

13 Syndromes of the Head and Neck

Chris Jo, DMD

This chapter addresses:

- Cleft Lip and Palate
- Nonsyndromic Craniosynostosis
- Apert Syndrome
- Crouzon Syndrome
- Hemifacial Microsomia
- Obstructive Sleep Apnea Syndrome

A multitude of anomalies and syndromes occur in the head and neck, most of which are beyond the scope of this book. This chapter covers some of the most common anomalies or syndromes associated with the craniomaxillofacial region. Congenital anomalies include nonsyndromic craniosynostosis, hemifacial microsomia, and cleft lip and palate (CLP). Congenital syndromes include Apert and Crouzon syndromes. Unlike the other syndromes in this section, which are congenital, obstructive sleep apnea syndrome (OSAS) is included here because the pathogenesis and manifestation of this syndrome are based on anatomical anomalies of the head and neck. Oral and maxillofacial surgeons are uniquely trained and play an integral role in the surgical management of these patients, whether or not syndromic.

The intent of this chapter is to familiarize readers with the pathogenesis, presentation, and management strategies of such anomalies. The chapter is structured so that key features of each syndrome or anomaly are emphasized. The reconstructive strategies and rationale for treatment are discussed. Because of the complexity of craniofacial deformities involved in the growing child, the reconstructive efforts are generally staged. There is no consensus on the best timing of each stage, but the general guidelines are presented. Various surgical strategies, depending on the surgeons' preference and clinical situation, can be used. These are presented in the chapter along with their rationale of treatment. Table 13-1 outlines the characteristics of some craniofacial syndromes.

Table 13-1. Characteristics of Some Craniofacial Syndromes

Syndrome	Prevalence/ Inheritance	Genetic	Synostosis/Orbit	Limbs	Central Nervous System
Apert syndrome	15.5:1 million, AD or sporadic or new mutation Males = females	Scr253Tyr or Pro253, Arg on Ig II or III of <i>FGFR3</i>	Early fusion of coronal suture, widely patent midline calvarial defect	Fusion of digits 2, 3, and 4 Less common is fusion of digits 2, 3, 4, and 5, with digit 1 free	Patient features (except coronal) and open synchondrosis, agenesis of septum pellucidum, agenesis of corpus callosum, cavum septum pellucidum, developmental delay
Crouzon syndrome	15-36:1 million, AD	15 mutations of Ig III domain of <i>FGFR3</i>	Variable suture involvement and skull shape, exotropia, exposed conjunctiva, and keratitis	None	Cerebellar tonsil herniation, jugular foramen stenosis, venous obstruction, and increased intracranial pressure Headache (29%), seizures (10%), mental retardation (5%)
Piccolier syndrome	16-40:1 million, AD	<i>FGFR1, FGFR2</i>	Pronounced exotropism	Broad thumbs	Three types: type 1, normal life expectancy and intelligence; type 2, cloverleaf skull, exotropism, elbow ankylosis, broad thumbs and great toes; type 3, same as type 2 without cloverleaf. Types 2 and 3 with ventriculomegaly, progressive hydrocephalus, and cerebellar herniation

AD, Autosomal dominant.

Syndromes Of The Head And Neck

**Raoul C. M. Hennekam, Ian D.
Krantz, Judith E. Allanson**



Syndromes Of The Head And Neck:

Syndromes of the Head and Neck Robert J. Gorlin, M. Michael Cohen Jr., D.M.D., Ph.D., Raoul C.M. Hennekam M.D., Ph.D., 2001-09-27 This classic text one of the true anchors of our clinical genetics publishing program covers over 700 different genetic syndromes involving the head and neck and it has established itself as the definitive comprehensive work on the subject The discussion covers the phenotype spectrum epidemiology mode of inheritance pathogenesis and clinical profile of each condition all of which is accompanied by a wealth of illustrations The authors are recognized leaders in the field and their vast knowledge and strong clinical judgment will help readers make sense of this complex and burgeoning field Dr Gorlin retires as editor in this edition and co editor Raoul Hennekam takes over Dr Hennekam is regarded as one of the top dysmorphologists and indeed one of the top clinical geneticists in the world Judith Allanson is new to the book but is a veteran OUP author and a widely respected geneticist and Ian Krantz at Penn is a rising star in the field Dr Gorlin's name has always been closely associated with the book and it has now become part of the title As in all fields of genetics there has been an explosion in the genetics of dysmorphology syndromes and the author has undertaken a complete updating of all chapters in light of the discoveries of the Human Genome Project and other ongoing advances with some chapters requiring complete rewriting Additional material has been added both in terms of new syndromes and in updating information on existing syndromes The book will appeal to clinical geneticists pediatricians neurologists head and neck surgeons otolaryngologists and dentists The 4th edition which published in 2001 has sold 2 600 copies **Gorlin's Syndromes of the Head and Neck**

