Index

Abdominal pain in Williams syndrome, 205
Adderall, 96
Adolphs, R., 239
Adrenarche in Klinefelter syndrome, 65
Alant, E., 385
Altshul-Stark, D., 90
American Association on Mental Retardation, 392
Amino acid metabolism, 298–303
Androgen-deficiency in Klinefelter syndrome, 62–64
Animals and hypothyroidism, 272–273
Anticipatory guidance and genetic counseling, 374–378
Anxiety
  and hyperarousal in fragile X syndrome, 82–83, 91–92
  in Williams syndrome, 241
Aqueductal stenosis, 141
Ashkoomoff, N. A., 231
Asperger syndrome in Turner syndrome, 27
Assistive technology, 482
Atkinson, J., 232
Attention and executive function
  congenital hypothyroidism and, 281–282, 284
  DS22q11.2 and, 178, 185–188
  fragile X syndrome and, 90–91
  Klinefelter syndrome and, 58–59
Attention deficit/hyperactivity disorder (ADHD)
  congenital hypothyroidism and, 281–282
  DS22q11.2 and, 168, 178
  fragile X syndrome and, 82, 85, 91–92
  phenylketonuria and, 301
  Williams syndrome and, 206, 241–242
Autism
  Duchenne muscular dystrophy and, 121
  fragile X syndrome and, 83–85
  interventions, 96–97
  mercury and, 351–352
  Turner syndrome and, 27
  Williams syndrome and, 242
Autoimmune thyroiditis, 22

