adverse reactions, 1293
Affected sib pair(s), 281
African American, 495
Age at onset, 839
Alcohol dehydrogenase, 1147
Alcoholism, 1147
Alkaptonuria, 1316
Allele
 Risk, 326 (L)
 Sharing, 871
Allelic association, 1484 (L)
Alopecia, 1323
 α B-crystallin, 685
 α -Mannosidase, 77
ALPS, 1002
Alu element, 446
Alu-mediated deletion, 1340
Alzheimer disease, 290 (L), 832, 839
Amblyopia, 600
Amyloid, 290 (L)
Androgen-receptor methylation, 1445
Angelman syndrome, 70, 385, 397
Anhidrotic/hypohidrotic extodermal dysplasia, 651 (L)
Anticipation, 586, 1580, 1594, 1646
APC, 1228 (L)
APC I1307K, 378
APC mutations, 653 (L)
Apert syndrome, 446
Apoptosis, 1282
ARSACS (see Autosomal recessive spastic ataxia of Charlevoix-Saguenay), 768
Arthritis, 136
Ascertainment, 1243 (L), 1246 (L)
Ashkenazi Jews, 949, 1071, 1233 (L), 1241 (L)
Association, 259, 641, 895 (L), 1194, 1485 (L), 1785 (L)
 Tests, 910 (L)
Asymmetry, 313 (L)
Ataxia-telangiectasia, 46 (E), 1617
ATM, 46 (E), 1617
ATP-binding-cassette transporter, 739
Atrichia, 1323
Atrioventricular-canal defect, 1119
Atypical Usher syndrome, 1221 (L)
Autoimmunity, 1002, 1096
Autonomy, 16 (R)
Autosomal dominant, 136, 189, 471, 556, 594, 788, 904 (L)
Autosomal recessive, 526
Autosomal recessive spastic ataxia of Charlevoix-Saguenay, 768
AV-canal defect, 1119

Bardet-Biedl syndrome, 900 (L)
BBS5, 900 (L)
Behçet disease, 1406
Berbers, 232
Biallelic markers, 629
Bias, 268
Biased distributions, 1248
Bicistronic mRNA, 698
Bicistronic transcript, 706
Bilateral cataract, 586
Bilirubin, 739
Binary data, 886
Bioelectric impedance, 196
Bipolar affective disorder, 210, 1243 (L), 1246 (L), 1670
Bivariate analysis, 1686
Bloom syndrome, 1241 (L)
Body-mass index, 196
Bone density, 1661
Books reviewed, author(s)/editor(s):
 Applegarth, D. A., 330
 Arking, R., 1788
 Dimmick, J. E., 330
 Freshney, R. I., 1488
 Hall, J. G., 330
 Lange, K., 1787
 Ravid, K., 1488
Books reviewed, title:
 Biology of Aging: Observations and Principles, 1788
 DNA Transfer to Cultured Cells, 1488
 Mathematical and Statistical Methods for Genetic Analysis, 1787
 Organelle Diseases, 330
BRAC1, 1228 (L), 1427
BRAC2, 1228 (L)
Brachydactyly, 570
 Type B, 578
BRCA1, 300 (L), 943 (E), 963, 1259, 1371

