

## Index

### a

- acute lymphatic leukemia (ALL) 95, 103, 394, 407
  - children 424, 425
  - HFE 401
  - MAPK–RAS pathway 92, 94
- acute myeloid leukemia (AML) 137, 309, 394
  - children 447, 448
  - MAPK–RAS pathway 92
- adenocarcinomas 90, 187, 208, 320
  - family cancer syndromes 43, 44, 109, 112
  - metastases 17
  - ovarian 207
  - pancreatic 166, 209, 341
- adenomas 250, 254, 445
  - Carney triad 465
  - family cancer syndromes 43, 44, 109
  - gastric cancer 309, 323–325
  - HNPCC 281–283
  - liver 357–359
  - *see also* familial adenomatous polyposis (FAP)
- adenomatous polyposis coli (APC) 7, 62, 63, 366
- adenomatous polyposis syndromes 269
- adenosine 269
- adhesion 7, 133, 263, 313, 315–317
  - cell–cell 3, 7, 13
  - cell–matrix 3, 7
  - metastases 3, 15, 17, 18
- adrenal tumors 137
- adrenocortical carcinomas 76, 89
  - BWS 456, 457
  - children 441, 448
  - LFS 399, 444
- aflatoxins 356, 366
- agammaglobulinemia 447, 448
- age at onset 416
  - brain tumors 109, 110, 112, 116
  - breast cancer 184, 190, 193, 194
  - children 441, 448
  - counseling 453, 463
  - family cancer syndromes 43, 44, 109, 112
  - FAP 257–261
  - gastric cancers 309–321, 323–325
  - GISTs 295–301
  - Gorlin syndrome 96
  - HNPCC 281–283, 251
  - JPS 272–274
  - leukemia 394
  - LFS 499, 444
  - liver tumors 355, 356
  - lung cancer 183–185
  - lymphoma 377, 378
  - MAPK–RAS pathway 92
  - melanoma 411–413
  - MRR 239
  - NBS 377
  - neurofibromatosis 123–125, 127, 134
  - ovarian and endometrial cancer 211
  - overgrowth syndromes 88–91
  - pancreatic cancer 341, 345, 346
  - PJS 270–272
  - predisposition 25–30, 34
  - prostate cancer 210, 215
  - RCC 245–250
  - retinoblastoma 140, 147–150
  - RTS 98
  - schwannomatosis 124
  - thyroid cancer 178, 184, 185
  - Wilms tumor 231, 232, 235
  - XP 415, 419
- Alagille syndrome 365
- alcohol 163, 164, 361
- alpha1-antitrypsin (AAT) deficiency 360
- alfetoprotein (AFP) 89

- Amsterdam criteria 28, 60, 283–287
    - FGC 309, 310, 312, 313, 324–328, 330
    - HNPCC 208–213, 251, 258, 262, 281–283, 286, 289, 290, 292, 293, 309, 312, 313
  - angiofibromas 53, 57, 66, 71
  - angio-immunoblastic type T-cell lymphoma 385
  - angiogenesis 5, 6, 8, 12, 14–16
  - angioliomas 45, 92
  - angiomyolipomas 57, 72, 284
  - angiosarcomas 356, 423, 457
  - aniridia (AN) 236
    - WAGR 231–233, 236, 445, 446
  - antihormonal therapy 489
  - antioxidants 222
  - aplastic anemia 394–396, 405
  - apoptosis 8, 461, 502–508
    - brain tumors 109
    - breast cancer 193
    - DNA repair 377, 378
    - family cancer syndromes 43, 44, 109, 112
    - leukemia 386, 394
    - LFS 460, 461
    - liver tumors 356
    - lung cancer 183
    - MAPK–RAS pathway 92
    - melanoma 411
    - PHTS 274
    - predisposition 34, 35
    - prostate cancer 215
    - retinoblastoma 150
    - Wilms tumor 232
  - astrocytomas 57, 70, 71
    - brain 109, 110, 112, 113, 116, 117
    - children 441–445
    - NF1 127, 131, 444, 461
    - SEGAs 117, 118
    - XP 415
  - ataxia telangiectasia (AT) 347, 382, 394
    - breast cancer 193
    - children 441
    - lymphoma 377, 378, 382
  - attenuated FAP (AFAP; AAPC) 259, 262
    - HNPCC 281
    - MAP 269
  - atypical teratoid/rhabdoid tumors (AT/RT) 75, 116
  - autosomal dominant inheritance
    - predisposition 29
  - autosomal recessive inheritance
    - predisposition 30, 36
  - axin 366
- b**
- Bannayan–Riley–Ruvalcaba syndrome (BRRS) 45, 64, 92
    - PTEN hamartoma tumor syndrome 123, 258, 259, 274
  - Bannayan–Zonana syndrome 101
  - basal cell carcinomas 96, 113, 423
    - family cancer syndromes 43, 44, 109, 112
    - Gorlin syndrome 109
    - RTS 97
    - XP 415
  - base excision repair (BER) 404, 410, 428
  - B-cells 383, 385, 388, 404
    - lymphocytic leukemia (B-CLL) 388
    - lymphomas 378, 384–386, 388, 389
  - Beckwith–Wiedemann syndrome (BWS) 238, 456
    - children 441, 442
    - GSD 364
    - MAPK–RAS pathway 92
    - overgrowth 88–91
    - Wilms tumor 232, 235
  - benign serous cystadenoma 350
  - benzopyrenes 356, 366
  - Bethesda criteria 284, 474
    - endometrial cancers 287
    - gastric cancers 318, 319
    - HNPCC 281–283
  - bile duct tumors 62, 356
    - HNPCC 281–283, 286
  - Birt–Hogg–Dubé syndrome (BHD) 70, 250
  - bladder cancer 48, 166, 187
    - children 441–445
    - Costello syndrome 445, 446
    - family cancer syndromes 43, 44
    - HNSCC 163–166
    - MAPK–RAS pathway 92
    - RCC 245
    - retinoblastoma 147
  - bleomycin 165
  - Bloom syndrome (BS) 30, 447, 448, 460
    - leukemias 393, 394
    - predisposition 26, 30
    - Wilms tumor 232
  - bone marrow failure 4, 5, 11, 12, 393–396, 405, 406, 448
    - leukemias 386, 393
  - bone tumors 441, 453–455
  - bowel cancer 61, 320
  - brain tumor–polyposis syndrome type 1 (BPT1) 61
  - brain tumors 8, 35, 109, 110, 112, 395

