

A case report of fatal familial insomnia with cerebrospinal fluid leukocytosis during the COVID-19 epidemic and review of the literature

Zheng Wang , Yueqi Huang, Shuqi Wang, Jiefang Chen, Gesang Meiduo, Man Jin, and Xiaoying Zhang

Affiliated Mental Health Center & Hangzhou Seventh People's Hospital, Zhejiang University School of Medicine, Hangzhou, China

ABSTRACT

Fatal familial insomnia (FFI) is a rare autosomal dominant genetic neurodegenerative disease. Generally, FFI patients will develop rapidly progressive dementia, sleep disturbance, autonomic dysfunction, and so on. Cerebrospinal fluid examination of FFI patients normally shows no obvious abnormalities. Here, we report a young male patient who was diagnosed with FFI during the COVID-19 epidemic. Clinical symptoms include psychobehavioral abnormality, cognitive decline, sleep disturbance, and autonomic dysfunction. No abnormalities were found in routine examinations after admission. However, the number of white blood cells in the cerebrospinal fluid increased. Though the patient was treated with anti-infection and immunotherapy, the symptoms were not relieved. A lumbar puncture was performed again, and it was found that the total Tau protein in the cerebrospinal fluid was elevated, and PET results showed that brain metabolism decreased. Finally, a genetic test was used to confirm the diagnosis of FFI. This case suggests that patients with FFI may also have elevated white blood cells in cerebrospinal fluid and timely detection of Tau protein in cerebrospinal fluid is helpful for early identification of FFI. And precise diagnosis relies on genetic testing.

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KEYWORDS

Clinical features; CSF analysis; fatal family insomnia; PET-CT; PRNP; Tau protein

Introduction

FFI, a rare autosomal dominant disease, is associated with the D178N-129M missense mutations of the prion protein gene (PRNP) on chromosome 20, and the main pathological feature is scrapie prion protein (PrP^{Sc}) deposition in the brain [1,2]. The average onset age of FFI is about 51 years old, and the course of the disease ranges from 8 to 72 months [3]. Clinical symptoms of FFI patients are variable. Typically, FFI patients are accompanied with rapidly progressive dementia, sleep disturbance, autonomic dysfunction, and so on. Eventually, it will lead to death [4].

In this article, we report one case of a young male patient diagnosed with FFI who had been vaccinated against COVID-19 during the COVID-19 pandemic. This patient had no positive family history but with severe sleep disturbances, progressive cognitive decline, significant changes in autonomic nervous function, and psychobehavioral abnormality. Early detection of cerebrospinal fluid showed increased white blood cells. Multiple magnetic resonance imaging (MRI) and electroencephalography (EEG) examination showed no abnormalities, and the effects of anti-infection and immunotherapy treatment were poor. Finally, the patient was diagnosed with FFI by genetic testing.

Case presentation

The patient, a 28-year-old courier, was admitted to the hospital in July 2021, because of 'Memory impairment and psychotic behaviour disorder for more than one month'. The patient suffered from memory loss one month before admission without obvious incentives. He often made repeated mistakes at work and forgot whether he had eaten. There are often involuntary movements of limbs and frequent changes in his body position during sleep. Later, hallucinations and delusion gradually appeared. When his brother was not at home, he claimed to have seen him. He also said that his child was adopted, and he had another one. The patient had seen a doctor in another hospital before coming to our department. He was diagnosed with 'anxiety disorder' and was treated with 'Olanzapine Tablets 2.5mg/day'. After treatment, the patient's sleep quality has slightly improved, but he often moves his hands and feet around during sleep and tears the quilt with his hands and teeth.

When admitted to the hospital, the patient's body temperature was normal, his consciousness was clear, his facial expression was stolid, and his feeling of time orientation was not good. His short-term memory was impaired. He could not recall three things (ball, flag,

CONTACT Xiaoying Zhang  xyzhong004@163.com  Department of Neurology, Hangzhou Seventh People's Hospital, 305 Tianmushan Road, Hangzhou 310013, China

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Scientific Report On Fatal Familial Insomnia

