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ASSOCIATED XLID); CK SYNDROME; CLARK-BARAITSER SYNDROME; COFFIN-LOWRY SYNDROME; CORNELIA DE LANGE SYNDROME, X-LINKED; CRANIOFACIOSKELETAL SYNDROME
CREATINE TRANSPORTER DEFICIENCYDUCHENNE MUSCULAR DYSTROPHY; DYSKERATOSIS CONGENITA; EPILEPSY-INTELLECTUAL DISABILITY IN FEMALES (EIDF); FITZSIMMONS SYNDROME; FLNA-ASSOCIATED XLID; FRAGILE X SYNDROME; GIUFFRE-TSUKAHARA SYNDROME; GLYCEROL KINASE DEFICIENCY; GOLABI-ITO-HALL SYNDROME: (SEE ALSO RENPENNING SYNDROME); GOLDBLATT SPASTIC PARAPLEGIA SYNDROME; GOLTZ SYNDROME; GRAHAM ANOPHTHALMIA SYNDROME; GUSTAVSON SYNDROME; HALL OROFACIAL SYNDROME; HEREDITARY BULLOUS DYSTROPHY, X-LINKED; HOLMES-GANG SYNDROME (SEE ALSO ATRX-ASSOCIATED XLID); HOMFRAY SEIZURES-CONTRACTURES; HYDE-FORSTER SYNDROME
HYDRANENCEPHALY WITH ABNORMAL GENITALIA (SEE ALSO ARX-ASSOCIATED XLID)HYDROCEPHALY-CEREBELLAR AGENESIS SYNDROME; HYDROCEPHALY-MASA SPECTRUM; HYPOPARATHYROIDISM, X-LINKED; INCONTINENTIA PIGMENTI; JUBERG-MARSIDI-BROOKS SYNDROME; KANG SYNDROME; LENZ MICROPHTHALMIA SYNDROME; LESCH-NYHAN SYNDROME; LISSENCEPHALY AND ABNORMAL GENITALIA, X-LINKED (SEE ALSO ARX-ASSOCIATED XLID); LISSENCEPHALY, X-LINKED; LOWE SYNDROME; LUJAN SYNDROME; MARTIN-PROBST SYNDROME; MEHMO SYNDROME; MENKES SYNDROME; MIDAS SYNDROME; MILES-CARPENTER SYNDROME; MOHR-TRANEBJAERG SYNDROME; MONOAMINE OXIDASE-A DEFICIENCY
MUCOPOLYSACCHARIDOSIS IIAMYOTUBULAR MYOPATHY; N-ALPHA-ACETYLTRANSFERASE DEFICIENCY; NANCE-HORAN SYNDROME; NORRIE DISEASE; OPITZ FG SYNDROME; OPTIC ATROPHY, X-LINKED; ORAL-FACIAL-DIGITAL SYNDROME I; ORNITHINE TRANSCARBAMOYLASE DEFICIENCY; OTOPALATODIGITAL SYNDROME I (SEE ALSO FLNA-ASSOCIATED XLID); OTOPALATODIGITAL SYNDROME II (SEE ALSO FLNA-ASSOCIATED XLID); PAINE SYNDROME; PALLISTER W SYNDROME; PARTINGTON SYNDROME (SEE ALSO ARX-ASSOCIATED XLID); PELIZAEUS-MERZBACHER SYNDROME; PERIVENTRICULAR NODULAR HETEROTOPIA (SEE ALSO FLNA-ASSOCIATED XLID); PETTIGREW SYNDROME
PHOSPHOGLYCERATE KINASE DEFICIENCY

Sommario/riassunto

The Atlas of X-Linked Intellectual Disability Syndromes is a comprehensive and up-to-date summary of the clinically distinctive disorders caused by genes on the X chromosome. Clinical and laboratory data on 150 syndromes are presented in a concise and consistent manner. Each syndrome is defined and information is provided on somatic features, growth and development, neurological signs, cognitive performance, imaging and other laboratory findings, and when possible, the nature and localization of the responsible gene. Craniofacial and other somatic findings are extensively illustrated. A differ