

Index

- 1000 genomes project 112–116
12(S)-hydroxyeicosaintetraenoic acid (12(S)-HETE) 132
2,3,7,9 tetrachlorodibenzo-p-dioxin (TCDD) 466–467 (See also TCDD and Dioxin)
24-hour recall 190
32P-post-labeling assay 49–57
8-hydroxy-2'-deoxyguanosine (8-OHdG) 354, 393–394, 464
8-oxodG 194
Accelerator mass spectrometry (AMS) 49, 53
Acquired (adaptive) immunity 216–233
Additive interaction 294–298
Adductomics 495
Adipokines 193, 199–201
Adiponectin 200–205, 369, 442–447
Adipose tissue 199, 201, 205, 442–448
Aflatoxin (AFB1) 52, 55–56, 68, 192, 340
Airways diseases 395–399, 481
Albumin adducts 55–56
Aliquoting 30–32
- Allele -based analyses, 289–290 -specific PCR, 78 (See also Polymerase chain reaction)
Alzheimer's disease (AD) 407–419
Amyotrophic lateral sclerosis (ALS) 410–411
Analysis of GWAS data 19, 290–293, 351, 367
Analytical variability 31, 167
Androgens 193, 201
Aneuploidy 68, 82, 104, 319, 489, 464
Antibody arrays 128–129
Antigen detection 175–176
ApoE genotypes with LDL-C 377–378
Arbitrarily-primed PCR (AP-PCR) 181
Aromatic amines 53–55, 171–172
Arrayed primer extension (APEX) 75, 77, 80–81
Arsenic and urothelial cancer 342
Asbestos-associated fibrosis (asbestosis) 388
Asia cohort consortium 250
Association studies 106, 116, 261–280 using families (See Family-based association studies)
Asthma 220, 224, 233, 272–279, 395–397, 475–488
- Atherosclerosis 443–444
Atomic absorption (AA) 46
Attenuation bias 164
Autoantibodies 225–226, 230–233
Autoimmune disorders 223
Automated systems 32–34
Bacterial plasmid analyses 181
Barker hypothesis 462 (See also Developmental origins)
Banking specimens 12
Bayesian approach 265, 291, 326
B-cells 217–218 malignancies, 226
Bead-array technology 109
BeadArray platforms 123
BEAMing 80
Benzene 83, 84, 125, 129, 170, 171, 172, 243, 245 and leukaemia, 341–342
Berylliosis 390
Bias 143–161 in prevalence surveys, 186 in the magnitude of the association, 186
Biobanks 90, 498

- Biochemical biomarkers 219
- Bioinformatics 37, 115, 121–141, 498–499
- Biological plausibility 327–328
- Biomarker characteristics, 165 validation, 350–351 validity, 167 of effect, 44, 304 of exposure, 44, 190–196, 304 of susceptibility, 44, 304
- Biomarkers analysis of genetic variation, 99–120 analytical methods, 43–62 assessment of genetic damage, 63–98 biosample management, 23–42 cellular –, 216 Consortium, 317 measurement errors, 143–162 of absolute intake, 190–196 of correlated intake, 190–196 of female and male puberty, 453–457 of female reproductive function, 456–459 of fetal and infant development, 459–460 of intermediate endpoints, 193 of internal dose, 43–57 of male reproductive function, 462–465 of obesity and cancer, 446–447 platforms for analysis, 121–142
- Biorepository 24–39, 113–114
- Biosafety 38
- Biospecimen collection, processing, and storage, 34, 40, 190, 241–242, 247–253 resource, 23
- Bisphenol A 462
- Blood 26, 27–33, 125, 132, 166, 170, 202, 203–204, 243, 306 pressure, 363, 367–379
- Body mass index (BMI) 201, 276, 342, 376
- Breast cancer 51–53, 126, 132, 133, 146, 156, 193, 194, 200–201, 263, 308, 314–319, 447, 496
- Bronchiolitis obliterans syndrome (BOS) 387, 388, 397–399
- C-reactive protein (CRP) 374, 375
- Canalization 375 (See also Developmental adaptation)
- Cancer 337–362
- Candidate plasma biomarkers in CHD, 367 susceptibility genes and investigation, 341, 410, 461
- Capillary electrophoresis 49, 88, 110, 123, 130
- Carcinoembryonic antigen (CEA) 225, 308, 389
- Carcinogen–DNA adducts 49–57, 481
- Case–cohort design 248–249, 282
- Case–control design 250–251, 282
- Case-only and other study designs 255–256, 285, 296, 461
- Causality 184–185, 338–339, 351
- CCR5 receptor 393, 438
- CD3 217
- CD4 217–221
- CD8 218, 221
- CD19 218
- CD36 445–446
- CD45 218, 228, 229
- Cell culture systems 182
- Cellular biomarkers (See Biomarkers)
- Centers for Disease Control and Prevention (US) and blood spot cards, 30 and integration of genetic variation in population-based research, 13 information management at the –, 37 NHANES, 165 PulseNet, 424
- Central melanocortin system 442
- Cerebrovascular disease 443
- Cervical cancer 183, 184, 186, 310, 432
- CFSUM1 and 2 133
- Chemotherapy 52, 85, 169, 308, 310–315, 435
- Childhood cancer 347, 454, 461, 475, 481
- Children 478–492
- Chlamydophila pneumoniae 434
- Cholecystokinin 442
