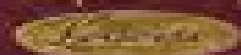


PHYSICIANS' GUIDE TO RARE DISEASES

Edited by Jess G. Thorne, M.D.

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Physicians Guide To Rare Diseases

**Michael H. Davidson, Lane
Benes, Anthony S. Wierzbicki**



Physicians Guide To Rare Diseases:

Physicians' Guide to Rare Diseases Jess G. Thoene, Doris C. Smith, National Organization for Rare Disorders, 1992
Guide to diagnosing and treating rare diseases one affecting fewer than 200 000 Americans **Physicians' Guide to Rare Diseases** Jess G. Thoene, 1995 This classic reference now with almost 900 disease descriptions is edited by nationally known pediatrician Jess G Thoene M D provides a quick organized approach to identifying puzzling unusual symptoms giving patients the opportunity for early diagnosis treatment Special features include A full color atlas of visual clues cross referenced to disease listings Thirteen separate specialty sections with an expert overview to introduce each section Eminent clinicians from the outstanding research centers in rare diseases give you the current status of research practical approaches to diagnosis therapy of the rare diseases in that specialty Full reference to more than 250 orphan drugs Each drug is cross referenced to the appropriate disease description Clear language for the Layperson Though written for physicians the descriptions of diseases symptoms treatments are clear understandable for patients educators others outside the medical profession Each disease description provides symptomatology etiology affected population a comparison of related disorders Each disease description contains a list of other resources to consult voluntary agencies units of the National Institutes of Health research centers around the country etc Addresses phone numbers are provided in a central reference Library Journal
PHYSICIAN S GUIDE is recommended for medical collections public libraries with consumer health collections

Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases N. Blau, M. Duran, M.E. Blaskovics, K.M. Gibson, 2012-12-06 Delay and mistakes in the diagnosis of inherited metabolic diseases may have devastating consequences Reference laboratory data are scattered and clinical descriptions of rare conditions are hard to locate This book describes 298 disorders grouped into 35 chapters according to the type of condition Within each group of disorders chapters provide tables of pertinent clinical findings as well as reference and pathological values for crucial metabolites Relevant metabolic pathways and diagnostic flow charts are included There are four indices to make the book as user friendly as possible Disorders index Signs and symptoms index Organs index and Tests index The Physician s Guide provides paediatricians and other physicians with a unique aid to help them select the correct diagnosis from a bewildering array of complex clinical and laboratory data *Physician's Guide to the Treatment and Follow-Up of Metabolic Diseases* Nenad Blau, Georg F. Hoffmann, J.V. Leonard, Joe T. R. Clarke, 2006-01-16 The greatest difficulty in life is to make knowledge effective to convert it into practical wisdom Sir William Osler The inborn errors of metabolism as a group of metabolic diseases are relatively rare and are sometimes called orphan diseases As a group they account for about 1 in 2 500 births Applegarth et al 2000 and as a cumulative group reaching 20 years of age their prevalence is about 40 cases per 100 000 population In terms of patient days of continuous supervision and care hundreds of thousands of such days are involved per generation of these patients Although experience with these diseases as a class may be small and people expert in their management may be relatively

