MOVEMENT DISORDERS

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STATUS DYSTONICUS IN CHILDREN- CASE REPORTS

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INTRODUCTION: Status dystonicus (SD) is a medical emergency leading to a significant morbidity and mortality. It can affect either patients with primary or secondary dystonia. SD is characterized by episodes of generalized muscle contraction sustained and refractory to medical treatment. Usually is possible to identify triggers as fever, infections or abrupt discontinuation of treatment. From a therapeutic point of view there are no evidence-based management guidelines in SD.

OBJECTIVE: we report on four patients with status dystonicus describing their clinical manifestations, treatments and evolution.

MATERIALS AND METHODS: we reviewed medical records of patients admitted to our department with a diagnosis of status dystonicus from January 2009 to June 2012.

RESULTS: we present four male patients with secondary dystonia with usual drug treatment. Range of age at diagnosis was 4-13 years. Fever was the most frequent trigger. Three patients required intravenous benzodiazepines without mechanical ventilation needed; two of which showed unfavorable response, and required implementation of baclofen intrathecal infusion pump, achieving the resolution of the SD and decline in basic medication. Only one patient was managed optimizing oral medication.

CONCLUSION: Status dystonicus is a very difficult therapeutic management condition. Early use of surgical techniques, such as baclofen intrathecal infusion pump should be considered before treatment failure or relapse with intravenous benzodiazepines. We emphasize the importance of preventing and controlling triggers. There is an urgent need to develop evidence-based treatment guidelines.

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HYPOMYELINATION WITH ATROPHY OF THE BASAL GANGLIA AND CEREBELLUM - CASE REPORT

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INTRODUCTION: Recently, leukoencephalopathies associated with hereditary disorders of myelin formation and metabolism became focus of the study of congenital causes of white-matter diseases. Among the non-classified leukoencephalopathies, the group hypomyelination diseases stand out as the largest category.

CASE REPORT: Female patient, 4 years old, presented motor and speech delay which was noticed after 18 months of life and evolved with movement disorder and loss of motor skills. It was observed dystonic posturing of limbs, asymmetric intent dystonia, head wobble and bilateral intention tremor of the upper limbs, those associated with global cerebellar ataxia. The patient had a brain magnetic resonance imaging (MRI), performed 7 months earlier, which demonstrated abnormalities of the periventricular and subcortical white matter - involving the U-fibers, with hyperintense signal on T2 and FLAIR and cerebellar atrophy. Another MRI performed in our service showed the same features, now with reduced volume in both of the putamens. Imaging findings fulfil the criteria of hypomyelination, with the characteristic changes of HABC.

DISCUSSION: The first description of this entity was made in 2002 and, since then, the diagnosis is based on neuroimaging criteria. The clinical picture is heterogeneous, including developmental delay, ataxia and extrapyramidal signs. Greater severity was associated with a higher degree of atrophy of the basal ganglia, with description of complete disappearance of putamen. Recently been associated with de novo mutation of the β - tubulin gene, TUBB4A, which was related to congenital dystonia.

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PAROXYSMAL DYSKINESIAS IN CHILDHOOD

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Introduction: Paroxysmal Dyskinesias are involuntary intermittent movements, manifested with dystonia, chorea, athetosis, tribalism or a combination of them. They are secondary to ion channels defects.

Objectives: The aim of this study is to define the most frequent forms of presentation in our population and the management response.

Materials and Methods: Retrospective study of medical records of 15 patients with paroxysmal dyskinesias treated at our center between 1991-2010

Results: 15 patients, 6 women and 9 men were evaluated, all of them presented normal development and no other pathologies. 4 had family history of abnormal movements (3 were brothers, who also had febrile seizures history). The average age of onset was 3.9 years. Dystonia: 6 patients generalized, 3 patients hemidystonia (6 nonkinesigenic and 3 kinesigenic). Choreoathetosis: 5 generalized and 1 hemichorea, 4 of them were nonkinesigenic and 2 kinesigenic. All patients had normal imaging, electrophysiological and metabolic studies. 7 patients received medical treatment with carbamazepine and 4 with acetazolamide, all of them with good clinical response.

Conclusion: In our study we found a male predominance in all dyskinesic patients. Dystonia were more common than choreoathetosis. Kinesigenic dyskinesia was predominant. There was a good response to symptomatic treatment. Three patients corresponded to a familial form of paroxysmal choreoathetosis with febrile seizures. We found no secondary causes.

Discussion: Although dyskinesias are often sporadic or familial and presented frequently spontaneous remission, it is important to look for secondary causes and discard epilepsy as the main differential diagnosis.