Bagnato, S. J., 445
Bailey, D. B., 82, 91, 400, 453
Baker, K., 190
Baranek, G. T., 91
Barth syndrome, 322, 425–430
Bayley Scales of Infant Development, 277
Bearden, C. E., 177
Bechar, T., 223
Becker muscular dystrophy, 106
Behavioral characteristics
  congenital hypothyroidism, 285, 286f
  DS22q11.2 and, 452–453
  Duchenne muscular dystrophy, 120–121, 125–127
  early intervention and, 447–448
  fragile X syndrome, 82–85, 453–454
  Klinefelter syndrome, 59–60
  mental retardation and adaptive, 391–393, 403–404
  Williams syndrome, 208–210, 217, 234–240
Bellinger, D., 337
Bellugi, U., 221, 223, 225, 230, 233, 238
Belser, R. C., 82, 87
Bender, B. G., 29
Bernard, S., 351
Bertrand, J., 220, 230
Bickel, H., 303
Billard, C., 119
Billard, C., 119
Billingsley, R. L., 149, 150
Bishop, D., 224
Bjogstad, K. B., 306
Blacher, J., 407
Bleeding, gastrointestinal, 22
Boccia, M. L., 82
Boutet, L., 89
Brain development
  Duchenne muscular dystrophy and, 112–113
  fragile X syndrome and, 76–81
  inborn errors of metabolism and, 300–301, 324–326
  Klinefelter syndrome and, 49, 60–64, 64
Brain development (cont.)
lead poisoning and, 336–339
manganese effect on, 353–354
mercury effects on, 346
neurofibromatosis and, 136–137, 141–142, 146–149
phenyketonuria and, 300–301
thyroid hormone and, 272
tumors and, 147
Turner syndrome and, 24–25
Brewer, V. R., 145
Bruno, E., 245
Campbell, D., 347
Carbohydrate metabolism disorders
galactosemia, 310–312, 313–314t
pyruvate dehydrogenase deficiency, 312
Cardiovascular disorders
Turner syndrome and, 20
Williams syndrome and, 206, 245
Carey Temperament Scales, 32
Cayler cariofacial syndrome, 164
Celiac disease in Turner syndrome, 21
Central hypothyroidism, 270
Central nervous system
creatine disorders and, 321
neurofibromatosis and, 141–142
Chelation, 342
Chen, T. Y., 407
Chess, S., 234
Child-focused practices in early intervention, 446–448
Cholesterol biosynthesis disorders, 323
Chromosome 22q11.2 deletion syndrome
attention in, 168, 178
cognitive and academic manifestations of, 170–173, 191–193
cognitive experimentation studies, 179–188
definition, 163–164
diagnosis, 166, 168–170
early intervention and special education for, 449–453
enumeration in, 182–183
executive function and inhibition in, 185–188
feeding problems, 451–452
incidence of, 164–165
IQ in, 173–174, 450
language skills in, 175, 176–177, 451
magnitude estimation and mathematical reasoning in, 183–185
memory in, 176, 177–178
motor abilities in, 178–179, 452
multidisciplinary assessment and service coordination, 452
neural and genetic correlates, 188–191
neuropsychological profile, 173–174
physical manifestations, 165–166, 167f
preschool services for children with, 453
psychiatric disorders associated with, 167–170
reading and spelling skills in, 175–176, 177
schizophrenia and, 168, 189–191
visual-spatial attention in, 181–182
Chromosomes
Duchenne muscular dystrophy abnormalities in, 106–107
imprinting and, 11
Klinefelter syndrome abnormalities in, 50–51
Turner syndrome abnormalities in, 5, 6f, 7t
Civil rights and mental retardation, 398–399
Classroom placement, 483–484
Cognition. See Neurocognition
Concerta, 96
Congenital hypothyroidism. See Hypothyroidism, congenital
Connective tissue abnormalities and Williams syndrome, 245
Coordination, visual-motor, 89
Cornish, K., 85, 89
Coyle, J. T., 304
Creatine deficiency syndromes, 320–321
Creswell, C., 27
Crohn’s disease, 22
Cross, G., 85
Cunningham, C. C., 401
Cystathionine beta-synthase deficiency, 303–305
Cystic hygroma, 13f
Davies, W., 11
Dental problems in Williams syndrome, 204
Devenny, D. A., 229
Dexedrine, 96
Diabetes mellitus in Turner syndrome, 23
Diagnosis
autism, 83–84
chromosome 22q11.2 deletion syndrome, 166, 168–170
Duchenne muscular dystrophy, 106
Duchenne muscular dystrophy abnormalities in, 106–107
Duchenne muscular dystrophy immediately before and after, 367–370, 374–378
food poisoning, 340–341
inborn errors of metabolism, 298, 299t
interpretation by children, 382–383
Klinefelter syndrome, 47–49
lead poisoning, 340–341
learning disability as a risk, not a, 422–424
learning to talk about, 379–382
mental retardation, 393, 395–396, 401–402
neurofibromatosis (NF), 135–137