- children 442, 429, 445, 447
  - enchondromatosis 457
  - family cancer syndromes 43–45, 50, 52, 54, 56, 58, 60, 62, 70, 72, 74, 76, 78, 80
  - Fanconi anemia 448
  - FAP 445
  - HNPCC 281, 282, 289
  - LFS 399, 444, 460
  - NF1 129, 134
  - NF2 134, 137
  - ovarian cancer 207
  - tuberous sclerosis 445
  - Turcot syndrome 258, 445
  - VHL 247
  - Wilms tumor 232
  - XP 415
  - breast cancer 44, 46, 92, 193, 194, 197–206, 326, 332, 333, 487
    - AT 383
    - counseling 187, 193, 194, 198, 463, 469–471, 473, 474, 477
    - Cowden syndrome 259, 274, 280
    - family cancer syndromes 43, 44, 109, 112, 210, 221
    - gastric cancer 309–315, 318, 321, 323–327, 330–334
    - genes 27, 33, 207, 210, 211, 217, 219, 235, 269
    - GISTs 295
    - LFS 77, 399, 444, 461, 474
    - lung cancer 183, 184
    - men 65, 215, 479
    - metastases 3, 15, 17, 18, 24
    - NF1 128
    - ovarian cancer 198–200, 207, 210, 212, 213
    - PJS 270
    - predisposition 27, 29, 30, 32, 207, 221, 251
    - psycho-oncological aspects 487, 488, 490, 492, 494, 495, 497
    - retinoblastoma 148
  - Burkitt lymphoma 378, 385, 387, 388
- C**
- cadherins 7, 16, 156, 263
    - gastric cancer 43, 309, 310, 312–315, 318–320, 323, 325
  - café-au-lait spots (CLS) 31, 119, 479
    - family cancer syndromes 109, 112, 210
    - GISTs 295, 296
    - NF1 127, 128, 130, 131, 136, 462
  - calcitonin 170, 173–175
  - carboplatin 212, 241, 242
  - cardiac cancer 310
  - cardio-facio-cutaneous (CFC) syndrome 92, 93
  - Carney complex 47, 68, 176
  - Carney syndrome 47, 50, 54
  - Carney–Stratakis syndrome 295, 303, 459
  - Carney triad (CT) 295, 299, 458, 459
  - caspsases 504, 512, 513
  - catenins 7, 82, 327
    - FAP 268, 269, 445
    - Wilms tumor 231, 232
  - caveolin 9
  - celecoxib 269
  - cell death regulator 156
  - cell-type specificity 34–36
  - central nervous system (CNS) tumors 109, 276, 424, 428, 442
    - children 441–443
    - family cancer syndromes 43, 44
    - MRR 239, 241
    - NF1 127, 444
    - NF2 114–116, 444
    - FSC 113
    - Turcot syndrome 118, 258, 259
    - XP 415
  - cerebro-oculo-facio-skeletal syndrome (COFS) 437
  - cervical cancer 75, 100, 138, 209
  - chemotherapy 241, 289, 482, 504
    - AT and NBS lymphoma 383–385
    - brain tumors 110
    - breast cancer 15, 193, 199
    - FAP 261
    - gastric cancer 309, 323
    - GISTs 296, 297, 304
    - HNSCC 163
    - lung cancer 184, 185, 187
    - myeloid leukemia 191
    - NF1 134
    - ovarian cancer 207
    - retinoblastoma 147
    - targeted 501, 508, 509
  - cholangiocellular carcinoma (CCC) 356
  - cholangitis 356, 359
  - cholesterol 364
  - chondromas 457
  - chondrosarcomas 444–446
  - chromatin 155
  - chromophobe carcinomas 45
  - chromophobe RCC 245, 246, 250, 252
  - chronic lymphocytic leukemia (CLL) 402
  - chronic myelogenous leukemia (CML) 298
  - cirrhosis 356, 357, 359, 361–363, 365

- cisplatin 380, 508
  - clause tables 198
  - clear cell cancer 248
    - family syndromes 43
    - RCC 245, 246, 248, 250, 252, 253
  - Cockayne syndrome (CS) 379, 382, 421, 427
    - DNA repair 421, 425, 433
  - colectomy 261, 266, 267, 274, 288, 289
  - collagen 4, 8, 13, 66
  - colocalization of mechanisms of
    - predisposition 34, 36
  - colon cancer 210, 346
    - AT 380
    - breast cancer 193
    - counseling 469, 470
    - DNA repair 377, 378
    - endometrial cancer 207, 208, 210
    - family cancer syndromes 44, 45
    - FAP 365
    - HNPCC 251, 281, 282
    - HNSCC 163
    - metastases 3, 15, 17
    - ovarian cancer 207, 209, 347
    - predisposition 36
    - Turcot syndrome 118
  - colonoscopy 266, 274
    - HNPCC 281, 282
  - colorectal cancers 272, 281
    - adenocarcinomas 208
    - adenomas 252, 256, 257
    - AFAP 262
    - carcinomas 299
    - CS and BRRS 274, 275
    - endometrial cancer 207, 211
    - family cancer syndromes 43, 44
    - FAP 257–259, 261, 262, 365, 445
    - gastric cancer 309, 310, 318, 324, 325
    - JPS 272
    - lung cancer 183
    - metastases 15, 17
    - PJS 270
    - predisposition 26, 28, 30
    - Turcot syndrome 118
    - *see also* HNPCC
  - common variable immunodeficiency syndrome 448
  - comparative genomic hybridization (CGH) 154
  - computed tomography (CT) scans 169, 260, 340
    - renal tumors 245, 247, 249
  - congenital hypertrophy of the retinal epithelium (CHRPE) 261
  - constitutional chromosome 3 translocations 247
  - copper 358, 359
  - Costello syndrome (CS) 48, 87, 94, 445, 446
  - cotinine 185
  - counseling 453, 469–474
    - breast cancer 193, 194, 197, 460, 461, 475, 476
    - gastric cancer 309, 310
    - ovarian cancer 210, 460, 471, 475, 476, 480
    - pancreatic cancer 345
    - psycho-oncological aspects 497
    - renal tumors 250
    - risk calculation 474, 475
    - Wilms tumor 238, 241
  - Cowden syndrome (CS) 63, 274
    - family cancer syndromes 45
    - FNMTC 169
    - overgrowth syndromes 87, 89
  - cryotherapy 149, 253
  - Cushing syndrome 67
  - cutaneous malignant melanoma (CMM) 416
  - cyclin 389, 412–414, 456
  - cyclophosphamide 185, 242
  - cystadenoma 247
  - cysteine 8, 429, 504
  - cystic fibrosis (CF) 345, 346
  - cytarabine 390
  - cytosine 368
  - cytostatic therapy 377, 379
- d**
- dactinomycin 241, 242
  - death receptors (DR) 504, 505
  - Denys–Drash syndrome (DDS) 231, 232, 237, 445
  - desferrioxamine 358
  - desmoids 49, 261, 269
    - AFAP 262
    - FAP 261, 265, 266, 268, 269, 455
    - Gardner syndrome 262
  - detoxification 164, 183, 185, 186, 189, 404
  - diabetes 46, 71, 349, 351, 357
  - Diamond–Blackfan anemia (DBA) 394, 397, 448
  - diepoxibutan (DEB) 395
  - diet 183, 222, 334, 359, 363
  - diffuse large B-cell lymphoma (DLBCL) 385
  - Di George/velocardiofacial syndrome (DGS/VCFS) 240, 241