- Cholesterol 102, 155, 441 (See also High-density lipoprotein cholesterol, Low-density lipoprotein cholesterol, Total cholesterol) and colon cancer, 154 and CHD mortality, 370, 371 as a marker of intermediate endpoints, 193 dietary –, 378 LDL –, 244, 363, 366 HDL –, 342 total –, 375
- Chromatin immunoprecipitation (ChIP) 113, 126, 495
- Chromosome aberrations 73, 81–85, 353, 355
- Chronic beryllium disease (CBD), 387–391 lymphocytic leukemia (CLL), 226–227 obstructive pulmonary disease (COPD), 388, 397–399
- Circadian rhythm 206
- Classification and regression tree (CART) 297

- Clinical**
- endpoint, 308, 311
 - medicine, 303–319
 - treatment, 421–434
 - trials, 310–312
- Clonal expansion**
- 217, 218, 222, 223, 227
- Cluster of differentiation (CD) number**
- 216, 389
- Clustering**
- Bayesian –, 291
 - familial –, 364
 - genotype –, 285, 286, 287
 - behaviour, 131
 - for classification, 135
- Coal workers' pneumoconiosis (CWP)**
- 387, 391–393, 394, 399
- Coefficient of variation**
- 158, 167, 209, 462
- Cohort**
- consortium, 248
 - Asia –, 250
 - NCI –, 351
 - design, 246, 248, 256, 263
 - studies, 245
- Collaboration**
- 330
- Comet assay**
- 354, 394, 464
- Commensals and pathogens**
- 426, 434–435
- Common rule**
- 13, 14
- Communicating test and study results**
- 10, 14, 15–17, 486–487
- Comparative genome hybridization (CGH)**
- 123, 306, 319, 348
- Complement fixation**
- 219–220
- Complex phenotypes**
- 272
- Concentration biomarkers**
- 190, 196
- Confidentiality of data**
- 10, 14, 15, 19, 116, 264, 467, 483, 486
- Confounding**
- 11, 16, 184, 225, 253, 268, 272, 273, 291, 326, 327, 329, 345, 349, 350, 351, 370, 373, 375, 443, 461
- Conicity index**
- 448
- Consortia**
- 323, 330
- Copy number variations (CNVs)**
- 103–104
- Coronary heart disease (CHD)**
- 363–379
- Correlated error**
- 157–159
- C-reactive protein**
- 193, 197, 221, 369, 372, 374, 375, 448
- Cross-sectional studies**
- 169, 242–245, 351, 447, 495, 496
- CRP (See C-reactive protein)**
- Cryopreservation**
- 31, 33, 249, 251, 253
- Cultivation**
- 175, 176, 177, 179
 - assays, 181–182
- Cysteine**
- 131
- Cytokine IL17**
- 218
- Cytokines**
- 220, 443, 481
- Data**
- base for SNPs, 102
 - mining methods, 297–298
 - normalisation, 134
 - standardization, 330
- dbSNP (See Database for SNPs)**
- Dementia**
- 408, 412
- Dermal exposures**
- 477
- DES**
- 460
- Design issues**
- 146, 346, 351
- Detection of mutations**
- 74–78
- Developmental adaptation**
- 375 (See also Canalization)
 - defect, 123, 476
 - disorders, 232, 454, 460, 461, 475, 493
 - exposures, 477
 - origins, 462 (See also Barker hypothesis)
 - studies, 25
 - toxicity, 464, 477, 482, 483
- Diabetes**
- 200, 201, 247, 319 (See also Type 1 –, Type 2–, TEDDY)
- Diagnostics**
- 130, 303, 305, 308, 438
- Diagnostics prenatal**
- 82
- Diesel exhaust**
- 172, 479
- Dietary**
- antioxidants, 194, 408
 - biomarkers, 195, 196
 - factors, 107, 189, 378
- Difference gel electrophoresis (DIGE)**
- 127
- Differential**
- measurement error, 144, 145, 146, 147, 150, 154, 155, 159
 - misclassification, 150, 169, 246, 251, 252, 285, 286, 328
- Diffusion-sink device**
- 205
- Dimension reduction**
- 134, 297
- Dioxins**
- 165, 218, 243, 252, 462, 466–467 (See also TCDD)
- Disease course**
- 45, 303, 350, 421, 434, 435
- Disease susceptibility locus (DSL)**
- 264, 271, 283
- Disorders of the immune system**
- 223
- DNA**
- adducts in excretion, 52
 - and protein adducts, 48, 56, 57, 192
 - damage, 52, 63, 65, 66–71, 86, 194–196, 339, 344, 345, 347, 350, 353, 354, 464, 482
 - diagnostics, 126
 - extraction, 31–32, 75, 76, 178, 282, 286
 - microarrays, 124, 126, 128, 129
 - repair, 341, 343, 344, 345, 353, 355, 477, 481, 494
 - sequencing, 74, 89, 111–112, 121–124
- Dose-response models**
- 352
- Doubly labelled water**
- 195
- Dried blood spot (DBS)**
- 175, 176, 227, 459
- Drug**
- development, 130, 310, 311–312, 315, 317, 319
 - discovery, 310–311
- Duplicates**
- 105, 108, 110, 114, 285, 287
- Elston-Stewart algorithm**
- 265–266

- Endocannabinoids** 461, 462, 465, 475, 476, 478, 481, 488
Endogenous
 DNA damage, 52 (See also DNA damage)
 hormones, 27, 33, 190, 200
 (See also Hormones)
Environmental
 163–174, 337
 exposures, 172, 230, 254, 343, 345, 456, 460, 475, 483, 494, 497
 exposure markers, 170
 neurotoxicants, 410