preparation, 369–370
recurrence risk and, 384–385
sibling of affected patients, 385–386
Turner syndrome, 16–18, 34–36
Williams syndrome, 206–208
Diamond, A., 300
Differential Ability Scales (DAS), 212, 213f, 215t
DiGeorge, Angelo, 163
Digit Span, 227
Dilts, C., 239
Division for Early Childhood of the Council for Exceptional Children, 443–444
Dorman, C., 118–119
Downey, J., 28
Down syndrome
attention deficit/hyperactivity disorder (ADHD) and, 85
memory and, 88
mental retardation and, 394, 395, 400
portrayed in the media, 398–399
prenatal diagnosis, 402
Doyle, T. F., 235
Drawing abilities and Williams syndrome, 230–231
Drosophila NF-1 protein, 140
DS2q11.2. See Chromosome 22q11.2 deletion syndrome
Dubowitz, V., 118
Duchenne, Guillaume-Benjamin-Amand, 106
Duchenne muscular dystrophy
academic skills in, 118–119
autism and, 121
and Becker muscular dystrophy, 106
behavioral characteristics, 120–121, 125–127
brain development and, 112–113
cognitive skills in, 114–117, 125–127
diagnosis, 106
discovery of, 106
dystrophin and, 107–108
gene, 106–107
IQ scores and, 113–114, 115–117
medical management of, 110–112
physical presentation, 108–110
prevalence, 105, 106
quality of life in older individuals with, 124–125
stress and, 122–123
Due process, 487–488
Dunst, C. J., 446
Dykens, E. M., 90, 233, 240, 452, 463
Dysgenesis, thyroid, 267–268
Dysglobulinemia, thyroid, 268–270
Dyslexia, 149, 150
Dystrophin, 107–108, 112–113
Early Childhood Outcomes (ECO), 442–443
Early intervention and early childhood special education (ECSE)
assessment for planning and monitoring, 439, 444–445
child-focused practices in, 446–448
determining eligibility for, 439
family-centered approach to, 445–446
family stressors and, 438
fragile X syndrome, 455–458
Individualized Family Service Plan, 439–440
interdisciplinary models, 448–449
Klinefelter syndrome, 460–464
outcomes, 441–443
purpose of, 437, 464–465
22q11.2 deletion syndrome, 449–453
recommended practices, 443–449
service delivery, 440–441
Turner syndrome, 458–460
Early Social Communication Scales, 220
Einfeld, S., 240
Elasint arteriopathy in Williams syndrome, 205, 245
Eligibility, individualized education plan, 476
Encephalopathy, mitochondrial, 316
Endocrinology of Klinefelter syndrome, 51–52
Engel, P., 234
English as a Second Language classes, 481–482
Enumeration tasks and DS22q11.2, 182–183
Epidemiology
Klinefelter syndrome, 50
neurofibromatosis (NF), 134
Estrogen therapy in Turner syndrome, 26
Event-related potentials, 60–61
Everyday Memory Questionnaire, 283–284
Executive function. See Attention and executive function
Expressive Vocabulary Test (EVT), 217–218
Extended school year (ESY), 482–483
Eye problems
Turner syndrome, 21
Williams syndrome, 203
Family. See also Parents
-centered approach to early intervention, 445–446
composition and mental retardation, 404–406
Farran, E. K., 230
Fatty acid oxidation disorders, 316–318
Feeding problems
DS22q11.2 deletion syndrome, 451–452
Turner syndrome, 21
Feinstein, C., 168
Fidler, D. J., 239
Finucane, B. M., 451
504 plans, 486
Fivush, R., 382
Fluorescent in situ hybridization (FISH) studies, 5
Fragile X syndrome
attention deficit/hyperactivity disorder and, 82, 85, 91–92, 93–97
autism and, 83–85, 96–97
behavioral considerations in, 82–85, 453–454
cognitive profile, 86
cognitive strengths and difficulties, 86–91, 424–425
early intervention and special education, 455–457
family support and, 457–458
genetic and brain-level considerations, 76–81
Fragile X syndrome (cont.)
importance of recognizing comorbidities in, 91–92
IQ and, 426–430
learning to talk about, 379–382
memory in, 88–89
mental retardation and, 396–397
number processing in, 90
pharmacologic interventions, 96, 97, 454–455, 457
physical characteristics, 73, 74f, 75–76t
prevalence, 73
repetitive speech in, 87–88
social anxiety and hyperarousal in, 82–83
speech and language in, 86–88
tangential and perseverative language in, 87
visual-motor coordination in, 89
Frigerio, E., 238
Frith, U., 432
Frontal brain structural malformation, 141–142
Functional behavioral assessments (FBAs), 475
Functional disomy, 11
Functional imaging studies of neurofibromatosis, 149–151
Fusco, J., 407

