



Conclusions

In most previous reports as in our case, the “belly dancing” epileptogenic zone correspond to the mesial frontal and/or parietal areas around the central sulcus, in agreement with the trunk somatotopic representation. The limited representation of abdomen and the overlapping representations of body parts over the sensory/motor homunculus, may account for the rarity of isolated epileptic “belly dancing” phenomenon.

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IRF2BPL nonsense mutation associated with adult onset myoclonic epilepsy and cerebellar ataxia: A case report

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Background and aims

The Interferon Regulatory Factor 2 Binding Protein-Like (IRF2BPL) is a recently discovered gene, whose mutations are associated with heterogeneous neurological phenotypes, in most cases developmental and epileptic encephalopathies.

We present the case of an IRF2BPL mutation associated with adult-onset myoclonic epilepsy and cerebellar ataxia.

Methods

A 22 years-old female patient with mild learning disabilities developed a progressive decline in cognitive and motor functions, manifesting mental slowness, forgetfulness, unsteady gait with frequent falls, mild speech ataxia, action tremor, myoclonic jerks, and a single generalized tonic-clonic seizure. Family-history was

negative, and brain imaging together with laboratory tests were normal. Prolonged EEG showed diffuse epileptiform abnormalities, frequent multifocal myoclonus, and a photo-paroxysmal response. The myoclonic jerks improved with Levetiracetam and Clonazepam, while cognitive and motor impairment persisted.

Results

Whole-exome-genome sequencing showed a never-before-described heterozygous nonsense mutation (c.364C>T, p. Gln122Ter) in the IRF2BPL gene.

Leading to the insertion of a premature STOP codon, this mutation is considered potentially pathogenic according to updated recommendations.

Conclusions

IRF2BPL mutations are mostly associated with severe neurological regression. Our patient showed a different phenotype, resembling the clinical features of Progressive Myoclonic Epilepsy (PME): myoclonic jerks, seizures and progressive neurologic impairment. Although the most common PME phenotypes are attributed to specific genetic defects, almost 30% of PMEs are of undetermined cause and this is the first report of an IRF2BPL mutation.

Our report needs confirmation by further studies; however, it contributes to the knowledge of the heterogeneity of the IRF2BPL-mutations phenotype and suggests that it should be considered in patients with PME.

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Role of intensity of electrical stimulation (ES) in the cerebellar antiepileptic effectiveness under conditions of diazepam administration

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Background and aims

ES of brain structures combined with antiepileptic drugs is a promising approach for better pharmacological control of resistant seizures. Aims. To investigate the dynamic of penicillin-induced foci in cats' brain cortex under conditions of paleocerebellar electrical stimulation (ES) delivered at different intensities and diazepam administration.

Methods

Via penicillin solution (20.000 IU/ml) application upon posterior sigmoid gyrus epileptic foci have been created in anesthetized, myorelaxed, and artificially ventilated cats. ES of paleocerebellum (100 Hz, 1 ms, duration of trial 1 s) have been delivered to VI lobula of paleocerebellum with two bandwidths of intensities: 80–150 mcA (lower) and 200–500 mcA (higher) ones. Diazepam was administered in dose 2.0 mg/kg, i.v. The monopolar EEG was registered, and i.v. saline administration to animals was used as control.

Results

The frequency of epileptic discharge generation at the maximal level of epileptic activity was 25–40 discharges per min. During ES delivered at a lower intensity, the decrease of discharges frequency by two-three times was observed. Higher frequency of ES induced activation of spikes generation up to 50–75 per min. In 30.0 min from the diazepam administration and 20 min from the moment of restoration of focal activity via additional penicillin application, lower intensity ES did not modulate spikes generation, while higher

intensity caused up to twice reduction of spikes frequency or complete suppression of focal activity.

Conclusions

Modulative effects of paleocerebellar ES on cortical penicillin foci depend on its intensity. Diazepam in high dose abolished effects of low-intensity ES while higher intensity induced suppression of epileptic activity.

