1. Record Nr. UNINA9910893549003321

Titolo	Jahresbericht / Universität Stuttgart : Berichtszeitraum ... = Annual report / Universität Stuttgart : period covered by report: .
Pubbl/distr/stampa	Stuttgart, : Univ., 2009-
Descrizione fisica	Online-Ressource
Disciplina	370
Soggetti	Zeitschrift
Lingua di pubblicazione	Tedesco
Formato	Materiale a stampa
Livello bibliografico	Periodico
Note generali	Gesehen am 06.10.20

2. Record Nr. UNINA9911019144803321

Titolo	Lysosomal storage disorders : a practical guide / / edited by Atul Mehta, Bryan Winchester
Pubbl/distr/stampa	Chichester, West Sussex, : Wiley-Blackwell, 2012, c2013
ISBN	9781118514672 111851467X 9781283644419 128364441X 9781118514696 1118514696 9781118514641 1118514645
Descrizione fisica	1 online resource (209 p.)
Altri autori (Persone)	MehtaAtul B WinchesterBryan
Disciplina	571.6/55
Soggetti	Lysosomal storage diseases Metabolism - Disorders
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa

Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Lysosomal Storage Disorders: A Practical Guide; Copyright; Contents; List of Contributors; Preface; Foreword; Part 1 General Aspects of Lysosomal Storage Diseases; 1 The Lysosomal System: Physiology and Pathology; Introduction; The greater lysosomal system; Lysosomal diseases; References; 2 Clinical Aspects and Clinical Diagnosis; Introduction; Clinical presentation; References; 3 Laboratory Diagnosis of Lysosomal Storage Diseases; Referral to specialist laboratory; Preliminary screening tests on urine or blood; Diagnosis of lysosomal enzyme defects LSDs due to defects in non-enzymatic proteinsNeuronal ceroid lipofuscinoses (NCLs); Molecular genetic testing; Prenatal diagnosis; Prospects; Acknowledgements; References; 4 Genetics of Lysosomal Storage Disorders and Counselling; Introduction; Genes, proteins, stored substrates, clinical phenotypes and diagnosis; Incidence and prevalence; Populations at a high-risk; Burden of illness; Population screening and diagnostic methods; Counselling issues; References; 5 Classification of Lysosomal Storage Diseases; Basis of classification of lysosomal storage diseases; Acknowledgements Part 2 The Individual Diseases6 Gaucher Disease; A representative case history; Gaucher disease; Epidemiology; Etiology and pathogenesis: genetic basis; Clinical forms; Diagnosis; Biomarkers; Routine follow-up of patients; Enzyme replacement therapy (ERT) for Gaucher disease: alglucerase and imiglucerase; Dosing regimens; Malignancies; Global shortage of imiglucerase (June 2009); Other treatment options: substrater eduction therapy (SRT); Summary; References; 7 Fabry Disease; Epidemiology; Genetic basis; Pathophysiology; Clinical presentation; Natural history; Laboratory diagnosis; Treatment Treatment guidelinesFurther reading; 8 The Gangliosidoses; References; 9 Metachromatic Leukodystrophy and Globoid Cell Leukodystrophy; MLD and GLD; Case studies; Epidemiology; Genetics; Pathophysiology; Clinical presentation; Diagnosis by MRI; Laboratory diagnosis; Treatment; Natural history studies; References; 10 Types A and B Niemann-Pick Disease; Representative case histories; Epidemiology; Genetics; Pathophysiology; Clinical presentation; Natural history; Laboratory diagnosis; Treatment; Acknowledgements and conflicts of interest; References; 11 Niemann-Pick Disease Type C Case historiesEpidemiology; Genetic basis; Pathophysiology; Clinical presentation; Natural history; Laboratory diagnosis; Treatment; References; 12 The Mucopolysaccharidoses; Epidemiology; Genetic basis; Pathophysiology; Clinical presentation; Natural history; Laboratory diagnosis; Treatment; Acknowledgements; References; 13 Pompe Disease; Case histories; Confusing nomenclature; Epidemiology; Genetic basis; Pathophysiology; Clinical presentation; Natural history; Enzymatic and molecular diagnosis; Treatment; Acknowledgment; Selected literature; 14 Glycoproteinoses; Epidemiology Pathophysiology
Sommario/riassunto	Awareness of lysomal storage disorders needs to be raised and there is very substantial pharmaceutical interest to do so. The disorders are often viewed as obscurities but in fact they are treatable. Enzyme replacement therapy is available for four of the disorders and will be available for a further three disorders in the course of the next year. Substrate reduction therapy is licensed for one of them but in the course of the next 12 months it will be licensed for two others and a new form of substrate reduction therapy is being introduced. These

diseases present to a very wide range