Gagliardi, C., 237
Galaburda, A. M., 64
Galactosemia, 310–312, 313–314t
Gastroesophageal reflux (GERD), 203, 205
Gastrointestinal disease in Turner syndrome, 21–22
Geffner, M. E., 31–32
Geier, D. A., 351
Geier, M. R., 351
General Conceptual Ability (GCA) scores, 212, 215f
Generalized anxiety disorder (GAD), 241
Genetic counseling
anticipatory guidance and, 374–378
beyond the diagnosis, 378–379
children’s interpretation of, 382–383
collection/review of family medical history, 371
components of, 370–374
contracting, 371
diagnostic process and, 368–370
discussion and formulation of diagnostic plan, 373
discussion of clinical suspicion and findings, 371–373
following diagnosis of genetic disorder, 374–378
follow-up, 378–379
medical genetics evaluation and, 368–370
recurrence risks and, 384–385
referral to the medical genetics clinic for, 368
siblings and, 385–386
Genetic mutations
DS22q11.2, 188–191
Duchenne muscular dystrophy, 106–107
fragile X syndrome, 76–81, 89
neurofibromatosis, 137–139
Williams syndrome, 242–248
Gerdes, M., 450–451
Gerner, G., 28
Geschwind, N., 64, 463
Glidden, L. M., 405, 406, 407
Glutaric acidemia type I, 306–307
Glycine encephalopathy, 318–319
Gonadal dysgenesis, 19–20
Good, C. D., 10
Gopnik, A., 220
Gosch, A., 234
Gothelf, D., 190
Grace, R. J., 60
Grammatical ability in Williams syndrome, 223–224
Grant, J., 226
Gratton effect, 186
Greenwood, R. S., 149
Greer, M. K., 239
Growth hormone, 19–21
GTF2I gene, 246–247
Gunther, D. F., 7
Guthrie, R., 303
Hagerman, R., 84, 90, 454
Harper, D. C., 121
Hart, S. J., 25
Hartman, D. E., 344
Hatton, D. D., 82, 85, 91, 394, 456
Havercamp, F., 21
Hayes, A., 84
Hearing loss
in Turner syndrome, 21
in Williams syndrome, 204
Henry, A. E., 28
Hepburn, S. L., 84
Hepworth, S., 283
Hernias, inguinal, 203
Heyerdahl, S., 287
Hobart, H. H., 243
Hoehn-Saric, R., 82
Hoffman, J. E., 232
Homeobox genes, 9
Homocystinuria, 303–305
Hooper, S. R., 31–32, 91
Hormonal factors
in Klinefelter syndrome, 51–52, 58, 59–60, 62–64
in Turner syndrome, 26
HSP27 gene, 246
Hyperarousal in fragile X syndrome, 82–83
Hypercalcemia, 203
Hyperglycinemia, nonketotic, 318–319
Hyperphenylalaninemia (HPA), 303
Hypersensitivity in fragile X syndrome, 82–83
Hypersocialibility in Williams syndrome, 238–239
Hypertension
in Turner syndrome, 20
in Williams syndrome, 205
Hypertrophic scar formation, 23
Hypothyroidism
central, 270
congenital
attention in, 281–282
behavior problems and, 285, 286f
cognitive abilities, 278–284
eye diagnosis, 290
etiologies of, 267–270
executive function in, 284
influence of disease and treatment-related
variables, 287–289
intelligence and, 276–278, 288–289
language skills and, 279
memory and, 283–284
motor abilities and, 280–281
neuropsychological profile in children with, 276
phenotype-genotype correlations, 289
physical features, 265–266
prevalence, 265
school achievement and, 284–285
thyroid dysgenesis and, 267–268
thyroid dyshormonogenesis and, 268–270
thyroid hormone resistance and, 270
visuospatial abilities and, 279–280
studies
with animals, 272–273
with humans, 273–274
transient, 271, 287
in Turner syndrome, 22
Hypotonia disorders, 321–322
Imprinted genes, 11
Inactivation, X, 8–9
Inborn errors of metabolism (IEM)
Barth syndrome, 322
carbohydrate, 310–314
cholesterol biosynthesis disorders, 323
classifications, 297
creatinine deficiency syndromes, 320–321
cystathionine beta-synthase deficiency, 303–305
fatty acid oxidation, 316–318
galactosemia, 310–312, 313–314t
glutaric acidemia type I, 306–307
hypotonia and, 321–322
maple syrup urine disease, 305–306
medium chain acylCoA dehydrogenase
deficiency, 317–318
methylmalonic and propionic acidemia, 308–310
mitochondrial, 315–316
molybdenum cofactor deficiency, 319–320
neurotransmitter disorders, 322–323
nonketotic hyperglycinemia, 318–319
organic acid, 308–310
phenylketonuria (PKU), 298–303
protein, 298–308
pyruvate dehydrogenase deficiency, 312
respiratory chain disorders, 315–316
seizures and, 318–321
system effects of, 297
tyrosinemia type II, 305
urea cycle disorders, 307–308
Inclusive education, 483–484
Independent educational evaluations (IEEs), 475
Individualized Education Program (IEP), 438, 471–472
annual goals, 478
assistive technology and, 482
Child Study Teams, 474
classroom placement, 483–484
current performance section, 477–478
definition, 473
due process and, 487–488
eligibility criteria, 476
evaluation process, 474–476
extended school year and, 482–483
history of, 472–473
identification of children with disabilities and, 473–474
lack of available resources for, 485–486
for limited English speakers, 481–482
parent role in, 479, 485
504 plans and, 486
preparing classroom peers and, 484
related services, 480–481
role of parents in, 479
self-contained classroom placements, 485
specialized training for staff, 481
testing areas, 474–476
timeline, 488–489
transition from secondary to postsecondary
education, 483
writing, 477
Individualized Family Service Plan (IFSP), 438–439, 439–440
Individuals with Disabilities Education Act
(IDEA), 416, 437, 471–473. See also Early
intervention and early childhood special
education (ECSE)
Inflammatory bowel disease in Turner syndrome, 22
Inhibition in DS22q11.2, 185–188
Intelligence and IQ. See also Mental retardation
congenital hypothyroidism and, 276–278, 288–289
cystathionine beta-synthase deficiency and, 303–304
DS22q11.2, 173–174, 450
Duchenne muscular dystrophy, 113–114, 115–117
fragile X syndrome, 86
galactosemia and, 311
Intelligence and IQ. See also Mental retardation (cont.)
Klinefelter syndrome, 56
lead poisoning effects on, 339
maple syrup urine disease and, 305–306
mental retardation classification and, 391–392
neurofibromatosis, 143
nonverbal learning disability and, 425–430
Turner syndrome, 25
Williams syndrome, 210–212, 246–247
Interdisciplinary models in early intervention, 448
Interventions
Duchenne muscular dystrophy, 125–127
fragile X syndrome, 92–97
learning disability, 431–432
planning and monitoring, 439
Itti, E., 62

