article index

Subject Index

Aberrant splicing, spectrum and frequency of mutations of exon 10b of *NF1* gene, 248

Abstracts, 1999 Annual Clinical Genetics Meeting, 11 (Jan/Feb)

ACMG: see American College of Medical Genetics

African Americans

phenotypic differences in, 49 with Prader-Willi syndrome, 49

Aganglionosis, colonic, laryngeal anomalies, heart defects, and preaxial polydactyly, description of unique syndrome, 104

Allele frequencies, differences in interleukin-4 receptor polymorphisms, ethnic group comparison, 267

Alpha-fetoprotein, prenatal triple marker screening program and, costs and effectiveness of, 199

Alzheimer disease, HMO and academic genetic center involvement in testing for, 272

American Board of Medical Genetics, 1999 diplomates, 307 American College of Medical Genetics

1999 Annual Člinical Genetics Meeting, abstracts and program, 11 (Jan/Feb)

College News, 61, 119, 173, 238, 345

continuing education program (editorial), 127

CPT coding revisions, 112

cytogenetics guidelines improvements (letter), 67

development of journal (editorial), 2

folic acid fortification/supplementation, policy statement, 66

mosaicism detection in amniotic fluid and, 94 Policy Statement

gene patents and gene testing accessibility, 237 re-contact duties and responsibilities, 171

presidential address, 56

Amniocentesis, prenatal triple marker screening program and, costs and effectiveness of, 199

Amniotic fluid, mosaicism detection in, 94

Angelman syndrome

gene imprinting analysis and, 141

maternal transmission of, 262

Apache Indians, genetic disorders among, 90

Ashkenazism, negative *BRCA1* mutation test results, psychological impact, 74

Athabaskan severe combined immunodeficiency, description and background information, 90

Atresia, intestinal, chorionic villus sampling and, literature review, 315

Autism, hyper-IgE syndrome, mental retardation, and, analphoid marker chromosome in, 213

Beckwith-Wiedemann syndrome, gene imprinting analysis and, 141

Binomial distribution, mosaicism detection in amniotic fluid, 94

Birth defects, genetics counseling and screening inconsistencies and, 286

Bowel disease, inflammatory, chromosome 12 and, linkage evidence, 194

Brain, structural anomalies in, cognitive function in neurofibromatosis type 1 and, 136

Brainstem dysgenesis, Athabaskan, description and background information, 90

BRCA1 testing, psychological impact of negative results (editorial), 69

Breast cancer

BRCA1 testing for susceptibility, psychological impact of negative results (editorial), 69

HER-2/neu gene amplification in, FISH detection, 98 HMO and academic genetic center involvement in testing for, 272

negative BRCA1 test results, psychological impact of, 74

Cancer: see also specific type

pediatric, gene imprinting analysis and, 141

Carnitine

primary deficiency: *see* Primary carnitine deficiency urinary transport of, primary carnitine deficiency detection and, 34

Cell growth/differentiation, neurofibromatoses treatment approaches and (CME/Invited Review), 158

Chorionic villus sampling, malformations in children exposed to, literature review, 315

Chromosomal material, aberrant, comparative genomic hybridization and, 4

Chromosome

analphoid marker, in hyper-IgE syndrome, autism, and mental retardation, 213

marker

comparative genomic hybridization and, 4 multicolor FISH utility in, 181

Chromosome 12, Crohn's disease and, linkage evidence, 194 Chromosome mapping, Palestinian family with monilethrix and, 109

Chromosome 15q11-q13

abnormalities, imprinted gene analysis in, 141 maternal transmission of Angelman syndrome and, 262

Cigarette smoking, maternal, trisomy 21 risk factors and, 80 CME exam

Antenatal treatment for classic 21-hydroxylase forms of congenital adrenal hyperplasia and the issues, 231 Treatment of neurofibromatosis, 165

CME/Invited Reviews

Antenatal treatment for classic 21-hydroxylase forms of congenital adrenal hyperplasia and the issues, 224 Emerging approaches toward the treatment of neurofibromatosis, 158

A new age in the genetics of deafness, 295

Cobalamin, methylmalonic acidemia and homocystinemia and, long-term outcome, 146

Cognitive problems, neurofibromatosis type 1 and, neuroimaging studies, 136

Colitis, ulcerative: see Ulcerative colitis

Colon cancer, HMO and academic genetic center involvement in testing for, 272

Colorectal cancer, Navajo Indians and (letter), 304

Computed tomography, structural anomalies in neurofibromatosis type 1 and, 136

Congenital adrenal hyperplasia, antenatal treatment for (CME/Invited Review), 224