Raoul C. M. Hennekam, Ian D. Krantz, Judith E. Allanson, 2010 This classic text covers over 700 different genetic syndromes affecting body structures and has established itself as the definitive comprehensive work on the subject The discussion covers the phenotype spectrum epidemiology mode of inheritance pathogenesis and clinical profile of each condition all of which is accompanied by a wealth of illustrations The authors are recognized leaders in the field and their vast knowledge and strong clinical judgment will help readers make sense of this complex and burgeoning field New syndromes have been added to this edition and existing syndromes have been updated This book will appeal to clinical geneticists pediatricians neurologists head and neck surgeons otolaryngologists and dentists **Syndromes of the Head and Neck** Robert J.

Gorlin, Jens Jørgen Pindborg, 1964 **Syndromes of the Head and Neck, An Issue of Atlas of the Oral & Maxillofacial Surgery Clinics** Dean M. DeLuke, 2014-09-28 This issue of the Atlas of the Oral and Maxillofacial Surgery Clinics of North America serves as an aid to identification of 70 of the more important head and neck syndromes and organized using the same nosology as in the landmark text of Robert Gorlin As such it should be useful to a varied audience including not only the oral and maxillofacial surgeon but also the oral pathologist the practitioner of oral medicine and members of the craniofacial team the orthodontist pediatric dentist speech and language specialist geneticist plastic surgeon ENT specialist pediatrician nurse coordinator and others Areas covered include Craniosynostosis Syndromes Syndromes Affecting Bone Metabolic and

Autoimmune Syndromes Syndromes Affecting Skin and Mucosa Hamartoneoplastic Syndromes Branchial Arch Syndromes Clefting Syndromes Syndromes Affecting the Central Nervous System Chromosomal Syndromes Syndromes with Unusual Facies and Syndromes with Unusual Dental Findings or Gingival Components **The Clinician's Guide to Dermatologic Differential Diagnosis** Paul I. Schneiderman, Marc E. Grossman, 2022-05-14 Over a decade has passed since the original edition of The Clinician's Guide to Dermatologic Differential Diagnosis first came out This massive two volume second edition undertaking features over 4 000 new photographs of rare and puzzling symptoms 98 new chapters and presents a complete update on all references and new findings since the original publication This two volume set is designed to assist in the diagnosis of skin disease by presenting a unique and comprehensive approach to differential diagnosis Organized alphabetically by physical finding each book in the set is meticulously put together for utility and usability Volume 1 features a comprehensive list of the variety of physical findings a patient might have It is the text of the work Volume 2 serves as the atlas with which to cross reference a physical finding It includes over 10 000 patient photos and strives to feature the rarer more difficult to encounter pictures to arm the reader with knowledge beyond the basic Created purely with the reader in mind The Clinician's Guide to Dermatologic Differential Diagnosis 2nd Edition strives to enhance the learning and teaching of differential diagnosis within dermatology These texts are for the patient that defies diagnosis going through checklists of diseases ensures that no clue is missed in aiding the patient The sincere hope is that these companion volumes will be helpful in all clinical setting from medical clinic and other specialty clinics to the Dermatology clinic from medical students to attendings and from the nurses to the physicians assistants whether in the academic setting or in the community environment anywhere in the world Syndromes of the Head and Neck Robert J. Gorlin, Meyer Michael Cohen, Raoul C. M. Hennekam, 2023 This work on dysmorphology covers most genetic syndromes that affect body structure The 4th edition takes into account the explosion in molecular genetics and a new chapter on the eye has been added *Congenital Malformations of the Head and Neck* Lisa M. Elden, Karen B. Zur, 2013-12-02 Congenital Malformations of the Head and Neck offers a unique conceptual and visual approach to children with congenital malformations of the head and neck Developed by renowned leaders in the field this title is richly illustrated with a wealth of patient photos radiology and endoscopic images of malformations Starting with the genetics of common congenital syndromes Congenital Malformations of the Head and Neck goes on to comprehensively cover malformations of the ear nose nasopharynx oral cavity oropharynx cleft lip and palate larynx trachea and neck Easy to read and an indispensable reference and teaching resource this title will serve as an invaluable reference for clinicians neurologists pediatricians otolaryngologists and head and neck surgeons It should also be of great interest to fellows and residents **Cummings Otolaryngology - Head and Neck Surgery E-Book** Paul W. Flint, Bruce H. Haughey, Valerie J. Lund, John K. Niparko, K. Thomas Robbins, J. Regan Thomas, Marci M. Lesperance, 2014-11-28 Now in its 6th edition Cummings Otolaryngology remains the world's most detailed and trusted