 tobacco smoke (ETS), 54, 170–171, 347, 352, 477, 481
Enzyme-linked immunosorbent assay (ELISA) 49, 53, 55, 57, 148, 149, 183, 390
EPIC-Heart 378
Epidemiology of infectious diseases 422, 426, 427
Epigenetic mechanisms 462
Epigenetics 348–353
Estrogens 53, 200, 201, 205, 412, 457
Ethical issues 9–22, 10, 460, 483, 488
Ethnicity 291, 293, 346, 365, 368, 441, 457
ETS exposure (See Environmental tobacco smoke)
European Prospective Study Into Cancer and Nutrition (EPIC) 191, 339, 342, 345, 352, 378
Evaluation of interactions 295, 296
Exogenous hormones 199, 200
Exposome 172, 494
Exposure
 assessment ,11, 163, 165, 169, 172, 241, 246, 251–252, 256, 285, 294, 345, 392,413, 461, 466, 494, 499
 assessment and misclassification, 246
 biomarkers, 22, 165, 190, 196, 243, 252, 305, 411, 415, 465
in utero, 230, 347, 354, 460,
variability, 167–168
Expression profiling 113, 124, 125, 126, 331, 497
F2-isoprostane 444, 445
Family-based
 association studies, 268, 273, 274, 276
 association tests, 269, 277
 designs, 261–280, 327
FBAT approach 269–277
FBAT-GEE statistic 273
Federal Drug Administration (US FDA) 310, 313, 317, 318
 biomarkers approved by the –, 308
 list of valid genomic biomarkers, 309
Female libido 456
Fetal and infant development 459–460
Fetal lung maturation 459
FEV1 273, 398, 479
Fibrinogen studies collaboration 371, 372, 373, 374
Field validation 350
Finger prick 203, 204
Fingerprint
 carcinogen –, 67, 72, 340
 gene –, 114, 222, 285, 340, 422, 423
Fixed effects models 326 (See also random effects)
FlexTree method 298
Flow cytometry 80, 89, 216, 217, 218, 227, 389, 464
Fluorescence in situ hybridization (FISH) 66, 82, 83, 124, 315, 341, 464
Fold change 125, 127, 134
Follicle stimulating hormone (FSH) 455
Follow-up of cases to determine clinical outcomes 250, 251, 254
Food frequency questionnaires (FFQ) 189, 190, 191, 194, 195–196
Foodborne outbreak 423
Forward phase arrays 128
Fourier transform infrared (FTIR) 130
Framingham 276, 367, 375, 444
Fungal carcinogens 68, 192, 340
Future use of specimens 12, 13, 31, 37, 460
Gail model 496
Gas chromatography (GC) 48, 130, 133, 466, 467
Gender 346–347
Gene
 expression profiling, 113, 124, 125, 126, 331, 497
 fingerprint (See Fingerprint)
 methylation, 66
 -environment interactions, 277, 281–302, 342, 397, 410, 460
 -gene interaction, 255, 281, 285, 299, 301, 410
Genetic
 algorithm (GA), 135, 105, 265, 266
 and lifestyle factors, 377–378
 biomarkers, 222–223, 231, 306, 317, 332, 391
 lineages, 427, 429, 430, 432
 linkage analysis, 261, 264–266, 388, 395, 410
 polymorphisms, 102, 112, 281, 282, 344, 399, 412, 413, 415, 448
 susceptibility, 230, 252, 255, 281, 282, 287, 303, 343, 352, 355, 443, 445–447, 467, 478
 susceptibility loci, 264, 271, 281, 282, 283, 287, 293, 336, 359
 testing, 256, 307, 318, 319, 391
 variation, 99–120, 194, 199, 209, 252, 281–302, 324, 364, 494, 495

- Genome 99–120, 122, 123, 136, 137, 172, 193, 196, 248, 265, 282, 283, 319, 332, 346, 348, 430, 431, 494, 496, 497
 scan meta-analysis (GSMA), 332
 -wide association studies (GWAS), 19–20, 99–120, 194, 264–280, 324–330, 338, 346, 348, 367, 438, 448, 454, 494 (See also Meta-analyses)
 -wide association studies of BM, 448
- Genomic amplification, 124, 175, 178, 180
 approaches, 365
 imprinting, 306, 465
- Genomics 4, 28, 88, 113, 121–142, 317, 319, 324, 333, 355, 495, 496
- Genotoxic compounds 50, 52, 74, 86, 195, 244, 317, 339, 344, 464, 477, 478, 488
- Genotype analysis, 108–109
 -based analyses, 288–290
- Genotypic risk ratio 262
- Genotyping 88, 102–123, 180–182, 252, 265–268, 281–287, 323, 324, 327, 329, 330, 343, 345, 367, 411, 460
- Gestational neuroimmunopathology (GENIP) hypothesis 232–233
- Ghrelin 442
- Glutathione 68, 131, 343, 394, 478, 480
 peroxidase 1, 393–394, 444, 445
- Glycemic status 376
- Half-lives 165–168, 192, 455, 461
- Haplotype analyses, 269–272, 281, 290, 313
 blocks, 103, 268
 mapping, 122–123
 phase, 102, 271
 tagging SNPs (htSNPs), 268
- Haplotypes 103, 122, 230, 231, 265, 266, 267–271, 290
- Hardy-Weinberg equilibrium 105, 286
- HDL-C (See High-density lipoprotein cholesterol) (See also Cholesterol, Low-density lipoprotein cholesterol, Total cholesterol)
- Health Insurance Portability and Accountability Act (HIPAA) 10, 13, 486
- Helper T-cells (See T-cells)
- Haemoglobin adducts 54, 165, 170, 172, 173, 242, 347, 355