few in the years to come many caregivers will become involved This book offers help to them Until the mid twentieth century hereditary metabolic and other genetic diseases were considered to be purely genetic problems Destiny would take its course treatment did not exist and genetic counseling about recurrence risks was virtually all that could be offered Phenylketonuria PKU was then shown to be a treatable genetic disease in which early diagnosis and effective treatment prevented the disease mental retardation in PKU Other genetic diseases for which an environmental experience was an essential component of cause e g exposure to a dietary component or a drug were then seen to yield to treatment NORD Guide to Rare Disorders National Organization for Rare Disorders,2003 NORD Guide to Rare Disorders is a comprehensive practical authoritative guide to the diagnosis and management of more than 800 rare diseases The diseases are discussed in a uniform easy to follow format a brief description signs and symptoms etiology related disorders epidemiology standard treatment investigational treatment resources and references The book includes a complete directory of orphan drugs a full color atlas of visual diagnostic signs and a Master Resource List of support groups and helpful organizations An index of symptoms and key words offers physicians valuable assistance in finding the information they need quickly **Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases** Nenad Blau,Carlo Dionisi Vici,Carlos R. Ferreira,Christine Vianey-Saban,Clara D. M. van Karnebeek,2022-02-21 This updated and enlarged second edition is a unique source of information on the diagnosis treatment and follow up of metabolic diseases The clinical and laboratory data characteristic of rare metabolic conditions can be bewildering for clinicians and laboratory personnel alike reference laboratory data is scattered and clinical descriptions can be obscure The new Physician s Guide with the additional more than 600 diseases now featured documents 1200 conditions grouped according to type of disorder organ system affected e g liver kidney etc or phenotype e g neurological hepatic etc It includes relevant clinical findings and highlights the pathological values for diagnostic metabolites Guidance on appropriate biochemical genetic testing is also provided and established experimental therapeutic protocols are described with recommendations on follow up and monitoring The authors are acknowledged experts and the book is a valuable desk reference for all who deal with inherited metabolic diseases Chapter 73 is available open access under a Creative Commons Attribution 4 0 International License via link [springer.com](https://www.springer.com)

Physician's Guide to Terrorist Attack Michael J. Roy,2003-10-27 Leading medical experts from the military and other branches of government comprehensively review the possible infectious disease and chemical agents that can be used in terrorist attack and the proper diagnostic and therapeutic methods for dealing with them The book also discusses treating blast injuries the effects of nuclear and radiological weapons and the psychological effects of terrorist events Comprehensive and up to date Physician s Guide to Terrorist Attack provides physicians health care workers and emergency personnel with a practical handbook for confidently and effectively diagnosing and managing the victims of terrorist attack **The Busy Physician's Guide To Genetics, Genomics and Personalized Medicine** Kevin M. Sweet,Ron C. Michaelis,2011-04-23 In

the coming decade the focus of medicine will shift from a disease oriented approach where the physician prescribes according to the disease the patient has to a personalized approach in which the physician first considers the patient's individual biochemistry before prescribing a treatment. Personalized medicine has the potential to improve efficacy and safety in virtually all fields of medicine. Unfortunately few physicians feel confident in their ability to apply the principles of genetics and genomics upon which personalized medicine is based to their practice. This book is intended to help the practicing physician understand and apply the principles of genetic and genomic medicine regardless of his/her level of background in the field. It provides a thorough foundation review of classical genetic principles with an emphasis on how these principles apply to personalized medicine and common complex diseases. In addition it provides a wide ranging review of the inroads that personalized medicine has made into several fields including cancer, psychiatric disorders, cardiovascular disease, substance abuse, Alzheimer disease, respiratory diseases, type 2 diabetes and macular degeneration. Most importantly this book is intended to enable the practicing physician, physician assistants and their entire healthcare team to anticipate the developments that will emerge in the near future and stay current with the field as it expands.

The Physician's Guide to Delusional Infestation Gale E. Ridge, 2024-04-05. This unique ground breaking book is a comprehensive clinical guide to the psychiatric disorder of Delusional Infestation (DI). DI is a psychiatric illness characterized by patients holding a monothematic fixed belief of infestations on their skin, body or immediate environment, a belief that is not supported by objective medical evidence. Delusional beliefs may be shared among individuals and perceived pathogens include living organisms such as parasitic worms, bacteria, fungi and insects. Organic or non organic infiltration by fibres, threads or other inanimate particles or objects known or unknown to medical science are also described by patients. DI is severely underreported because of social stigma, limited physician awareness and patient doctor hopping. Written for physicians, the book offers a multidisciplinary, compassionate care model for patients burdened by this pernicious disorder. Other health professionals and administrators will find the work highly valuable as will professionals in fields such as veterinary medicine, pest management, entomology, parasitology and information services supported by academia. The Physician's Guide to Delusional Infestation assembles and organizes all current knowledge of DI and presents material in three parts. Part I introduces and reviews DI's historical background showing how our understanding of the condition has developed. Its concise sections include useful tables, illustrations, algorithms and reference lists for clinicians who work with DI patients. Part II covers differential diagnosis, treatment and ways to work effectively with DI patients. Since these patients are often high functioning, particular techniques of interviewing, joining with patients, sampling and laboratory testing are explained. International travel, physician fallibilities, possible alternative diagnoses, the Internet and veterinary and public health concerns are all addressed. Part III is a series of chapters written by experts in their fields. This section allows each author to present personal experiences from their respective professions, observations and opinions that show a variety of perspectives among the book's authors. Part III

