

INDEX

Note: Page numbers of article titles are in **boldface** type.

- Acquired immunodeficiency syndrome.
See *Human immunodeficiency virus infection*.
- Activated protein C and/or A, resistance to, **169-186**
etiology of, 171-173
factor V gene mutation in, 173-183
biochemistry of, 173-175
laboratory evaluation of, 176-183
multifactorial, 176
thrombosis risk in, 175-176
pathogenesis of, 170-171
- Adenosine deaminase deficiency, gene therapy for, 200-201
- Adrenal hyperplasia, congenital. See *Congenital adrenal hyperplasia*.
- AIDS. See *Human immunodeficiency virus infection*.
- Alzheimer's disease, apolipoprotein E polymorphism in, 111-112
- Amplicor Chlamydia test, evaluation of, 52-59
- Amplified fragment length polymorphism, in forensic testing, 190
- Androgens, excessive production of, in congenital adrenal hyperplasia, 125-126
- Angiocentric immunoproliferative lesion, DNA analysis in, 2
- Antibodies, intracellular, in gene therapy, 204
- Anticholesterol agents, blood lipid levels and, in apolipoprotein E polymorphism, 113
- Antisense oligonucleotides, in gene therapy, 203
- Antisense therapy, for neoplasia, 207-208
- Apolipoprotein E, polymorphism of, **105-123**
cholesterol reduction therapy and, 112-113
genotype analysis in, 114, 116
genotype frequencies in, 116-117
in Alzheimer's disease, 111-112
in atherosclerosis, 110-111
in cerebrovascular disease, 112
in coronary artery disease, 110-111
in hyperlipoproteinemia, 106, 108-110
in peripheral vascular disease, 112
lipoprotein receptor binding and, 106-107
phenotype analysis in, 113-114
posttranslational modification in, 106
- Apoptosis, bcl-2 gene rearrangements and, 30
- Aspergillosis, in immunodeficiency, 78
- Ataxia-telangiectasia, breast cancer in, 148
- Atherosclerosis, apolipoprotein E polymorphism in, 110-111
gene therapy for, 201-202
- ATM gene, DNA analysis of, direct, 154
mutations of, in ataxia-telangiectasia, 148
- Autolymphocyte therapy, in gene therapy, for neoplasia, 206
- Automation, of polymerase chain reaction, 217-219
- B lymphocytes, gene rearrangement in, in hematopoietic disorders, 1-21
- B-cell lymphoproliferative disorders, in transplantation, 13-16
- bcl-2 gene, **23-47**
IgH locus of, 27-28
normal, 26
protein of, function of, 29-31
rearrangements of, in lymphoid malignancies, 23-24
detection of, 31-39
diffuse lymphoma, 40
follicular lymphoma, 39-40
fusion with 2J_H, 28-29
Hodgkin's disease, 41
molecular events in, 25-26