DNA double strand breaks (DSB) 377, 381, 382–384, 390, 404  
 DNA repair 77, 183, 378, 393, 394, 404, 411, 421, 508  
 – breast cancer 193, 194  
 – deficiency 377–390  
 – LFS 460, 461  
 – liver tumors 356  
 – lung cancer 183–185  
 – melanoma 411, 412, 414  
 DNA single-strand breaks 378, 382  
 docetaxel 212, 508  
 dormancy 6, 10–12, 153  
 Down syndrome (trisomy 21) 398, 446  
 doxorubicin 241  
 duodenal polyposis 265, 268  
 duodenal tumors 51, 53, 60, 261, 268  
 Duncan’s disease 448  
 dyskeratosis congenital (DC) 394–396  
 dysplastic nevus syndrome 414

**e**

Ehlers–Danlos syndrome 189  
 electron microscopy (EM) 149  
 emphysema 360, 361  
 employment 478  
 enchondromatosis 454, 457  
 endocrine therapy 11  
 endocrine tumors 65  
 endolymphatic sac tumors (ELST) 246  
 endometrial cancer 48, 207–213, 287, 312, 471  
 – BRRS 259  
 – Cowden syndrome 274  
 – family cancer syndromes 43, 44, 48, 50, 53, 59, 60, 62  
 – galactosemia 362  
 – HNPCC 207–211, 251, 281, 282, 286, 289, 320  
 endoscopic retrograde  
 cholangiopancreatography (ERCP) 350  
 endoscopic ultrasound (EUS) 350  
 endoscopy 310, 319, 324, 330–334  
 – FAP 266–268  
 – JPS 272, 273  
 – PJS 271, 347  
 environmental factors 27, 28, 30, 36, 163, 458  
 – HNPCC 162–165, 281  
 – leukemia 400  
 – lung cancer 183–185  
 – melanoma 421  
 – PDB 458  
 – prostate cancer 216

ependymomas 133, 138  
 – brain tumors 109, 110, 112  
 – family cancer syndromes 54, 56, 57, 64, 70  
 epidermoid cysts 261  
 epithelial-mesenchymal transition (EMT) 13  
 Epstein–Barr virus (EBV) 167  
 esophageal cancer 11, 62, 166, 332, 341  
 – GISTs 295, 298  
 esophageal leiomyoma 459  
 esophago-gastro-duodenoscopy (EGD) 266, 268  
 ethnicity 27, 88, 166, 250, 294  
 – Ashkenazi jews 31, 220, 350, 351, 476  
 – gastric cancer 309, 312, 335  
 – leukemias 404  
 – liver tumors 355–358  
 – melanoma 411, 417  
 – prostate cancer 215–217, 219–222  
 etoposide 241, 383  
 Ewing sarcoma 55, 75, 458  
 excision repair cross-complementing (ERCC) 187  
 exomphalus-macroglossia-gigantism (EMG) syndrome 456  
 expressivity 152, 153  
 extracellular regulated kinase (ERK) 12  
 ezrin 114

## **f**

familial adenomatous polyposis (FAP) 35, 49, 60, 62, 257–265, 333, 365  
 – children 441, 443  
 – counseling 453, 467  
 – family cancer syndromes 44, 50, 60, 62  
 – FNMTc 169  
 – gastric cancer 259, 320, 323, 324  
 – gastrointestinal polyposis syndromes 249–251, 252–260  
 – HNPCC 262, 281  
 – pancreatic cancer 341  
 – sarcomas and bone tumors 453  
 – Turcot syndrome 118, 432  
 familial atypical multiple mole melanoma (FAMMM) 346, 421  
 – pancreatic cancer 69, 336, 341  
 familial clear cell renal cell cancer (FCRC) 248  
 familial gastric cancer 309, 312, 315, 324, 325  
 familial medullary thyroid carcinoma (FMTC) 69, 164–167

- familial non-medullary thyroid carcinoma (FNMTC) 169, 176, 177  
 familial pancreatic cancer (FPC) 335, 336, 338–341  
 familial papillary microcarcinoma 176  
 familial platelet disorder (FPD) 401  
 familial polyposis coli 334  
 Fanconi anemia (FA) 26, 380, 436  
 – breast cancer 189, 191  
 – leukemia 384, 385, 392  
 – Wilms tumor 232  
 ferritin 357  
 ferroportin 356  
 fibrin and fibrinolysis 8  
 fibroadenomas 202  
 fibrofolliculomas 45  
 fibromas 49, 62, 253, 444  
 fibromatosis 442, 443  
 fibronectin (FN) 8, 22  
 fibrosarcomas 35, 356, 423, 458  
 fleurettes 149, 150  
 Flexner–Wintersteiner rosettes 143, 144  
 fluorodeoxyglucose positron emission tomography (FDG-PET) 136  
 follicular thyroid cancer (FTC) 163  
 Frasier syndrome (FS) 231  
 fructosemia 263
- g**
- gain-of-function mutations 44, 93, 305  
 galactosemia 362  
 gall bladder cancer 285  
 gangliocytomas 64, 110  
 gangliomas 170  
 ganglioneuroblastomas 48, 94  
 ganglioneuromas 66, 92, 112  
 Gardner's fibroma 454, 455  
 Gardner's syndrome 35, 49, 60, 61, 72, 253, 454  
 – FAP 250, 251, 253  
 – FNMTC 169  
 gastrectomy 274, 315, 319, 322  
 gastric adenomas 252  
 gastric and gastrointestinal cancer 26, 99, 148, 326, 328–335  
 – AFAP 258  
 – family cancer syndromes 43–46, 49–51, 53, 54, 59, 60, 62–64, 67–69  
 – FAP 259, 320, 323, 324  
 – HNPCC 274, 280, 303, 304, 311–317, 330  
 – JPS 272, 323, 325  
 – metastases 3, 15, 17, 301  
 – PJS 270, 320, 323, 325  
 – upper digestive tract 164  
 gastric ulcers 325, 328  
 gastritis 325, 328  
 gastrointestinal polyposis syndromes 249–267  
 gastrointestinal stromal tumors (GISTs) 131, 132, 287–295, 485  
 – Carney triad 458, 459  
 – neurofibromatosis 450  
 gemcitabine 508  
 gender 200, 361, 401, 486  
 – ALL 390  
 – brain tumors 112  
 – breast cancer in men 64, 214, 463  
 – BRRS 266  
 – gastric cancer 301, 304, 310  
 – GISTs 295  
 – HFE 356  
 – HNPCC 274  
 – lung cancer 180  
 – PJS 270  
 – TSC 70  
 – VHL 73  
 – Wilms tumor 238  
 gene association studies 188  
 genetic testing 88, 164, 165, 436, 450, 474  
 – brain tumors 114, 129, 130  
 – breast cancer 187, 191, 192, 194  
 – counseling 458, 460, 462, 467, 469  
 – family cancer syndromes 44  
 – FAP 258  
 – gastric cancer 309, 315  
 – HFE 356  
 – melanoma 411  
 – psycho-oncological aspects 497  
 – RCC 245  
 – retinoblastoma 147  
 – XP 415  
 genitourinary anomalies (GU) 231, 236  
 genitourinary tumors 55, 64, 207  
 genodermatoses 68, 73  
 genome-wide association (GWA) 188, 196, 197  
 genotype 27, 30, 31, 64, 75  
 – FAP 257–262  
 – gastric cancer 309, 325  
 – GISTs 295–304  
 – Gorlin syndrome 95, 96  
 – JPS 272–274  
 – leukemia 394  
 – lung cancer 183–185  
 – melanoma 411–418

genotype/phenotype correlation 75  
 – HNPCC 312  
 – leukemia 385  
 gleevec 502  
 glial tumors 414  
 glioblastomas 61, 112, 119, 136  
 – brain tumors 109, 110, 112  
 – Turcot syndrome 258  
 glioma polyposis syndrome 119  
 gliomas 380  
 – brain tumors 109, 110, 112  
 – children 424  
 – family cancer syndromes 50, 51, 54, 61, 68  
 – NF1 127–139, 444  
 – retinoblastoma 147  
 globoid dysplasia 318, 319  
 glomus tumors 137  
 glycogen storage disease (GSD) 364  
 gonadal and germ-cell tumors 441  
 gonadoblastoma 89, 92, 237  
 Gorlin–Gotz syndrome 112  
 Gorlin syndrome 26, 49, 68, 95, 96, 109, 112, 113  
 gynecological tumors 208, 213  
 – *see also* cervical cancer; uterine cancer  
 gynecomastia 272