includes a chapter on common human parasites with their biology behaviours and distributions The book will serve as a comprehensive yet concise reference for physicians whose patients have unusual baffling or implausible medical histories and physical examinations A soon to be gold standard resource The Physician s Guide to Delusional Infestation equips and empowers all physicians and many others interested in the topic with an expert comprehensive understanding of this complex disorder

Informed Consent United States. Congress. House. Committee on Small Business. Subcommittee on Regulation, Business Opportunities, and Technology,1993 Directory, Resources for Deafness and Other Communication Disorders ,1994 **National Library of Medicine Current Catalog** National Library of Medicine (U.S.),1993 Genetic Breakdown Stacey Price Brown,2008-02-19

We all get sick from time to time and have to go to the doctor The doctor prescribes medication we go home and take the medication with trust in their judgment that first we were diagnosed properly and secondly that the medication will work without causing serious bodily harm or even death In 1999 a pharmaceutical company took on a common problem that affects one in three Americans arthritis There are over one hundred different types of arthritis but nevertheless it can be a very painful debilitating disease and in some cases it can cause death Many were treating the problem with aspirin but they could not tolerate it because it caused gastrointestinal bleeding After clinical trials the pharmaceutical company discovered that this new drug not only caused sudden cardiac arrest but it also caused a host of other medical problems The manufacturer decided to market the drug anyway and thousands of Americans died or were injured but sadly enough many don t know why This book answers that question and many more Many people suffer from arthritis which causes an inflammatory response and pain in the joints There are three common types of arthritis rheumatoid osteoarthritis and infectious arthritis although there are over one hundred different known categories There is no cure for arthritis This disease attacks the joints and causes pain and stiffness The difference between Osteo and Rheumatoid is that the latter will not only destroy the joints but it will also affect the vocal cords causing hoarseness When the disease is finished running its course by systemically destroying the joints and surrounding muscle it will destroy organs and tissue as well Osteoarthritis is limited to joint destruction It will break down cartilage primarily affecting the hands feet and spine Most people suffering with infectious arthritis already have an existing joint problem that has exacerbated into infectious arthritis You ve heard the expression no pain no gain You don t have to have arthritis to reach for an aspirin Just the pain from working out in the gym or a hard day at the job may require an aspirin Usually it begins with minor joint pain that is treated with aspirin When we have a minor ache or pain we take an aspirin When we have a headache we take an aspirin You can go anywhere in the world and this is one pharmaceutical that has gained worldwide recognition and deservedly so The chemical name for Aspirin is acetylsalicylic acid and has been used for years as an analgesic which means it will relieve minor aches and pains It will also reduce fever and inflammation Aspirin has been the wonder drug and the answer to pain fever and inflammation for years As we already know the human body is very complex After we ingest

