MICRO LABS SYNAPSE

Interesting cases NEUROLOGY

Vol 2|Issue 8|2021

Dear Doctor,

Greetings from Micro Labs Limited.

We are happy to bring you "Neurology case series", which contains latest and interesting cases in the field of Neurology.

This issue contains

1. Novel Mutation in CACNA1A Associated with Activity-Induced Dystonia, Cervical Dystonia, and Mild Ataxia
2. Takotsubo Cardiomyopathy After Traumatic Brain Injury- Case Report
3. Case report on Ice Pack Test Eased Ptosis in a Patient Presenting with a Possible Oculomotor Nerve Schwannoma

Should you require any specific article or medical information, please feel free to contact us at medicalservices@microlabs.in

Looking forward to your valuable feedback

Best Regards,

Medical Team,

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Novel Mutation in CACNA1A Associated with Activity-Induced Dystonia, Cervical Dystonia, and Mild Ataxia

Patient description:

A 30-year-old woman presented with the history of slowly progressive, activity-induced stiffness and pain in her right leg since age 15. Birth and early childhood history were unremarkable. Around age 15, the subject developed symptoms which began as a limp in her right leg. She denied any illnesses, exposures, or trauma around this time. At age 20 the subject experienced neck pain and stiffness with occasional neck spasms with decreased range of motion.

When the patient was evaluated in clinic she denied right leg stiffness or pain at rest and when she began to walk, her right foot turned in and her right leg, knee, proximal thigh, and hip stiffened up. She denied tingling, numbness, burning. right arm, left arm, or left leg symptoms, gait instability or feeling of imbalance.

She was using a cane and wearing left and right leg braces, which helped with her walking. She was finding difficulty with stairs and with walking long distances. No family history of similar symptoms.

Physical examination revealed that her strength, tone, and reflexes were normal in all extremities at rest. Mild retrocolis, right laterocolis, and right torticolis was observed at rest. Mild head titubation and very mild past pointing on finger-to- nose testing was observed. No other signs of dysmetria or ataxia. Lower extremity study was deferred because of receiving botulinum toxin injections to the right leg. A tentative diagnosis of activity-induced dystonia was made in light of her history, symptoms, and physical exam.

The subject was on trial with carbidopa/levodopa without improvement and was also receiving regular botulinum toxin injections for her cervical dystonia and right leg stiffness, with some improvement in her symptoms. She was also given with cyclobenzaprine as well, with some improvement in her right foot pain and neck spasms.

A dystonia panel was sent, and showed two potentially pathologic mutations, one in CACNA1A and the other in PNKP, along with a variant of unknown significance in ATP7B. The mutation in CACNA1A is C2324 G <A, protein W775X. She also has a heterozygous pathological mutation

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in PNKP, C1029 + 2 T <C, though this is autosomal recessive. After receiving genetic diagnosis, the subject was trialed on acetazolamide without improvement in her dystonia symptoms.

Sex	Female
Age of onset	15
Initial symptom	Stiffness and pain in her right leg with activity (started age 15)
Other symptoms	Neck pain and stiffness, with occasional neck spasms (started age 20)
Functional impact	Difficulty with stairs and with walking long distances
Personal medical history	Unremarkable birth and early childhood history
Family history	No family history of similar symptoms
Exam findings	Normal strength, tone, and reflexes in all extremities at rest Mild retrocolis, right laterocolis, and right torticolis at rest Mild head titubation and very mild past pointing on finger-to-nose testing
Imaging	Unremarkable MRI of the brain and spinal cord
Other diagnostic studies	EMG/NCS of the left arm was unremarkable
Attempted treatments	Carbidopa/levodopa: no improvement Botulinum toxin injections: symptomatic relief of cervical dystonia Cyclobenzaprine: symptomatic relief Acetazolamide: no improvement
Genetic testing	*CACNA1A*, C2324 G < A: novel mutation, heterozygous, autosomal dominant, computer modeling suggests pathogenicity *PNKP*, C1029 + 2 T < C: heterozygous, autosomal recessive, known pathological mutation *ATP7B*, C2544 C < T: autosomal recessive, variant of unknown significance

Clinical characteristics and genetic testing

Conclusion:

Patient's dystonia may be due in part to cerebellar dysfunction related to her CACNA1A mutation. This case study reports a novel CACNA1A variant associated with activity-induced dystonia of the right leg, cervical dystonia, and mild ataxia.