- bcl-2 gene (*Continued*)
 prognostic value of, 42-43
 protein function in, 29-31
 small noncleaved cell lymphoma, 40-41
 tissue localization of, 42
- BRCA1 gene, 139-141
 DNA analysis of, direct, 153-156, 159-160
 functional detection of mutations of, 158-159
 informed consent for, 162-163
 linkage analysis in, 152-153
 mutation screening methods for, 155-158
 tests for, 149, 151-152
 identification of, 142-143, 145-146
 in ataxia-telangiectasia, 148
 in Muir-Torre syndrome, 147
 penetrance of, 145
 protein of, characteristics of, 146
- BRCA2 gene, 146-147
 DNA analysis of, informed consent for, 162-163
- Breast cancer, hereditary, 139-167
 BRCA1 gene in. See BRCA1 gene.
 BRCA2 gene in, 146-147, 162-163
 clinical features of, 143-144
 epidemiology of, 144-145
 genotyping of, 140-141
 in males, 146-147
 molecular diagnosis of, direct DNA
 detection in, 153-155, 159-160
 functional detection of mutations in, 158-159
 future directions in, 161
 historical perspective of, 148-151
 informed consent in, 162-163
 linkage analysis in, 152-153
 mutation screening methods in, 155-158
 practical considerations in, 161-162
 tests for, 148-151
 phenotyping of, 140-141
 risk assessment in, 144-145
 syndromes associated with, 143-144, 147-148
 TP53 gene in. See TP53 gene.
 tumor suppressor gene inactivation in, 139
 tumors associated with, 143-144
- Burns, fungal infections in, 79
- Cancer, gene therapy for, 205-208
 Candidemia, clinical features of, 74-75
 Candidiasis, in immunodeficiency, clinical features of, 74-77
 DNA analysis of, 77-78, 80-82, 85
 nosocomial, epidemiology of, 82-85
 typing of, 80-85
- Candiduria, in immunodeficiency, 76-77
- Carbonic anhydrase II deficiency, genetic studies of, 213-214
- Cerebrovascular disease, apolipoprotein E polymorphism in, 112
- Chemical mismatch cleavage analysis, in breast cancer, 157-158
- Chemoprotection, in chemotherapy, multidrug-resistant gene modification in, 207
- Chlamydia trachomatis*, detection of, Amplicor product in, evaluation of, 52-59
 multiplex polymerase chain reaction in, 64-68
- Cholesterol, blood profiles of, in apolipoprotein E polymorphism, 107-110
 atherosclerosis and, 110-111
 therapy effects on, 112-113
 drugs lowering, apolipoprotein E polymorphism and, 113
- Chromogenix Coatest, in activated protein C resistance detection, 183
- Coagulation disorders, activated protein C and A resistance. See *Activated protein C and/or A, resistance to*.
- Congenital adrenal hyperplasia, 125-137
 clinical features of, 125-126
 diagnosis of, 126, 131-132
 gene involved in, 126-128
 genotype-phenotype relationships in, 132-135
 molecular genetics of, 128-131
- Conjunctiva, lymphoma of, 7-11
- Contraceptives, oral, thrombosis risk in, factor V gene mutation and, 176
- Contract negotiation, for pathology practice networking, 238-239
- Coronary artery disease, apolipoprotein E polymorphism in, 110-111
 gene therapy for, 201-202
- Cortisol, replacement of, in congenital adrenal hyperplasia, 126
- Cost effectiveness, of pathology practice networking, 237
- Cowden's disease, breast cancer in, 147-148
- Credentialing, standard, for pathology practice networking, 236
- CYP21 genes, in 21-hydroxylase deficiency, 127-131
- Cystic fibrosis, gene therapy for, laboratory implications of, 209
- Cytotoxic gene therapy, for neoplasia, 206-207
- Denaturing gradient gel electrophoresis, in breast cancer, 155, 157

- Dexamethasone, in congenital adrenal hyperplasia, 126
- Diabetes mellitus, gene therapy for, 201
- Diet, blood lipid levels and, in
apolipoprotein E polymorphism, 113
- Direct automatic sequencing, in breast cancer gene analysis, 154-155
- DNA, storage of, on Guthrie cards, 216
- DNA technology, apolipoprotein E polymorphism, 105-123
detection methods in, 218
future developments in, 213-222
guidelines for, 215-217
in activated protein C and A resistance, 169-186
in congenital adrenal hyperplasia, 125-137
in forensic testing, 187-196
in fungal infections, in immunodeficiency, 73-88
in gene therapy, 197-211
in hematopoietic disorders, B and T cell gene rearrangement, 1-21
in hereditary breast cancer, 139-167
in HIV infection, nucleic acid sequence-based amplification, 89-103
in 21-hydroxylase deficiency, 125-137
in lymphoid malignancies, bcl-2 gene rearrangements, 23-47
in sexually transmitted diseases, multiplex polymerase chain reaction, 61-71
proportion of total test volume, 214-215
role of, 49-60
enthusiasm for, 50-51
example of, Chlamydia Amplicor evaluation as, 52-59
skepticism about, 51-52
- Dot blot test, in breast cancer gene mutation detection, 160
in forensic testing, 189-190
- Electrochemiluminescence method, in nucleic acid sequence-based amplification, 94, 96-97
- Electronic teleconferencing, for pathology practice networking, 235-236
- Electrophoresis, contour-clamped homogenous gel, in fungal typing, 81-82
denaturing gradient gel, in breast cancer, 155, 157
- Enzyme-linked gel assay, in nucleic acid sequence-based amplification system, 94-95
- Esophagus, candidiasis of, in immunodeficiency, 75
- Extracellular transplantation, gene therapy in, 208
- Factor V gene mutation, in activated protein C and/or A resistance, 173-183
biochemistry of, 173-175
laboratory evaluation of, 176-183
multifactorial, 176
thrombosis risk in, 175-176
- Familial combined hyperlipidemia, apolipoprotein E polymorphism in, 110
- Fibroblasts, autologous, in gene therapy, for neoplasia, 206
- Financial system, standardized, for pathology practice networking, 237-238
- Forensic testing, DNA technology in, 187-196
history of, 187-188
legal aspects of, 192-193
methods for, 188-191
quality control in, 191-192
regulatory aspects of, 191-192
- Fungal infections, in immunodeficiency, DNA analysis of, 73-88
aspergillosis, 78
candidiasis, 74-78, 80-85
in burns, 79
in transplantation, 79
laboratory aspects of, 80-82
zygomycosis, 78-79
- FV R506Q gene. See *Factor V gene mutation*.
- Gene(s), rearrangement of, bcl-2, in lymphoid malignancies, 23-47
in hematopoietic disorders. See *Hematopoietic disorders*.
- Gene therapy, 197-211
antisense, 207-208
clinical applications of, 198-199
cytotoxic, 206-207
extracellular transplantation and, 208
in infectious disease, 202-205
in monogenic disorders, 199-201
in multigenic disorders, 201-202
in neoplastic disease, 205-208
laboratory impact of, 209-210
metabolic suicide, 216
monitoring of, 209
tissue engineering and, 208
tumor suppressor, 207
- Genetic suppressor elements, in gene therapy, 204
- Genotypes, of apolipoprotein E, analysis of, 114, 116
frequencies of, 116-117
of 21-hydroxylase deficiency gene, analysis of, 132-135