anything it goes directly to the stomach which is considered a hollow organ This organ contains gastric juices or enzymes which are prepared to digest and break down food liquids or medicine so that the human body can receive a benefit There are digestive enzymes present that break down whatever is in that stomach environment and changes it from large molecules into smaller ones so that they can move on into the smaller intestine Once there they can be absorbed by the human body In order for this to be achieved the stomach environment contains up to three liters of gastric juices Gastric juice is a hormone that comes from gastrin After it is broken down the molecules are called peptides or chemical bonds and then it is released directly into the bloodstream Think of the body as a computer It will detect that there are peptides in the stomach However if the body detects that there are too many peptides then the human computer will continue to send gastric juice and will not turn off until it feels that everything has been broken down The result is gastrointestinal bleeding Aspirin is in the class of nonsteroidal drugs The name Aspirin was first used by the German compa Encyclopedia of Special Education, Volume 1 Cecil R. Reynolds, Kimberly J. Vannest, Elaine Fletcher-Janzen, 2018-03-02 The only comprehensive reference devoted to special education The highly acclaimed Encyclopedia of Special Education addresses issues of importance ranging from theory to practice and is a critical reference for researchers as well as those working in the special education field This completely updated and comprehensive A Z reference includes about 200 new entries with increased attention given to those topics that have grown in importance since the publication of the third edition such as technology service delivery policies international issues neuropsychology and RTI The latest editions of assessment instruments frequently administered in special education settings are discussed Only encyclopedia or comprehensive reference devoted to special education Edited and written by leading researchers and scholars in the field New edition includes over 200 more entries than previous edition with increased attention given to those topics that have grown in importance since the publication of the third edition such as technology service delivery policies international issues neuropsychology and Response to Intervention Positive Behavioral Interventions and Supports PBIS Autism and Applied Behavior Analysis Entries will be updated to cover the latest editions of the assessment instruments frequently administered in special education settings Includes an international list of authors and descriptions of special education in 35 countries Includes technology and legal updates to reflect a rapidly changing environment Comprehensive and thoroughly up to date this is the essential A Z compilation of authoritative information on the education of those with special needs **Encyclopedia of Special Education, Volume 2** Cecil R. Reynolds, Kimberly J. Vannest, Elaine Fletcher-Janzen, 2018-03-06 The only comprehensive reference devoted to special education The highly acclaimed Encyclopedia of Special Education addresses issues of importance ranging from theory to practice and is a critical reference for researchers as well as those working in the special education field This completely updated and comprehensive A Z reference includes about 200 new entries with increased attention given to those topics that have grown in importance since the publication of the third edition such as technology service delivery policies international issues neuropsychology

and RTI The latest editions of assessment instruments frequently administered in special education settings are discussed Only encyclopedia or comprehensive reference devoted to special education Edited and written by leading researchers and scholars in the field New edition includes over 200 more entries than previous edition with increased attention given to those topics that have grown in importance since the publication of the third edition such as technology service delivery policies international issues neuropsychology and Response to Intervention Positive Behavioral Interventions and Supports PBIS Autism and Applied Behavior Analysis Entries will be updated to cover the latest editions of the assessment instruments frequently administered in special education settings Includes an international list of authors and descriptions of special education in 35 countries Includes technology and legal updates to reflect a rapidly changing environment Comprehensive and thoroughly up to date this is the essential A Z compilation of authoritative information on the education of those with special needs

Nutrition Management of Patients with Inherited Metabolic Disorders Acosta,2010-10-22 5 Stars Doody's Book ReviewWritten by the foremost nutrition experts in the United States Nutrition Management of Patients with Inherited Metabolic Disorders supplies the knowledge needed by nutritionists dietitians and other healthcare professionals who provide services to patients with inherited metabolic disorders IMDs Many IMDs have been managed by diet improving neurological and physical outcomes however nutrition problems still occur These problems could be due to the quality of the medical foods inadequate prescription by healthcare providers or poor diet adherence by the patient Nutrition Management of Patients with Inherited Metabolic Disorders examines these issues and helps medical food manufacturers medical geneticists nutritionists dietitians and other healthcare providers find alternative forms of supplying the nutrients that promote optimal health and nutrition for the patient Click here for a list of contributors This book is ideal for professionals working with patients with IMDs and it provides the basis for graduate level courses in dietetics nutrition genetics nursing genetics counseling and public health It is a valuable resource for medical students in their biochemistry courses and for postdoctoral fellows completing fellowships in genetics 2010 476 pages

Fast Facts: Familial Chylomicronemia Syndrome Michael H. Davidson,Lane Benes,Anthony S. Wierzbicki,2021-11-02 Familial chylomicronemia syndrome FCS is an ultra rare genetic disorder characterized by the abnormal build up of chylomicrons the largest type of lipoprotein which transport dietary fat from the gut to the rest of the body Patients with FCS often experience severe symptoms the most feared of which is acute potentially life threatening pancreatitis This resource is intended to raise awareness of FCS among all members of the healthcare team who come into contact with patients with FCS with the aim of earlier diagnosis and management thus preventing some of the more devastating physical neurological and cognitive symptoms of the disorder