Reference:

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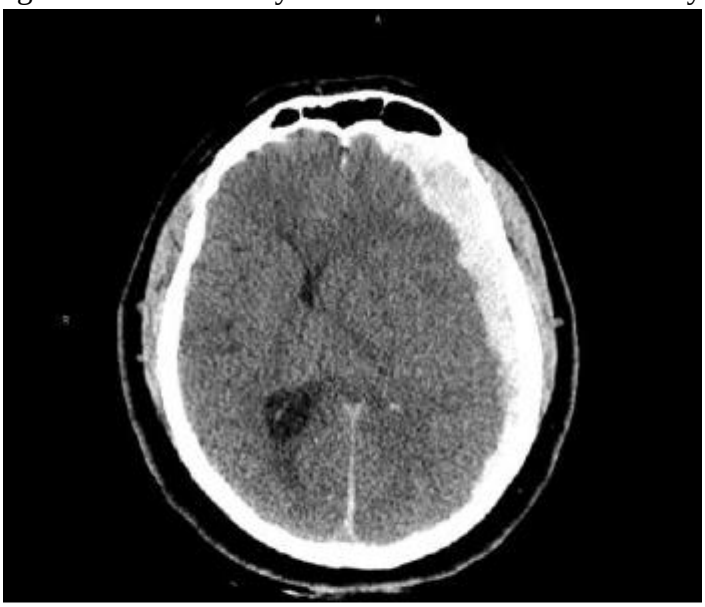
Stampfl B, Fee D. Novel Mutation in CACNA1A Associated with Activity-Induced Dystonia, Cervical Dystonia, and Mild Ataxia. Case Reports in Neurological Medicine. 2021 Aug 3;2021.

Takotsubo Cardiomyopathy After Traumatic Brain Injury- Case Report

Patient description:

A 30-year-old male patient presented with a history of alcohol use and was found unresponsive with unknown down time in his hotel room by the hotel staff. Glasgow Coma Score (GCS) was 13 his GCS rapidly declined after arriving in the trauma bay and was intubated for airway protection. Dilated pupil on the left side with GCS of 6T after a brief sedation interruption was revealed by Neurological examination.



Computed tomography head showed an acute left subdural hemorrhage with significant left-to-right midline shift.

Ecchymosis on the left chest, shoulder and knee was revealed by secondary trauma survey. Computed tomography (CT) scan of the head reaveled 19 mm left convexity acute subdural hematoma with 14 mm of midline shift as well as uncal herniation. Small pneumomediastinum and mild left-sided pneumothorax was observed in CT Scan. Toxicology screen was only positive for ethanol with a level of 45 mg/dL. For decompressive hemicraniectomy and hematoma evacuation the subject was taken immediately to the operating room.

To the bronchial trees or oesophagus, intra-operative bronchoscopy and esophagogastroduodenoscopy did not reveal additional injuries. The subject was admitted to neurotrauma ICU for close monitoring and his GCS improved to 10T, Postoperatively. The subject had a routine admission electrocardiogram on the post-trauma day (PTD) 1 which revealed non-specific T wave changes with a normal ejection fraction. On post-trauma day- 2 there was an improved GCS of 13 and the subject developed tachycardia and acute hypoxia on post-trauma day-

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3. Chest X-ray showed mild pulmonary edema. His oxygen supplementation was escalated to high flow nasal canula at 45 liters per minute. Following the injury hypoxia was attributed to pulmonary edema secondary to aggressive fluid resuscitation. With diuresis, and supplemental oxygen his respiratory status rapidly improved. T waive inversions on the night of post-trauma day -4 was noted. ECG revealed prolonged QTc interval and worsening TWI on leads I, II, aVL, and V2-6. The subject did not complain of chest discomfort or shortness of breath.

Troponin I was obtained given these new ECG changes and returned mildly elevated at 0.63 ng/mL. Troponin I downtrended to 0.54 ng/mL and then to 0.36 ng/mL by the next morning. Transthoracic echocardiography was done and showed that left ventricular apical ballooning with an estimated EF of 26%. Heart failure symptoms were not observed. Cardiology consultation recommended the initiation of lisinopril and metoprolol for the management of TCM. The subject was monitored in the telemetry unit until discharge to a rehabilitation facility for the remainder of his hospitalization. Follow-up of Transthoracic echocardiography on post-trauma day - 20 revealed improvement in ejection fraction to 50–55%.

Conclusion:

While rarely reported in subjects with traumatic brain injury, Takotsubo cardiomyopathy developed in an otherwise healthy young male following severe traumatic brain injury necessitating decompressive hemicraniectomy. Transthoracic echocardiography should be considered in subjects with traumatic brain injury who have cardio-pulmonary symptoms or unexplained ECG abnormalities.

Reference:

Wang F and Darby J (2021) Case Report: Takotsubo Cardiomyopathy After Traumatic Brain Injury. Front. Neurol. 12:727754. doi: 10.3389/fneur.2021.727754

Case report on Ice Pack Test Eased Ptosis in a Patient Presenting with a Possible Oculomotor Nerve Schwannoma

Patient description:

A 37-year-old man presented to the ophthalmologist with the complains of left-eye lacrimation left-eye lacrimation. The examination showed that ptosis, limitation of upward, downward, and inward gaze, and mydriasis, and the patient was diagnosed with complete third nerve palsy.