## **h**

hamartin 117, 248  
 hamartomas 64, 66, 304, 456  
 – brain tumors 106, 109, 112, 113  
 – BRRS 274, 275  
 – BWS 456, 457  
 – Carney triad 295  
 – family cancer syndromes 43, 44, 109, 112, 210, 221, 326, 399  
 – Gorlin syndrome 93  
 – PJS 347  
 hamartomatous polyposis syndromes 258  
 haploinsufficiency 90, 236  
 – MAPK–RAS pathway 92  
 head and neck cancer 163, 164, 166, 219  
 – rhabdoid 116  
 – squamous cell (HNSCC) 163  
*Helicobacter pylori* (HP) 310, 343, 345  
 hemangioblastomas 110, 247  
 hemangiomas 45, 57, 63, 444  
 hemochromatosis (HFE) 356–358, 400, 401  
 hemorrhagic telangiectasia 274  
 hepatic tumors *see* liver tumors hepatitis 355, 356  
 hepatobiliary cancer 61

hepatoblastoma 89, 356, 364, 365  
 – BWS 433, 434, 444  
 – children 430, 432–434  
 – family cancer syndromes 49, 61  
 – FAP 260, 354, 432  
 hepatocellular carcinoma (HCC) 92, 355, 445, 446  
 – overgrowth syndromes 87–89  
 herceptin 485  
 hereditary diffuse gastric cancer (HDGC) 26, 49, 309, 312, 313  
 hereditary leiomyomatosis renal cell cancer (HLRCC) 75, 249  
 hereditary non-polyposis colorectal cancer (HNPCC) 60, 258, 281, 312, 351, 380  
 – children 430, 432  
 – DNA repair 367  
 – endometrial cancer 207, 208, 210–213, 275, 282, 288, 312  
 – family cancer syndromes 43, 44, 109, 112  
 – gastric cancer 273, 288, 309–315, 318–320, 320  
 – NF1 127  
 – ovarian cancer 207–213, 239, 288  
 – pancreatic cancer 271, 274, 345  
 – predisposition 25, 27, 282, 283  
 – renal pelvis cancer 246, 251  
 – risk calculation 474, 475  
 – Turcot syndrome 118, 258  
 hereditary pancreatitis (HP) 345, 348, 349  
 hereditary papillary RCC (HPRCC) 245, 249  
 Hirschsprung disease (HSCR) 53, 67  
 HLA complex 402  
 Hodgkin lymphoma (HL) 57, 98, 385, 386, 394, 403  
 homologous recombination (HR) 381  
 hormone therapy 222  
 Horner Wright rosettes 149, 150  
 Hurthle cell carcinomas 170  
 hyperparathyroidism–jaw tumor syndrome (HPT–JT) 66, 239, 250, 253  
 hyperpigmentation 63, 70, 422  
 – GISTs 295–297  
 – PJS 257, 270  
 hyperplastic polyposis 258  
 hypoxia 15, 74  
 hysterectomy 211, 212, 288  
 hysteroscopy 213

## **i**

ifosfamide 185, 241  
 imatinib 295, 298, 389, 459

- GISTs 295–299
  - immunodeficiency 48, 382, 393, 394, 447, 448
  - immunohistochemical (IHC) analysis 283, 322
  - immunosuppression 183
  - immunotherapy 504
  - inductal papillary mucinous tumor (IPMT) 350
  - informed consent 469, 470, 471, 473, 478, 482
  - inhibitor of apoptosis proteins (IAPs) 507
  - insurance 478, 488
  - integrins 7, 8, 10, 12, 14–16
  - interferon 217, 423
  - interstrand crosslinks (ICLs) 381, 382
  - intravasation 3, 4, 8, 11, 13, 15
  - irinotecan 289
  - iron overload 356–358, 367, 400
  - irradiation 69
    - brain tumors 109, 112
    - breast cancer 193
    - DNA repair 377, 378, 382
    - Fanconi anemia 197
    - Gorlin syndrome 95
    - lung cancer 183
    - retinoblastoma 441
    - thyroid cancer 176
    - *see also* radiation therapy; ultraviolet radiation
  - isolated hemihyperplasia (IHH) 89
  - isoniazid 187
  - isotretinoin 425
- j**
- jaundice 362, 365, 366
  - jejuna tumors 52, 319
  - juvenile HFE 356
  - juvenile myelomonocytic leukemia (JMML) 93, 400, 402, 443
    - NF1 127, 131
  - Juvenile polyposis syndrome (JPS) 65, 272–274
    - gastric cancer 273, 309, 310
- k**
- keratoacanthomas 61, 284
  - kidney cancer *see* renal tumors
  - kinesin 155
  - Klinefelter syndrome 447
  - Kostmann syndrome 394, 395, 397, 402
- l**
- LAMB syndrome 47, 50
  - laminin 4, 7, 8
  - laparoscopy 175
  - laryngeal carcinoma 51
  - lasers 149
  - leiomyoblastomas 296
  - leiomyomas 249, 296
  - leiomyosarcomas 75, 296, 356
    - family cancer syndromes 43–45
  - LEOPARD syndrome (LS) 93
  - leucovorin 289
  - leukemias 16, 185, 386, 393, 502
    - AT and NBS 377, 382, 383, 385, 386
    - breast cancer 192
      - children 424, 441
      - family cancer syndromes 43–46, 48, 50, 52, 54, 56, 58, 60, 62, 64, 66, 68, 70, 72, 74, 76
      - gastric cancer 320, 324
      - LFS 397, 444
      - MAPK–RAS pathway 92
      - ovarian cancer 204
      - overgrowth syndromes 83, 88
      - RSTS 95
      - XP 415
    - Leydig cell tumors 75
    - Lhermitte–Duclos disease 64, 109, 259
    - Li-Fraumeni-like (LFL) syndrome 399
    - Li-Fraumeni syndrome (LFS) 76, 399, 460
      - family cancer syndromes 50, 74, 76
      - gastric cancer 304, 309, 316, 322–324
      - lung cancer 168, 332, 424
      - ovarian cancer 204
      - predisposition 25, 26, 32
      - risk calculation 474, 475
      - Wilms tumor 232
    - lipomas 110, 274, 301, 454
      - BRRS 259, 274
      - family cancer syndromes 45, 52, 62–64
      - overgrowth syndromes 87, 89
    - liver transplantation 362, 364, 379
    - liver tumors 88, 355, 374
      - children 424, 429, 441–452
      - metastases 169, 170, 216, 302, 304, 345, 353, 363
      - overgrowth syndrome 83, 87–89
    - lonarfarnib 138
    - loss-of-function mutation 140
    - loss-of-heterozygosity (LOH) 32, 33, 44, 365
      - family cancer syndromes 43–46, 48, 58, 64, 66, 68, 72, 74, 76
      - gastric cancer 309
      - leukemia 393
      - liver tumors 355, 356, 358
      - PDB 458
      - RCC 245
      - Wilms tumor 231–233