- BRCA2, 943 (E), 963
- C282Y, 1056
- CACNA1A, 89
- Calcium channel, 89
- Calcium-sensing receptor, 189
- Calmodulin, 189
- Calpain 3, 1524
- Calpainopathy, 1524
- Cancer, 292 (L)
- Brain, 776
 - Breast, 300 (L), 943 (E)
 - Breast, hereditary, 1371
 - Genetics, 328 (L), 921
 - Nonpolyposis colorectal, 329 (L)
 - Ovarian, 300 (L), 943 (E), 1228 (L)
 - Prostate, 776, 921, 1087
 - Syndrome, hereditary, 801
 - Testicular, 921
- Candidate genes, 788
- Cardiac arrhythmia, 1015
- Cardiac defects, 304 (L)
- Cardiomyopathy, 295 (L), 479, 685
- Cardiovascular disease, 685
- Carpal tunnel syndrome, 471
- Carrier
- Frequency, 949
 - Screening, 1241 (L)
- Cartilage, 1036
- Case report, 993
- Cataract, 1357
- Cell fusion, 1400
- Centenarians, 292 (L)
- Centromere, 1440
- Centromere inactivation, 1440
- Cerebellar ataxia, 89
- Cerebral palsy, 526
- CFTR glycoprotein, 1499
- Charcot-Marie-Tooth disease, 1580
- Chinese, 250
- Chondrocalcinosis, 136
- Chromosomal duplication, 1702
- Chromosome 10q, 1127
- Chromosome 10q21-22, 172
- Chromosome 10q23.3-24.2, 586
- Chromosome 10q24, 1646
- Chromosome 12, 939 (E)
- Chromosome 13, 768
- Chromosome 16, 939 (E)
- Chromosome 16p12, 1655
- Chromosome 17, 471, 1420, 1661
- Chromosome 19, 89
- Chromosome 1p36, 776
- Chromosome 1q25-q31, 518
- Chromosome 1q42.2, 1087
- Chromosome 1q43, 1087
- Chromosome 21q22, 210
- Chromosome 22, 594
- Chromosome 22q11, 747, 1076
- Chromosome 2q, 651 (L)
- Chromosome 2q31, 900 (L)
- Chromosome 3, 533, 538
- Chromosome 4, 904 (L)
- Chromosome 5p, 136
- Chromosome 5q35.3, 547
- Chromosome 6, 808, 1096
- Chromosome 6p21-22, 1400
- Chromosome 6p21.3, 146
- Chromosome 7, 310 (L), 556
- Chromosome 8, 563, 1119, 1411, 1679
- Chromosome 9, 570
- Chromosome 9q22, 578
- Chromosome 9q31, 1110
- Chromosome Xp22, 1604
- Chromosome(s)
- Abnormality, 1480 (L)
 - Breakpoints, 747
 - p53, 292 (L)
 - Segregation, 1480 (L)
- Class II HLA DRB1, 1709
- CLN2 protease, 1511
- Clustering, 1248
- Coagulation deficiency, 1071
- Coalescent time, 1071
- Cochleosaccular degeneration, 318 (L)
- Cockayne syndrome, 1259
- Cognition, 118
- Collagen, 62
- Collagen IX, 1036
- Color blindness, 1679
- Colorectal polyposis, 378
- Common fragile sites, 908 (L)
- Common origin, 1233 (L)
- Community review, 1719
- Complementation analysis, 1400
- Complex disease(s), 326 (L), 1243 (L), 1246 (L)
- Complex genetic diseases, 281
- Complex I deficiency, 1505
- Concerted evolution, 24
- Congenital, 600
- Anomaly disorder, 747
 - Blindness, 1225 (L)
 - Cataract, 1357
 - Heart disease, 659 (L)
 - Insensitivity to pain with anhidrosis, 1207 (L), 1570
 - Muscular dystrophy, 126
 - Nystagmus, 1141
- Connective tissue disease, 993
- Connexins, 1357
- Constrained tests, 1248

