

1. Record Nr.	UNINA9910453137303321
Autore	Stevenson Roger E. <1940->
Titolo	Atlas of X-linked intellectual disability syndromes // Roger E. Stevenson, Charles E. Schwartz, and R. Curtis Rogers
Pubbl/distr/stampa	New York : , : Oxford University Press, , [2012] ©2012
ISBN	0-19-997524-8 0-19-981186-5
Edizione	[Second edition.]
Descrizione fisica	1 online resource (363 p.)
Altri autori (Persone)	SchwartzCharles E RogersR. Curtis <1953-> (Richard Curtis) StevensonRoger E. <1940->
Disciplina	616.85/88042
Soggetti	X-linked mental retardation Intellectual disability Electronic books.
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Rev. edition of: X-linked mental retardation / Roger E. Stevenson, Charles E. Schwartz, Richard J. Schroer. 2000.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Cover; TABLE OF CONTENTS; FOREWORD; PREFACE; AARSKOG SYNDROME; ABIDI SYNDROME; ADRENOLEUKODYSTROPHY; AGENESIS OF THE CORPUS CALLOSUM, X-LINKED; AHMAD SYNDROME; AICARDI SYNDROME; ALLAN-HERNDON-DUDLEY SYNDROME; ALPHA-THALASSEMIA INTELLECTUAL DISABILITY (SEE ALSO ATRX-ASSOCIATED XLID); AP1S2-ASSOCIATED XLID; APAK ATAXIA-SPASTIC DIPLEGIA SYNDROME; ARMFIELD SYNDROME; ARTS SYNDROME; ARX-ASSOCIATED XLID; ATAXIA-DEAFNESS-DEMENTIA, X-LINKED; ATKIN-FLAITZ SYNDROME; ATRX-ASSOCIATED XLID; BERGIA CARDIOMYOPATHY; BERTINI SYNDROME; BORJESON-FORSSMAN-LEHMANN SYNDROME; BRANCHIAL ARCH SYNDROME, X-LINKED CANTU SYNDROME; CARPENTER-WAZIRI SYNDROME (SEE ALSO ATRX-ASSOCIATED XLID); CEREBRO-CEREBELLO-COLOBOMA SYNDROME; CEREBRO-OCULO-GENITAL SYNDROME; CEREBRO-PALATO-CARDIAC SYNDROME (SEE ALSO RENPENNING SYNDROME); CHARCOT-MARIE-TOOTH NEUROPATHY, COWCHOCK VARIANT; CHARCOT-MARIE-TOOTH

NEUROPATHY, IONASESCU VARIANT; CHASSAING-LACOMBE
CHONDRODYSPLASIA; CHRISTIAN SYNDROME; CHRISTIANSON
SYNDROME; CHUDLEY-LOWRY SYNDROME (SEE ALSO ATRX-
ASSOCIATED XLID); CK SYNDROME; CLARK-BARAITSER SYNDROME;
COFFIN-LOWRY SYNDROME; CORNELIA DE LANGE SYNDROME, X-
LINKED; CRANIOFACIOSKELETAL SYNDROME
CREATINE TRANSPORTER DEFICIENCY
DUCHENNE 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 II
AMYOTUBULAR 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