- lovastatin 138
- lung cancer 160, 166, 168, 183, 184, 210, 424, 449, 502
  - family cancer syndromes 50, 52, 54–56, 58, 60, 62
  - HNSCC 163
  - LFS 218, 373, 399
  - metastases 3, 17, 18, 157, 169, 170, 363
  - ovarian cancer 204
  - PJS 259, 270
  - retinoblastoma 147, 148, 184
  - XP 415
- lycopenes 222
- lymph nodes and vessels 4, 180
  - thyroid cancer 170
- lymphadenectomy 177, 211, 205, 310, 319
- lymphangiomas 45, 57
- lymphomas 91, 197, 378
  - children 424, 441
  - DNA repair 366, 377, 378, 380, 382–384
  - family cancer syndromes 45, 46, 48, 50, 52, 54, 56, 62, 64, 66, 74
  - LFS 399
- Lynch syndrome *see* hereditary non-polyposis colorectal cancer (HNPCC)
  
- m**
- Mafucci syndrome 457
- magnetic resonance imaging (MRI) 247, 357
  - brain tumors 109, 110, 112, 114, 116
  - NF1 127, 131, 132
  - NF2 133
  - RCC 245
  - schwannomatosis 127
  - XP 419
- magnetic resonance tomography (MRT) 202
- malignant peripheral nerve sheath tumors (MPNST) 461
  - NF1 113, 127–129, 131–134
- malignant renal rhabdoid tumor (MRR) 239, 242
- mammography 198–200
- mantel cell lymphomas(MCL) 388, 389
- Marfan's syndrome 189
- Marinesco–Sjögren syndrome 430
- mastectomy 200, 494
- medullary renal carcinoma (MRC) 251
- medullary thyroid carcinomas (MTC) 170
  - children 424, 441
  - family cancer syndromes 52, 66, 70
- medulloblastomas 109, 445
  - brain tumors 109, 114
  - children 424, 441
  - family cancer syndrome 48–50, 52, 54, 56, 58, 60, 62, 64, 68
  - Gorlin syndrome 95, 445
  - Turcot syndrome 258, 259, 262
- medulloepithelioma 149
- melanoma 153, 411–419, 427, 434, 471
  - breast cancer 197
  - Cowden syndrome 274
  - family cancer syndromes 46, 50–52, 54, 58, 62, 68, 70
  - GISTs 295
  - HNSCC 163
  - PC 345
  - retinoblastoma 147
  - XP 415, 419, 421
- melanoma–astrocytoma syndrome 52, 69, 70
- melanoma–pancreatic cancer syndrome (MPCS) 69, 346
- melanocortin 415
- melanotropin 415
- meningiomas 83, 100, 110, 138, 444, 460
  - children 441, 442
  - family cancer syndromes 45, 48, 50, 52, 54, 56, 58, 60, 64, 68, 70, 74
  - NF2 127, 129, 131, 133, 444
  - RSTS 97
- merlin 114, 132, 139
- mesenchymoma 49, 69
- mesoblastic nephromas 250
- mesothelioma 189
- metalloproteinases (MMPs) 8, 13
- metastases 3, 15, 17, 157, 169, 302
  - bone 170, 501
  - Carney triad 458, 459
  - gastric cancer 5, 11, 16, 18, 19, 309
  - GISTs 295–298
  - liver 175, 296, 304, 355, 356, 459
  - lungs 73, 170, 248
  - lymph nodes 177
  - omentum 296
  - PC 215, 216, 220
  - PHTS 274
  - RCC 240, 247, 250
  - retinoblastoma 147, 156
  - targeted therapy 501, 502
  - thyroid cancer 170, 176–181
- metastatic cascade 3–8, 13, 15, 17
- microsatellite instability (MSI) 17, 28, 60
  - FGC 310, 312, 313
  - HNPCC 281–284, 288, 312–313
- mismatch repair (MMR) 28, 281, 282, 379
  - family cancer syndromes 43, 57, 58, 60
  - FGC 324, 325

- HNPCC 251, 281–283, 320–325, 339
  - HNSCC 163
  - leukemia 394
  - NF1 128, 134, 135
  - ovarian and endometrial cancer 207–214
  - Turcot syndrome 119, 259
  - mitochondrial pathway 504, 505
  - mitogen activated protein kinase (MAPK)
    - 8, 92, 93, 280
  - RAS pathway 92–95
  - mitomycin 380, 375
  - mosaicism 88, 90, 152, 239
  - BWS 456
  - NF1 127, 128, 130, 461
  - NF2 127, 132–133
  - RCC 245
  - RTS 79
  - schwannomatosis 127
  - Wilms tumor 235
  - Muir–Torre syndrome (MTS) 53, 61, 80, 82
  - Mulibrey nanism (MUL) 239
  - multifocality 246, 302, 331
  - family cancer syndromes 43–46
  - GISTs 295–297
  - RCC 246, 248
  - multi-hit hypothesis 33
  - multiple endocrine neoplasia (MEN) 26,
    - 53, 65, 66, 137
  - children 441, 442
  - type 2A 170, 178–180
  - type 2B 178, 179, 443
  - multiple osteochondromas (MO) 454
  - MUTYH-associated polyposis (MAP) 258,
    - 269, 282
  - myelodysplastic syndrome (MDS) 93, 394
    - leukemia 385, 386, 388, 393, 394
  - myeloid leukemia 185, 239
  - myeloperoxidase (MPO) 187
  - myxomas 47, 67, 456
- n**
- NAME syndrome 47, 54
  - nasopharyngeal angiofibroma 454, 455
  - nasopharyngeal carcinoma (NPC) 166
  - nephrectomy 253
  - nephroblastomas 66, 76, 365
  - nephrogenic rests (NR) 232
  - nephrotic syndrome (NS) 231, 236, 259
  - neural tumors 70
  - neurinomas 110
  - neuroblastomas 92, 94, 444–446
    - overgrowth syndromes 83, 87–89
  - neuroendocrine tumors 323
  - neurofibromas 31, 33, 113, 114, 450
    - family cancer syndromes 48, 50, 54, 68
    - GISTs 295
    - NF1 127, 129–132
    - NF2 127
    - plexiform 135, 136
    - spinal 123, 130, 134
  - neurofibromatosis 54, 124–134, 295, 450,
    - 503
    - predisposition 26, 31, 32, 34
  - neurofibromatosis type 1 (NF1) 103, 123,
    - 127, 139–145, 444, 461
    - children 424, 441
    - family cancer syndromes 54
    - GISTs 295–297
    - leukemia 390, 393
    - MAPK–RAS pathway 92
    - predisposition 26, 34
    - Wilms tumor 232
  - neurofibromatosis type 2 (NF2) 124, 125,
    - 127, 130, 444
    - children 424, 441
    - family cancer syndromes 54
    - GISTs 295
    - predisposition 26
  - neurofibromatosis–Noonan syndrome 128,
    - 140
  - neurofibromin 113, 131, 132, 139, 295,
    - 462
  - neuromas 171
  - neurotrophin 156
  - neutropenia 394, 395, 397
  - nevroid basal cell carcinoma syndrome
    - (NBCCS) 68, 79, 84, 95, 103, 104
  - nibrin 383, 384
  - nicotine 183, 185
    - *see also* smoking
  - Nijmegen breakage syndromes (NBS) 55,
    - 394, 447
    - lymphoma 377, 378, 380, 382–390
  - non-Hodgkin lymphoma (NHL) 70, 98,
    - 385, 448
    - AT and NBS 383, 385, 386
  - non-homologous end joining (NHEJ) 381
  - non-medullary thyroid cancer (NMTC) 275
  - non-neoplastic genetic syndromes 445, 446
  - non-small lung cancer 166
  - non-steroidal anti-inflammatory drugs
    - (NSAIDs) 185
  - Noonan syndrome (NS) 92–94, 128, 400,
    - 407
  - Noonan-neurofibromatosis syndrome
    - (NFNS) 94
  - nucleotide excision repair (NER) 187, 379,
    - 390, 415, 421