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PRIMARY MYOCLONUS-DYSTONIA – OFTEN UNDERDIAGNOSED ENTITY: REPORT OF FOUR AFFECTED FAMILIES

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Introduction: Primary myoclonus-dystonia is a rare autosomal-dominant movement disorder characterized by a combination of myoclonic jerks and dystonia. We describe the clinical phenotype in the 4 affected families.

Methods: The case records of the four families affected with Primary myoclonus-dystonia attending the Neurology-Clinic were reviewed. The clinical features were analyzed and reported

Case description: All the affected children had onset in the first decade of life. The father of one child had symptom onset in third decade of life. Myoclonic jerks affecting the upper limbs (7/7), head/neck region (5/7), trunk (2/7) or lower limbs (2/7) were the main presenting features. Dystonias were subtle and included writer's cramp (5/7), cervical dystonia (1/7) and toe dystonia (1/7). None of the patients had behavioural problems at the point of presentation. The investigations for the secondary causes were unremarkable.

Conclusions: Primary myoclonus-dystonia is an underdiagnosed entity and raised awareness among the paediatric neurologists is required.

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MOVEMENT DISORDER IN THREE FILIPINO ADOLESCENTS WITH ANTI NMDA RECEPTOR ENCEPHALITIS: A CASE SERIES

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Objective: To report the clinical features of anti NMDA receptor encephalitis in 3 Filipino adolescents.

Background: Anti-N-Methyl-D-Aspartate (NMDA) Receptor Encephalitis represents a new category of immune-mediated disorders that typically present with fever, neuropsychiatric manifestations, movement disorder, aphasia and dysautonomia. It has been reported to commonly occur in young women with teratomas but has also been diagnosed in men and children without tumors.

The Cases: We report 3 previously healthy adolescents with serum and cerebrospinal fluid that tested positive for anti-NMDA receptor antibodies. The first case is a 14 year old female who presented with fever, seizures, disorientation, aphasia, hemiparesis, unresponsiveness, dystonia and choreoatheosis. The second case is a 14 year old male who presented with fever, prominent psychiatric symptoms, persistent hypertension, and orofacialdyskinesias at the onset of illness and then later developed global aphasia, catatonia, ileus, urinary and bowel incontinence. The 3rd case is a 15 year old male who presented with a 3 month history of bizarre behavior, followed by neck and truncal dystonia and orofacialdyskinesias. Work up for associated tumors was negative in all three patients. Immunotherapy was given to all three patients, IVIG in one and intravenous methylprednisolone in the other two. The dyskinesias resolved 5 to 7 days after treatment.

Conclusion: Abnormal posture and movements are important features of anti- NMDA receptor encephalitis. Its presence in a child or adolescent manifesting with behavioural changes, neuropsychiatric manifestations, seizures, and autonomic instability should alert the clinician to this condition.

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CHOREA AS A MANIFESTATION OF CNS VASCULITIS IN A 6-YEAR-OLD BOY WITH SYSTEMIC LUPUS ERYTHEMATOSUS

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Systemic Lupus Erythematosus (SLE) of pediatric onset represents 10% to 20% of all SLE cases. Neuropsychiatric manifestations are found in up to 50% of children with SLE within the first year from the time of diagnosis. Although chorea is a rare neuropsychiatric manifestation of SLE, when present, it is usually associated with the presence of antiphospholipid antibodies (aPL). Reports of patients with SLE chorea without aPL antibodies suggest that at least in some cases there is a different pathophysiologic mechanism. We describe the case of a six-year-old boy with a diagnosis of SLE that presented with lupus nephritis and subsequently experienced left hemichorea without MRI evidence of brain lesions. Antiphospholipid panel was negative. Brain SPECT revealed moderately decreased perfusion-metabolism in the right posterior parietal and occipital cortex suggestive of vasculitis. Patient was treated for a CNS vasculitis and symptoms of chorea, which resolved within several months. This case highlights the major clinical differences between adult and pediatric onset SLE and demonstrates that in SLE patients with negative aPL, CNS vasculitis may be the underlying etiology for the neuropsychiatric manifestations, including chorea.

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BOTULINUM TOXIN- A IN PEDIATRIC STIFF HIPS

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Objective: We aim to determine the functional and orthopedic contribution of botulinum toxin-A in the treatment of pediatric stiff hips.

Methods: Three patients with complicated developmental dysplasia of the hip after revision surgery and a case of idiopathic chondrolysis were given botulinum toxin injections in selected muscles as an adjunctive therapy to the standard orthopedic management.

Results: All patients experienced significant reduction in pain with significant improvement, in posture, range of motion and mobility.

Conclusion: Botulinum toxin- A injection when given to selected muscles appears effective in relieving pain and improving range of motion in patients with complicated developmental dysplasia of the hip and idiopathic chondrolysis.

Disclosure: The authors have no conflicts of interest whatsoever.