- Control region, 232
- Copula model, 886
- Coronary heart disease, 1453
- Costa Rica, 1670
- Covariance-structure modeling, 268
- COX deficiency, 1330
- Creutzfeldt-Jakob disease, 1063
- Crohn disease, 808
- CTG repeats, 360
- Culture, 1719
- Cyclic ichthyosis, 732
- Cystic fibrosis, 303 (L), 1499
 - Carriers, 303 (L)
- Deafness, 971, 1580
- Deficiency, 706
- Deletion 22q11.2, 659 (L)
- Dementia, 832
- Denys-Drash syndrome, 1778 (L)
- Dependent sib pairs, 1248
- Der(22) syndrome, 747
- Design effects, 1248
- Development, 538
- Developmental delay, 722
- Developmental dyslexia, 146
- DFNA17*, 318 (L)
- DFNB16*, 1238 (L)
- Diabetes mellitus
 - Type I (insulin-dependent), 793
 - Type II, 1127
- Diagnostic testing, 323 (L)
- Diffuse mesangial sclerosis, 1778 (L)
- DiGeorge syndrome, 659 (L)
- Disability insurance, 329 (L)
- Disease, 1071
 - Epidemiology, 871
 - Mapping, 1728
 - Susceptibility, 793, 1406
- Disequilibrium, 895 (L)
- DNA
 - Alphoid, 1440
 - Ancient, 250
 - Circulating, 218
 - Diagnosis, 51
 - Flexibility, 908 (L)
 - Methylation, 70, 385, 397, 667 (L)
 - Mutational analysis, 667 (L), 993
 - Polymorphism, 1063
 - Pooling, 1679
 - Repair, 1259, 1264, 1270, 1276, 1282
 - Testing, 328 (L)
- Dominant
 - Congenital cataract, 1357
 - Inheritance, 1563
 - X-linked inheritance, 1141
- Double mutant chromosomes, 422
- Double-strand breaks, 1264
- Dubin-Johnson syndrome, 739
- Duplication, 300 (L)
- Duty to recontact, 852
- Dyslexia, 157
- Dystonia-deafness peptide (*DDP*) gene, 759
- ECE-1, 304 (L)
- Ectrodactyly, 1646
- Education, 14 (R)
- Egypt, 1166
- Elastin, 118
- EM algorithm, 1186
- Endoplasmic reticulum, 1493
- Endothelial dysfunction, 673
- Endothelin, 304 (L)
- Endothelin receptor B (*EDNRB*), 1216 (L)
- Endothelium, 310 (L)
- Epidemiology, 963, 1147
- Epidermolytic hyperkeratosis, 732
- Epigenetic, 397
- Epimerase deficiency, 462
- Epiretinal membrane, 1230 (L)
- Error rates, 1194
- Erythrocyte membrane, 108
- Ethics, 1719
- Etiology, 526
- Eugenics, 335
- Evolution, 18, 24, 31, 40 (R), 1709
- Exon, 739, 1709
- Expansion, 323 (L)
- Expression, 1563
 - Studies, 1216 (L)
- Extracellular matrix proteins, 993
- Eye
 - Disease, 897 (L)
 - Movements, 600
- False-positive prediction error, 1739
- Familial
 - Adenomatous polyposis, 653 (L)
 - Analysis, 886
 - Combined hyperlipidemia, 1453
 - Dysautonomia, 1110
 - Hemiplagic migraine, 89
 - Hemophagocytic lymphohistiocytosis, 172
 - Mediterranean fever, 949
- Family size, 225
- FANCE*, 1400
- Fanconi anemia, 1400
- Fas/CD95/APO-1, 1002
- Fatty acid(s), 99
 - β -oxidation, 479
- FDB, 1378

- Fecundity, 225
 Feedback inhibition, 1563
 Fertility, 225
FGFR2, 446
 FH, 1378
 FHL, 165
 Fibroblast growth factor receptor 3, 722
 Finite sample, 910 (L)
 FISH, 1638
 Focal segmental sclerosis, 1778 (L)
 Folate, 1045
 Founder
 Effect, 180, 768, 1110
 Mutation, 1024
 Populations, 1728
 Fragile sites, 354
 Fragile X syndrome, 495
 Frameshift, 658 (L)
 Frasier syndrome, 1778 (L)
 FRAXA, 495
 FRAXE, 495
 Friedreich ataxia, 365 (R)
 Frontotemporal dementia, 414
 FTDP-17, 414
 Full-term placenta, 1445
 Functional disomy, 1783 (L)
 Fundus flavimaculatus, 1394

 G-bands, 908 (L)
 Galactosemia, 462
 GALE, 462
 Gaucher disease, 1233 (L)
 Gaussian model, 1464
 Gene, 165
 Assignment, 126
 -environment interaction, 326 (L), 871
 -family evolution, 40 (R)
 Loss, 18
 Mapping, 1238 (L)
 Regulation, 1365
 Genetic
 Analysis, 533
 Counseling, 290 (L), 385, 993, 1289 (E), 1293
 Diagnosis, 897 (L)
 Disease, 70, 99, 397
 Drift, 949, 1071
 Epidemiology, 871
 Heterogeneity, 172, 1210 (L), 1378
 Information, 335
 Instability, 346
 Linkage, 801, 1243 (L), 1246 (L)
 Linkage analysis, 1646
 Mapping, 594
 Testing, 335, 1289 (E)
 Variation, 1719

 Genetics, 1670
 Service providers, 852
 Genome scan, 196, 1464, 1478 (L), 1694
 Genomewide, 1485 (L)
 Scan, 900 (L), 1453
 Significance, 1739
 Genomic
 Disorders, 471
 Imprinting, 70, 385, 397
 Instability, 653 (L)
 Medicine, 1 (R)
 Record, 31
 Genotype reconstruction, 861
 Genotype-phenotype correlation, 435, 479, 1024, 1225 (L)
 Germ cells, 1638
 Germline, 986
 Mosaicism, 1475 (L)
 Mutation, 308 (L)
 Glaucoma, primary open-angle, in Korea, 1775 (L)
 Glomerulus, 51
 Glycoconjugates, 1563
 Golgi apparatus, 1493
 Gonad, 1632
 Gonadoblastoma, 921