source for superb guidance on all facets of head and neck surgery Completely updated with the latest minimally invasive procedures new clinical photographs and line drawings this latest edition equips you to implement all the newest discoveries techniques and technologies that are shaping patient outcomes Be certain with expert dependable accurate answers for every stage of your career from the most comprehensive multi disciplinary text in the field Consult this title on your favorite e reader conduct rapid searches and adjust font sizes for optimal readability Overcome virtually any clinical challenge with detailed expert coverage of every area of head and neck surgery authored by hundreds of leading luminaries in the field Experience clinical scenarios with vivid clarity through a heavily illustrated full color format which includes approximately 3 200 images and over 40 high quality procedural videos Get truly diverse perspectives and worldwide best practices from a multi disciplinary team of contributors and editors comprised of the world s leading experts Glean all essential up to date need to know information All chapters have been meticulously updated several extensively revised with new images references and content Stay at the forefront of your field with the most updated information on minimally invasive surgical approaches to the entire skull base vestibular implants and vestibular management involving intratympanic and physical therapy based approaches radiosurgical treatment of posterior fossa and skull base neoplasms and intraoperative monitoring of cranial nerve and CNS function Apply the latest treatment options in pediatric care with new chapters on pediatric sleep disorders pediatric infectious disease and evaluation and management of the infant airway Find what you need faster through a streamlined format reorganized chapters and a color design that expedites reference Manage many of the most common disorders with treatment options derived from their genetic basis Assess real world effectiveness and costs associated with emergent technologies and surgical approaches introduced to OHNS over the past 10 years Incorporate recent findings about endoscopic microscopic laser surgically implantable radiosurgical neurophysiological monitoring MR and CT imaging and other timely topics that now define contemporary operative OHNS Take it with you anywhere With Expert Consult you ll have access the full text video clips and more online and as an eBook at no additional cost

Paparella's Otolaryngology: Head & Neck Surgery Michael M Paparella, Sady Selaiman da Costa, Johan Fagan,

Otorhinolaryngology- Head & Neck Surgery Chris de Souza, 2017-12-31 Otorhinolaryngology Head endoscopic sinus surgery facial plastics head and neck cranial base and oncology laryngology otology In this new edition endoscopic sinus surgery is given its own section encompassing all aspects of this surgery and an entirely new section on otology is comprised of 37 chapters including otitis media and cochlear implants The facial plastics section provides information on dermabrasion chemical peels laser treatment botox and rhinoplasty amongst many other topics New topics in this edition include laryngopharyngeal reflux trauma and stenosis of the larynx and laryngeal cancer bringing the text firmly up to date Illustrated in full colour across 2000 pages this vast two volume set is an ideal source of reference for otorhinolaryngology practitioners and residents Key Points New edition of comprehensive two volume set covering all sub specialties in

