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Disorders A Practical

The last two decades have
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Lysosomal storage disorders, which has substantially extended our understanding of both the scientific and the clinical basis of these diseases. Lysosomal Storage Disorders: A Practical Guide is the fruit of an ambitious

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There are more than 40
lysosomal storage diseases

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(LSDs) of humans, many of which are also found in domestic animals. 3, 17, 20 Most are caused by a primary lysosomal hydrolase defect, but others ...

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Lysosomal Storage Disorders (LSDs) comprise a group of disorders causing defects at the organelle and sub-organelle level with a wide range of pathophysiologies

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Practical Guide and clinical consequences. Signs and symptoms of LSDs involve multiple organ systems. The

Practical management of lysosomal storage disorders (LSDs ...

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Lysosomal storage disorders are a group of more than 50 rare diseases. They affect the lysosome -- a structure in your cells that breaks down substances such as proteins, carbohydrates, and old...

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Lysosomal Storage Disorders:
Causes, Symptoms, Diagnosis
...

Lysosomal storage diseases
are a group of about 50 rare
inherited metabolic
disorders that result from

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defects in lysosomal

function. Lysosomes are sacs of enzymes within cells that digest large molecules and pass the fragments on to other parts of the cell for recycling. This process requires several critical

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Practical Guide enzymes. If one of these enzymes is defective due to a mutation, the large molecules accumulate within the cell, eventually killing it. Lysosomal storage disorders are caused by lysosomal

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Lysosomal storage disease - Wikipedia

In each case, lysosomal storage diseases are caused by an inborn error of metabolism that results in the absence or deficiency of

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an enzyme, leading to the inappropriate storage of material in various cells of the body. Most lysosomal storage disorders are inherited in an autosomal recessive manner.

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A Practical Guide is the
fruit of an ambitious
project aiming to review
both the scientific and the

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lysosomal storage disorders,
resulting in this accessible
volume, which gives an up-to-
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of lysosomal storage disorders
needs to be raised and there
is very substantial
pharmaceutical interest to

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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

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Lysosomal storage diseases
are a group of rare
inherited metabolic

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Lysosomal Disorders |

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Lysosomal storage disorders
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Awareness of lysosomal storage
disorders needs to be raised

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substantial pharmaceutical interest to do so. The disorders are often viewed as obscurities but in fact they are treatable. ...

Lysosomal storage disorders

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...

It is quite difficult to diagnose this disorder as the symptoms vary from one type to another type of Lysosomal Storage Disorders. There are a few diagnoses

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Practical Guide carried out in a specialized laboratory. Tissue biopsies are also used for diagnosis of Lysosomal Storage Disorder in the patient by analyzing the sample of tissue or cells collected from the patient's body for

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Lysosomal Storage Disease
-Types, Symptoms, Causes and
its ...

Lysosomal storage diseases
(LSDs) are due to the
inherited deficiency of one

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of over 40 lysosomal enzymes, and lead to accumulation of undegraded substrate in a range of organs and tissues. They are multisystem and progressive disorders.

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Practical Guide extended our understanding of both the scientific and the clinical basis of these diseases. Lysosomal Storage Disorders: A Practical Guide is the fruit of an ambitious project aiming to review both the scientific and the

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Practical Guide of lysosomal storage disorders, resulting in this accessible volume, which gives an up-to-date overview of the subject. There is substantial scientific interest in these diseases:

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Practical Guide new advances in small molecule therapy are likely to be useful in the near future, and trials are already underway. Lysosomal storage disorders offer a unique platform for teaching modern clinical science,

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from basic genetics through to clinical applications. The first part of the book reviews and classifies our current understanding of the physiology and pathophysiology of lysosomal storage disorders. The

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Second part of the book reviews individual diseases, and gives perspectives from patients and experts looking towards future therapeutic directions. Lysosomal Storage Disorders: A Practical Guide is the ideal

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Enables readers to gain both

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holistic and specific knowledge on the topic of Lysosomal Storage Disorders. In the past few decades, we have witnessed a 'golden age' of Lysosomal Storage Disorders (LSDs) clinical care, diagnosis, and

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Research and this book is a well-timed review of the rapidly developing subject of LSDs. Contributions from many leading scientists and clinicians in the field provide the reader with a completely comprehensive

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Practical Guide overview of the subject.

Topics covered in the book include: The general aspects of LSDs, with special attention paid to physiology and pathology Clinical and laboratory diagnosis, including newborn screening

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and the genetics of LSDs
Individual LSDs, such as the
various sphingolipidoses,
mucopolysaccharidoses,
glycogen storage disease,
glycoproteinoses,
galactosialidosis, and
neuronal ceroid

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Lipofuscinoses Other disorders of the lysosome, including those involving defects in the lysosomal membrane and in the emerging roles of the lysosome in cellular metabolism The existing and emerging

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Practical Guide treatments for LSDs Key patient issues such as availability and disease awareness, including in the Third World. For researchers and industry professionals in the field of LSDs, this book serves as a completely

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Practical Guide comprehensive reference work to understand both theoretical concepts and how they may be used in practical applications. For students of LSDs, it provides a solid base of foundational knowledge and

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Practical Guide serves as a practical guide.

This book presents an overview of lysosomal storage disorders, and provides the reader with an

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Practical Guide and prospective therapeutic strategies. The book is written by an expert in the field who has been engaged in both basic and clinical research, in addition to having extensive practical experience in patient care.

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It is written from the perspective of someone who entered the field just as treatment was being introduced, and who has been engaged in the seminal clinical trials and the development of therapeutic

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Practical Guide. It offers a broad perspective and should appeal to both novices and experts in the field who seek a single resource that provides a comprehensive picture of relevant topics on this subject. A multi-

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Practical Guide, the author addresses the issue of diagnosis and patient management, underlying mechanisms of disease, sources of morbidity and treatment options, covering issues of interest to both

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the basic scientist and the
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As clinical management of inherited metabolic diseases (IMDs) has improved, more patients affected by these

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Conditions are surviving into adulthood. This trend, coupled with the widespread recognition that IMDs can present differently and for the first time during adulthood, makes the need for a working knowledge of

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important than ever.

Inherited Metabolic Disease
in Adults offers an
authoritative clinical guide
to the adult manifestations
of these challenging and
myriad conditions. These

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Practical Guide include both the classic pediatric-onset conditions and a number of new diseases that can manifest at any age. It is the first book to give a clear and concise overview of how this group of conditions affects adult

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of haematology.

The neuronal ceroid lipofuscinoses are an extremely rare group of inherited neurodegenerative diseases that primarily

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affect children. Core symptoms of these conditions typically include epilepsy, cognitive decline and visual failure. These diseases are so rare that professionals who come into contact with them need a consultative

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lipofuscinoses will prove

useful for clinicians,

family physicians, research

scientists, diagnostic

laboratories, families

affected by the disease as

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well as by workers in
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In September of 2007 Gaucher Disease received a commendation in the Haematology category of the

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Competition! Although rare in the general population, Gaucher disease is the most prevalent of the lysosomal storage disorders, making research into this

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Particular orphan disorder an invaluable prototype for the diagnosis, research, and treatment of others. Like many rare conditions, Gaucher disease has benefited from the explosion of medical research in the

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