

Letter to the Editor

Neurocutaneous, Phakomatoses, and Hamartoneoplastic Syndromes: Mnemonic

Dear Sir,

We read the article "PHACES syndrome – A rare case report" on Medical Journal of Dr. D. V. Patil Vidyapeeth with great interest. Das reported a case of a female preschool child presenting with face hemangioma, delayed development, and seizures. The ophthalmologic examination revealed cataract, coloboma, and optic disc hypoplasia. Furthermore, a posterior fossa arachnoid cyst and central nervous system vascular abnormalities were seen. Then, a diagnosis of PHACES syndrome was made.^[1]

PHACES syndrome is primarily a cutaneous condition characterized by multiple congenital malformations. PHACES is an acronym of Posterior fossa alterations, facial Hemangioma, Arterial, Cardiac, Eye, and Sternal abnormalities. It is considered a nonhereditary syndrome, and the etiology is not clearly understood. In this context, phakomatoses also known as neuro-oculo-cutaneous syndromes or neurocutaneous disorders are a group of diseases characterized by

the involvement of structures that arise from the embryonic ectoderm. We would like to provide a mnemonic to remember PHACES syndrome and the majority of the other neurocutaneous diseases already known in the literature [Table 1] to help in the differential diagnosis of this group. It is worth mentioning that some of the diseases described in Table 1 are not considered for every author as a phakomatoses.^[2-3]

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Nil.

Conflicts of interest

There are no conflicts of interest.

Jamir Piton Rissardor^{1,2}, Ana Leticia Fornari Caprara^{1,2}

Departments of ¹Neurology and ²Medicine, Federal University of Santa Maria, Santa Maria, Rio Grande do Sul, Brazil

ORCID:

Jamir Piton Rissardor: <https://orcid.org/0000-0001-6179-2177>

Table 1: Neurocutaneous disorders mnemonic "NEUROCuTANIOLS PHAKoMATosis"

NEUROCuTANIOLS		PHAKoMATosis	
N	Neurofibromatosis type 1 and 2 Nevus comedonicus syndrome	P	PHACES syndrome (Pascual-Castroviejo type II syndrome)
E	Epidermal nevus syndromes (various types) Ehlers-Danlos type (mainly IV)		Phacomatosis pigmento-keratorica Pelliter-Hall syndrome Proteus syndrome Pigmentosa (xeroderma pigmentosa) Perry-Romberg syndrome (progressive hemifacial atrophy)
U	Wyburn-Mason syndrome		
R	Ravalcaba (Bannayan-Riley-Ravalcaba)		
O	Osler-Weber-Rendu syndrome		
C	Cerebrotendinous xanthomatosis Cerebellotrigeminal dermal dysplasia	H	Haberland syndrome Hafner (Garcia-Hafner syndrome) Abdallat-Davis-Farrage syndrome Crowden syndrome/cold syndrome Child syndrome Menkes disease Angora hair nevus Tuber cinereum hamartoma (hypothalamic hamartoma)
T	Tuberous sclerosis		
A	Ataxia telangiectasia (Louis-Bar syndrome)	A	
N	Neurocutaneous melanosis Nevoid basal cell carcinoma syndrome Nevoid melanosis	K	
I	Incontinentia pigmenti Ito (hypomelanosis of Ito)	M	
L	Lhermitte Duclos disease	A	
S	Sturge-Weber syndrome	T	
		S	Schimmelpenninck syndrome

Neurocutaneous Disorders Phakomatoses Hamartoneoplastic Syndromes

**Christos P. Panteliadis, Ramsis
Benjamin, Christian Hagel**



Neurocutaneous Disorders Phakomatoses Hamartoneoplastic Syndromes:

Neurocutaneous Disorders Martino Ruggieri, Ignacio Pascual Castroviejo, Concezio Di Rocco, 2009-10-01 The book provides an authoritative source of knowledge about these problematic disorders It bridges the gap between clinical recognition and the new molecular medicine The editors distinguished clinicians and geneticists assembled an internationally renowned group of collaborators many of them the experts who first described a particular disorder or established its present accepted definition They have written a practical comprehensive guide to the recognition investigation and management of more than 60 recognised phakomatoses *Neurocutaneous Disorders* Martino Ruggieri, Ignacio Pascual Castroviejo, Concezio Di Rocco, 2010-11-16 The book provides an authoritative source of knowledge about these problematic disorders It bridges the gap between clinical recognition and the new molecular medicine The editors distinguished clinicians and geneticists assembled an internationally renowned group of collaborators many of them the experts who first described a particular disorder or established its present accepted definition They have written a practical comprehensive guide to the recognition investigation and management of more than 60 recognised phakomatoses Neurocutaneous Disorders, 2008

The Causes of Epilepsy Simon D. Shorvon, Frederick Andermann, Renzo Guerrini, 2011-04-14 Causation is an aspect of epilepsy neglected in the scientific literature and in the conceptualization of epilepsy at a clinical and experimental level It was to remedy this deficiency that this book was conceived The book opens with a draft etiological classification that goes some way to filling the nosological void The book is divided into four etiological categories idiopathic symptomatic cryptogenic and provoked epilepsies Each chapter considers topics in a consistent fashion dealing with the phenomenon of epilepsy in each etiology including its epidemiology clinical features and prognosis and any specific aspects of treatment The book is a comprehensive reference work a catalogue of all important causes of epilepsy and a clinical tool for all clinicians dealing with patients who have epilepsy It is aimed at epileptologists and neurologists and provides a distillation of knowledge in a form that is helpful in the clinical setting *Retina* Atul Kumar, 2018-05-31 This book is a comprehensive guide to the diagnosis and management of retinal diseases and disorders Divided into nine sections text begins with an introduction to basic sciences and diagnostics The following sections provide in depth coverage of both medical and surgical treatment of various disorders retinal degeneration and fundal dystrophies macular disease and degeneration retinal vascular disorders paediatric retinal disease choroidal and retinal inflammations trauma and problems requiring surgical intervention The final section discusses miscellaneous conditions such as ocular tumours toxic and photic retinopathies and phakomatoses Highly illustrated with clinical photographs OCT and angiography images and tables the book provides online access to a selection of videos demonstrating surgical procedures covered in the text Key points Comprehensive guide to diagnosis and management of retinal diseases Covers both medical and surgical treatments Highly illustrated with clinical photographs OCT and angiography images and tables Provides online access to videos demonstrating surgical procedures

Absolute Pediatric Neurology Yasser M. Awaad, 2018-07-24 This practical book features more than 1000 questions and answers with illustrations for pediatric neurologists adult neurologists general pediatricians and students taking their initial board examination and maintenance of certification All questions are in multiple choice format and followed by the correct answer with a full explanation and appropriate references Chapters are sectioned by different topics in pediatric neurology including Epilepsy Metabolic Disorders and Movement Disorders and other topics Timely and thorough this is a handy and succinct resource

Malformations of the Nervous System Harvey B. Sarnat, Paolo Curatolo, 2007-09-26 This volume deals with brain development malformations of the central nervous system showcasing a unique approach that furthers research through systematic integration of exciting new developments from fields including molecular genetics neuroimaging and neuropathology By integrating data and research from these disciplines better conceptualization of the mechanisms of the developmental processes is achieved Clinicians will find invaluable insights into complex issues including midline hypoplasias disorders of segmentation of the neural tube and hamartomatous disorders of cellular lineage amongst others The clinical manifestations of central nervous system malformations are also discussed along with new advancements in MRI techniques and analysis including volumetric morphology spectroscopy and functional neuroimaging Sections dedicated to management and treatment are also included in an effort to aid clinicians in their goal of providing better care for individuals affected by these types of malformations A single source that encompasses the various aspects of cerebral malformations A unique approach that furthers research through systematic integration of exciting new developments from fields including molecular genetics neuroimaging and neuropathology New diagnostic tools management protocols and treatments for patient care