- Guthrie cards, in DNA storage, 216
- Hematopoietic disorders, B and T cell gene rearrangement in, 1-21
 angiocentric immunoproliferative lesion, 2-4
 large granular lymphocytosis, 16-19
 lymphoma, of mucosa-associated lymphoid tissue, 5-7
 of skin, 11-13
 small cell cleaved, 7-11
 post-transplant B-cell lymphoproliferative disorder, 13-16
- Hemostasis, protein C in, 170-171
- Hereditary breast cancer. *See Breast cancer, hereditary.*
- Heteroduplex analysis, in breast cancer, 157
- HIV infection. *See Human immunodeficiency virus infection.*
- Hodgkin's disease, bcl-2 gene rearrangements in, 41
 in HIV infection, 4
- Human immunodeficiency virus infection, angiocentric immunoproliferative lesion in, 2
 diagnosis of, nucleic acid sequence-based amplification in, 94-101
 in qualitative detection, 94-95
 in quantitative detection, 95-98
 performance of, 98-100
 single-tube, 95-98
 esophageal candidiasis in, 75
 Hodgkin's disease in, 4
- Human papillomavirus, detection of, multiplex polymerase chain reaction in, 64, 68-69
- 21-Hydroxylase deficiency, 125-137
 clinical features of, 125-126
 diagnosis of, 126, 131-132
 gene involved in, 126-128
 genotype-phenotype relationships in, 132-135
 molecular genetics of, 128-131
 treatment of, 126
- Hypercholesterolemia, apolipoprotein E polymorphism in, 109-110
- Hyperlipoproteinemia, apolipoprotein E polymorphism in, 106, 108-110
- Immunization, intracellular, in gene therapy, 202-205
- Immunodeficiency, fungal infections in. *See Fungal infections.*
 severe combined, gene therapy for, 200-201
- Immunoglobulin G, heavy chain of, in bcl-2 gene rearrangement, 27-28
- Immunohistochemical methods, in bcl-2 gene rearrangement detection, 38
- Immunotherapy, for neoplasia, in vitro, 205-206
 in vivo, 206
- In situ hybridization, in bcl-2 gene rearrangement detection, 38-39
- Infections, gene therapy for, 202-205
- Inflammatory pseudotumors, of orbit, vs. lymphoma, 9
- Information systems, for pathology practice networking, 235
- Isoelectrofocusing, in apolipoprotein E polymorphism phenotype analysis, 113-114
- J_H gene, fusion with bcl-2 gene, 28-29
- Laboratory management, networking in, 227-241
 legal aspects of, 239-240
 model for, 233-239
 practice size and, 230-233
 sociopolitical background for, 227-230
 of hospital-based practices, current health care trends and, 225-226
- Large granular lymphocytosis, DNA analysis of, 16-19
- Legal aspects, of DNA technology, in forensic testing, 192-193
 of networking in laboratory management, 239-240
- Leukemia, bcl-2 gene rearrangements in, 24
- Li-Fraumeni syndrome, breast cancer in, 147
- Ligase chain reaction, 90
 in sexually transmitted disease diagnosis, 62-63
- Linkage analysis, in breast cancer detection, 152-153
- Lipid disorders, apolipoprotein E polymorphism and. *See Apolipoprotein E, polymorphism of.*
- Lipoprotein receptor binding, apolipoprotein E polymorphism and, 106-107
- Liver, candidiasis of, in immunodeficiency, 75
- Lymphocytes, modified with tumor necrosis factor, in gene therapy, for neoplasia, 206
- Lymphocytosis, large granular, DNA analysis of, 16-19