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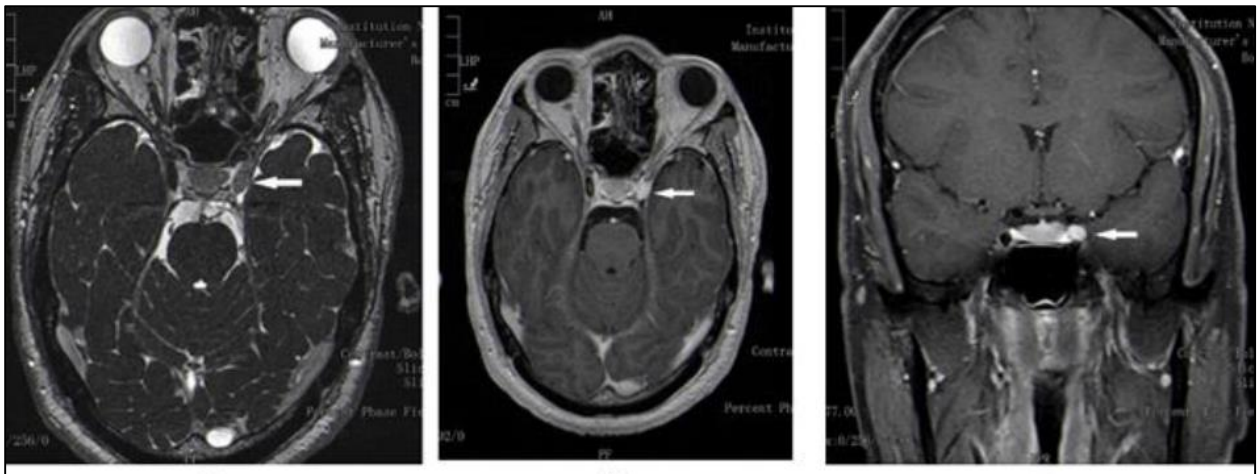
Fundus examination and intraocular pressure was normal. Fluorescein angiography, indocyanine green chorioangiography, and optical coherence tomography were all negative. The subject reported a history of mild left-sided head trauma which was experienced 1.5 years earlier. No other medical history was observed and there was no family history of metabolic, neurological, or autoimmune disorders.

The subject was referred to neurological clinic where left oculomotor nerve palsy was confirmed. No other neurological deficit was found. He reported that he experienced fluctuation of his left-eyelid droop during daytime and improvement was observed when washing his face with cold water. The ptosis was developed slowly over the past 4–5 months. The ice pack test (IPT) was performed to exclude myasthenia gravis (MG) associated with the oculomotor nerve palsy. An ice pack was applied to the subjects left eyelid for 2 min and his ptosis improved significantly. Although the improvement was only temporary which lasted for 2 minutes but the patients left eye opened nearly completely.



(a) T2-weighted axial magnetic resonance imaging (MRI) brain without Gadolinium. (b) T1-weighted axial brain MRI with gadolinium (c) T1-weighted coronal brain MRI with gadolinium

Plain and post-contrast brain magnetic resonance imaging (MRI) showed a 10 × 6× 7mm nodular lesion on the left cavernous sinus lateral wall. Neuroradiologists supposed a schwannoma of the left third cranial nerve. Neuroradiologists supposed a schwannoma of the left third cranial nerve. Due to the lesion's small size and localization stereotactic biopsy was not performed. There were

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also no radiological signs of raised intracranial pressure, vascular ischemia, haemorrhage, or cavernous sinus infection or trauma. Brain and neck computed tomographic angiography was performed as oculomotor nerve schwannoma (ONS) are extremely rare and ruled out a thrombosed aneurysm of the posterior communicating artery and a thrombosis of the left cavernous sinus. Dislocation of the left internal carotid artery was revealed by the brain MRI angiography but a Doppler ultrasound study showed completely normal blood flows. On physical examination there was no evidence of any specific skin lesions.

The final diagnosis, based on clinical and MRI findings was left third nerve palsy caused by a possible sporadic ONS in the left cavernous sinus. Neurosurgeons decided and referred the patient for Gamma Knife radiosurgery (GKRS). This method was chosen due to the small tumour size (<3 cm), the difficult location for surgical approach, and because it can destroy such lesions.

Conclusion:

This case study showed that IPT can also be positive in neurogenic (non-myasthenic) ptosis, but its usefulness in other disorders associated with muscle weakness and fatigability remains questionable. IPT eased ptosis in a patient presenting with a possible oculomotor nerve schwannoma (ONS), is interesting because it shows that the IPT can give a positive result in neurogenic (non-myasthenic) ptosis.

References:

Buechner S, Capone L. Ice Pack Test Eased Ptosis in a Patient Presenting with a Possible Oculomotor Nerve Schwannoma: A Case Report. Neurology International. 2021 Oct;13(4):510-6.