- Henle-Koch's postulates 183, 422
- Hepatitis A 182, 423, 424, 435
- Hepatitis B 56, 178, 182, 340, 425, 433, 434, 435
- Hepatitis C 178, 182, 425, 433, 434
- Herceptin 314
- Heritability 261, 282
- Herpes virus 8 425, 432, 433, 434
- Heterocyclic amines (HCAs) 191, 192
- Heteroduplex mobility assay (HMA) 181
- Heterogeneity, 325–327
 disease –, 293, 414
 genetic –, 196
 etiologic –, 343, 255
- Heterogeneity-based genome search meta-analysis (HEGESMA) 332
- Hierarchical-Bayesian methods 293, 298
- High performance liquid chromatography (HPLC) (See Liquid chromatography)
- High-density lipoprotein cholesterol, 372, 374, 375
 SNP detection, 109–110, 267
- High-order interactions 297–298
- High-throughput sequencing 71, 80–81, 124, 126, 436, 496, 112, 121
- HIPAA (See Health Insurance)
- Historical cohort approach 263
- HLA-DP β 1 387, 391, 399
- Hormonal variation 206
- Hormone assays 204, 208
- Hormones 199–213
- Hospital-based studies 250, 251, 253, 254, 282, 287
- Host defence system 176, 182, 216, 219, 220, 221, 224
- HPV serotypes 16 and 18 435
- Huber-White variance correction procedure 263
- HuGENet (See Human genome epidemiology network)
- Human chorionic gonadotropin (hCG), 457, 458, 459
 epidermal growth factor receptor 2 (HER2), 305, 312, 314, 315, 317
 genome epidemiology network (HuGENet), 324, 325, 327, 330
 immunodeficiency virus (HIV), 215, 429, 430, 432, 435, 438, 480
 leukocyte antigens (HLA) (See also HLA-P β 1), 221, 222, 230, 231, 438
 microbiome project (HMP), 113
 papillomavirus (HPV), 39, 178, 183, 184, 186, 187, 426, 432, 435
- I2 325, 326, 329
- Identifiability of biological specimens 14
- IgA 220, 221
- IgD 220
- IgE 219, 220–224, 479, 481
- IgG 182, 185, 220, 221, 232, 233, 306, 369
- IgM 182, 185, 220, 221, 306

- Immune**
- biomarkers in animal models, 224
 - biomarkers of neurodevelopmental disorders, 232
 - deficiency disorders, 223–229
 - proliferative disorders, 224
 - reactive disorders, 223–224
 - system, 215–240
- Immunoaffinity chromatography (IAC)**
- 48
- Immunoassays**
- 48, 49, 53, 57, 177, 178, 183, 208, 218, 345
- Immunodiffusion**
- 183
- Immunogenic exposures**
- 223–224
- Immunoglobulin**
- 219–226
- Immunohistochemistry (IHC) test**
- 49, 57, 314, 315
- Immunopathology**
- 216, 223, 232
- In utero**
- 230, 233, 347, 460, 461, 462, 465, 475–488
 - and childhood periods, 476, 477, 488
- Indirect detection (See Serological methods)**
- Individual variability**
- 167
- Inductively coupled plasma-optical emission spectrometer (ICP-OES)**
- 46
- Infectious**
- causes of cancer, 175–188, 421, 433, 434
 - diseases, 175–188, 421–440
- Infertility/early pregnancy loss**
- 200, 454, 456, 457–488
- Inflammation**
- 216, 441–445
- Inflammatory biomarkers**
- 44, 193
- Informatics**
- 34–37
 - system security, 35–37
- Information**
- bias, 144, 326–327
 - management (See also *Laboratory information management*), 23–42
- Informed consent**
- 12–13, 486
- Innate immunity**
- 216, 221
- Institutional review board (IRB)**
- 10–20, 486
- Insulin**
- resistance, 193, 200–201, 444–446
 - like growth factor I (IGF-I), 201, 209–210, 342
 - like growth factor binding protein 3 (IGFBP-3), 201, 210, 342
- Intake assessment methods**
- 190
- Interleukins**
- 218–220, 374, 389, 393
- Intermediate**
- biologic effects, 243–245
 - endpoints, 242–245
- International HapMap project**
- 102, 104, 108, 110, 111, 116, 122, 283, 291
- International Agency for Research on Cancer (IARC)**
- biological resource centre guidelines, 27, 28, 29, 31
 - classification of carcinogens, 71
 - p53 database, 70, 73
- International Organization for Standardization (ISO)**
- 38
- International Society for Biological and environmental repositories (ISBER)**
- best practices, 27, 28, 29, 31, 34, 37, 38
- Interpretation of biomarker data**
- 16
- Interpreting the results**
- 15, 327
- Interstitial lung diseases**
- 388, 391, 399
- Intervention strategies**
- 353–354, 434, 436, 479, 483
- Intraclass correlation coefficient**
- 143, 155–157, 158, 159, 167, 209, 245
- Intramethod reliability**
- 155–156
- Intrauterine growth retardation**
- 459–460
- Inventory control**
- 36
- Irinotecan**
- 312–314
- Iron**
- 191, 369, 389, 392
- Kadoorie prospective study in China**
- 378
- Kaposi's sarcoma (KS)**
- 425, 432, 433
- Kappa in reliability studies**
- 143, 158
- Kin-cohort approach**