Pediatric Neurology, Part I Olivier Dulac, Maryse Lassonde, Harvey B. Sarnat, 2013-04-24 The child is neither an adult miniature nor an immature human being at each age it expresses specific abilities that optimize adaptation to its environment and development of new acquisitions Diseases in children cover all specialties encountered in adulthood and neurology involves a particularly large area ranging from the brain to the striated muscle the generation and functioning of which require half the genes of the whole genome and a majority of mitochondrial ones Human being nervous system is sensitive to prenatal aggression is particularly immature at birth and development may be affected by a whole range of age dependent disorders distinct from those that occur in adults Even diseases more often encountered in adulthood than childhood may have specific expression in the developing nervous system The course of chronic neurological diseases beginning before adolescence remains distinct from that of adult pathology not only from the cognitive but also motor perspective right into adulthood and a whole area is developing for adult neurologists to care for these children with persisting neurological diseases when they become adults Just as pediatric neurology evolved as an identified specialty as the volume and complexity of data became too much for the general pediatrician or the adult neurologist to master the discipline has now continued to evolve into so many subspecialties such as epilepsy neuromuscular disease stroke malformations

neonatal neurology metabolic diseases etc that the general pediatric neurologist no longer can reasonably possess in depth expertise in all areas particularly in dealing with complex cases Subspecialty expertise thus is provided to some trainees through fellowship programmes following a general pediatric neurology residency and many of these fellowships include training in research Since the infectious context the genetic background and medical practice vary throughout the world this diversity needs to be represented in a pediatric neurology textbook Taken together and although brain malformations H Sarnat P Curatolo 2007 and oncology W Grisold R Soffietti are covered in detail in other volumes of the same series and therefore only briefly addressed here these considerations justify the number of volumes and the number of authors who contributed from all over the world Experts in the different subspecialties also contributed to design the general framework and contents of the book Special emphasis is given to the developmental aspect and normal development is reminded whenever needed brain muscle and the immune system The course of chronic diseases into adulthood and ethical issues specific to the developing nervous system are also addressed A volume in the Handbook of Clinical Neurology series which has an unparalleled reputation as the world's most comprehensive source of information in neurology International list of contributors including the leading workers in the field Describes the advances which have occurred in clinical neurology and the neurosciences their impact on the understanding of neurological disorders and on patient care

Epidemiology of Brain and Spinal Tumors Jahangir Moini, Nicholas Avgeropoulos, Mohtashem Samsam, 2021-04-09 Epidemiology of Brain and Spinal Tumors provides a single volume resource on imaging methods and neuroepidemiology of both brain and spinal tumors The book covers a variety of imaging techniques including computed tomography CT MRI positron emission tomography PET and other laboratory tests used in diagnosis and treatment Detailed epidemiology various imaging methods and clinical considerations of tumors of the CNS make this an ideal reference for users who will also find diverse information about structures and functions cytology epidemiology including molecular epidemiology diagnosis and treatment This book is appropriate for neuroscience researchers medical professionals and anyone interested in a complete guide to visualizing and understanding CNS tumors Provides the most up to date information surrounding the epidemiology biology and imaging techniques for brain and spinal tumors including CT MRI PET and others Includes full color figures photos tables graphs and radioimaging Contains information that will be valuable to anyone interested in the field of neurooncology and the treatment of patients with brain and spinal tumors Serves as a source of background information for basic scientists and pharmaceutical researchers who have an interest in imaging and treatment

