LEMBAR

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Mechanical thrombectomy (MT) for acute ischemic stroke (AIS) in COVID-19 Indui Jurnal

pandemic: a systematic review

Penulis Artikel Ilmiah

Status Pengusul

Aditya Kurnianto*, Dodik Tugasworo, Yovita Andhitara, Retnaningsih, Rahmi

Ardhini and Jethro Budiman

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Prof. Dr. dr Tri Nur Kristina, DMM, M.Kes.

NIP 195905271986032001

: Fakultas Kedokteran Unit kerja : Ilmu Kedokteran Bidang Ilmu : Guru Besar Jabatan/Pangkat

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23 Desember 2021

Prof. Dr. dr. Hardhono Susanto, PAK

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Current Issue

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Table of Contents



RSS

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Cortisol level in depressed patients and its relation with suicidal risk and anhedonia

Context In recent times, there has been an increased interest in research related to anhedonia. Nevertheless, its linkage to major depressive disorders and underlying neurobiology are still no...

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Original Article

Visual dysfunction and neurological disability in multiple sclerosis patients in correlation with the retinal nerve fiber layer and the ganglion cell layer using optical coherence tomography



Aim The aim of this study was to measure retinal nerve fiber layer (RNFL) and ganglion cell layer (GCL) complex thickness with Cirrus optical coherence tomography (OCT) in Egyptian multiple sc...

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Correlation between median nerve conduction studies and ultrasonography in cases of carpal tunnel syndrome



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Carotid artery stenting in high-risk patients: a single-center experience



Background Carotid artery stenting (CAS) is a valid alternative to carotid endarterectomy in selected patients. Periprocedural risk of complications is the main determinant for CAS validity. C...

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Background Early stages of amyotrophic lateral sclerosis (ALS)-like syndrome when only one limb is affected can be missed by physicians suggesting other unrelated disorders especially with evi...

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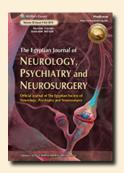
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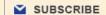


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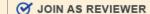
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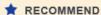












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Iyus Yosep, Henny Suzana Mediani, Linlin Lindayani, Aat Sriati

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Ondine's curse, a fatal infarction diagnosed by polysomnography and saved by ventilation: a case report

Hany Aref, Tamer Roushdy, Amr Zaki, Nevine El Nahas

Article keywords

Ondine's curse, Lateral medullary infarct, Posterior circulation stroke, Sleep apnea

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<u>Risk homozygous haplotype regions for autism identifies population-specific ten genes for numerous pathways</u>

Swati Agarwala, Nallur B. Ramachandra

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Homozygosity, Recessive genes, Autism, Mutations, Haplotype blocks

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Mechanical thrombectomy (MT) for acute ischemic stroke (AIS) in COVID-19 pandemic: a systematic review

Aditya Kurnianto, Dodik Tugasworo, Yovita Andhitara, Retnaningsih, Rahmi Ardhini, Jethro Budiman

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Nikita Mohan, Muhammad Ali Fayyaz, Christopher del Rio, Navpreet Kaur Rajinder Singh Khurana, Sampada Sandip Vaidya, Esteban Salazar, John Joyce, Amrat Ayaz Ali

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SARS-CoV 2, Stroke, Neuro-invasive, COVID-19, Neuroimaging

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THE EGYPTIAN JOURNAL OF NEUROLOGY, PSYCHIATRY AND NEUROSURGERY (MAY 2021)

<u>Telephone-based assessment of multiple sclerosis patients at Ain Shams University Hospital in the coronavirus disease 2019 pandemic</u>

Mohamed A. Abdel Hafeez, Dina A. Zamzam, Mahmoud S. Swelam, Alaa Abo Steit, Janet Masoud, Azza Abdel Nasser, Ahmed Hazzou, Eman Hamid, Hany Aref, Magd F. Zakaria, Mohamed M. Fouad

Article keywords

COVID-19, Multiple sclerosis, Telephone calls, Hauser ambulation index, Multiple sclerosis neuropsychology questionnaire, Symptoms of multiple sclerosis scale

Abstract+

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RESEARCH Open Access

Risk homozygous haplotype regions for autism identifies population-specific ten genes for numerous pathways



Swati Agarwala and Nallur B. Ramachandra *

Abstract

Background: Recessive homozygous haplotype (rHH) mapping is a reliable tool for identifying recessive genes by detecting homozygous segments of identical haplotype structures. These are shared at a higher frequency amongst probands compared to parental controls. Finding out such rHH blocks in autism subjects can help in deciphering the disorder etiology.

Objectives: The study aims to detect rHH segments of identical haplotype structure shared at a higher frequency in autism subjects than controls to identify recessive genes responsible for autism manifestation.