- pathway 418, 421
- XP 415

**o**

- odontomas 261
- oligodendroglioma 97
- oligonucleotides 433, 508
- Ollier's disease 457
- oncocytomas 45, 70, 246, 250, 252, 254
- ophthalmology 434
- optic pathway tumors 136
- oral cancer 167
- oral contraceptives (OC) 214, 261
- osteoblastomas 455
- osteochondromas 454, 457
- osteogenic sarcomas 35
- osteomas 62, 259, 454, 455
  - FAP 257–259
  - Gardner syndrome 258, 262
- osteosarcomas 98, 458, 460
  - children 441–452
  - family cancer syndromes 50, 56, 58, 72, 76
- ovarectomy 199, 200, 498
- ovarian cancer 31, 46, 202–208, 213, 239, 282
  - breast cancer 190, 193, 201, 204, 206, 207
  - counseling 201, 463, 467, 469, 470
  - family cancer syndromes 44–46, 48–50, 52, 54, 58, 60, 62, 64
  - galactosemia 362
  - Gorlin syndrome 95
  - HNPCC 207, 208, 210, 251, 281–283
  - LFS 444
  - overgrowth syndromes 88, 89
  - pancreatic cancer 209, 210, 341, 342
  - PJS 259
  - psycho-oncological aspects 467, 487, 488, 490
- overgrowth syndromes 87–89, 227, 231, 445
- oxaliplatin 289

**p**

- paclitaxel 185, 211
- Paget's disease of bone (PDB) 458, 465
- pancreatectomy 351
- pancreatic adenocarcinomas 166, 211, 341
- pancreatic cancer (PC) 69, 166, 209, 210, 345–346, 403, 473
  - breast cancer 193
  - counseling 331, 453

- family cancer syndromes 43, 45, 48, 50, 52, 57, 62, 64, 66, 68, 72
- HNPCC 281, 339, 340
- JPS 259
- ovarian cancer 203, 204, 347, 352
- PJS 259, 336, 347
- VHL 245
- pancreatoblastoma 445, 446
- pancreatitis 348, 349, 351
- papillary renal cell carcinoma 231, 245–249, 251
- papillary thyroid carcinoma (PTC) 163, 169, 176, 365
- papillary thyroid carcinoma with associated renal neoplasia (FPTC-PRN) 250
- parafibromin 239
- paragangliomas (PGLs) 248, 302, 248
  - Carney triad 458, 459
  - GISTs 295–297
- parathyroid adenomas 45, 50, 52, 53, 64, 66, 70, 250
- parotid neoplasms 90
- parotid monomorphic adenomas 65
- parotid oncocytomas 45, 70
- pedigree documentation 469, 474
- penetrance 152, 193, 403, 404
  - breast cancer 184, 190, 193
  - enchondromatosis 454
  - endometrial cancer 207
  - family cancer syndromes 43, 62, 72, 74, 76
  - FAP 257, 445
  - gastric cancer 288, 309
  - GISTs 295–297
  - Gorlin syndrome 95
  - HFE 356, 400
  - HNPCC 281
  - leukemia 393
  - lung cancer 183
  - melanoma 411, 412–418
  - MRR 239
  - MTC 169
  - NMTC 176
  - ovarian cancer 202
  - pancreatic cancer 341
  - predisposition 27, 28, 30, 31, 35, 36
  - prostate cancer 210, 215, 216
  - RCC 245, 246
  - retinoblastoma 140, 147, 441
  - risk calculations 474
  - schwannomatosis 127
  - Wilms tumor 225, 231, 232
- penicillamine 359

- Perlman syndrome 238, 445
- Peutz–Jeghers syndrome (PJS) 55, 61–63, 257–259, 270–272
  - gastric cancer 273, 320, 323, 325
  - pancreatic cancer 271, 274, 341
  - pharmacogenomics 508
- pharyngeal cancer 164
- phenotype 8, 11, 30, 31, 88–90, 508
  - AAT deficiency 360, 361
  - AFAP 258
  - AML 390
  - breast cancer 204
  - BWS 456
  - Carney triad 458
  - DNA repair 367
  - family cancer syndromes 60–62, 73, 74
  - FAP 257–260, 275
  - gastric cancer 316, 317, 323, 325
  - GISTs 295–298
  - Gorlin syndrome 95
  - HNPCC 281
  - MAP 259
  - MAPK–RAS pathway 92, 93
  - melanoma 411, 412
  - MRR 239, 241
  - NF1 127, 129, 130
  - ovarian cancer 204
  - pancreatic cancer 341, 342, 345, 346
  - PJS 270, 347
  - predisposition 27, 29, 30, 32, 34
  - prostate cancer 215, 216, 218
  - RCC 245–248
  - Wilms tumor 231
  - XP 415, 419, 421
  - *see also* genotype/phenotype correlation
- pheochromocytomas 36, 87
  - Carney triad 458
  - family cancer syndromes 48, 52, 54, 58, 66, 72, 74
  - MTC 170–172, 174
  - NF1 124, 126, 131, 132, 449
  - RCC 245, 246
- phlebotomy 358
- pineoblastoma 149
- plasmin 8–10
- plasminogen activator inhibitors 8, 9–13
- platinum 187, 199, 211
- pneumothorax 45, 70
- Pollitt syndrome 430
- poly(ADP–ribose) polymerase (PARP)
  - inhibitor 212, 482
- polymorphisms 16, 164, 165, 183–185, 334, 366, 461, 478, 502
  - DNA repair 367
  - HNSCC 163
  - leukemia 382, 390
  - LFS 460
  - liver tumors 355, 357
  - lung cancer 183–189
  - melanoma 405, 406
  - MRR 234
  - pancreatic cancer 345
  - predisposition 30, 31
  - prostate cancer 211, 215
- polyostatic Paget disease 454
- polyposis syndromes 62, 309, 312, 313, 334
  - gastric cancer 304, 317, 320, 323, 324, 325
- predictive diagnostics 489–491, 497, 500
- predisposition or susceptibility 25–27, 43, 44, 59–76, 473–483
  - brain tumors 112, 113, 124, 132
  - breast cancer 26, 28, 30, 178, 187, 216
  - BS 441
  - children 346
  - counseling 457, 469
  - endometrial cancer 203
  - GISTs 290, 294
  - HNPCC 26, 28, 274, 276
  - HNSCC 158–160
  - leukemia 383, 389, 390, 392–394
  - lung cancer 177, 178, 180, 182, 183
  - melanoma 401, 402, 404, 406, 407
  - MRR 233
  - ovarian cancer 201, 204
  - pancreatic cancer 336
  - prostate cancer 210, 211–217
  - RCC 238, 243
  - STS 441
  - Wilms tumor 232, 233
- pregnancy 261, 363
- prevention 189, 463, 482
  - breast cancer 187, 192–196
  - counseling 458, 466, 468, 469
  - FAP 260
  - lung cancer 183
  - melanoma 407
  - ovarian and endometrial cancer 206, 207
  - prostate cancer 215, 216
- primary pigmented (micro) nodular adrenocortical disease (PPNAD) 67
- primitive neuroectodermal tumors (PNETs) 59, 69, 76
- Pringle Bourneville disease 117
- prostate cancer 16, 178, 209–217
  - breast cancer 192
  - family cancer syndromes 46, 50, 73