Jackendoft, R., 219
Johnson, M. H., 173
Jordan, H., 232
Judgment of Line Orientation test, 145–146
Jugular vein, 9

Karayiorgou, M., 187
Karmiloft-Smith, A., 218, 226, 236
Karyotype abnormalities
Klinefelter syndrome, 50–51
Turner syndrome, 5, 6f, 7t
Kaufman Brief Intelligence Test (KBIT) and
Williams syndrome, 210–212, 213t, 214f
Kemper, M. B., 90
Keysor, C. S., 82
Kiley-Brabeck, K., 168, 179, 187
Klein, B. P., 226
Klein-Tasman, B. P., 235, 242
Klinefelter, H., 47
Klinefelter syndrome
academic abilities in, 56–57, 419
adrenarche in, 65
attention and executive function in, 58–59
brain development in, 49, 60–64
diagnosis and identification, 47–49
eyearly intervention and special education, 460–464
epidemiology, 50
hormonal factors influencing development in, 62–64
IQ in, 56
karyotypes and genetic mechanisms, 50–51
language abilities in, 57–58
microcephaly in, 61–62
motor function in, 52–55
neurocognition in, 55–59
physical characteristics of, 47–49, 52, 53–55f
prevalence, 47, 50
social development in, 59–60
testosterone levels in, 51–52, 58, 59–60, 62–64
Koeberl, D. D., 32
Kogan, C. S., 89
Kooistra, L., 280
Kozma, C., 174, 192
Kraemer, U., 337
Kuntsi, J., 27

Lactic acidosis, 316
Laing, E., 220
Lajiness-O’Neill, R. R., 176
Landau, B., 221, 222, 232
Language and speech abilities
congenital hypothyroidism and, 279
DS22q11.2 and, 175, 176–177, 451
fragile X syndrome and, 86–87
Klinefelter syndrome and, 57–58
relations between cognitive abilities and, 225–227
tangential and perseverative, 87
Williams syndrome and, 208–210, 219–227
Laws, G., 224
Lead poisoning
diagnosis, 340–341
government regulations and, 342–343
lessons learned from, 356
neurodevelopmental effects of, 338–339
socioeconomic status and, 337–338
treatment, 342–343
vulnerability of children to, 336–338
Learning and academic achievement
congenital hypothyroidism and, 284–285
DS22q11.2 and, 170–173, 191–193
Duchenne muscular dystrophy and, 118–119
Klinefelter syndrome and, 462–464
neurofibromatosis and, 143–145, 150–151, 156–157
Learning disability (LD)
biological basis of, 418
classification of, 416–417
definitions, 415–418
generalizing group findings to individuals with, 424–425
genetic disorders as models of, 418–422
intervention and identification, 431–432
IQ and, 425–430
nonverbal, 27, 29t, 415, 425–431
as a risk, not a diagnosis, 422–424
Lenhard, W., 400
LeRoux, C., 305
Lesniak-Karpiaik, K., 31, 82, 88
Levitin, D. J., 233, 241
Levy, Y., 223
Leyfer, O. T., 240
Liebowitz, D., 118
LIMKI gene, 247
Liver
enzymes in Turner syndrome, 22
methylmalonic and propionic acidemia and, 309
transplantation in urea cycle disorders, 307–308
Lopez, S., 407
Lunsky, Y., 406
Lymphatic development in Turner syndrome, 9–10
Lymphedema, 14f, 15, 18, 20
Lyon, M. F., 9
Macrocephaly in neurofibromatosis, 148–149
Magnetic resonance imaging
of congenital hypothyroidism, 274
of glutaric acidemia type I, 306–307
of maple syrup urine disease, 306
of neurofibromatosis, 136–137, 146–147, 147–149, 149–151
of phenylketonuria, 301
Magnitude estimation and DS22q11.2, 183–185
Manganese poisoning, 352–356
Maple syrup urine disease (MSUD), 305–306
Masataka, N., 219, 220
Mathematics skills
Barth syndrome and, 322
DS22q11.2 and, 180–181, 183–185
fragile X syndrome and, 90
Turner syndrome and, 28
Mazzocco, M. M. M., 28, 82, 88, 90, 144
McCauley, E., 30–31
McDonald-McGinn, D. M., 451, 452
McLeod, D. R., 82
McMillan, D. L., 404
McWilliam, R. A., 445, 448
Mediation, 487–488
Medium chain acylCoA dehydrogenase deficiency, 317–318
Meltzoff, A. N., 220
Memory
congenital hypothyroidism and, 283–284
DS22q11.2 and, 176, 177–178
Duchenne muscular dystrophy and, 115
fragile X syndrome and, 88–89
Klinefelter syndrome and, 464
Williams syndrome and, 226–229
Mental retardation. See also Intelligence and IQ adaptation during childhood, 402–403
adaptive behaviors and, 391–393, 408
behavioral phenotypes, 403–404
civil rights and, 398–399
cystathionine beta-synthase deficiency and, 303–304
destitutionalization and, 397
diagnosis of, 399–400, 401–402
diagnostic and etiological challenges, 393
dimensions of classification, 396–397
diverse etiology, 393–394
family characteristics and resources, 404–406
fragile X syndrome and, 81, 82
heterogeneity of functioning and, 391–393
hypotonia and, 321–322
mainstreaming and inclusion and, 398
medical treatment and, 397–398
methylmalonic and propionic acidemia and, 309–310
mild, 403
moderate/severe/profound, 403
multicultural issues and, 407
neonatal screening for, 399–400
phenylketonuria and, 300
prenatal screening for, 399
range in functioning of those with, 400–401
seizures and, 324
social support for, 406
stigmatization of, 395
uncertainty of prognosis in, 395–396
Mercury
autism and, 351–352
clinical effects of, 346, 347–352
elemental, 345–346
fetal effects of, 350–351
high level effects of, 347–349
historical uses and misuses of, 344
methyl-, 346–352
moderate to low level effects of, 349–351
Merrick, John, 133–134
Metabolic syndrome in Turner syndrome, 23
Methylmalonic and propionic acidemia, 308–310
Methylmercury, 346–352
Methylphenidate/Ritalin, 96
Meyer-Lindenberg, A., 222, 239, 242, 247
Microcephaly
Klinefelter syndrome, 61–62
Williams syndrome, 203
Miller, L. J., 82
Mirrett, P. L., 456, 457
Mishkin, J. V., 25
Mismatch negativity (MMN) signal, 187
Mitochondrial disorders, 315–316
Molybdenum cofactor deficiency, 319–320
Morris, C. A., 247
Mosaicism, 7, 9–10, 81
Motor function
congenital hypothyroidism and, 280–281
DS22q11.2 and, 178–179, 452
Klinefelter syndrome and, 52–55
Williams syndrome and, 204–205
Mullen Scales of Early Learning (MSEL), 30, 213t, 217
Multicultural issues and mental retardation, 407
Multiple schwannomatosis. See Neurofibromatosis (NF)
Munir, F., 85, 88
Murphy, D. G., 28
Murphy, M. M., 90
Musicality and Williams syndrome, 233–234
Myelin abnormalities, 300–301, 324–326