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The effects of antiepileptic drugs on high-frequency oscillations in somatosensory evoked potentials

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Background and aims

High frequency oscillations (HFOs) of Somatosensory evoked potentials (SEPs) reflect the activity of thalamo-cortical and cortical neurons from the sensory pathway. Antiepileptic-drugs (AEDs) reduce seizures acting on the balance between excitation and inhibition. We aimed to study the effect of AED mono and polytherapy on SEP-HFO's components.

Methods

Twenty-five patients with focal epilepsy were enrolled for the purpose of this study. Patients were divided in 3 groups according to the number of AEDs (1, 2 or 3 AEDs). Patients in group 1 underwent SEP-HFOs recording in drug naïve condition and at 1 month after AED titration. HFOs were compared in duration, amplitude and latency among the three groups.

Results

The amplitude and duration of late HFOs of the affected hemisphere (AH) are different between groups and inversely correlated with the number of AEDs. In naïve patients monotherapy reverts the asymmetry in totHFOs (total HFOs) duration.

Conclusions

Our results demonstrate that SEP-HFOs are sensitive to the action of AEDs on cortical excitability. This effect seems to affect mainly the cortical component of HFOs in the AH and it is related to the number of AEDs taken. Significance: SEP-HFOs might be a viable tool to probe cortical excitability changes induced by AEDs. © 2020 International Federation of Clinical Neurophysiology.

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The use of optically pumped Magnetoencephalography (OP-MEG) in epilepsy

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Background and aims

Magnetoencephalography (MEG) has been used in specific cases of refractory epilepsy due to its inherent properties of excellent spatial resolution and relative immunity to muscle artefact. However current MEG systems are high maintenance and restrictive for the patient, meaning that recording sessions are usually brief (1–2 h), thereby limiting its availability in the clinical setting. Our aim was to demonstrate the first use of Optically Pumped Magnetoencephalography (OP-MEG) in two epilepsy patients with unrestricted head movement. Unlike conventional MEG that uses a superconducting SQUID system, here we use a different type of sensor (OPM), which operates at room temperature and crucially can be placed directly on the patient's scalp, permitting free head movement.

Methods

We performed three OP-MEG recording sessions in two patients with refractory focal epilepsy (right posterior temporal focus in patient 1 and left frontal focus in patient 2), who were able to perform natural movements within their environment. We then compared these recordings with clinical scalp EEG performed earlier. In the second patient we also incorporated motion capture in order to regress out head movement from the MEG signal.

Results

OP-MEG was able to identify analogous interictal activity to scalp EEG including spikes, polyspikes, sharp waves and spike and wave activity. We were able to source localise this activity using both linearly constrained minimum variance (LCMV) beamformer and equivalent current dipole (ECV) methods to an appropriate brain region.

Conclusions

This is the first application worldwide of OP-MEG in human epilepsy. Future directions include simultaneous EEG/OP-MEG recording and prolonged OP-MEG telemetry.

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Pioglitazone intensifies antiseizure effects of cerebellar transcranial direct current stimulation (TDCS) upon kindled seizures

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Background and aims

The suppression of epileptic activity follows TDCS. It was shown that blocking peroxisomal proliferator-activated γ -receptors (PPAR γ) with BADGE, (100 mg/kg, i.p.) prevented antiseizure effects caused with cerebellar TDCS (Godlevsky L.S., Pervak M.P., 2019). Aim. To investigate effects of cerebellar TDCS under conditions of treatment with the agonist of PPAR γ pioglitazone upon pentylenetetrazol (PTZ) - kindled seizures.

Methods

In rats the kindling was induced via PTZ (35.0 mg/kg, i.p.) for three weeks. TDCS was performed at 250 μ A intensity for 10.0 min, with cathode on the skull surface oriented to the cerebellar cortex.