- gastric cancer 304, 310
- GISTs 292
- ovarian cancer 203, 204, 337
- prostate specific antigen (PSA) 216
- prostatectomy 215, 216, 222
- Proteus-like syndrome 64, 90, 274
- Proteus syndrome (PS) 63, 64, 88, 90, 274
- psychology and psychosocial concerns 473–483
  - counseling 458, 459, 465, 467–469
- psychosomatics 489, 491–493
- psychotherapy 487, 492, 493, 496, 497
- PTEN hamartoma tumor syndrome (PHTS) 92, 258, 259, 274
  - Cowden syndrome 111
  - gastric cancer 323, 325
- pulmonary chondroma 295
- purine 368
- pyrimidine 298

**q**

- quantitative multiplex polymerase chain reaction (QM-PCR) 155

**r**

- radiation therapy 110, 211, 216, 242, 296, 508
  - AT and NBS lymphoma 383, 385
  - brain tumors 109, 110, 112, 130, 136
  - endometrial cancer 207
  - FAP 260
  - GISTs 295
  - HNSCC 163
  - lung cancer 183
  - MRR 239
  - prostate cancer 210, 216
  - retinoblastoma 147, 150
- radiosurgery 116, 138
- radioiodine ablation 170
- recombination repair (RR) 381
- recruitment hypothesis 33
- rectal cancer 60, 211, 284
  - *see also* colorectal cancer
- recurrence of tumors 488
  - Carney triad 458
  - FAP 260
  - MTC 169
  - NMTC 176
  - RCC 245
  - retinoblastoma 147
  - Wilms tumor 231
- red cell aplasia 395
- red flags 28

- Reed syndrome 56, 75
- renal cancer and renal cell carcinoma (RCC) 16, 73, 245–251
  - breast cancer 193
  - children 424
  - Cowden syndrome 274
  - family cancer syndromes 43–45
  - oncocytomas 246
  - overgrowth syndromes 87
  - parenchymal tumors 251
  - prostate cancer 216
  - rhabdoid tumors 117
  - Wilms tumor 231, 443
- renal pelvis cancers 52, 246, 284
- respiratory tract tumors 46, 63
  - *see also* lung cancer
- retinal angiomas 73
- retinal hemomangioblastomas 246
- retinoblastoma 55, 69, 147, 413, 444
  - children 424
  - HNSCC 163
  - liver tumors 355
  - lung cancer 166, 183
  - predisposition 25, 26, 30, 32, 34, 35
- retinomas 150, 151
- reversion repair (RER) 380
- rhabdoid predisposition syndrome 56
- rhabdoid tumors 116, 231
- rhabdomyosarcoma 48, 51, 54, 55, 75, 89, 94, 365
  - BWS 364, 456
  - children 424, 441
  - FAP 445
  - MAPK–RAS pathway 92
  - MRR 239
  - NF1 444, 461
- rofecoxib 269
- Rothmund–Thomson syndrome (RTS) 71, 97, 460
- Rubinstein–Taybi syndrome (RSTS) 97

**s**

- Sabinas syndrome 430
- salpingo-oophorectomy 200, 201, 205, 211, 288
- sarcomas 75, 76, 441, 444, 453
  - brain tumors 109
  - family cancer syndromes 43, 44–48, 50, 54, 58, 68, 72, 74–78
  - gastric cancer 309, 318–324
  - GISTs 295–297
  - LFS 399, 444
  - overgrowth syndromes 87
  - XP 415

- schwannoma 114–116, 124, 126, 127, 129, 130–133, 135
  - family cancer syndromes 45–48, 50, 54, 56, 64, 66, 68
  - vestibular (VS) 124, 127, 129, 13, 137
  - XP 415
- schwannomatosis 116, 125, 126, 129, 132
- Schwachman–Diamond syndrome (SDS) 394
- screening 44, 62, 89–92
  - breast cancer 184, 190, 191, 194, 197, 198
  - colon cancer 346
  - counseling 453, 463, 467
  - FAP 347
  - FNMTC 169
  - gastric cancer 309–324
  - HFE 356
  - HNPCC 281–284
  - MTS 53
  - JPS 272
  - NPC 166
  - pancreatic cancer 335, 340
  - prostate cancer 210, 215, 216, 218
  - RCC 245–250
  - retinoblastoma 147
  - Wilson’s disease 358
  - XP 415
- sebaceous tumors 61
- Seckel syndrome 380
- selenium 222
- serine 8, 417
- serpins 10
- Sertoli cell tumors 70, 286
- severe combined immunodeficiency (SCID) 473, 474
- severe congenital neutropenia (SCN) 417, 418, 420, 429
- sex cord tumors with annular tubules (SCTAT) 65, 285
- sickle cell trait 265
- sigmoidoscopy 280, 305
- Simpson–Golabi–Behmel syndrome (SGBS) 94, 251, 445
- single nucleotide polymorphism (SNP) 194, 232, 426, 433
  - lung cancer 173, 175, 192
- Sipple syndrome 55
- skin cancer 439, 444, 446–450, 452, 453
  - Cockayne syndrome 401, 404
  - endometrial cancer 218
  - family cancer syndromes 45, 46, 52, 54, 56, 58, 60, 62, 66, 68, 70, 72
  - RTS 100
  - TTD 421
  - XP 415, 419, 421
- small bowel cancer 61, 282, 320
  - GISTs 295–300
  - HNPCC 213, 251, 258, 262, 281, 309
- small intestine cancer 50, 63, 65
- smoking 163–165, 334, 348, 354, 503
  - lung cancer 173, 175, 192
- soft tissue sarcoma (STS) 44, 114, 153, 210, 441, 443, 444
  - children 407, 424, 441
  - family cancer syndromes 45, 46, 52, 54, 56, 58, 60, 62, 66, 68, 70, 72
  - LFS 399, 444, 460
  - neurofibromatosis 109, 402, 443
- sonography 201, 213
  - *see also* ultrasound
- Sotos syndrome (SS) 87, 90, 445
- spinal neurofibromatosis (SNF) 128, 143
- spinal tumors 130, 441
- spindle cell sarcomas 457
- squamous cell carcinoma 21, 52, 57, 59
  - head and neck 161–166
  - RTS 100
  - XP 415, 419, 421
- statins 138
- stomach cancer 341
  - breast cancer 170
  - endometrial cancer 207, 208
  - GISTs 295, 296
  - JPS 259
  - ovarian cancer 363
- subependymal giant cell astrocytoma (SEGA) 117, 119, 446
- succinate dehydrogenase (SDHB) 248, 295, 303
- suicide gene therapy 504
- sulindac 268, 269
- sun exposure 418, 422
  - *see also* ultraviolet radiation
- survivin 507
- surgery 11, 282, 464
  - brain tumors 109, 112
  - breast cancer 184, 190, 193, 482, 483
  - Carney triad 458
  - counseling 453, 463, 467
  - endometrial cancer 207, 208
  - FAP 257, 258, 458, 466
  - FNMTC 169
  - gastric cancer 11, 288, 309, 310, 315, 322
  - GISTs 295–297
  - HNPCC 262, 281
  - HNSCC 163
  - JPS 272