Neisworth, J. T., 445
NEO Five Factor Inventory, 405
Netley, C., 55, 60–61

Neurocognition
congenital hypothyroidism and, 278–284
DS22q11.2 and, 170–173, 179–188
Duchenne muscular dystrophy and, 114–117, 125–127
fragile X syndrome and, 86–91
galactosemia and, 311
Klinefelter syndrome and, 55–59
neurofibromatosis and, 142–146
relations between language and, 225–227
Turner syndrome and, 10, 23–30
Williams syndrome and, 208–210, 227–234, 248–249

Neurofibromas, 139–140
Neurofibromatosis (NF)
brain structure/function correlates, 146–147
brain tumors and, 147
case studies, 151–156
clinical features, 135–136
definition, 133–134
diagnosis, 135–137
epidemiology, 134
functional imaging studies of, 149–151
genetic counseling and, 375–377
influences on central nervous system morphology, 141–142
intellectual functioning in, 143
learning and academic achievement profile in, 143–145, 150–151, 156–157, 419, 420–422
macrocephaly and, 148–149
MR hyperintensities and, 147–148
neurocognitive status of children with, 142–146
NF-1 gene, 137–139
systemic impact of, 139–140
visual-spatial processing in, 145–146

Neuroimaging
functional, 62
structural, 61–62
Williams syndrome, 204

Neuropsychological profiles of DS22q11.2, 173–174
Neurotoxic metals
lead, 336–344
manganese, 352–356
mercury, 344–352
Neurotransmitter disorders, 322–323

No Child Left Behind Act, 441–442, 473
Nonketotic hyperglycinemia, 318–319
Nonverbal learning disability (NLD), 415
etiologies and discrepant profiles, 425–431
Turner syndrome and, 27, 29f
Number processing in fragile X syndrome, 90

Occupational therapy
Duchenne muscular dystrophy, 111–112
fragile X syndrome, 95
O'Neill, M. J., 11
Online Mendelian Inheritance of Man (OMIM), 11, 368
Opperman, S., 385
Organic acid metabolism disorders
methylmalonic and propionic acidemia, 308–310
Orthodontic problems in Turner syndrome, 14f, 22–23
Osborne, L. R., 247
Otitis media, chronic, 203–204
Ovarian function in Turner syndrome, 10

Pagani, B., 232
Pagon, R. A., 239
Pankau, R., 234

Parents. See also Family
due process for, 487–488
role in IEPs, 479, 485
Parker, S. K., 352
Pattern construction and Williams syndrome, 231–232
Pavlakis, S. G., 316
Peabody Picture Vocabulary Test (PPVT-III), 214f, 217
Peers, classroom, 484
Percutaneous umbilical blood sampling, 369–370
Peregrine, E., 225
Peroxosomal disorders, 322

Personality. See Behavioral characteristics
Pharmacologic therapy
Duchenne muscular dystrophy, 110–112
fragile X syndrome, 96, 97, 454–455, 457
Phenylketonuria (PKU), 298–303, 418
Philosky, A., 84

