

## Subject Index

## a

- Abstracts**, 2000 Annual Clinical Genetics Meeting, 1
- ACMG**: *see* American College of Medical Genetics
- ACMG/ASHG statement**
- genetic testing for colon cancer, 362
  - tandem mass spectrometry in neonatal screening, 267
  - technical and clinical assessment of fluorescence in situ hybridization: technical considerations, 356
- American College of Medical Genetics**
- 2000 Annual Clinical Genetics Meeting, 1
  - brief history of, 255
  - College News, 153, 205, 259, 304, 369
  - future directions in, 255
  - guidelines, expert witness testimony for the specialty of medical genetics, 367
  - policy statement
    - informed consent for medical photographs, 353
    - universal newborn hearing screening, 149
  - presentations at medical meetings, colorblindness and (letter), 350
  - presidential address, 255
  - recommendations for standards for interpretation of sequence variations, 302
- Androgen receptor**, women with abnormal X chromosome, 136
- Angelman syndrome**, interstitial duplications, phenotype correlations for, 131
- Asplenia**, heterotaxy and, prevalence in newborns, 157
- Athletes**, female, gender verification for, 249
- Autism**, interstitial duplications and, 131

## b

- Berlin diagnostic criteria**, Marfan syndrome, dural ectasia in, comparison with Ghent criteria, 278
- Biochemical genetics**, diagnosis and management of genetic disease and, 328
- BRCA1 mutation test**, negative result, mammography behavior after, 307
- Breast cancer**, mammography behavior after negative *BRCA1* mutation test result and, 307

## c

- Cancer**: *see also* specific type
- genetic testing validation and (editorial), 155

- Cardiovascular malformations**, heterotaxy and, prevalence in newborns, 157
- CFTR gene**, cystic fibrosis and, interactivity between, 124
- CGG repeat dynamics**, fragile X syndrome, similarity in siblings, comparison with unrelated patients, 242
- Chorionic villus sampling**, malformations in children exposed to (letter), 300
- Chromosome 3q**, optic atrophy and, clinical heterogeneity of, 283
- CLN gene**, late-infantile neuronal ceroid lipofuscinosis, heterogeneity of, 312
- Colorblindness**, presentations at medical meetings and (letter), 350
- Colorectal cancer**, genetic testing for, ACMG/ASHG statement, 362
- Commentaries**
- Challenges in communicating genetics: A public health approach, 198
  - Some ethical implications of the Human Genome Project, 193
- Computed tomography**, Marfan syndrome, dural ectasia in, 173
- Congenital anomalies**
- glycosylation disorders, prevalence and pathogenesis, 329
  - women with abnormal X chromosome and, 136
- Congenital heart defects**, heterotaxy and, prevalence in newborns, 157
- Coronary heart disease**, genetic testing validation and (editorial), 155
- C282Y mutation**, HFE genotypes contribution to iron overload disease and, 271
- Cystic fibrosis**
- CFTR gene and, interactivity between, 124
  - psychological effects and reproductive uncertainty in (letter), 203

## d

- Deafness**, universal newborn screening and, ACMG policy statement, 149
- Depression**, interstitial duplications and, 131
- Developmental delay**, global, interstitial duplications and, 131
- Diabetes**
- genetic testing validation and (editorial), 155
  - maternal, heterotaxy and, prevalence in newborns, 157
- Duplication**, interstitial, characteristics, 131
- Dural ectasia**, in Marfan syndrome
- Berlin vs. Ghent diagnostic criteria, 278
  - MR and CT findings and criteria, 173
- Dyspraxia**, verbal, in galactosemia, outcomes analysis, 142