Pediatric Neurology Part I Flores-Sarnat Laura, 2013-04-24 Epidermal nevus syndrome ENS is an inclusive term for a heterogeneous group of congenital disorders characterized by the presence of epidermal nevi associated with systemic involvement These disorders as are all primary neurocutaneous syndromes are neurocristopathies The epidermal nevi that follow the lines of Blaschko and most systemic anomalies in skeletal ocular cardiovascular endocrine and orodental tissues as well as lipomas are due to defective neural

crest The most important and frequent anomaly in the brain in all forms of epidermal nevus syndromes ENSs is hemimegalencephaly HME This malformation often is not recognized despite being the principal cause of neurological manifestations in ENSs They consist mainly of epilepsy and developmental delay or intellectual disability The onset of epilepsy in ENS usually is in early infancy often as infantile spasms Several syndromic forms have been delineated I propose the term Heide s syndrome for those distinctive cases with the typical triad of hemifacial epidermal nevus ipsilateral facial lipoma and hemimegalencephaly Most ENSs are sporadic The mechanism is thought to be genetic mosaicism with a lethal autosomal dominant gene Specific genetic mutations PTEN FGFR3 PIK3CA and AKT1 have been documented in some patients The large number of contributors for over more than a century and a half to the description of these disorders precludes the use of new author eponyms

The genetics of human mendelian skin disorders Jia Zhang,Yiran Guo,Ming Li,Wei Hsum Yap,2023-01-19 **Neuroimaging, An Issue of Neurologic Clinics** Laszlo Mechtler,2014-02-28 With each passing year neuroimaging becomes more and more central in the diagnosis of neurologic disease as its capacity to detect subtle changes in nervous system tissue increases This issue of Neurologic Clinics features twelve articles that address the key clinical applications of the several radiologic modalities Topics include Neuro Imaging of Infectious Disease Neuro Imaging of Dementia Imaging of Traumatic Brain Injury 3 MR Guided Focused Ultrasound A New Technology for Clinical Neurosciences Novel Multi Modality Imaging Techniques for the Diagnosis and Evaluation of Arteriovenous Malformations Neuroimaging in the Critical Care Environment Imaging of Cerebral Ischemia From Acute Stroke to Chronic Disorders Imaging of Cancer Related Neurotoxicity Neurocutaneous Syndromes Imaging of Chiari Malformation and Hydrocephalus and others

Management of Adrenal Masses in Children and Adults Electron Kebebew,2016-10-07 This book is designed to present a comprehensive and state of the art approach to the management of adrenal neoplasms that provides a resource to the broad group of providers that will encounter such a patient Sections address issues that are faced by providers who encounter a patient with an adrenal neoplasm These areas include an overview of the genetic basis and familial cancer syndrome associated with adrenal neoplasms pathobiology advanced and tumor specific imaging approaches and technologies biochemical analysis standard medical and surgical therapies and emerging technology and treatment approaches to benign and malignant adrenal neoplasms Written by experts in the field each of these sections address level of clinical evidence and provide recommendations and treatment algorithms Extensive illustrations make this an interactive text Management of Adrenal Masses in Children and Adults will serve as a very useful resource for all providers dealing with and interested in this common but challenging tumor It will provide a concise yet comprehensive summary of the current status of the field that will help guide patient management and stimulate investigative efforts

IARC Monographs on the Evaluation of Carcinogenic Risks to Humans ,1988 **Neurología** Juan José Zarranz Imirizaldu,2018-05-24 6a Edici n de la obra de referencia en Neurolog a tanto para estudiantes de la asignatura de neurolog

a como para residentes y profesionales en sus primeros años de práctica Cuenta con la participación de los mejores expertos de España en cada uno de los temas tratados El texto mantiene la estructura básica de las ediciones anteriores comenzando con capítulos sobre la anamnesis y la exploración ya que a pesar de los grandes avances en las técnicas de diagnóstico sobre todo por la imagen la orientación clínica adecuada del problema del paciente neurológico por la anamnesis y la exploración siguen siendo esenciales Incorpora numerosas novedades Además de la revisión completa del texto las tablas y los cuadros se han renovado muchas figuras se han añadido algoritmos de decisión y se ha actualizado la bibliografía Se han añadido cuatro capítulos nuevos tres sobre temas de gran actualidad las encefalitis autoinmunes las enfermedades priónicas y las enfermedades mitocondriales