Physical presentation
chromosome 22q11.2 deletion syndrome, 165–166, 167f
Duchenne muscular dystrophy, 108–110
fragile X syndrome, 73–76
Klinefelter syndrome, 47–49, 52, 53–55f
Turner syndrome, 12–17, 35f, 36
Williams syndrome, 201, 202f
Picker, J. D., 304
504 plans, 486
Pléh, C., 227
Plesa-Skwerer, D., 236
Plomin, R., 420, 423, 424
Posttraumatic stress disorder (PTSD) and fragile X syndrome, 83
Prader-Willi syndrome, 233
Pragmatics in Williams syndrome, 224–225
Prepulse inhibition (PPI), 187
Preschool services
DS22q11.2 and, 453
Prevalence rates
- chromosome 22q11.2 deletion syndrome, 164–165
- Duchenne muscular dystrophy, 105, 106
- fragile X syndrome, 73
- Klinefelter syndrome, 47, 50
- Turner syndrome, 7–8
- Propionic and methylmalonic acidemia, 308–310

Protein metabolism disorders
- cystathionine beta-synthase deficiency, 303–305
- glutaric acidemia type I, 306–307
- maple syrup urine disease, 305–306
- phenylketonuria, 298–303
- tyrosinemia type II, 305
- urea cycle disorders, 307–308

Pseudoautosomal region (PAR), 3

Psychiatric disorders associated with DS22q11.2, 167–170

Psychosocial phenotypic expression
- congenital hypothyroidism, 274–276
- DS22q11.2, 452–453
- interventions for fragile X syndrome, 93–94
- Klinefelter syndrome, 59–60
- Turner syndrome, 23–24, 30–33
- Williams syndrome, 234–242

Puberty
- adrenarche in, 65
- Klinefelter syndrome and, 47–49, 65
- Turner syndrome and, 19–20
- Pyruvate dehydrogenase deficiency, 312