- 263
- K-nearest neighbors (KNN)**
- 135
- Laboratory information management system (LIMS)**
- 110, 115
- Lab-on-a-chip**
- 88–89, 96
- Lander-Green-Kruglyak (LGK) method**
- 265–266
- Leptin**
- 203, 342, 369, 442, 443, 444, 445, 447, 455
- Libido**
- 454, 456, 462, 463
- Lifestyle exposures**
- 373–378
- Linkage**
- analyses, 264–268, 276, 388, 398
 - disequilibrium (LD), 102–103, 106, 265, 267, 268, 272, 283, 327, 376
- Liquid chromatography (LC)**
- 48, 75, 77, 79, 127, 130, 132
 - High performance – (HPLC), 48, 49, 52, 53, 54, 55, 57, 79, 132, 170
 - Ultra-performance – (UPLC), 130, 498
- Liver cancer**
- 45, 47, 52, 56, 79, 192, 193, 226, 340, 341, 344, 434
- LOD score**
- 264–266
- Logistical issues in children's studies**
- 483
- Longitudinal investigation of fertility and the environment (LIFE) study**
- 458
- Low-density lipoprotein (LDL-C) cholesterol**
- 244, 363, 364, 366, 372, 375, 377, 408 (See also Cholesterol, High-density lipoprotein cholesterol, Total cholesterol)

- Low-level exposures**
 352, 482, 499
Lp-PLA2
 371, 372, 376
 studies collaboration, 371
Lung cancer
 56, 67, 86, 132, 171, 172, 226,
 338–340, 347, 348, 349,
 352, 353, 388, 390, 478,
 481
Luteinizing hormone (LH)
 455
Lyme disease
 434
Lymphocytes
 86, 217, 244, 253, 391, 394,
 480
 subsets, 217
Lymphokines
 220, 221
Major histocompatibility complex (MHC)
 (transplantation) antigens, 221
 genes, 222
Malaria
 422, 432
Male
 hormones, 463
 libido, 462–463
Manganese superoxide dismutase (MnSOD)
 193
Mapping by admixture of linkage disequilibrium (MALD) (See also *Linkage disequilibrium*)
 106
Mass spectrometry (MS)
 48, 79, 124, 127–128, 495, 497
 (See also *Tandem – (MS/ MS)*, 79, 127
Mast cells
 219
Matrix-assisted laser desorption/ionization time-of-flight (MALDI-TOF)
 87, 109, 128 (See also *Surface-enhanced laser*)
Measurement error
 143–162, 184
Measuring DNA adducts
 49–57
Mechanisms of mutagenesis
 64–72
Membrane EIAs
 177, 183
Mendelian randomization
 373–375, 461
Menstrual cycle
 205–206, 454, 456–457
Mesothelioma
 388–390
Meta-analyses
 323
 of genome-wide association studies, 330–332 (See also *Genome-wide association studies*)
 of individual participant data (MIPD), 329–330
Metabolome
 122–123, 130, 497
Metabolomics
 121, 129–133, 337, 497
Methicillin resistant Staphylococcus aureus (MRSA)
 427, 428, 435
Methyl mercury
 46, 460, 465
Methylation
 86–88, 126, 348–349
Methylenetetrahydrofolate reductase (MTHFR) gene
 88, 343–344
Mexico City Prospective Study
 378
Microarray
 platforms, 104, 125, 129, 332, 431
 -based gene expression profiling, 331
Microarrays
 83, 124, 125, 126, 128, 180, 219, 332–333, 350, 367, 431, 496
Microbial genome
 180, 181
 Program, 431
Microbiota
 432, 436, 438
Microfabricated genetic analysis systems
 88
Micronuclei
 85–86, 351, 497
Microsatellite instability
 76, 86
 panels, 265
Microscopic examination
 82, 175–176, 463
Minor allele frequency (MAF)
 101, 110, 283, 364, 377, 413
Misclassification (See also *Measurement error*)
 147, 148, 150, 186, 246, 251
 bias, 144
 due to random within-person variation, 246
Mitochondrial genome
 63, 73
Mixtures of carcinogen exposure
 49, 190, 410, 456, 461, 499
Mobile genetic elements
 424, 426, 427, 428–429, 436
Model
 of parallel tests, 156
 -dependent methods, 266, 267
 -free methods, 266, 273
Molecular
 cancer epidemiology, 337–338
 epidemiology, 1, 1–7, 9–22,
 323–324, 242
 fingerprints, 421–423
Monoclonal B-cell lymphocytosis (MBL)
 226–227
Monte-Carlo Markov chain (MCMC) algorithms
 266
Müllerian inhibiting substance (MIS)
 455
Multidimensional protein identification technology (MuDPIT)
 124, 127
Multi-factorial dimension reduction (MDR)
 297–298
Multilocus haplotype
 271
Multimarker
 FBATs, 272
 tagging, 283
Multiple food records
 189
Multiplicative interaction
 294–298, 467
Multistage designs
 263, 274, 282, 284, 293, 352
Multistep carcinogenesis
 72–74
Mutation
 patterns, 70–71
 -enriched PCR, 77–78 (See also *Polymerase chain reaction*)
Mutations
 63–98
 in mitochondrial DNA, 73
Mycotoxins
 192, 410
Myocardial infarction
 129, 364–367, 444, 445
Nanotechnology
 388, 495, 496

- National Cancer Institute (US) iv, 23, 27, 37, 194, 299, 308
 informatics at the – , 37
 Cancer genetic markers of susceptibility (CGEMS), 194
 FDG-PET, 308
- National Centers for Children's Environmental Health 483, 488