otorhinolaryngology Previous edition published 2009 9788184486797 New sections on endoscopic sinus surgery and otology New topics include laryngopharyngeal reflux trauma and stenosis of the larynx and laryngeal cancer **Scott-Brown's Otorhinolaryngology and Head and Neck Surgery, Eighth Edition** John Watkinson, Ray Clarke, 2018-07-17 Scott Brown's Otorhinolaryngology is used the world over as the definitive reference for trainee ENT surgeons audiologists and trainee head and neck surgeons as well as specialists who need detailed reliable and authoritative information on all aspects of ear nose and throat disease and treatment Key points accompanied by a fully searchable electronic edition making it more accessible containing the same content as the print edition with operative videos and references linked to Medline highly illustrated in colour throughout to aid understanding updated by an international team of editors and contributors evidence based guidelines will help you in your clinical practice features include key points best clinical practice guidelines details of the search strategies used to prepare the material and suggestions for future research new Endocrine section Scott Brown will provide trainee surgeons ENT and Head and Neck audiologists and ENT physicians with quick access to relevant information about clinical conditions and provide them with a starting point for further research The accompanying electronic edition enhanced with operative videos will enable both easy reference and accessibility on the move

Diagnostic Pathology: Familial Cancer Syndromes - E-Book Vania Nosé, 2025-06-02 This expert volume in the Diagnostic Pathology series is an up to date comprehensive diagnostic support tool for pathologists oncologists and other physicians who diagnose and treat patients with cancer An excellent point of care reference for practitioners at all levels of experience and training the third edition of Diagnostic Pathology Familial Cancer Syndromes offers clinically useful information on hereditary cancer syndromes including differential diagnosis and management Richly illustrated and easy to use this volume is ideal as a one stop resource for day to day reference or as a reliable training tool Helps physicians recognize syndromes and syndrome associated neoplasms and advise treating physicians patients and their families on the possibility of a familial syndrome and their risk of developing other tumors Addresses a wide range of inherited tumor syndromes to increase understanding of the gross and histologic features of syndromic associated neoplasms as well as associated manifestations Part I includes nearly 100 detailed chapters describing diagnoses by organ and system associated with familial cancer syndromes Part II contains more than 80 chapters with detailed descriptions of major inherited syndromes cross referenced with diagnoses and Part III features an updated Molecular Factors Index that includes a complete description of each known gene associated with a familial cancer syndrome Includes substantial updates throughout with more than a dozen new chapters new images and references updated guidelines and classifications based on the 2025 WHO Classification of Tumours Genetic Tumour Syndromes details on the newest familial cancer syndromes and more Contains hundreds of tables organized by body part and or organ system that include selected existing genetic and familial cancers and identified associated genes clinical features recognized manifestations and staging images microscopic

examples Features nearly 2 300 print and online images including clinical and radiologic images algorithms graphics gross pathology and histology images and a wide range of special and immunohistochemical stains and molecular markers all carefully annotated to highlight the most diagnostically significant factors Employs consistently templated chapters bulleted content key facts annotated images and an extensive index for quick expert reference at the point of care Any additional digital ancillary content may publish up to 6 weeks following the publication date

A Clinician's Guide to Dermatologic Differential Diagnosis, Volume 1 Paul Schneiderman, Marc Grossman, 2006-06-28 *Head and Neck Imaging E-Book* Peter M. Som, Hugh D. Curtin, 2011-04-11 Head and Neck Imaging by Drs Peter M Som and Hugh D Curtin delivers the encyclopedic and authoritative guidance you've come to expect from this book the expert guidance you need to diagnose the most challenging disorders using today's most accurate techniques New state of the art imaging examples throughout help you recognize the imaging presentation of the full range of head and neck disorders using PET CT MRI and ultrasound Enhanced coverage of the complexities of embryology anatomy and physiology including original color drawings and new color anatomical images from Frank Netter help you distinguish subtle abnormalities and understand their etiologies Compare your imaging findings to thousands of crystal clear examples representing every type of head and neck disorder Gain an international perspective from global authorities in the field Find information quickly with a logical organization by anatomic region Master the latest approaches to image guided biopsies and treatments Utilize PET CT scanning to its fullest potential including head and neck cancer staging treatment planning and follow up to therapy Visualize head and neck anatomy better than ever before with greatly expanded embryology physiology and anatomy content including original drawings and new color anatomical images Grasp the finer points of head and neck imaging quickly with more images more detail in the images and more anatomic atlases with many examples of anatomic variants Access the complete content and illustrations online at www.expertconsult.com fully searchable