Quality of life and Duchenne muscular dystrophy, 124–125

Quigley, C. A., 31–32

Rabensteiner, B., 234
Raefski, A. S., 11
Ratcliffe, S., 55, 61
Recurrence risks, 384–385
Religious beliefs, 405
Renal malformations in Turner syndrome, 20
Respiratory chain disorders, 315–316
Rey-Osterreith Complex Figure Test, 59
Robbins, I., 83
Roberts, J., 82, 90, 91
Robinson, B. F., 226, 228
Rogers, S. J., 84
Rogol, A., 461
Role play, 95
Ross, J., 459, 460
Ross, M. T., 10, 26–27, 33, 82
Rourke, B. P., 27
Rovet, J., 28, 55, 459, 463
Rowe, M. L., 221
Rutter, M., 337
Sadler, L. S., 245
Salbenblatt, J. A., 60
Samango-Sprouse, C., 461
Sarimski, K., 239
Savendahl, L., 35
Scales of Independent Behavior-Revised (SIB-R), 216f, 217
Schizophrenia and DS22q11.2, 168, 189–191
School achievement. See Learning and academic achievement
Schoolcraft, S. A., 405, 406
Schowalter, J. E., 28
Schrimsher, G. W., 146
Seizure disorders, 318–321, 324
manganese and, 355
Self-contained classroom placement, 485
Semantic development and Williams syndrome, 221–222
Sener, R. N., 305
Shafer, J. W., 25
Shapiro, J., 407
Shen, D., 61
Shiloh, S., 367
Short stature homeobox-containing (SHOX) gene
- Klinefelter syndrome and, 52
- Turner syndrome and, 9, 16–17, 18–19
- Shprintzen, Robert, 163, 174
- Siblings of affected patients, 385–386, 404–405
- Single photon emission computed tomography (SPECT), 62
Skin disorders in Turner syndrome, 23
Skinner, M., 82
Skotko, B. G., 399
Skuse, D. H., 11, 27
Smith-Lemi-Opitz syndrome (SLOS), 323, 418
Smith-Magenis syndrome, 239–240
Sobin, C., 168, 179, 187
Social support and mental retardation, 406
Socioeconomic status
- of families with mentally retarded children, 405–406
- lead poisoning and, 337–338
Solot, C. B., 175, 177
Speech and language abilities. See Language and speech abilities
Spinal meningocoels, 141
Spinal muscular atrophy (SMA), 114
Stature
- Klinefelter syndrome and, 47–49
- Turner syndrome and, 9, 16–17, 18–19
- Williams syndrome and, 203
Stiers, P., 192
Stiles, J., 231
Strabismus
- Turner syndrome, 21
- Williams syndrome, 203
Stress and Duchenne muscular dystrophy, 122–123
Stroke-like episodes, 316
Structural neuroimaging, 61–62
Sudhalter, V., 82, 87, 394
Sullivan, K., 236, 237
Supravalvar aortic stenosis (SVAS), 205
Swilen, A., 30, 168
Symons, F. J., 456
Tager-Flusberg, H., 236, 237
Tang, C. S., 407
Temple, C. M., 223
Tercyak, K., 383
Test for Reception of Grammar (TROG), 218
Testicular failure in Klinefelter syndrome, 47–49
Test of Relational Concepts (TRC), 218
Test of Visuomotor Integration, 230
Testosterone levels and Klinefelter syndrome, 51–52, 58, 59–60, 62–64
Thomas, A., 234
Thyroid. See also Hypothyroidism
disease in Turner syndrome, 22
dysgenesis, 267–268
dyshormonogenesis, 268–270
hormone
brain development and, 272
resistance, 270–271
Timeline, IEP, 488–489
Toddler Turner Study Group, 31–32
Tomc, S. A., 234
Transient hypothyroidism, 271
Transition from secondary to postsecondary education, 483
Trivette, C. M., 446
Tumors, brain, 147
Turk, J., 83, 85
Turner syndrome
brain imaging in, 24–25
cardiovascular malformations and hypertension in, 20
comprehensive assessment strategies in, 33–34
definition, 3–5
diabetes mellitus in, 23
diagnosis in adulthood, 16
diagnosis in childhood, 15–18
early identification enhancement, 34–36
early intervention and special education, 458–460
functional disomy and, 11
gastrointestinal disease in, 21–22
gonadal dysgenesis in, 19–20
hearing loss in, 21
heterogeneity of expression in, 32–33
imprinting and, 11
karyotype abnormalities, 5, 6f, 7t
key physical findings, 12–17
learning disability and, 419
lymphatics in, 9–10
lymphedema in, 14f, 15, 18, 20
medical issues associated with, 18–30
neurocognition in, 10, 23–30
neurocognitive manifestations in early childhood, 29–30
neurocognitive manifestations in school-age children and adolescents, 25–28, 29t
orthodontic problems in, 14f, 22–23
ovarian function in, 10
pathophysiology, 8–10
physical characteristics in, 3–5, 35f, 36
prevalence of, 7–8
psychosocial manifestations of, 23–24, 30–33
renal malformations in, 20
short stature and, 18–19
SHOX gene and, 9, 16–17, 18–19
skin disorders in, 23
stature in, 9
strabismus in, 21
x-linked recessive disorders and, 11
Tyrosinemia type II, 305
Ungerleider, L. G., 25
Urea cycle disorders, 307–308
Urinary tract malformations in Williams syndrome, 205–206
Uttal, D. H., 424
Van Leishout, C. F. M., 234
Vicari, S., 219, 227
Vignos, P. J., 118
Vineland Social-Emotional Early Scales, 32
Visual aids, 95, 482
Visual-motor coordination in fragile X syndrome, 89
Visual-spatial impairment
congenital hypothyroidism and, 279–280
DS22q11.2 and, 181–182
neurofibromatosis and, 137, 144–146
Williams syndrome and, 229–233
Volterra, V., 223
Von Arnim, G., 234
Von Recklinghausen, Friedrich Daniel, 133
Walker, S. O., 420, 423, 424
Walsh, V., 222
Wang, P. P., 174, 177
Warwick, M. M., 62
Wassink, T. H., 27
White, S. M., 234
Williams syndrome
attention deficit/hyperactivity disorder and, 206, 241–242
autism spectrum disorder and, 242
cardiocvascular disease and, 245
definition, 199
deleted genes expressed in the brain, 247–248
deletion length, 246
drawing abilities and, 230–231

Index
early diagnosis, 206–208
early language and communicative development
in, 219–221
experimental studies of personality in, 235–239
genetics and genotype-phenotype correlations in,
242–243
as a genomic disorder, 243
intelligence in, 210–212, 246–247
language, cognition, personality, and behavior in,
208–210
medical problems and therapeutic
recommendations, 201, 203–206
memory in, 226–229
musicality in, 233–234
neurocognition in, 208–210, 227–234, 248–249
pattern construction and, 231–232
physical features, 201, 202f
prevalence of symptoms of, 200–201t
problem behaviors in, 239–240
psychopathology and, 240–242
questionnaire studies of personality and
temperament, 234–235
relations between language and cognitive abilities
in, 225–227
semantic development, 221–223
short deletions, 246–247
spatial cognition in, 229–230
standardized assessments and, 210–219
variability in phenotype, 243–245
Williams Syndrome Cognitive Profile (WSCP), 214
Winneke, G., 337
Wolery, M., 447
Woodhead, M., 83
Woodin, M. F., 177
Worden, D. K., 118
Wu, J. Y., 121

X-inactivation, 8–9
X-linked recessive disorders, 11

Yap, S., 304
Yorifujii, T., 27

Zackai, E., 451
Zukowski, A., 218, 222