- Lymphoid hyperplasia, vs. lymphoma, 9-10
- Lymphoid malignancies. See also *Lymphoma*.
- bcl-2* gene rearrangements in. See *bcl-2* gene, rearrangements of.
- Lymphoma, *bcl-2* gene rearrangements in, 23-24
- diffuse, 40
- follicular, 39-40
- prognostic value of, 42-43
- small noncleaved cell, 40-41
- DNA analysis in, of mucosa-associated lymphoid tissue, 5-7
- of skin, 11-13
- small cell cleaved, 7-11
- Lymphoproliferative disorders, B-cell, in transplantation, 13-16
- Lysosomal storage diseases, gene therapy for, 200-201
- Managed care, pathology services in, networking in. See *Laboratory management, networking in*.
- Marketing, for pathology practice networking, 238
- Mitochondrial DNA sequencing, in forensic testing, 190-191
- Molecular pathology. See *DNA technology*.
- Mucopolysaccharidosis, gene therapy for, 200-201
- Mucormycosis, in immunodeficiency, 78-79
- clinical features of, 78-79
- Mucosa-associated lymphoid tissue, lymphoma of, DNA analysis in, 5-7
- Muir-Torre syndrome, breast cancer in, 147
- Multidrug-resistant gene, modification of, in chemoprotection, 207
- Muscular dystrophy, gene therapy for, 200-201
- Mycosis fungoides, DNA analysis of, 11-13
- Neisseria gonorrhoeae*, detection of, multiplex polymerase chain reaction in, 64-68
- Neoplasia, gene therapy for, 205-208
- Networking, in pathology services. See *Laboratory management, networking in*.
- Nomenclature, standardized, for pathology practice networking, 237
- Nosocomial infections, candidiasis, epidemiology of, 82-85
- Nucleic acid sequence-based amplification, 89-103
- amplification in, 92, 94
- in HIV infection, 94-101
- in qualitative detection, 94-95
- in quantitative detection, 95-98
- performance of, 98-100
- single-tube, 95-98
- in sexually transmitted disease diagnosis, 62-63
- methods for, 90-91
- nucleic acid isolation in, 91-92
- product detection in, 94
- Oligonucleotides, antisense, in gene therapy, 203
- Oral cavity, candidiasis of, in immunodeficiency, 75
- Oral contraceptives, thrombosis risk in, factor V gene mutation and, 176
- Orbit, lymphoma of, 7-11
- Osteopetrosis, in carbonic anhydrase II deficiency, genetic studies of, 213-214
- Ovarian cancer, hereditary breast cancer and, 140, 143, 145-147
- Papillomavirus, detection of, multiplex polymerase chain reaction in, 64, 68-69
- Peripheral vascular disease, apolipoprotein E polymorphism in, 112
- Peritonitis, candidal, in immunodeficiency, 76
- Polymerase chain reaction, 90
- automation of, 217-219
- costs of, 52
- enthusiasm for, 50-51
- in apolipoprotein E polymorphism genotype analysis, 114, 116
- in *bcl-2* gene rearrangement detection, 35-39
- in breast cancer gene detection, 149-150
- in candidiasis diagnosis, 77-78
- in *Chlamydia trachomatis* detection, evaluation of, 52-59
- in factor V gene mutation detection, 178-180
- in forensic testing, 188-189
- in 21-hydroxylase deficiency diagnosis, 128-129, 131-132
- miniaturization of, 217
- multiplex, in sexually transmitted disease diagnosis, 61-71
- Chlamydia trachomatis*, 64-68
- commercial tests for, 62-63
- human papillomavirus, 64, 68-69
- Neisseria gonorrhoeae*, 64-68
- principles of, 63