Diagnostic Imaging: Head and Neck - E-Book

Bernadette L. Koch, Surjith Vattoth, Philip R. Chapman, 2021-12-12 Covering the entire spectrum of this fast changing field Diagnostic Imaging Head and Neck fourth edition is an invaluable resource for neuroradiologists general radiologists and trainees anyone who requires an easily accessible highly visual reference on today's head and neck imaging Dr Philip R Chapman and his team of highly regarded experts provide up to date information on recent advances in disease identification imaging techniques and tumor staging to help you make informed decisions at the point of care The text is lavishly illustrated delineated and referenced making it a useful learning tool as well as a handy reference for daily practice Serves as a one stop resource for key concepts and information on head and neck imaging including a wealth of new material and content updates throughout Features more than 2 800 illustrations including radiologic images full color illustrations clinical and gross pathology photographs and histology photographs as well as an additional 2 200 digital images online Features numerous new chapters and updates from cover to cover including changes to staging of HPV related p16 oropharyngeal squamous cell

carcinoma new metastatic disease imaging recommendations protocols and treatments and the latest knowledge on the genetics of various congenital conditions and syndromes Reflects new Lugano and WHO classifications for staging lymphomas updates in the AJCC Cancer Staging Manual 8th Edition and updates from the 2018 ISSVA Classification regarding avoidance of outdated and inappropriate terminology and nomenclature that can lead to misdiagnosis or inappropriate treatments Uses bulleted succinct text and highly templated chapters for quick comprehension of essential information at the point of care Essentials of Head and Neck Oncology Lanny Close,David Lee Larson,Jatin P. Shah,1998 Cancer of the head and neck is a complex disease which when understood can often be treated very effectively rendering the patient disease free while preserving or restoring both form and function Offering the unique perspective of the three specialties involved in head and neck cancer otolaryngology plastic surgery and general surgery here is a comprehensive overview of the field today ESSENTIALS OF HEAD AND NECK ONCOLOGY provides full coverage of oncologic evaluation treatment and results all distilled into a practical reference that fits into the lab coat pocket This is the ideal textbook for board examination preparation or practitioners looking to update their knowledge in a particular area **Diagnostic Pathology: Head and Neck,E-Book** Lester D. R. Thompson,Susan Müller,Brenda L. Nelson,2022-05-02 This expert volume in the Diagnostic Pathology series is an excellent point of care resource for practitioners at all levels of experience and training Covering all areas of head and neck pathology it incorporates the most recent clinical pathologic and molecular knowledge in this challenging field to provide a comprehensive overview of all key issues relevant to today s practice Richly illustrated and easy to use the third edition of Diagnostic Pathology Head and Neck is a one stop reference for accurate complete pathology reports ideal as a day to day reference or as a reliable teaching resource Provides a clear framework for a better understanding of the clinical and histopathologic appearances of more than 325 new or evolving entities of the head and neck and endocrine organs Features updates on recent changes in the field and current information from the AJCC Cancer Staging Manual 8th Edition and the World Health Organization Classification of Head and Neck Tumours 5th Edition Covers numerous new diagnostic entities including intraductal carcinoma sclerosing polycystic adenoma microsecretory adenocarcinoma adamantinoma like Ewing sarcoma SMARCB1 and SMARCA4 deficient carcinomas and HPV related multiphenotypic sinonasal carcinoma Contains major updates on sinonasal tract salivary gland and odontogenic tumors oropharyngeal squamous cell carcinoma and its correct classification and staging the recent significant reclassification of neuroendocrine tumors and changes in terminology and identification for head and neck soft tissue tumors Suggests supporting studies in a realistic clinical context with tables and immunohistochemical charts that assist with management decisions and prognostic outcome predictions Features more than 3 200 images including full color drawings of head and neck anatomy development pathway drawings staging protocol illustrations and genetic pathway graphics radiologic images clinical photographs and gross images and histology histochemistry immunohistochemistry in situ hybridization and

molecular images as well as more than 2 600 additional online only images Employs consistently templated chapters bulleted content key facts and annotated images for quick expert reference at the point of care Includes the enhanced eBook version which allows you to search all text figures and references on a variety of devices Handbook of Tumor Syndromes