- National children's study 462, 488
- National health and nutrition examination Survey (NHANES) 47, 165, 166, 203, 444
- National Institute of Environmental Health Sciences (NIEHS) 73, 137, 455, 483, 488
- Natural killer (NK) cell 217
- NBS (See Newborn bloodspot screening)
- NCI cohort consortium 248, 351
- Nested case-control study 56, 148, 169, 251, 282, 339, 437
- Networks pathways and – , 281, 298 consortia and – , 317, 324, 329–330, 355 transmission – , 426, 430
- Neurobehavioural disorders 233, 482–487
- Neurodegenerative disease 407–419
- Neuromental disorders (NMDs) 232, 233
- New infectious agents 176, 182, 421, 422, 428, 429, 432–433
- Newborn bloodspot screening (NBS) 227
- Newborn screening 233, 307 for immune disorders, 216, 227 for Type 1 diabetes, 231
- NHANES (See National Health and Nutrition)
- NMDs (See Neuromental disorders)
- Nondifferential misclassification (See also Misclassification) 148–154, 246, 251
- Non-lymphoid cells 218
- Non-specific markers 171, 216, 220, 224
- Nuclear magnetic resonance 49, 57, 124, 130, 132, 133, 349, 498
- Nucleic acid detection 177–185
- Nurses health study 203, 415
- Nutrient intake 195
- Nutritional factors, 342, 346–347, 354, 460, 478 status biomarkers, 189, 190
- Obesity 193, 200, 441–452
- Occupational carcinogen exposure 51–52
- “Oomics” 122, 134–136, 319, 496–498
- “Omic” technologies 348–350, 497
- Open reading frames (ORFs) 431
- Opportunistic infection 435
- ORFs (See Open reading frames)
- Origin of an epidemic 426–427
- Outbreak investigations 422–426
- Oxidative damage 353, 387, 393, 398 stress 194, 349, 388, 393, 394, 409, 410, 443–444, 464 – biomarker 8-oxoguanosine, 131
- p53 aflatoxin and – , 340 amplichip – microarray, 47 and apoptosis, 394 and asbestos exposure, 389–390 autoantibodies, 389–390 mutations, 71, 76, 305, 339, 341, 351 smoking and – , 51
- PAH-DNA adducts 50, 51, 56, 195, 339, 340, 346, 347, 353, 354, 478, 480, 482
- Pairs statistic 267
- Pairwise tagging 283
- Parent compounds and metabolites 46, 192, 481
- Parkinson's disease (PD) 409–413
- PCR (See Polymerase chain reaction (PCR))
- Pedigree-based association test (PBAT) 276, 277
- Penetrance 262–264, 266, 343, 346
- Persistent bioaccumulative toxics, 460 organic pollutants, 165, 460
- Personalized medicine 100, 123, 126, 304, 319, 496
- Pesticide exposure 413, 480, 482, 487
- Phenotypic sample mean 272
- Phthalates 460
- Phylogenetics 181, 424, 429, 430
- Physical activity 193, 194
- Physicians' health study 193, 399
- Plasmodium 432
- Pneumoconioses 387, 388, 398
- Policy 40, 337, 356, 476, 488, 498 public health and environmental – , 354
- Polycyclic aromatic hydrocarbons (PAHs) 67, 72, 171–172, 191, 338, 477, 480, 487
- Polymerase chain reaction (PCR) 66, 74, 432 allele-specific – , 78 measurement of rearrangements by – , 83–84 multiplex–, 179 mutation-enriched – , 77–78 real-time–, 83, 180 quantitative (Q-PCR) – , 229 single-template – , 89 sensitivity and specificity, 75–76 -RFLP, 181, 392 -single stranded conformation polymorphism (SSCP), 181
- Polymicrobial infections 436

- Population**
 genetics, 99, 100, 105–107, 292
 stratification, 105–106, 276, 291–294, 327–329
 structure, 291–293
 substructure, 106, 271, 274, 291
 –based case-control studies, 250–255
 –based studies, 250
- Precautionary principle** 18
- Pre-eclampsia** 459
- Pregnancy outcomes** 460–461
- Preventive medicine** 303, 305, 317
- Principal component analyses** 105–106, 131, 291, 327
- Privacy of subjects** 10, 13, 14, 15, 19, 25, 40, 246, 264
- PROCAM risk calculator** 375
- Progesterone** 204–205, 457–458
- Prognosis and prognostics** 308
- Promoter methylation** 86, 348–349
- Prospective**
 cohort, 241
 cohort studies, 245–254, 282, 483, 495–496
 studies collaboration, 370–373
- Prostate, lung, colorectal and ovarian (PLCO) study** 193–194
- Protein**
 biomarker profiles, 413
 microarrays, 128
 quantitation, 127–128
- Proteome** 122–126, 130, 135, 499
 human – organization, 27
- Proteomics** 28, 121–142, 317, 331, 349–351, 407, 438, applications, 497
- Protocol development** 11
- Puberty onset** 453–456
- Pubertal status** 455
- Public health ethics** 18–19
- Publication bias** 327, 355
- Q statistic** 325
- QRISK** 375–376
- Quality**
 assurance, 37, 25, 115, 167
 control, 25, 37, 90, 104, 110, 113–116, 285–287, 291, 466