 H63D, 1056
 Hairless gene, 1323
 Hairpin formation, 346
 Hamartoma, 308 (L)
 Haplolethality, 1702
 Haplotypes(s), 508, 1728
 Analysis, 1056, 1063, 1110, 1233 (L)
 Hardy-Weinberg equilibrium, 326 (L)
 HDL cholesterol, 608, 1686
 Health insurance, 329 (L)
 Heart defect, 1119
 Heat shock factors, 685
 Hemochromatosis, 1056
 Hemolytic anemia, 108
 Hereditary
 Breast cancer, 1371
 Cancer risk, 1228 (L)
 Cancer syndrome, 801
 Hearing impairment, 318 (L)
 Hearing loss, 1238 (L)
 Lymphedema, 547
 Nephritis, 62
 Nonpolyposis colorectal cancer, 329 (L)
 Sensory and autonomic neuropathy, 1110
 Sensory and automatic neuropathy type IV, 1570
 Spastic paraplegia, 563
 Tubular disorder, 180
 Hermansky-Pudlak syndrome, 658 (L)
 Heterogeneity, 556, 578, 1411, 1594

- Heteroplasmy, 1158, 1330
 Heterotaxy, 712
 Heterozygosity screening, 1340
 Heterozygote testing, 1241 (L)
 Hibernian fever, 939 (E)
 HIBM, 1420
 High-affinity receptor for nerve growth factor, 1570
 High-resolution mapping, 1076
 Hirschsprung disease, 304 (L), 1216 (L)
 HLA, 896 (L)
 HLA-B, 895 (L), 1406
 HLA-B17, 895 (L)
 hMRE11-hRAD50-NBS1 protein complex, 1264
 HNPCC, 329 (L)
 Homo sapiens, 1709
 Homocysteine, 1045
 Homogentisate 1,2 dioxygenase, 1316
 Homologous recombination, 471
 Homozygosity
 By descent, 900 (L)
 Mapping, 126, 165, 526, 651 (L), 1400, 1679
 HPS, 658 (L)
HRPT2, 518
 HSAN type IV, 1207 (L)
 Human
 Evolution, 619, 1166, 1427
 Sperm karyotypes, 1480 (L)
 Subjects, 1719
 Huntington disease, 323 (L), 1289 (E), 1293
 Hutterites, 225
 Hypercholesterolemia, 1378
 Hyperostosis, 1661
 Hyperparathyroidism, 518
 Hyperparathyroidism–jaw tumor syndrome, 518
 Hypertension, 1694
 Hypocalciuria, 189
 Hypomagnesemia, 180

IBM3, 1420
 Identity by descent, 1464, 1728
 Ideology, 16 (R)
 Idiopathic, 1141
 Generalized epilepsy, 1411
 Male infertility, 1638
 IgA deficiency, 1096
 Immotile cilia syndrome, 313 (L)
 Immunodeficiency, 659 (L)
 Immunoglobulin class switch, 1270
 Inborn errors of metabolism, 1316
 Inbreeding, 225
 Inclusion-body myopathy, 1420
 Infertility, 928
 Informed consent, 1719
 Inner-ear malformation, 318 (L)
 Intestinal inflammation, 808

 Intracellular drift, 1158
 Intron, 1709
 Iron, 1388
 Metabolism, 365 (R)
 Overload, 1388
 Ischemia, 685
 Italians, 949

 Japanese, 250
 Jaw tumor, 518
 Jervell-Lange-Nielsen syndrome, 1015
 Jomon, 250
 Juvenile hemochromatosis, 1388