Congenital anomalies

methylmalonic acidemia and homocystinemia and, longterm outcome, 146

unique constellation of heart defects/laryngeal anomalies/ preaxial polydactyly/colonic aganglionosis, 104

Contraceptives, oral, trisomy 21 risk factors and, 80

Costs/benefits, prenatal triple marker screening program,

CPT coding revisions, 112

Crohn's disease, chromosome 12 and, linkage evidence, 194 CT: see Computed tomography

Cystic fibrosis

carrier screening (editorial), 125 carrier testing in high-risk population, 323 prenatal screening issues, 129

Cytogenetics

ACMG guidelines and (letter), 67 molecular karyotype and, 254

multicolor FISH utilization (editorial), 178 mosaicism detection in amniotic fluid, 94

nulticolor FISH utility in, 181

elemedicine consultation and, 328

afness, genetics of (CME/Invited Review), 295

Death, sudden, medium-chain acyl-CoA dehydrogenase deficiency and, brief report, 293

Demographics, rheumatoid arthritis in Pima Indians, genealogic approach, 187

Disease, genetic components of, primary care physician education and, 13

Disomy, maternal, gene analysis in Prader-Willi syndrome and chromosome 15 anomalies, 141

DNA methylation, maternal transmission of Angelman syndrome and, 262

Down syndrome

maternal risk factors for, 80

prenatal screening impact on birth status, 22

prenatal triple marker screening program and, costs and effectiveness of, 199

Duplication, intrachromosomal, comparative genomic hybridization and, 4

vsmorphology, online database prototype, 207

Itorials

ACMG's continuing medical education program: A new initiative, 127

Conception, gestation, and birth of a journal: The future of genetics in medicine includes Genetics in Medicine, 2

Genetics: Not just in there somewhere, but at the very center of medicine, 3

Genetics and the practice of medicine: The future is here, 1 Human Genome epidemiology: Translating advances in human genetics into population-based data for medicine and public health, 71

Molecular cytogenetics: Show me the colors, 178

Offering CF carrier screening: Who set the goal, and what is the goal?, 125

The psychological impact of a negative BRCA1 test: A wolf in sheep's clothing?, 69

In search of the Holy Grail: NF1 mutation analysis and genotype-phenotype correlation, 245

The virtues of the virtual world, 177

Education

ACMG's continuing medical education program (editori-

medical genetics and (presidential address), 56

Epidemiology

genome, translation of genetics advances and (editorial),

medium-chain acyl-CoA dehydrogenase deficiency, 332 rheumatoid arthritis in Pima Indians, genealogic approach,

trisomy 21 risk factors, 80

Ethnicity

allelic frequency differences in interleukin-4 receptor polymorphism, 267

and GALT, 40

Athabaskan severe combined immunodeficiency, description and background information, 90

genetic disorders among Apache Indians, 90

negative BRCA1 mutation test results in Ashkenazism, psychological impact, 74

rheumatoid arthritis in Pima Indians, genealogic approach,

Familial studies: see also Genetic studies/testing

heart defects, preaxial polydactyly, laryngeal anomalies, colonic aganglionosis and, description of unique syndrome, 104

monilethrix, molecular analysis of Palestinian family with, 109

Fatty acid

medium-chain acyl-CoA dehydrogenase deficiency and, epidemiology review, 332

oxidation

medium-chain acyl-CoA dehydrogenase deficiency and, sudden death from, brief report, 293 primary carnitine deficiency detection and, 34

Fetus

Down syndrome, prenatal screening impact on birth status, 22

trisomy 21 risk factors, 80

FISH: see Hybridization, fluorescence in situ

Folic acid, FDA fortification/supplementation requirements. ACMG statement on, 66

LT, 40

Galactosemia, 40

Galactose 1-phosphate uridyltransferase (GALT), 40

348

Gastrochisis, chorionic villus sampling and, literature review, 315

Gene deletion

in lissencephaly, FISH analysis with *LIS1* probes and, 29 NF1 gene, cognitive problems in neurofibromatosis type 1 and, 136

Gene discovery, transition to medical practice (editorial), 71 Gene expression, analysis in Prader-Willi syndrome and chromosome 15 anomalies, 141

Gene mutation

detection, ethnic-related differences in interleukin-4 receptor polymorphisms and, 267

exon 10b of NF1 gene, spectrum and frequency of mutations, 248

hHb6 gene, molecular analysis of Palestinian family with monilethrix and, 109

Gene patents, ACMG policy statement, 237

Genealogic studies, rheumatoid arthritis in Pima Indian population, 187

Genetic centers, academic, involvement in genetic testing for common disorders, 272

Genetic diseases

among Southwestern Athabaskan Amerindians, 151 among Southwestern Athabaskan Amerindians (letter), 304