 bioinformatics –, 136
 genetic data –, 285–287
 microarray –, 134
- Quantitative phenotypes** 265, 272–273
- Quartile-quartile plots (Q-Q plots)** 293–294
- Radioimmunoassay (RIA)** 48, 129, 183, 199, 208
- Random**
 effects models (*See also* Fixed effects), 326
 Forest procedure, 297
 genetic drift, 107
- Randomly amplified polymorphic DNA (RAPD)** 181
- Rapid ascertainment of cases** 250, 252
- Rapid detection of infectious agents** 434
- Recovery biomarkers** 190
- Recruiting participants** 12, 230, 365, 444, 461, 463
- Recurrence risk** 262
 relative –, 262
- Reliability**
 coefficient, 155–158
 study, 144, 146–147, 155–159
- Restriction endonuclease analysis** 181
- Restriction fragment length polymorphism (RFLP)** 75, 77, 108, 181
- Retentate chromatography-mass spectrometry (RC-MS)** 124, 128
- Reticulocytes** 85
- Reverse phase arrays** 128
- Reynolds algorithm** 375
- Risk assessment**
 46, 184
 individual –, 16, 496
 and distinct risk profiles, 253
 and early-life exposures, 465
 for cancer-causing agent, 57
 for low level exposures, 352–353, 184
 of complex diseases, 116
 process, 44
- Risk prediction algorithms** 294, 375–376
- Rolling circle amplification (RCA)** 114
- Saliva** 30–31, 190, 204–205
 /buccal cell collection, 30–31
- Sample**
 collection, 23–42, 205–208
 cost of –, 247
 feasibility of –, 247
 invasive or complicated –, 461
 method of –, 249
 problems in –, 484, 184
 protocols of –, 349
 timing of –, 167, 173, 199, 252
 heterozygosity, 286–287
 processing, 26, 110, 207
 size, 153, 154, 283–285
 in GWAS, 415 (*See also* under Genome-wide)
- Screening cohorts** 249
- Security systems for biospecimen facilities** 38–39
- Segregation analysis** 264–266
- Selection bias** 268, 326, 327
 – in case-control studies, 250, 251, 251
- Selective reporting bias** 327, 328, 329
- Selenium** 191

- Sensitivity**
- beryllium –, 387
 - in error quantification –, 143, 147–150, 159
 - insulin –, 193, 201, 442, 446
 - PCR –, 75–76, 78, 83
 - of analytical techniques, 25, 45, 46, 47
 - of DNA assays, 49, 53, 57, 90, 177, 178, 186
 - of mutation detection, 74–75, 76, 80, 82, 88
 - of omic analyses, 498
- Sequence analysis**
- 104, 110, 113, 115
- Serological methods**
- 176, 182
- Severe acute respiratory syndrome (SARS) (See New infectious agents)**
- Severe combined immune deficiency (SCID)**
- 228, 229
- Sex steroids**
- 199, 200
- Short**
- oligonucleotide mass analysis (SOMA), 75, 79
 - tandem repeats (STRs), 105, 285
 - term longitudinal biomarker studies, 242
- Silicosis**
- 393, 394
 - characteristics of –, 392
 - and coal workers' pneumoconiosis, 388, 392, 399
- Single**
- gene disorders, 307
 - nucleotide polymorphisms (SNPs), 101–103, 104, 106, 109, 110, 111
 - the – consortium, 267
 - in GWASs, 123,
 - and microsatellites, 265, (See also dbSNP, tag SNPs)
- Sources of measurement error in biomarkers**
- 144, 145, 147
- Specificity**
- antibody –, 220
 - antigen –, 217, 220, 222
 - assay – and deviation, 106
 - carcinoembryonic antigen test –, 308
 - in error quantification, 143, 147–150, 159
 - method, 87, 88
 - PCR – and sensitivity, 75
 - sequence – of DNA, 177
 - and Hill's criteria, 184
- Specimen**
- collection, 25, 26, 27–39
 - and errors, 145–146, 157, 159, 208
 - and molecular tools, 437
 - and TCDD exposure assessment, 466
 - less-invasive methods of – for children studies, 479
 - processing, 31–32, 146, 176
 - tracking, 35, 37
- Spectral karyotyping (SKY)**
- 83
- Sperm chromatin structure assay (SCSA)**
- 464
- Spontaneous mutations**
- 69, 223, 427
- Spot urine (See Urine)**
- Spotted microarrays**
- 124
- Standard operating procedures**
- 33, 35, 36, 37, 106, 114, 179
- Standardization**
- data, 330
 - of terms, 37
 - International Organization for –, 38
 - method –, 46
- Statistical**
- evaluation of interaction, 294–295
 - heterogeneity, 325
 - power, 248, 272, 273, 283, 284, 415
- Storage**
- 23–42,
 - alternate technologies for –, 39
 - at the NCI, 37
 - conditions, 34
 - options, 207–208
 - requirements, 202, 203
 - system maintenance, 34
- STROBE-ME**
- 327
- Study design**
- 11, 169
 - basic principles in –, 281–302
 - common epidemiological –s, 447
 - family-based –, 261–280
 - issues in –, 351
 - population-based –, 241–260
- Study results**
- interpreting and communicating –, 15–17
- Subject error**
- 150, 155