KCNQ1, 1015
KGFR, 446
KvLQT1, 1015

 Large pedigrees, 839
 Late-infantile neuronal ceroid lipofuscinosis, 1511
 Late-onset sensorineural deafness, 1604
 Laterality, 712
 LDL cholesterol, 608
 Leaky scanning, 698
 Leber congenital amaurosis, 1225 (L)
 Left-right asymmetry, 712
Lefty, 712
 Leiomyomatosis, 62
 Lens, 1357
 Life insurance, 328 (L), 329 (L)
 Limb mammary syndrome, 538
 Limb-girdle muscular dystrophy, 556, 1524
LIMK1, 118
 Linkage, 157, 165, 172, 268, 310 (L), 556, 578, 594,
 651 (L), 895 (L), 904 (L), 1036, 1357, 1411,
 1453, 1485 (L), 1754, 1785 (L)
 Analysis, 126, 136, 180, 196, 210, 281, 586, 608,
 641, 788, 808, 861, 871, 1378, 1388, 1394
 Disequilibrium, 259, 1147, 1186, 1194, 1427,
 1670, 1728, 1754, 1765
 Disequilibrium analysis, 793
 Mapping, 900 (L), 1096, 1655
 Lipoprotein lipase, 608
 Locus, 1388
 LOD score(s), 281
 Log-linear model, 1186
 Long QT syndrome, 1015
 Low copy repeats, 1076
 Lymphedema
 Early-onset, 547
 Hereditary I, 547
 Primary congenital, 547
 Lymphoma, 1002
 Lysenko, 16 (R)
 Lysosomal storage disorder, 77

- Macrophage, 165
- Macular
 Degeneration, 422, 1394
 Dystrophy, 1394
- Magnesium reabsorption, 180
- Major histocompatibility complex, 793, 1096, 1406
- Male infertility, 928
- Malformations, 1702
- Malignant fibrous histiocytoma, 801
- Mapping, 313 (L), 533, 629
- Maternally inherited, 295 (L)
- Maximum likelihood, 886
- McAlpine, Phyllis J., 1253
- Mediterranean fever, 939 (E)
- Medullary cystic disease, 1655
- Memorial, 1253
- Mental retardation, 462, 1119
- Methylenetetrahydrofolate reductase, 1045
- Mexican Americans, 1127
- MHC, 896 (L), 1709
- MICA*, 1406
- Microdeletion, 118
 Syndrome(s), 659 (L), 1076, 1475 (L)
- Microdissection, 1445
- Microencephaly, 1119
- Microinterstitial deletion, 1604
- Microsatellite(s), 629
 Markers, 1406
 Mutation, 793, 1388, 1473 (L)
- Microtubule assembly, 414
- Microtubule-associated protein tau, 414
- Migration, 1166
- Milroy disease, 547
- Minicore disease, 1420
- Minisatellite mutation, 1473 (L)
- Missing data, 1186
- Missplicing, 479
- Mitochondria, 295 (L), 365 (R)
- Mitochondrial
 Complex I, 1505
 Cytopathy, 1330
 Encephalomyopathies, 1158
- Mitochondrion, 971
- Mixed-effects model, 1754
- Model systems, 691 (R)
- Molecular
 Evolution, 18, 24, 31, 40 (R)
 Pathology, 1330
- Molybdenum cofactor, 698, 706
- Molybdopterin-synthase, 698
- Monte Carlo Markov chain, 839
- Monte Carlo tests, 1484 (L)
- Mosaicism, 653 (L), 986, 1632
- Moyamoya disease, 533
- MRP2/cMOAT*, 739
- mtDNA, 232, 250, 295 (L), 1158, 1166, 1330
 Repair, 1276
- Multiallelic TDT, 1484 (L)
- Multidrug resistance-associated protein, 739
- Multiple comparison, 1739
- Multiple epiphyseal dysplasia, 1036
- Multipoint analysis, 259, 839, 1127
- Muscle-eye-brain disease, 126
- Mutation(s), 51, 77, 295 (L), 300 (L), 323 (L), 385, 414, 479, 706, 971, 1036, 1071, 1210 (L), 1221 (L), 1323, 1357, 1524, 1617, 1632
 Analysis, 290 (L), 658 (L), 1775 (L)
 Ancestral, 570
 Detection, 939 (E)
 Intragenic, 1340
 Microsatellite, 793
 Minisatellite, 1473 (L)
 Missense, 422
 Rates, 508
 Recurrence, 89
- Mutational
 Hot spot, 1316
 Spectrum, 1541
- Myopathy, 788
- Myosin, 1420
- Myotonic dystrophy, 360
- N370S mutation, 1233 (L)
- Nail aplasia, 570
- Native American(s), 619, 817
- Neocentromere, 1440
- Neonatal
 Hypertrypsinemia, 303 (L)
 Screening, 303 (L)
- Neoplasm/genetics, 667 (L)
- Nephritis, hereditary, 62
- Nephropathy, 1778 (L)
- Nephrotic syndrome, 51
- Nerve growth factor, 1570
- Networks, 232
- Neural tube defects, 1045
- Neurofibromatosis, 1230 (L)
- Neuronal migration disorder, 126
- New *TIGR/MYOC* mutations, 1775 (L)
- NF2 mutation, 1230 (L)
- Nipple, 538
- Nitric acid, 673
- Nitric oxide synthase, 310 (L)
- Nondisjunction, 1638
- Nonne-Milroy lymphedema, 547
- Nonparametric, 259
 Linkage, 281
- NTRK1 mutation, 1207 (L)
- Nystagmus, 600

