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 Anxietv 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 Billingsley, R. L., 149, 150 Bishop, D., 224 Bjugstad, K. B., 306 Blacher, J., 407 Bleeding, gastrointestinal, 22 Boccia, M. L., 82 Boutet, I., 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 mercurv 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 - 107imprinting 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 Daniels, M., 168, 179 Davenport, M. L., 31-32, 35, 430, 459 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 - 170Duchenne muscular dystrophy, 106 genetic counseling beyond, 378-379 genetic counseling immediately before and after, 367-370, 374-378 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 prenatal genetic, 369-370 recurrence risk and, 384–385 and siblings 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 Dovle, 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 Dyshormonogenesis, 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 Elastin 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 Eve 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

500

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 GTF21 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 early 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 creatine 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

phenyketonuria (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 Jackendoff, R., 219 Johnson, M. H., 173 Jordan, H., 232 Judgment of Line Orientation test, 145-146 Jugular vein, 9 Karaviorgou, M., 187 Karmiloff-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 early intervention and special education, 460-464 endocrinology of, 51–52 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-Karpiak, K., 31, 82, 88 Levitin, D. J., 233, 241 Levy, Y., 223 Leyfer, O. T., 240 Liebowitz, D., 118 LIMK1 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 - 10Lymphedema, 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, 147-148, 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 deinstitutionalization 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 phenyketonuria 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 Mervis, C. B., 219, 223, 225, 226, 228, 230, 231, 235.238 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 clinical 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 - 142intellectual 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, 29t 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 - 23Osborne, 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 Peroxisomal 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 Philofsky, 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 - 165Duchenne 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 DS22g11.2, 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 - 125Quigley, 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, 221-223 Sener, R. N., 305 Shaffer, 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 - 123Stroke-like episodes, 316

Structural neuroimaging, 61-62 Sudhalter, V., 82, 87, 394 Sullivan, K., 236, 237 Supravalvar aortic stenosis (SVAS), 205 Swillen, 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 - 30neurocognitive 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 prenatal diagnosis, 13, 18 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 - 242autism spectrum disorder and, 242 cardiovascular disease and, 245 definition, 199 deleted genes expressed in the brain, 247-248 deletion length, 246 drawing abilities and, 230-231

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 - 210medical 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 Yorifuji, T., 27

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