- MTC 169–171
- neurofibromatosis 127, 128, 130
- ovarian cancer 189, 435
- pancreatic cancer 341, 342
- PJS 248, 249
- prostate cancer 199, 210
- RCC 245
- retinoblastoma 140
- Wilms tumor 231
- sympathetic nervous system tumors 441

**t**

- talin 7, 114
- tamoxifen 198–200, 268
- Tay syndrome 430
- T-cells 39, 383
  - lymphoblastic lymphoma (T-LBL) 385
  - lymphoma 240, 377, 378, 380
  - prolymphocytic leukemia (T-PLL) 410
  - receptors 399, 313, 371
- telangiectasias 98, 422
  - *see also* ataxia telangiectasia (AT)
- telomeres 401, 417, 418, 421, 428
- testicular tumors 89, 90, 268
  - family cancer syndromes 50, 52, 56, 58, 64, 66, 68, 70
- threonine 8, 429
- thrombocytopenia 401
- thyroid cancer 91, 170–177, 275, 460, 462
  - Cowden syndrome 259, 274, 280
  - family cancer syndromes 45, 48–51, 53, 55, 58, 59, 62–64, 66–68, 70, 71
  - FAP 269, 470
  - neurofibromatosis 138
  - non-medullary 176, 178, 274, 275
  - Wilms tumor 469
  - *see also* medullary thyroid cancer
- thyroid tumors with cell oxyphilia (TCO1) 176
- thyroidectomy 175–177
- thymoma 190
- thymine 380
- tipifarnib 140
- toremifene 268
- TRAIL receptors 500–502, 503
- transferrin receptor (TFR) 356, 357
- transitional cell cancers 94, 250–255
- trichilemmoma 111
- trichodiscomas 45, 70
- trichothiodystrophy (TTD) 380, 382, 436
  - DNA repair 432, 436, 437, 439, 440, 442
  - XP 440, 441
- triethylene tetramine (TETA) 359
- trisomy 7 247

- trisomy 8 399
- trisomy 13 239
- trisomy 18 239, 460
- trisomy 21 393, 398, 399, 460
- Triton tumors 475
- trypsin 348, 359
- tuberin 117, 134, 247
- tuberous sclerosis (TS) 33, 119, 120, 134, 247, 459, 460
- tuberous sclerosis complex (TSC) 58, 72–74, 247, 251
- tumor necrosis factor (TNF) 510, 511
- Turcot syndrome 59, 63, 119, 120, 257, 258, 260, 261
  - children 458
  - family cancer syndromes 59, 62, 63, 71
  - predisposition 34
- Turner syndrome 460
- twin studies 184
- “two-hit” hypothesis 32, 33, 43, 89, 317, 388, 400, 442
  - NF1 131, 133
  - predisposition 25, 31–34
  - Wilms tumor 231
- tyrosine 362, 363, 366, 399
- tyrosine kinase (TK) 294, 300, 511–513
- tyrosinemia 362, 363, 459, 460

**u**

- ultrasound 94, 175, 176, 201, 285, 319, 350
  - BWS 364
  - overgrowth syndromes 88–91
  - RCC 244, 245, 248, 250
  - transvaginal (TVU) 213, 285
- ultraviolet radiation (UV) 414–416, 431–433
  - XP 424, 425, 431
- uniparental disomy (UPD) 236, 398–400, 455
  - overgrowth syndromes 88, 89
- u-PA system 6, 8, 9–11, 13, 14
- u-PAR system 6, 8–10, 11–13, 15
- ureter cancer 64, 249, 281, 286
- urinary cancer 44, 249, 280, 286
- urine 244, 245, 250, 358, 359
- urogenital tumors 46, 62, 286
- urothelial cancer 207, 208, 249, 250
- uterine cancer 56, 63, 199, 484

**v**

- vascular tumors 35
- vincristine 241

vinyl chloride 355  
vitronectin 4, 7, 8, 10  
von Hippel Lindau (VHL) syndrome  
120–123, 138, 244  
– family cancer syndromes 59, 74–75  
– predisposition 26, 34  
– RCC 244, 245, 247, 249–251  
von Recklinghausen disease 55, 297

**W**

Wagenmann–Froboese syndrome 54  
Watson syndrome 129  
Weaver syndrome (WS) 90, 91  
Werner syndrome (WS) 54, 60, 71, 72,  
379, 460  
Wilms tumor 16, 66, 231–241, 247, 361,  
395, 457  
– BWS 459–461  
– children 459, 457, 459–461  
– family cancer syndromes 46, 50, 51, 66,  
74, 76  
– overgrowth syndromes 88–91  
– predisposition 26, 32

Wilms tumor–aniridia–urogenital  
abnormalities–mental retardation  
(WAGR) 231, 232, 235, 236  
– children 457  
Wilson's disease 359, 360  
Wiskott–Aldrich syndrome 459

**X**

xanthoma 364  
xeroderma pigmentosum (XP)  
60, 187, 430, 431, 437–443  
– Cockayne syndrome 443–446  
– DNA repair 377, 378, 437–443  
– NER pathway 446–448  
– plus neurological abnormalities 441–443  
– predisposition 26, 30, 35  
– TTD complex 445, 446  
X-ray cross complementing group 1  
(XRCC1) 187, 188

**Z**

zinc 360  
Zollinger–Ellison syndrome 66