- Obesity, 70, 196
 Ocular
 Albinism, 1604
 Motility, 600
 Oldest old, 832
 Oligogenic, 839
 Osteoarthritis, 904 (L)
 Overgrowth syndromes, 372 (E)

 Paracentric inversion, 1480 (L)
 Paraplegia, 563
 PARP, 1282
 Paternal migration(s), 817
 PCR-SSCP, 1775 (L)
 Penetrance, 326 (L), 1002, 1228 (L)
 Peopling of the Americas, 619
 Periodic fever, 939 (E)
 Permutation test(s), 1754
 Peroxisomal disorders, 99
 Peroxisomes, 99
 Pfeiffer syndrome, 446
 Phenotype, 1230 (L)
 Phenotypic variation, 1221 (L)
 Phenylthiocarbamide, 1478 (L)
 PHKA2, 1541
 Photoreceptor degeneration, 897 (L)
 Pingelap, 1679
 Pleiotropic disorders, 1550
 Pleiotropy, 46 (E)
 PMP22 mutation, 1580
 Podocytes, 51
 Poly (ADP-ribose) polymerase, 1282
 Polygenic inheritance, 1216 (L)
 Polyglutamine diseases, 339
 Polymerase chain reaction, 218
 Polymorphism(s), 290 (L), 292 (L), 323 (L), 378, 508, 619, 1427
 Polyposis syndromes, 308 (L)
 Population genetics, 1147, 1765
 Population study, 414
 Positional cloning, 533
 Possible triangle, 871
 Power, 281, 1739
 Analysis, 861
 Approximation, 1177
 Calculations, 1194
 Prader-Willi syndrome, 70, 397
 Predictive testing, 1293
 Predisposition, 378
 Preeclampsia, 310 (L)
 Pregnancy, 218
 Pregnancy-induced hypertension, 310 (L)
 Premature-termination codon, 479
 Premutation, 495

 Prenatal diagnosis, 218
 Presenilin 1, 290 (L)
 Presidential Address, 1 (R)
 Presymptomatic testing, 328 (L)
 Prevalence, 495, 1221 (L)
 Prion disease, 1063
 PRNP mutation, 1063
 PROP, 1478 (L)
 Propylthiouracil, 1478 (L)
 Protective factors, 832
 Protein
 Biosynthesis, 1499
 Misfolding, 339
 Synthesis, 1493
 Translation, 108
 Proteinuria, 51
 Proteoglycans, 372 (E)
 Pseudodominant inheritance, 422
 Psoriasis, 895 (L), 896 (L)
 Psychosocial aspects, 1293
 PTC, 1478 (L)

 Quantitative trait locus/loci, 146, 157, 268, 608, 1686, 1754, 1765
 Quebec, 768

 Reading disability, 146, 157
 Rearrangement disorders, 1076
 Recombination, 40 (R), 62
 Recombination suppression, 1427
 Relatives, 1464
 Relaxed replication, 1158
 Renal magnesium loss, 180
 Repetitive sequences, nucleic acid, 62
 Reproductive compensation, 225
 Reproductive freedom, 335
 RET proto-oncogene, 1216 (L)
 RetGC1 mutations, 1225 (L)
 Retina, 897 (L), 1024, 1394
 Retinal
 Disease, 422
 Dystrophies, 1024
 Hamartoma, 1230 (L)
 Retinitis pigmentosa, 971
 Retinoblastoma, 667 (L)
 Rett syndrome, 1783 (L)
 Rh-deficiency syndrome, 108
 Rh50 glycoprotein, 108
 Ribosomal RNA, 295 (L)
 Riley day syndrome, 1110
 Risk
 Assessment, 667 (L)
 Factor, 292 (L)
 Modification, 1371
 RNA CUG repeats, 360