Methods: In the present study, 426 unrelated autism genotyped probands with 232 parents (116 trios) were obtained from Gene Expression Omnibus (GEO) Database. Homozygosity mapping analyses have been performed on the samples using standardized algorithms using the Affymetrix GeneChip® 500K SNP Nsp and Sty mapping arrays datasets.

Results: A total of 38 homozygous haplotype blocks were revealed across sample datasets. Upon downstream analysis, 10 autism genes were identified based on selected autism candidate genes criteria. Further, expressive Quantitative Trait Loci (QTL) analysis of SNPs revealed various binding sites for regulatory proteins *BX3*, *FOS*, *BACH1*, *MYC*, *JUND*, *MAFK*, *POU2F2*, *RBBP5*, *RUNX3*, and *SMARCA4 impairing essential autism genes CEP290*, *KITLG*, *CHD8*, and *INS2*. Pathways and processes such as adherens junction, dipeptidase activity, and platelet-derived growth factor—vital to autism manifestation were identified with varied protein-protein clustered interactions.

Conclusion: These findings bring various population clusters with significant rHH genes. It is suggestive of the existence of common but population-specific risk alleles in related autism subjects.

Keywords: Homozygosity, Recessive genes, Autism, Mutations, Haplotype blocks

Background

Autism is a heritable, neurodevelopmental condition affecting information processing in the brain, heterogeneous with < 15% known genetic causes. It has a world-wide prevalence rate of 1 in 59 children being affected [1]. It alters connections and organization of nerve cells and their synapses, impairing the overall cognition, emotional, social, and physical health of the affected individuals [2].

The study of recessive risk gene loci is performed through extended runs of homozygosity (ROH) as a genomic feature, useful to map recessive disease genes in outbred populations [3, 4].

The authors expect to find an unusually higher number of affected individuals in complex disorders to have the identical haplotype in the region surrounding a disease [5, 6]. Therefore, a rare pathogenic variant and surrounding haplotype are often enriched in frequency in a group of affected individuals compared to a cohort of unaffected controls [7]. These variants in the haplotypes

Department of Studies in Genetics and Genomics, University of Mysore, Mysuru, Karnataka 06, India



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CASE REPORT Open Access

Ondine's curse, a fatal infarction diagnosed by polysomnography and saved by ventilation: a case report



Hany Aref, Tamer Roushdy , Amr Zaki and Nevine El Nahas

Abstract

Background: Lateral medullary syndrome causing Ondine's curse is a rare yet fatal brainstem infarction. Any patient presenting with lateral medulla infarction ought to be well observed and a polysomnography must be ordered for him.

Case presentation: A patient presenting with Ondine's curse is dealt with through polysomnography as a diagnostic procedure that was followed by tracheostomy with portable ventilator and cardiac pacemaker as a therapeutic maneuver which ultimately preserved his life.

Conclusion: Lateral medullary syndrome infarct could be a life-threatening stroke if not diagnosed and managed properly.

Keywords: Ondine's curse, Lateral medullary infarct, Posterior circulation stroke, Sleep apnea

Background

The medulla oblongata controls vasomotor and respiratory functions. It is considered the primary respiratory control center, as it sends signals from the respiratory central pattern generators to muscles controlling breathing [1].

Stroke involving the lateral medulla oblongata is named Wallenberg syndrome (lateral medullary syndrome) [2].

Lateral medullary syndrome (LMS) typically presents with hiccup, vertigo, nystagmus, vomiting, dysphagia, dysarthria, and dysphonia that usually resolves without fatal sequel. Two to 6% of LMS presents with disturbance in vital functions causing central apnea, bradycardia, and hypoventilation (Ondine's curse) that is fatal if not properly identified and managed [3].

Ondine's curse is named after a mythical story of a man who was doomed to a life where he keeps breathing only while awake and conscious. So there was always a choice between sleeping and remaining alive. In the current case report, we present a case of Ondine's curse due to LMS.

Case presentation

The case includes a 57-year-old male, diabetic, hypertensive, with recurrent cerebrovascular strokes dating 2013 and 2016 (modified Rankin score (mRS) 1).

In August 2018, he presented to the emergency room (ER) with sudden onset, 11 h duration of swaying of gait to the left, vertigo, nausea, and vomiting followed by dysphagia to fluids with nasal regurgitation, hoarseness of voice, and partial ptosis of left eye secondary to partial involvement of descending sympathetic tract (National Institute of Health Stroke Scale (NIHSS) 5).

MRI brain with diffusion-weighted imaging (DWI) revealed left lateral medullary infarction (Fig. 1).

Few hours after admission, he suffered from tachypnea with drop of oxygen saturation to 60%, which was followed by bradycardia (17 beats per minute) and gasping. The patient was rushed to the intensive care unit (ICU) and intubated for assisted ventilation using bi-

^{*} Correspondence: Tamer.roushdy@med.asu.edu.eg Neurology Department, Faculty of Medicine, Ain Shams University, Cairo, Egypt