Genetic services/counseling

consanguineous couples and offspring, need for practice guidelines, 286

cystic fibrosis carrier testing in high-risk population, 323 hemochromatosis diagnosis in proband's family members and, 89

Internet role in, 177

population-based surveillance of (letter), 305

prenatal, minimum guidelines for (letter), 233

primary care physician utilization and perceptions of, 13 primary care physicians and, 13

re-contact duties and responsibilities, 171

Genetic studies/testing

accessibility of, ACMG policy statement, 237

consanguineous couples and offspring, need for practice guidelines, 286

CPT coding revisions, 112

Crohn's disease and chromosome 12, 194

cystic fibrosis, implementation issues, 129

cystic fibrosis carrier screening (editorial), 125

deafness and (CME/Invited Review), 295

HMO and academic genetic center involvement in, 272 Internet role in, 177

monilethrix, molecular analysis of Palestinian family with,

rheumatoid arthritis in Pima Indian population, genealogic approach, 187

Genetics

history and future of (presidential address), 56 importance in medical practice (editorial), 3 practice of medicine and (editorial), 1, 2

Genetics in Medicine

development of (editorial), 2 informatics and, 52 need for (letter), 118 reviewer acknowledgment, 349 staying on top of the revolution, 53

Genodermatoses, genetic disorders among Southwestern Athabaskan Amerindians, 90

Genome, advances in genetics, translation to epidemiology data (editorial), 71

Genome project, after, 56

Genomic hybridization, comparative, clinical application of,

Genotype, NF1 mutation analysis (editorial), 245

Genotyping, HFE, hemochromatosis diagnosis in proband's family members and, 89

Harland Sanders Award, introduction, 219

Health maintenance organizations, involvement in genetic testing for common disorders, 272

Hearing, genetics of (CME/Invited Review), 295

Heart defect, laryngeal anomalies, preaxial polydactyly, colonic aganglionosis and, description of unique syndrome, 104

Hemochromatosis, diagnosis in proband's family members, 89

HER-2/neu gene, amplification in breast cancer, FISH analysis, 98

HFE mutation, hemochromatosis diagnosis in proband's family members and, 89

Hirschsprung disease, heart defects, preaxial polydactyly, laryngeal anomalies and, description of unique syndrome, 104

Homocystinemia, methylmalonic acidemia and, long-term outcome, 146

Hybridization, fluorescence in situ

comparative genomic hybridization and, 4

HER-2/neu gene amplification in breast cancer, 98

karyotype refinement and, 254

lissencephaly analysis by, 29

multicolor

utility in cytogenetics, 181

utilization in molecular cytogenetics (editorial), 178

21-Hydroxylase deficiency, congenital adrenal hyperplasia caused by, antenatal treatment for (CME/Invited Review), 224

Hypoglycemia, nonketotic, primary carnitine deficiency and,

Immunodeficiency, Athabaskan severe combined, description and background information, 90

Immunoglobulin E, hyper-IgE syndrome, autism, and mental retardation, analphoid marker chromosome in, 213

Immuunodeficiency, hyper-IgE syndrome, autism, and mental retardation, analphoid marker chromosome in, 213

Imprinting

genetic

analysis in Prader-Willi syndrome, 141

Prader-Willi syndrome and chromosome 15 anomalies, 141

genomic, maternal transmission of Angelman syndrome, 262

In Memoriam

Greenberg, Frank, 117 Kaplan, B. F., 65 Shapira, E., 64

Weiss, Lester, 64

Incest, genetics counseling and screening inconsistencies and, 286

Informatics, for the genetic clinician, 52

Interleukin-4 receptor polymorphism, allelic frequency differences, ethnic group comparison, 267

International Federation of Human Genetics Societies, history, purpose, and structure of (letter), 235

Internet

Online London Dysmorphology Database, 207 role in research, evaluation, and counseling, 177

Iron overload, hemochromatosis diagnosis in proband's family members and, 89

Karyotype, molecular refinement of, 254

Kinship coefficient, rheumatoid arthritis in Pima Indians and, genealogic approach, 187

Laboratory testing, CPT coding revisions, 112

Laryngeal anomalies, heart defects, preaxial polydactyly, colonic aganglionosis and, description of unique syndrome, 104

Letters to the Editor, 67, 118, 170, 233, 235, 304, 305

LIS1 gene, FISH analysis, 29

Lissencephaly, FISH analysis of, 29

London Dysmorphology Database, online accessibility, 207

Magnetic resonance imagine, structural anomalies in neurofibromatosis type 1 and, 136

Managed care organizations, involvement in genetic testing for common disorders, 272