Saif Hameed,Zeeshan Fatima



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Neuroimmune Pharmacology and Therapeutics Howard E. Gendelman, Tsuneya Ikezu, 2024-12-26 Bridging neuroscience immunology and pharmacology and bringing together the foremost authorities Neuroimmune Pharmacology and Therapeutics 3rd edition is an invaluable reference and textbook The text discusses the immunology of the nervous system New chapters are offered on innate humoral and cellular immune responses innate and adaptive immunity The additions join each component of the immune response in descriptions for how each affects nervous system function in both health and disease Next discussions of neuropharmacology now include both drug development and delivery into brain subregions to optimize clinical responses This edition features greatly expanded coverage of therapeutics The new extensions have blossomed into focused therapies engaging the immune system directly deploying it for drug delivery attracting the newly evolving field of genetics vaccinations and bioengineering ultimately leading to improved therapeutic disease outcomes All of the revisions in this edition are designed to bring an early trainee together with a skilled clinical and translational scientist to discuss the state of the art in each part of the emerging field of immunity as it affects the nervous system during steady state and disease and how it can be harnessed for therapeutics and clinical benefit *Pathogenicity and Drug Resistance of Human Pathogens* Saif Hameed, Zeeshan Fatima, 2020-01-23 The book comprehensively discusses the mechanisms of pathogenesis and drug resistance current diagnostics landscape of four key human pathogens bacterial fungal protozoans and viral which are the causes of major infectious diseases It also assesses the emerging technologies for the detection and quantification of these pathogens Further it discusses the novel opportunities to fight against these infectious diseases and to identify pertinent drug targets with novel methodologies It also reviews the current and future insights into the control elimination and eradication of these infectious diseases Importantly the book discusses the epidemiological characteristics and various challenges in combating Ebola and Influenza diseases Finally the book highlights the growing role of nanotechnology and bioinformatics resources for combating the infectious diseases In summary the book provides the mechanistic insight of the pathogenicity drug resistance therapeutic strategies and identification of the novel drug targets of Mycobacterium tuberculosis Plasmodium Candida Hepatitis C and emerging viral infections Neuroimaging of Sleep and Sleep Disorders Eric Nofzinger, Pierre Maquet, Michael J. Thorpy, 2013-03-07 This up to date superbly illustrated book is a practical guide to the effective use of neuroimaging in the patient with sleep disorders There are detailed reviews of new neuroimaging techniques including CT MRI advanced MR techniques SPECT and PET as well as image analysis methods their roles and pitfalls Neuroimaging of normal sleep and wake states is covered plus the role of neuroimaging in conjunction with tests of memory and how sleep influences memory consolidation Each chapter carefully presents and analyzes the key findings in patients with sleep disorders indicating the clinical and imaging features of the various sleep disorders from clinical presentation to neuroimaging aiding in establishing an accurate diagnosis Written by neuroimaging experts from

around the world Neuroimaging of Sleep and Sleep Disorders is an invaluable resource for both researchers and clinicians including sleep specialists neurologists radiologists psychiatrists psychologists *The Best American Science and Nature Writing 2020* Michio Kaku,Jaime Green,2020 Presents an anthology of the best science and nature writing published in the previous year selected from American periodicals *The Encyclopedia of Neuropsychological Disorders* Chad A. Noggle,Raymond S. Dean,Arthur MacNeill Horton,2012 Print CourseSmart *Edinburgh Companion to Science Fiction and the Medical Humanities* Donna McCormack,Anna McFarlane,Gavin Miller,2025-03-31 The medical humanities are becoming increasingly important as their first wave is interrogated by a critical approach that aims to uncover the wider possibilities of the field In conversation with this debate this volume explores the ways in which science fiction studies can contribute to such discussions Science fiction challenges techno optimism and offers a non realist avenue for the expression of illness experience Science fiction also estranges its readers from their societies and the medical possibilities inherent in those societies inviting consideration of how medicine may be complicit with or opposed to other structures of power By engaging these concerns this Companion volume offers a unique viewpoint on the power of the future to shape the present