## e

**Editorials**

Clinical validation of genetic tests, 155

From the Editor's desk, 123

Lessons from genetic discrimination, 207

**Education**, genetics, health professionals and, 226

**Environmental effects**, genetic discrimination and (editorial), 207

**Epidemiology**

CFTR gene and cystic fibrosis interactivity, 124

congenital glycosylation disorders, prevalence and pathogenesis, 329

genetic testing, population-based registry use in validation of, 186

heterotaxy, associated conditions and prevalence in newborns, 157

osteoporosis, familial risk profile for, 222

**Ethical issues**

gender verification of female athletes, 249

Human Genome Project, ethical implications of (commentary), 193

## f

**Familial clustering**, fragile X syndrome, similarity in siblings, comparison with unrelated patients, 242

**Familial studies**: *see also* Genetic studies/counseling

family history-taking in family practice, 180

osteoporosis, familial risk profile for, 222

**Fatty acid transport disorders**, metabolic evaluation, 338

**Fetal screening**, chorionic villus sampling, malformations in children exposed to (letter), 300

**Fluorescence in situ hybridization**, technical and clinical assessment of, ACMG/ASHG statement, 356

**Folic acid**, neural tube defect prevention, college women's awareness and consumption of, 209

**Fragile X syndrome**, similarity in siblings, comparison with unrelated patients, 242

## g

**Galactosemia**, verbal dyspraxia in, outcomes analysis, 142

**Gender verification**, female athletes and, 249

**Genetic discrimination**

(editorial), 207

by health insurers, patient fear of, legal protections and, 214

**Genetic disease**, biochemical genetics role in diagnosis and management, 328

**Genetic services/counseling**

family history-taking in family practice, 180

osteoporosis, familial risk profile for, 222

**Genetic studies/testing**

clinical validation of (editorial), 155

family history-taking in family practice and, 180

fragile X syndrome, similarity in siblings, comparison with unrelated patients, 242

genetic testing for colon cancer, ACMG/ASHG statement, 362

inborn errors of metabolism, molecular diagnosis of, 345

mammography behavior after negative *BRCA1* mutation test result and, 307

Paget disease of bone and limb-girdle muscular dystrophy, clinical and molecular studies in family with, 232

population-based registry use in validation of, 186

sequence variation interpretation, ACMG recommendations for standards for, 302

**Genetics**

CFTR gene and cystic fibrosis interactivity, 124

expert witness testimony, ACMG guidelines, 367

Internet resources and (letter), 351

public communication of findings in (commentary), 198

**Genetics education**

health professionals and, 226

resources on the Internet, 296

**Genetics in Medicine**, report on journal (editorial), 123

**Ghent diagnostic criteria**, Marfan syndrome, dural ectasia in, comparison with Berlin criteria, 278

**Global**, interstitial duplications and, 131

**Glycosylation**, congenital disorders of, prevalence and pathogenesis, 329

**G<sub>M2</sub> gangliosidosis**, locus-specific databases and, 319

**GM2A gene**, locus-specific databases in **G<sub>M2</sub> gangliosidosis** and, 319

## h

**H63D mutation**, HFE genotypes contribution to iron overload disease and, 271

**Health care**

CFTR gene and cystic fibrosis interactivity, 124

family practice, family history-taking, genetic screening implications, 180

**Health care workers**, genetics education and, 226

**Health insurance**, genetic discrimination by, patient fear of, legal protections and, 214

**Health professionals**, genetics education and, 226

**Hearing screening**, universal newborn, ACMG policy statement, 149

**Hemochromatosis**, HFE genotypes contribution to, 271

**Heterogeneity**, genetic, late-infantile neuronal ceroid lipofuscinosis, heterogeneity of, 312

**Heterotaxy**, associated conditions and prevalence in newborns, 157

**HEXA gene**, locus-specific databases in **G<sub>M2</sub> gangliosidosis** and, 319

**HEXB gene**, locus-specific databases in **G<sub>M2</sub> gangliosidosis** and, 319

**HFE genotypes**, iron overload disease and, 271

**Human Genome Project**

ethical implications of (commentary), 193

genetic testing validation and (editorial), 155

**Hydroxymethylbilane synthase gene**, mutations in, acute intermittent porphyria and, 290

# i

- In Memoriam**, Haun, R. L., 204
- Informed consent**, medical photographs, ACMG policy statement, 353
- Internet**
  - genetics resources on, 296
  - genetics resources on (letter), 351
- Iron overload disease**, HFE genotypes contribution to, 271
- Isomerism**, heterotaxy and, prevalence in newborns, 157