Maternal transmission, Angelman syndrome, 262

Medical genetics, clinical objectives for undergraduate medical students, 54

Medicine practice

genetics and (editorial), 1, 2

genetics importance in (editorial), 3

Medium-chain acyl-CoA dehydrogenase deficiency

human genome epidemiology review, 332 sudden death and, brief report, 293

Membrane transport, primary carnitine deficiency detection and 34

Mental retardation

genetics counseling and screening inconsistencies and, 286 hyper-IgE syndrome, autism, and, analphoid marker chromosome in, 213

Methylmalonic acidemia, homocystinemia and, long-term outcome, 146

Molecular analysis, neurofibromatoses treatment approaches and (CME/Invited Review), 158

Monilethrix, molecular analysis of Palestinian family with, 109

Mosaicism, detection in amniotic fluid, 94

MRI: see Magnetic resonance imaging

Mutations, in the GALT gene, 40

Myopathy, primary carnitine deficiency and, 34

Native Americans, genetic disorders among, 90 Navajo Indians, genetic disorders among, 90

Neonatal screening: see also Prenatal screening/diagnosis

ethnic-related differences in interleukin-4 receptor polymorphisms and, 267

Neural tube defects

folic acid fortification/supplementation and, 66 prenatal triple marker screening program and, costs and effectiveness of, 199

Neurofibromatoses

emerging treatment approaches (CME/Invited Review), 158

treatment of (CME exam), 165

Neurofibromatosis type 1

brain structural anomalies in, neuroimaging studies, 136 spectrum and frequency of mutations of exon 10b of *NF1* gene, 248

Neuroimaging, structural anomalies in neurofibromatosis type 1 and, 136

Neuropathy, Navajo, description and background information, 90

NF1 gene

exon 10b, spectrum and frequency of mutations in, 248 mutation analysis (editorial), 245

neuroimaging detection of structural anomalies and, 136 **Nomenclature,** appropriateness of (letter), 170

Pediatrics

cancer, gene imprinting analysis and, 141 malformations related to chorionic villus sampling and, literature review, 315

Phenotyping

differences in African Americans with Prader-Willi syndrome, 49

hemochromatosis diagnosis in proband's family members and, 89

NF1 mutation analysis (editorial), 245

Pima Indians, rheumatoid arthritis in, genealogic approach, 187

Poikiloderma, Navajo, description and background information, 90

Polydactyly, preaxial, laryngeal anomalies, heart defects, and colonic aganglionosis and, description of unique syndrome, 104

Polymerase chain reaction, reverse transcription, imprinted gene analysis in Prader-Willi syndrome, 141

Polymorphism, spectrum and frequency of mutations of exon 10b of *NF1* gene and, 248

Prader-Willi syndrome

imprinted gene analysis in, 141

phenotypic differences in African Americans, 49

Pregnancy

elective termination, Down syndrome diagnosis and, 22 triple marker screening program and, costs and effectiveness of, 199

Prenatal diagnosis/screening: see also Neonatal screening California triple marker program, cost and effectiveness of, 199

congenital adrenal hyperplasia and (CME/Invited Review), 224

consanguineous couples and offspring, need for practice guidelines, 286

cystic fibrosis

carrier screening (editorial), 125

implementation issues, 129

Down syndrome, prenatal screening impact on birth status, 22

genetics, minimum guidelines for (letter), 233

21-hydrolase deficiency and (CME/Invited Review), 224 mosaicism detection in amniotic fluid, 94

Primary care, genetics services utilization and perceptions in, 13

Primary carnitine deficiency, urinary carnitine transport and, 34

Psychological considerations

negative BRCA1 mutation test results, 74 negative BRCA1 test results (editorial), 69

Renal resorption, primary carnitine deficiency detection and, 34

Research

International Federation of Human Genetics Societies and (letter), 235

Internet role (editorial), 177

Resource utilization, genetics services, primary care physicians and, 13

Rheumatoid arthritis, genealogy construction in Pima Indians, 187

Risk factors

33

congenital adrenal hyperplasia due to 21-hydroxylase deficiency (CME/Invited Review), 224 cystic fibrosis, carrier testing in high-risk population, 323 trisomy 21, maternal origin, 80

Telemedicine, cytogenetic analysis and, 328

Terminology, appropriateness of (letter), 170

Translocation, unbalanced, comparative genomic hybridization and, 4

Triple marker screening, prenatal, program for, cost and effectiveness of, 199

Trisomy 21, risk factors for, 80

Ulcerative colitis, chromosome 12 and, linkage evidence, 194 **Undergraduate medical students,** clinical objectives in medical genetics, 54

Vascular disruption defects, chorionic villus sampling and, literature review, 315

World Wide Web: see Internet