Advancing Prion Science Institute of Medicine,Medical Follow-up Agency,Committee on Transmissible Spongiform Encephalopathies: Assessment of Relevant Science,2004-04-04 In *Advancing Prion Science* the Institute of Medicine s Committee on Transmissible Spongiform Encephalopathies Assessment of Relevant Science recommends priorities for research and investment to the Department of Defense s National Prion Research Program NPRP Transmissible spongiform encephalopathies TSEs also called prion diseases are invariably fatal neurodegenerative infectious diseases that include bovine spongiform encephalopathy commonly called mad cow disease chronic wasting disease scrapie and Creutzfeldt Jakob disease To develop antemortem diagnostics or therapies for TSEs the committee concludes that NPRP should invest in basic research specifically to elucidate the structural features of prions the molecular mechanisms of prion replication the mechanisms of TSE pathogenesis and the physiological function of prions normal cellular isoform *Advancing Prion Science* provides the first comprehensive reference on present knowledge about all aspects of TSEs from basic science to the U S research infrastructure from diagnostics to surveillance and from prevention to treatment **Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease, Seventh Edition** Roger N. Rosenberg,Juan M. Pascual,2024-10-04 Rosenberg s *Molecular and Genetic Basis of Neurologic and Psychiatric Disease Seventh Edition* provides a comprehensive introduction and reference to the foundations and key practical aspects relevant to neurologic and psychiatric disease This volume has been thoroughly revised and includes newly commissioned chapters on ethics genetic counselling and genet therapy for the central nervous system disorders A favorite of over four generations of students clinicians and scholars this new edition retains and expands the informative concise and critical tone of the previous edition This is an essential reference for general medical practitioners neurologists psychiatrists geneticists and related

professionals and for the neuroscience and neurology research community at large Both volumes combined provide a comprehensive coverage on the neurogenetic foundation of neurological and psychiatric disease This volume provides a detailed introduction on both the clinical and basic research implications of molecular and genetics surrounding the brain Includes new chapters on genomics of human neurological disorders CRISPR and genome engineering Sleep Disorders Pasquale Montagna,Sudhansu Chokroverty,2011 The second part of a two part work in the Handbook of Clinical Neurology series on sleep disorders Handbook of Laboratory Animal Science Jann Hau,Steven J. Schapiro,Gerald L. Van Hoosier Jr.,2004-11-29 Building upon the success of the Handbook of Laboratory Animal Science and completing Volumes I and II of the Second Edition Handbook of Laboratory Animal Science Second Edition Animal Models Volume III provides the final component to present a comprehensive overview of animal models in biomedical research As with Volume II this new volume add *Ebook: The Science of Psychology: An Appreciative View* King,2016-09-16 Ebook The Science of Psychology An Appreciative View **Aging and Age-related Diseases** Michał Karasek,2006 Ageing has become a great problem for many countries Due to world wide life prolongation the number of people over 60 years old has grown rapidly into a ten percent piece of the world population The growing age of the world population raises many social economical and medical problems The proportion of people in the economically active age groups to those who are over 65 is constantly decreasing A major consequence of the increasing numbers of individuals in advanced age groups is increasing numbers of patients suffering from age related diseases The aim of this book is to present the basic data on human ageing as well as on age related diseases *Incredible Consequences of Brain Injury* Alexander R. Toftness,2022-11-11 Incredible Consequences of Brain Injury The Ways your Brain can Break explains the acquired brain disorders that can suddenly change a person s life Underlining the intricate workings of the human brain and the amazing things it does every day this book examines what happens when the brain stops functioning as it should Through the use of case studies and historical examples this concentrated collection of different neuropsychological conditions provides the reader a glimpse into the lived experiences of each disorder Each chapter is firmly rooted in relevant neuropsychological literature combined with easy to understand explanations and guided reflection In its essence this book is a celebration of the human brain and the myriad factors that make it up serving to maintain hope in recovering from brain conditions and to marvel at the intricate workings of the brain This valuable compendium is essential for anyone who wants to learn more about how the brain functions and dysfunctions and will be equally useful for students instructors and healthcare workers It will further be of use to individuals with brain conditions and their dear ones and for the individuals who are interested in learning more about the human brain *Sleep Disorders Part II* Pasquale Montagna,Sudhansu Chokroverty,2012-11-27 Sleep Disorders II covers various aspects of sleep disorders These include the different classification of sleep disorders the genetic influences of sleep disorders abnormality in the sleeping pattern and the circadian rhythm sleep disorder A sleep disorder is a medical disorder that affects the sleeping

patterns of humans and sometimes animals The disruptions in sleep can be caused by different factors such as teeth grinding night terrors and the like The book also discusses different perspectives on insomnia and hypersomnia According to the International Classification of Sleep Disorders insomnia is a sleep that is low in quality or a difficulty in sleeping On the other hand hypersomnia is a sleeping disorder characterized by excessive daytime sleepiness EDS or prolonged nighttime sleep The book discusses narcolepsy a syndrome that is characterized by excessive daytime sleepiness that is associated with cataplexy and other REM sleep phenomena The different medicines for this disease are also discussed People who are practicing neurology and internal medicine especially those in pulmonary cardiovascular gastrointestinal renal and endocrine specialties will find this book valuable A comprehensive resource for the study of sleep science sleep medicine and sleep disorders Fascinating noninvasive neuroimaging studies that demonstrate marked changes during different sleep states A state of the art reference that summarizes the clinical features and management of many of the neurological manifestations of sleep disorders