- RNU2, 1427
 Romano-Ward syndrome, 1015
 RP2, 1210 (L)
 RPE65 mutations, 1225 (L)
- SADDAN, 722
SALL1 mutations, 435
 Sample size, 1485 (L)
SCA10, 594
SCA7, 1594
 Science, 16 (R)
 Sclerosteosis, 1661
 Screening, 943 (E)
 Segregation distortion, 1646
 Selection, 268
 Selective advantage, 949
 Sequence deletion, 62
 Serine, 971
 Short arm of chromosome X, 1141
 Sialic acid, 1563
 Sib pair(s), 196, 259, 268, 1694
 Sib-pair methods, 146
 Siberians, 619
 Sibling, 1785 (L)
 Sibling disequilibrium test, 1785 (L)
 Silent mutations, 1216 (L)
Situs, 712
Situs inversus, 313 (L)
 Skeletal dysplasia, 801
 Skin, skeletal, brain dysplasia, 722
 Smith-Magenis syndrome, 471
SMN1 dosage testing, 1340
SOLAR, 1686
 Solid-phase minisequencing, 993
 Somatic, 1632
SOX2, 538
 Spanish patients, 1233 (L)
 Spastic ataxia, 768
 Spastic paraparesis, 586
 Spasticity, 563
 Specificity, 691 (R)
 Sperm chromosomes, 1480 (L)
 Spina bifida, 1045
 Spinal muscular atrophy, 1340, 1365
 Spinal-cord disorder, 563
 Spinocerebellar ataxia, 594
 Splenomegaly, 1002
 Splicing, 1036, 1617
 Split-hand/split-foot, 1646
 Spontaneous abortion, 934
 Sporadic mutations, 653 (L)
 Standard of care, 852
 Standard of practice, 852
 Stargardt disease, 1394
 Statistical genetics, 629
- Statistics, 641, 910 (L)
 STGD, 1024
 Stress-response proteins, 685
 Suicide, 1289 (E)
 Suppressor mutation, 108
 Supravalvular aortic stenosis, 118
 Survival motor neuron gene, 1340
 Syndactyly, 446
- T lymphocyte, 165
 t(11;22), 747
T-box genes, 1550
 Taste, 1478 (L)
TBX3, 1550
 Testis tissue, 1638
 Therapy, 1365
 Thermolabile variant, 1045
 Threshold model, 1127
 Townes-Brocks syndrome, 435
 Transcription factor(s), 435, 1365
 Transforming growth factor- β , 691 (R)
 Translation inhibition, 1330
 Transmanifesting carriers, 934
 Transmission distortion, 1646
 Transmission/disequilibrium test, 326 (L), 668 (L),
 793, 861, 1177, 1186, 1194, 1485 (L), 1754,
 1785 (L)
 Triglyceride, 608
 Trinucleotide, 1594
 Trinucleotide repeat(s), 323 (L), 360
 Triplet repeats, 346
 Triplolethality, 1702
TRKA, 1570
 tRNA, 971
 True-positive prediction error, 1739
TSC1, 986, 1305
TSC2, 986, 1305
 Tuberous sclerosis, 1305
 Tuberous sclerosis complex, 986, 1632
 Tumor-suppressor gene, 308 (L), 776, 801
 Twins, 157
 Type I error, 1739
 Tyrosine kinase receptor, 1570
- UDP-galactose-4-epimerase, 462
 Ulcerative colitis, 808
 Ulnar-mammary syndrome, 1550
 Unesterified cholesterol, 1686
USH2A gene, 1221 (L)
 Usher syndrome, 1221 (L)
- van Buchem disease, 1661
 Variance components, 146, 259, 1127
 Vascular disorders, 678
 Vascular remodeling, 673

- Vavilov, 16 (R)
Velo-cardio-facial syndrome, 659 (L), 747, 1076
VLCAD deficiency, 479
- WD-40 repeats, 1604
Western Eurasians, 232
Wide genome screening, 586
Williams syndrome, 118
Williams-Beuren syndrome, 1475 (L)
WT1, 1778 (L)
- X activation, 759
X chromosome, 508, 808, 897 (L), 934
- X inactivation, 1445
X-linked, 600
 Liver glycogenosis, 1541
 Mental retardation, 759
 Retinitis pigmentosa, 1210 (L)
Xeroderma pigmentosum, 1259
XXY karyotype, 1781 (L)
- Y chromosome, 218, 508, 619, 921, 928
Yayoi, 250
- Zinc finger, 435