Communicating Rare Diseases and Disorders in the Digital Age Costa, Liliana Vale,Oliveira, Sónia,2020-01-03 A primary concern of rare disease diagnosis is the lack of accurate information that may lead to delayed interventions administering inaccurate treatments and social consequences Health communication continues to be one way and rely on the expertise from the health

practitioner In such a broad spectrum of rare diseases patients may find it difficult to obtain timely information accurate diagnosis and appropriate treatments surgeries medications or psychological counseling in their own countries The use of information and communication technologies can create new communication channels that address this lack of knowledge Communicating Rare Diseases and Disorders in the Digital Age is an essential reference source that uses computer mediated communication to improve patient knowledge when afflicted or dealing with rare health conditions Featuring research on topics such as support networking eHealth management and social computing this book is ideally designed for health practitioners physicians patients medical administrators nurses surgeons infectious disease educators hospital directors world health organizations academicians students and researchers seeking coverage on current advances in health communication computer science and epidemiology

Handbook of Rural Health Sana Loue, Beth E. Quill, 2013-11-11 This book integrates the expertise of profession tion available on the various health concerns and als from a broad array of disciplines anthro subpopulations and by the numerous method pology health services research epidemiology ological complexities in compiling the neces medicine dentistry health promotion and so sary data Recognition of the nuances within and cial work in an examination of rural health across rural populations as recommended here care and rural health research This investiga will allow us to provide care more efficiently tion includes an inquiry into issues that are uni and effectively and to prevent disease or ame versal across rural populations such as public liorate its effects Reliance on some of the newer health issues and issues of equity in health care technologies and approaches discussed here Several chapters explore the health care issues such as distance learning and broad based com that confront specified subpopulations includ munity wide health initiatives will facilitate ing for instance migrant workers and Native disease treatment and prevention in relatively Americans while others provide a more focused isolated areas Ultimately all of us must work approach to diseases that may disproportionately to ensure the availability of adequate health care have an impact on residents of rural areas such to even the most isolated communities for as as specific chronic and infectious diseases

Concise Encyclopedia of Special Education Cecil R. Reynolds, Elaine Fletcher-Janzen, 2004-01-16 The Concise Encyclopedia of Special Education Second Edition is a comprehensive resource for those working in the fields of special education research and practice Featuring reviews of assessment instruments and teaching approaches legal issues overviews of specific learning disabilities dozens of biographies and more this complete desk reference is an indispensable guide for professionals academics and students alike Named an American Library Association Top 25 Reference of the Year in its First Edition The Concise Encyclopedia serves as an important reference for the education of handicapped and other exceptional children Written and edited by highly regarded and respected experts in the fields of special education and psychology this authoritative resource guide provides a reference base for educators as well as professionals in the areas of psychology neuropsychology medicine health care social work and law Additionally this acclaimed reference work is essential for administrators psychologists diagnosticians and

school counselors as well as parents of the handicapped themselves What s new in this edition Comprehensive coverage of new legislation such as Individuals with Disabilities Act IDEA and the Americans with Disabilities Act Cultural competence in Special Education including new material on culturally linguistically diverse students Many new entries including notable biographies new service delivery systems special education laws new assessment instruments cross cultural issues neuropsychology and use of the Internet in research and service delivery Some of the topics covered Academic assessment Achievement tests Addictions Bilingual education Child and adolescent behavior management Counseling with individuals and families with disabilities Early childhood education Gifted education Intelligence tests Mathematics disabilities Psychoeducational methods Rehabilitation Socioeconomic status Special education parent and student rights Traumatic brain injury

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web standard 1 2 3 of the code states that if any of the substances are contained in food listed in box 1 below no matter how small the amount which may be present as an ingredient or an ingredient of a compound ingredient or the substance is used as a food additive or an ingredient or component of such a substance or a substance or food us

how to declare food allergens on food products correctly - Jul 01 2022

web august 24 2022 any severe reaction to food that elicits an immunological response is defined as a food allergy in today s world food allergies are very common with their prevalence spreading to many countries around the globe affecting most children 4 6 compared to adults 1 2