Dongyou Liu, 2020-05-08 Tumors cancers are characterized by uncontrolled growths of abnormal cells that extend beyond their usual boundaries and disrupt the normal functions of affected organs and systems While about 75% 80% of tumors cancers arise sporadically without a family connection 20% 25% appear to be familial including 10% 15% nonhereditary familial tumors or familial tumors and 5% 10% hereditary familial tumors or hereditary tumors As nonhereditary and hereditary familial tumors often show both tumor related and non tumor related or syndromic symptoms they are referred to as tumor syndromes or cancer susceptibility syndromes In comparison with sporadic tumors cancers tumor syndromes 300 described so far tend to occur at a younger age involve multiple organs and systems produce multiple often in a distinct spectrum and bilateral lesions form multiple hamartomatous benign or precursor lesions locate in specific sites display unique syndromic features and affect multiple members generations of a family This book provides state of art and authoritative coverage of nearly 100 tumor syndromes with chapters presenting overviews of individual tumor syndromes in relation to their biology epidemiology pathogenesis clinical features diagnosis treatment and prognosis Featuring contributions from oncologists clinicians and specialists the book offers a reliable comprehensive reference on tumor syndromes for scholars and students of medicine dentistry pharmacology nursing public health and other biomedical disciplines Key Features Reviews the biology epidemiology pathogenesis and clinical features of tumor syndromes Contains up to date information on the diagnosis and treatment of tumor syndromes Includes expert coverage from leading oncologists and clinicians Related Titles J R McIntosh Understanding Cancer An Introduction to the Biology Medicine and Societal Implications of This Disease ISBN 978 0 8153 4535 0 P S T Shanmugam Understanding Cancer Therapies ISBN 978 1 1381 9815 9 J M Baehring and J M Piepmeier eds Brain Tumors Practical Guide to Diagnosis and Treatment ISBN 978 0 3673 9022 8 D Liu Tumors and Cancers Endocrine Glands Blood Marrow Lymph ISBN 978 1 4987 2975 8 Dongyou Liu PhD has worked at several research and clinical laboratories in Australia and the United States for the past three decades with a focus on molecular characterization of microbial pathogens and detection of human genetic disorders and tumors cancers He is the primary author of more than 50 original research and review articles in various peer reviewed international journals the contributor of 197 book chapters and the editor of more than 10 books **Practical Head and Neck Pathology** Danielle Elliott Range, Xiaoyin "Sara" Jiang, 2019-11-14 Practical Head and Neck Pathology Frequently Asked Questions centers around frequently asked questions but it is not meant to be comprehensive in its approach Instead it aims to touch on all major or commonly encountered entities Each chapter begins with a list of questions typically ordered from basic histologic knowledge to inflammatory processes primary tumors and secondary tumor types Strategies for the diagnosis of salivary

gland metastatic head and neck cancers and thyroid disease using ancillary testing is highlighted Small biopsy dilemmas in undifferentiated tumors of the sinonasal and nasopharyngeal tracts are undertaken with emphasis on differential diagnosis and ancillary techniques The tables are the foundation of the book offering a quick reference guide for salient features of various entities and differential diagnoses A comprehensive set of references after each question can be used to supplement the information provided and give the reader a foundation for further learning This book answers some of the difficult prevailing and emerging questions in head and neck pathology that arise in everyday practice While such challenges may not always be frequent they have tremendous impact on clinical practice Clinically pertinent molecular advances in treatment and diagnosis for the practicing and junior pathologist are covered *Otolaryngology and Head and Neck Surgery* ,2006

Unveiling the Energy of Verbal Artistry: An Mental Sojourn through **Syndromes Of The Head And Neck**

In some sort of inundated with monitors and the cacophony of quick connection, the profound power and emotional resonance of verbal beauty frequently fade in to obscurity, eclipsed by the continuous onslaught of sound and distractions. However, set within the musical pages of **Syndromes Of The Head And Neck**, a interesting perform of literary elegance that impulses with natural thoughts, lies an unique journey waiting to be embarked upon. Composed with a virtuoso wordsmith, that magical opus courses viewers on a mental odyssey, delicately exposing the latent possible and profound influence stuck within the delicate internet of language. Within the heart-wrenching expanse of this evocative analysis, we can embark upon an introspective exploration of the book is main subjects, dissect its interesting writing design, and immerse ourselves in the indelible impression it leaves upon the depths of readers souls.

https://archive.kdd.org/results/publication/Documents/The_Essential_James_Bond_The_Revised_Authorised_Guide_To_The_World_Of_007.pdf

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Syndromes Of The Head And Neck Introduction

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