- Surface-enhanced laser desorption ionization time-of-flight mass spectrometry (SELDI-TOF-MS)**
- 128, 133, 341 (See also Matrix-assisted laser)
- Surrogate endpoint**
- 304, 308
- Surveillance**
- 2, 424–425
 - in outbreak investigation, 428
 - medical –, 391
 - of vaccines, 184
 - population-based – systems, 19
- Survival study**
- 249, 251, 254, 316
- Susceptibility**
- 2, 16
 - biomarkers of –, 44, 45, 193–194, 220, 222
 - in case-control studies, 252
 - loci, 282–283
- System interoperability**
- 36
- Systematic reviews**
- 323, 324–325, 326, 327
 - of published and unpublished studies, 369
- Systematic variation normalization (SVN)**
- 134
- Systems biology**
- 122, 135
 - studies, 378
- Tag SNPs**
- 283, 296
- TagSNPs**
- 103, 104, 109, 290
- Tandem mass spectrometry (MS/MS)**
- 79, 127–129, 498 (See also Mass spectrometry)
- Target tissue**
- 145, 192, 195, 339

- TCDD (*See also* Dioxin)
466–467
half-life, 165
- T-cells
217–218, 223, 224
development, 228
malignancies, 226
helper –, 218, 221
- T-cell receptor (TR)
217, 220, 222
excision circle (TREC), 228
genes, 222
- Technical validation
350
- TEDDY (The Environmental Determinants of Diabetes in the Young)
230–231
- Temporal variability
167
- Testicular dysgenesis syndrome (TDS)
460
- Testosterone
201, 204, 207, 208, 455, 456, 463
- TH1 response
218
- TH2 response
218
- TH17 pathway
218
- The emerging risk factors collaboration (ERFC)
371–373
- Therapeutics
308–310
- Time integration
247
- Tissue
banks, 195, 318
collections, 28–29, 32
- Tobacco smoke
as an exposure marker,
170–172
and lung cancer, 338–340
- Toll-like receptors (TLRs)
221
- Total cholesterol (*See also* Cholesterol, High-density lipoprotein cholesterol, Low-density lipoprotein cholesterol)
370, 371, 375
- Toxic exposures
476, 481, 483, 496
- Toxicogenomics
136, 348, 497
- Transcript profiling
125
- Transcriptome
113, 122, 123, 496, 497
- Transcriptomics
121–142, 317, 319, 438, 496, 497
- Transgenerational health effects
461
- Translational research
310
- Transmission system
425, 427, 436
- Transmission/disequilibrium test (TDT)
268
- Tricarboxylic acid cycle (TCA)
132
- Triglycerides
as a marker of obesity, 441
in literature reviews, 369, 372
in risk prediction algorithms,
375
PUFA in –, 53
- Tumour-associated antigen (TAA)
225–226
- Tumour-specific antigens (TSAs)
225
- Twin registries
261
- Two-dimensional polyacrylamide gel electrophoresis (2D PAGE)
127
- Two-phase sampling design
255
- Two-stage testing strategy
273, 275
- Type 1 diabetes (T1D)
229, 230, 231
- Type 2 diabetes (T2D)
132, 223, 441–452, 477 (*See also* Diabetes, TEDDY)
- Ultra-performance liquid chromatography (UPLC) (*See also* Liquid chromatography)
- United Kingdom Biobank
26, 28, 345
Ethics and Governance Framework, 12
study, 378
- U.S. Environmental Protection Agency (EPA)
455
acknowledgement, 356
method standardization, 46
–’s cancer guidelines, 476
- Urinary
mutagenicity, 195, 196
nitrogen, 190, 195
- Urine
collection, 29–30, 190, 457
specimens, 349
24-hour –, 204
morning –, 30, 204, 207
spot –, 202, 204
- Vaccine
422, 426
candidates, 421, 434, 435
induced autism, 468
HPV –, 432
HPV – trial, 39
HBV – trials, 184
programmes, 430
tumour –s, 226
US federal – Injury Compensation Program, 468
- Validity
coefficient, 143, 150–151
of a biomarker test ,155
studies, 11, 143, 144, 150, 154
design of – and reliability studies, 146–147, 155
clinical –, 16
relation of reliability to –, 156–157
- Variation
coefficient, 158
- Venice criteria
328, 345
- Vitamin C
191
- Vitamin D
191
–/calcium-related pathway genetics, 194
metabolites, 200
receptor, 209
association of – and several cancer sites, 351
variation of – concentrations, 206
- Vitamin E
191
- Waist
circumference, 445, 448
to-height ratio, 448
- West Nile virus
encephalitis, 433
and dengue, 422
in the United States, 427, 433
- White blood cells (WBC)
50, 202, 203, 218, 249, 253, 339, 480, 481

Whole-genome
amplification (WGA), 30, 109,
114, 248,
research, 13, 19-20, 110, 123,
124, 276, 285
Windows of susceptibility
456, 465
Within-person variability
195
biomarker –, 159, 246, 247, 370
over time, 208
Xenobiotic
131, 464
chemicals, 230
metabolism, 398, 494
activation and detoxification
of –s, 478
exposure to a –, 16, 43, 44,
242, 464
markers of –, 304
X-ray fluorescence (XRF)
46, 410
Zinc
195, 196
superoxide dismutase, 410