- Polymerase chain reaction (*Continued*)
Ureaplasma urealyticum, 69
 sequence-specific, in factor V gene mutation detection, 180-183
 skepticism about, 51-52
- Primer-mediated restriction endonuclease site generation, in factor V gene mutation detection, 180
- Protein A, activated, resistance to. See *Activated protein C and/or A*.
- Protein C, activated, resistance to. See *Activated protein C and/or A*.
- Protein S, in hemostasis, 171
- Protein truncation testing, in breast cancer, 159
- Pseudotumors, inflammatory, of orbit, vs. lymphoma, 9
- Q-beta replicase assay, 90
 in sexually transmitted disease diagnosis, 62-63
- Quality assurance, in forensic testing, 191-192
 standardized, for pathology practice networking, 236-237
- Regulatory aspects, of DNA technology, in forensic testing, 191-192
- Renal tubular acidosis, in carbonic anhydrase II deficiency, genetic studies of, 213-214
- Reporting procedures, standardized, for pathology practice networking, 237
- Restriction endonuclease fingerprinting, in breast cancer, 157
- Restriction endonuclease site generation, primer-mediated, in factor V gene mutation detection, 180
- Restriction enzyme analysis, in fungal typing, 81-82
- Restriction fragment length polymorphism, in apolipoprotein E polymorphism genotype analysis, 116
 in factor V gene mutation detection, 178-180
 in forensic testing, 188
 in fungal typing, 82
- Ribozymes, in gene therapy, 203
- RNA, as decoy, in gene therapy, 204
- Salt wasting, in congenital adrenal hyperplasia, 134
- Schizophrenia, gene therapy for, 202
- Sequence-specific polymerase chain reaction, in factor V gene mutation detection, 180-183
- Severe combined immunodeficiency, gene therapy for, 200-201
- Sexually transmitted diseases, diagnosis of, multiplex polymerase chain reaction in, 61-71
Chlamydia trachomatis, 64-68
 commercial tests for, 62-63
 human papillomavirus, 64, 68-69
Neisseria gonorrhoeae, 64-68
 principles of, 63
Ureaplasma urealyticum, 69
- Single-stranded conformation polymorphism analysis, in breast cancer, 155
- Skin, lymphoma of, DNA analysis of, 11-13
- Slot blot test, in breast cancer gene mutation detection, 160
- Sly disease, gene therapy for, 200-201
- Southern blot analysis, in bcl-2 gene rearrangement detection, 31-37
 in breast cancer, 158
- Stroke, apolipoprotein E polymorphism in, 112
- T lymphocytes, gene rearrangement in, hematopoietic disorders. See *Hematopoietic disorders*.
- Telangiectasia-ataxia, breast cancer in, 148
- Teleconferencing, for pathology practice networking, 235-236
- Thrombophilia, in activated protein C and A resistance, 171-173
- Thrombosis, multifactorial, gene mutations in, 176
- Tissue engineering, gene therapy in, 208
TP53 gene, DNA analysis of, direct, 153-156
 functional detection of mutations of, 158-159
 tests for, 149, 151-152
 mutations of, in ataxia-telangiectasia, 148
 in hereditary breast cancer, 142, 147-148
- Transplantation, B-cell lymphoproliferative disorder in, 13-16
 extracellular, gene therapy in, 208
 fungal infections in, 79
- Triglycerides, blood levels of, in apolipoprotein E polymorphism, 107
- Tumor necrosis, in lymphocyte modification, in gene therapy, for neoplasia, 206
- Tumor suppressor genes, in gene therapy, for neoplasia, 207
 inactivation of, in breast cancer, 140

Ureaplasma urealyticum, detection of,
multiplex polymerase chain reaction
in, 69

Utilization review, standardized, for
pathology practice networking, 236

Vaccination, intracellular, in gene therapy,
202-205

Vascular disease, apolipoprotein E
polymorphism in, 112

Virilization, in congenital adrenal
hyperplasia, 126, 134

Wound infections, candidal, in
immunodeficiency, 77

Yeast functional assay, in breast cancer,
159

Zygomycosis, in immunodeficiency, 78-79
clinical features of, 78-79