SÍNDROMES NEUROPEDIÁTRICOS: Origen, fundamentos para el diagnóstico e importancia Joaquín A. Peña, 2022-09-05 Este libro revisa con actualidad y precisión los diversos signos y síntomas reflejos y síndromes neuropedítricos y examina el entorno histórico de sus descubridores Est destinado a llenar un vacío en la formación académica y la visión humanística de la medicina Se trata de un texto valioso para estudiantes residentes médicos y todos los profesionales involucrados en el tratamiento y rehabilitación de los pacientes neurológicos Escrito en forma sencilla y esquemática con criterio clínico excelente manual de referencia y fuente de información Joaquín A. Peña es neuropediatra investigador y profesor universitario En este libro aporta su experiencia personal fruto de su prolongado trabajo profesional y docente

Canpolat-Karakükcü-Dursun-Poyrazoğlu Pediatric Temel Bilgiler Mehmet CANPOLAT, Musa KARAKÜKCÜ, İsmail DURSUN, M. Hakan POYRAZOĞLU, 2025-05-27 **Emergencia y decadencia de la reciente medicina española** Ignacio Pascual Castroviejo, 2014-02-10 El Doctor Ignacio Pascual Castroviejo 1934 ha dedicado toda su vida profesional a la investigación y a la práctica de la Medicina especialmente a la Neurología Pediátrica publicando varios libros algunos en Inglés y más de 300 trabajos científicos en revistas de gran prestigio internacional así como participando y dando conferencias en las principales reuniones y congresos internacionales En este libro el doctor Pascual Castroviejo relata desde su niñez hasta hoy sus vivencias y anécdotas así como su visión personal de la Medicina Social Pública y Universitaria de España así como de los actores principales que contribuyeron de una forma u otra a su desarrollo e innovación Neurocutaneous Disorders

Christos P. Panteliadis, Ramsis Benjamin, Christian Hagel, 2022-02-03 This book provides extensive data on the more common and many of the more rare congenital and hereditary syndromes that manifest in the nervous system and skin Though often complex and multi systemic these disorders can frequently be diagnosed using a combination of simple visual inspection and sound clinical expertise Drawing on fully referenced information from thousands of articles the international editorial team has prepared a comprehensive overview that includes historical perspectives clinical features the pathogenesis and diagnostic and therapeutic strategies In addition it addresses the biochemical molecular and genetic basis of the disorders The book is divided into four main sections Starting with general aspects of aetiology diagnostics and therapy the first part then covers the genetics neuro imaging neuropathology ocular manifestations and surgical management The second part

discusses developmental malformations such as Sturge Weber syndrome Ataxia Telangiectasia Hypomelanosis of Ito and other rare syndromes including haemangiomas The focus of the third part is on tumour suppressor DNA repair disorders the most common of which is Neurofibromatosis 1 It also describes Neurofibromatosis 2 Schwannomatosis Tuberous sclerosis von Hippel Lindau disease Naevoid basal cell carcinoma and others The book s fourth and final section covers defects in enzymes and structural proteins which manifest as Cerebrotendinous xanthomatosis Ehlers Danlos syndrome Menkes syndrome Refsum disease

Neurocutaneous Syndromes Monica P. Islam,E. Steve Roach,2015-11-09 Neurocutaneous Syndromes provides the most updated and comprehensive resource on the disorders that lead to the growth of tumors in various parts of the body those caused by the abnormal development of cells in an embryo and characterized by the presence of tumors in various parts of the body and eyes including the nervous system and by certain differences in the skin The most common neurocutaneous syndromes include neurofibromatosis Sturge Weber syndrome tuberous sclerosis ataxia telangiectasia and von Hippel Lindau disease Symptoms vary widely and while present early may not express until later in life As molecular medicine and genetic science is continuing to impact our understanding of neurocutaneous syndromes this book also includes the latest molecular and genetic science Provides a comprehensive coverage of neurocutaneous syndromes Details the latest molecular and genetic science related to neurocutaneous syndromes Presents a focused reference for clinical practitioners and the neuroscience clinical neurology and neurogenetics research communities Includes updated sections on the latest molecular and genetic science

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