# l

- Laterality defects**, heterotaxy and, prevalence in newborns, 157
- Legal issues**
  - expert witness testimony in medical genetics, ACMG guidelines, 367
  - genetic discrimination by health insurers, patient fear of, 214
- Letters to the Editor**, 203, 300, 350
- Locus-specific databases**,  $G_{M2}$  gangliosidoses, 319

# m

- Magnetic resonance imaging**, Marfan syndrome, dural ectasia in, 173
- Mammography**, after negative *BRCA1* mutation test result, 307
- Marfan syndrome**
  - dural ectasia in, MR and CT findings and criteria, 173
  - dural ectasia influence in, Berlin vs. Ghent diagnostic criteria, 278
- Mental retardation**, women with abnormal X chromosome and, 136
- Metabolic evaluation**, fatty acid transport and mitochondrial oxidation disorders, 338
- Metabolism**, inborn errors of, molecular diagnosis and, 345
- Mitochondrial oxidation disorder**, metabolic evaluation, 338
- Molecular diagnosis**, inborn errors of metabolism and, 345
- Muscular dystrophy**, limb-girdle, Paget disease of bone and, clinical and molecular studies in family with, 232
- Mutation analysis**, late-infantile neuronal ceroid lipofuscinosis, heterogeneity of, 312
- Mutation pattern**, fragile X syndrome, similarity in siblings, comparison with unrelated patients, 242

# n

- Neonatal screening**
  - fatty acid transport and mitochondrial oxidation disorders, 338
  - hearing screening, universal, ACMG policy statement, 149

tandem mass spectrometry in, ACMG/ASHG statement, 267

- Neonate**, heterotaxy, associated conditions and prevalence, 157
- Neural tube defects**, folic acid prevention of, college women's awareness and consumption of, 209
- Neuronal ceroid lipofuscinosis**, late-infantile, heterogeneity of, 312

# o

- Obesity**, interstitial duplications and, 131
- Optic atrophy**, clinical heterogeneity of, 283
- Osteoporosis**, familial risk profile for, 222

# p

- Paget disease of bone**, limb-girdle muscular dystrophy and, clinical and molecular studies in family with, 232
- Patient photographs**, informed consent for, ACMG policy statement, 353
- Pediatrics**, chorionic villus sampling, malformations in children exposed to (letter), 300
- Pervasive developmental disorder**, interstitial duplications and, 131
- Polysplenia**, heterotaxy and, prevalence in newborns, 157
- Porphobilinogen deaminase**, hydroxymethylbilane synthase gene mutations, acute intermittent porphyria and, 290
- Porphyria**, acute intermittent, mutations in hydroxymethylbilane synthase gene, 290
- Prader-Willi/Angelman syndrome**
  - interstitial duplications, phenotype correlations for, 131
  - phenotype for (letter), 301
- Pregnancy**, neural tube defect, folic acid for prevention, college women's awareness and consumption of, 209
- Primary care**, family history-taking in family practice, 180
- Public health**, public communication of findings in genetics (commentary), 198

# r

- Research**
  - genetic testing, population-based registry use in validation of, 186
  - medical photographs, informed consent for, ACMG policy statement, 353

# s

- Self-injurious behavior**, interstitial duplications and, 131
- Sequence variations**, interpretation of, ACMG recommendations for standards for, 302

**Sex testing**, genetic, female Olympic athletes, 249  
**Sexual differentiation**, genetic, female Olympic athletes, 249  
**Situs ambiguous**, heterotaxy and, prevalence in newborns, 157  
**Situs inversus**, heterotaxy and, prevalence in newborns, 157  
**Spectrometry**, tandem mass, in neonatal screening, ACMG/ASHG statement, 267  
**Speech delays**, developmental, interstitial duplications and, 131  
**Spinal deformity**, Marfan syndrome, dural ectasia in, MR and CT findings and criteria, 173  
**Strabismus**, interstitial duplications and, 131  
**Stroke**, genetic testing validation and (editorial), 155

## t

**Telemedicine**, impact on medical practice (letter), 351

## v

**Verbal disabilities**, verbal dyspraxia, in galactosemia, outcomes analysis, 142

## w

**World Wide Web**: *see* Internet

## x

**X chromosome**, abnormal, X inactivation pattern in women with, 136