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web in february 2021 new plain english allergen labelling requirements were introduced for how certain foods known to be common allergens are declared these changes will mean food allergen information is clearer and easier to find on food labels food businesses have three years to update their labels to meet the new requirements

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web the use of precautionary allergen labelling there will be occasions during the food manufacturing process where it is not possible to avoid the unintended presence of allergens to address this precautionary allergen labelling pal was introduced to communicate the risk of unintended presence of allergens in foods to consumers

fda issues guidances on food allergen labeling requirements - Sep 03 2022

web november 29 2022 today the u s food and drug administration fda issued two guidance documents about food allergen labeling requirements to help the food industry meet the requirements to

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web allergen declarations refers to required information on food packaging where certain allergens are present e g in the ingredient list and or a contains statement while pal and advisory labelling refers to statements that indicate the possible presence of allergens as a consequence of allergen cross

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web 31 may 2021 european union comments on codex circular letter cl 2021 9 ocs fl request for comments information on allergen labelling revision of the general standard for the labelling of prepackaged foods cxs 1 1985

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web allergen advice statements can also be used on the product label to explain how allergen information is presented on a label for example allergen advice for allergens see ingredients

eu 1169 2011 guide allergen labelling requirements menutech - Nov 05 2022

web dec 31 2020 the list above contains the correct naming of all 14 allergens as well as the allergen s letter code as recommended by the codex commission it is possible to mark dishes e g on a restaurant menu with a codex letter code or a food allergen icon it is however necessary to provide an adjacent information sheet

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web food allergen declarations new food labelling requirements for declaring allergens on 25 february 2021 the food standards code was changed to introduce new requirements for allergen labelling as set out in standard 1 2 3 external site and schedule 9 external site

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web may 16 2023 letter to industry food safety risks of transferring genes for proteins that are food allergens to new plant varieties used for food april 2023 guidance for industry recommendations for the

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web jan 10 2023 food allergies occur when the body s immune system reacts to certain proteins in food food allergic reactions vary in severity from mild symptoms involving hives and lip swelling to severe

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web may 9 2023 the new requirements will help ensure that mandatory food allergen declarations are clear and consistent for consumers these requirements include declaring allergen information in a specific format and location on food labels and using simple plain english terms for allergen declarations

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web declaration by patient visitor 1 do you have a fever cough shortness of breath blocked or runny nose sore throat or cannot smell no yes 2 have you been tested covid 19 positive pcr or art in the last 7 days for fully vaccinated persons or 14 days for unvaccinated or partially vaccinated persons no yes 3

example declaration stating allergen free gluten free and non gmo - Mar 29 2022

web sep 29 2014 example declaration stating allergen free gluten free and non gmo posted in allergen management can anyone help on how to write a declaration to show that our food products are allergen free gluten free and non gmo we are producing rtes

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web the following known food allergens must be declared on food labels for more information on our labelling requirements

read our labelling guidelines for food importers and manufacturers in addition sfa adopts a science based risk assessment and management approach that is consistent with international standards

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web food allergens that need to be declared on your label if your food or drink contains a common allergen or ingredient sourced from it you must state this clearly on your label the allergens that must be declared are peanuts almonds brazil nuts cashews hazelnuts macadamias pecans pine nuts pistachios walnuts crustacea molluscs

code of practice on food allergen management for food business - May 11 2023

web with the increasing health burden posed by food allergens comes the expectation that fbos take steps to accurately declare the presence of allergenic ingredients minimize the risk from and where possible prevent unintended allergen presence and that competent authorities provide guidance and oversight where necessary to fbos on food aller

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web declaration of allergens date 18th july 2019 to whom it may concern product loctite lb 8014 known as food grade antiseize referenced document regulation eu no 1169 2011 on the provision of food information to consumers declaration to the best of our knowledge loctite lb 8014 known as food grade antiseize does not contain any

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