Mandell, Douglas, and Bennett's Principles and Practice of Infectious Diseases E-Book John E. Bennett, Raphael Dolin, Martin J. Blaser, Gerald L. Mandell, 2009-10-19 After thirty years PPID is still the reference of choice for comprehensive global guidance on diagnosing and treating the most challenging infectious diseases Drs Mandell Bennett and Dolin have substantially revised and meticulously updated this new edition to save you time and to ensure you have the latest clinical and scientific knowledge at your fingertips With new chapters expanded and updated coverage increased worldwide perspectives and many new contributors Mandell Douglas and Bennett's Principles and Practice of Infectious Diseases 7th Edition helps you identify and treat whatever infectious disease you see Consult this title on your favorite e reader conduct rapid searches and adjust font sizes for optimal readability Compatible with Kindle nook and other popular devices Get the answers to questions you have with more in depth coverage of epidemiology etiology pathology microbiology immunology and treatment of infectious agents than you ll find in any other infectious disease resource Find the latest diagnoses and treatments for currently recognized and newly emerging infectious diseases such as those caused by avian and swine influenza viruses Put the latest knowledge to work in your practice with new or completely revised chapters on influenza new pandemic strains new Middle East respiratory syndrome MERS virus probiotics antibiotics for resistant bacteria antifungal drugs new antivirals for hepatitis B and C Clostridium difficile treatment sepsis advances in HIV prevention and treatment viral gastroenteritis Lyme disease Helicobacter pylori malaria infections in immunocompromised hosts immunization new vaccines and new recommendations and microbiome Benefit from fresh perspectives and global insights from an expanded team of international contributors Find and grasp the information you need easily and rapidly with newly added chapter summaries These bulleted templates include diagnosis therapy and prevention and are designed as a quick summary of the chapter and to enhance relevancy in search and retrieval on Expert Consult Stay current on Expert Consult with a thorough and regularly scheduled update program that ensures access to new developments in the field

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Principles and Practice of Clinical Virology Arie J. Zuckerman,2009-03-12 Principles and Practice of Clinical Virology is the bible for all working in the field of clinical virology from the trainee to the expert because there s always something new to learn As before the book provides a detailed account of the diagnosis and treatment of virus infections with a stronger emphasis on clinical expertise and management Each chapter deals with a single virus or group or viruses and is written by leading international experts in the field What s new in this edition Showcases the wealth of new knowledge acquired on virus infections and reflects the discovery of newly recognized emerging infections the improvement or development of new vaccines and an increasing repertoire of antiviral agents for treatment All chapters have been thoroughly revised and there are a number of new contributors joining the cadre of internationally recognized experts Includes a new chapter on vaccinology covering the principles relating to the development and use of vaccines generally which complements the specific vaccines described in the other chapters The two chapters on nosocomial infections have been enlarged and will be particularly useful for those having to advise on the management of hospital acquired infections Emphasizes the rapid accumulation of new information in such fields as retroviruses particularly HIV SARS hepatitis C and influenza including avian influenza **Sleep Disorders Medicine** Sudhansu Chokroverty,2017-05-02 Since publication of the first edition in 1994 the second edition in 1999 and the third edition in 2009 many new advances in sleep medicine have been made and

warrant a fourth edition This comprehensive text features 19 additional chapters and covers basic science technical and laboratory aspects and clinical and therapeutic advances in sleep medicine for beginners and seasoned practitioners With the discovery of new entities many new techniques and therapies and evolving basic science understanding of sleep Sleep Disorders Medicine Fourth Edition brings old and new knowledge about sleep medicine together succinctly in one place for a deeper understanding of the topic Neurologists internists family physicians pediatricians psychiatrists psychologists otolaryngologists dentists neurosurgeons neuroscientists intensivists as well as those interested in advancing their knowledge in sleep and its disorders will find this edition to be an invaluable resource to this burgeoning field

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