Acknowledgement: The authors thank Dr.Jaffar Ali, Senior Editor, Research and Publication Center, KFMC, for reviewing the manuscript.

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EFFICACY OF FINASTERIDE IN THE TREATMENT OF ADULTS WITH REFRACTORY TOURETTE DISORDERS.

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The enzyme steroid 5α reductase (S5α R) catalyzes the conversion of Δ^4-3-ketosteroid precursors--such as testosterone, progesterone and androstenedione--into their 5α-reduced metabolites. The 5α-reduced metabolite of testosterone, 5α-dihydrotestosterone (DHT), is the most potent androgen and stimulates prostatic growth led to the development of S5α R inhibitors with high efficacy and tolerability. Two of these agents, finasteride and dutasteride, have received official approval for the treatment of benign prostatic hyperplasia and are being tested for prevention of prostate cancer. Over the last decade, converging lines of evidence have highlighted the role of 5α-reduced steroids and their precursors in brain neurotransmission and behavioral regulation. Some preliminary data suggest that S5 R inhibitors may elicit therapeutic effects in a number of disorders associated to dopaminergic hyperreactivity, including psychotic disorders, Tourette and impulse control disorders. In the past year, we tried finasteride (5mg/day) in 5 male and 1 female adults with severe tics that resist to neuroleptics and other medicines for more than 10 years. Their tics reduced significantly within 1 to 4 weeks in 4 patients including the female one. These 4 patients maintained finasteride well for 6 to 12 months. The 3 males had no abnormal erection, ejaculation, libido, gynecomastia, and any malignancy. The only female had no health problem too. Regular liver ultrasound examination and blood checkup for CBC, PT/APTT, AST, ALT, Bilirubin (T/D), ALK-P, albumin and globulin every 3 months did not show any abnormal changes. Finasteride deserves a further evaluation of clinical efficacy in Tourette disorders.

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A GILLES DE LA TOURETTE (TS) CASE FULLY RECOVERED BY TOPIRAMATE

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Introduction: Gilles de la Tourette syndrome is characterized by involuntary vocal and/or motor tics. It affects 1-2% of school children and is the most common movement disorder in pediatric group. Spontaneous remission rate is high, and treatment is required for those who failed to improve and experience social problems. Here a 11 year-old girl, who failed different treatments and refused going to school due to tics but showed a dramatic improvement with topiramate, is presented.

Case: She has complaints of bending her neck and body to right side, and making a high-pitched cry for four years, and used, respectively, risperidone (1 year), risperidone + fluoxetine (6 months), pimozide (3 months) and amisulperid (3 months). When she applied with symptoms, she used no medicine, had normal EEG and brain MRI. Second EEG was also normal. Topiramate was initiated, her complaints resolved from the second week on (2 mg/kg/day), and remained tics-free during 5 months follow-up period.

Discussion: The pathophysiology of TS is not clearly understood yet. The most common therapeutic agents include typical narcoleptics (haloperidol, pimozide), atypical narcoleptics (risperidone, aripiprazole, quetiapine, ziprasidone), alpha-adrenergic agonists (clonidine, guanfacine), benzamides (sulpiride, tiapride), selective noradrenaline reuptake inhibitors (atomoxetine) and oxcarbazepine. An optimum treatment guideline could not be established. Controlled studies are not sufficient in the literature. Recommendations include specialists' experiences and preferences (2). Topiramate was reported to reduce tics in some patients.

This case is valuable to highlight topiramate as the treatment option in TS patients who failed conventional pharmacotherapy.

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REDEFINING THE CLINICAL PHENOTYPE OF PSYCHOMOTOR DISABILITIES WITH X-LINKED MCT8 DEFICIENCY: IMPLICATIONS FOR IMPROVED THERAPIES.

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INTRODUCTION: Monocarboxylate Transporter 8 (MCT8) is a thyroid hormone transporter expressed in various human organs, including brain tissue. Loss-of-function mutations in the MCT8 gene, located on the X chromosome, manifest as mild-to-moderate intellectual disability and moderate-to-severe psychomotor impairment in boys with pleasant dispositions and elevated serum T3 levels. While affected boys present with marked axial hypotonia and quadriparesis, they have surprisingly little spasticity early in the disease course. Rather, their hypotonia tends to be accompanied by subtle involuntary movements, specifically dystonia. The lack of spasticity represents a challenge for their rehabilitation and explains why spasticity-directed therapies have produced suboptimal responses. Our goal was to better define the spectrum of motor disabilities in MCT8 deficiency in order to improve the rehabilitation results of patients with this condition.

METHODS: To accomplish our goal, we evaluated a multi-national cohort of patients with MCT8 mutations. Direct clinical evaluations or clinical video recordings (n=6), were reviewed by our pediatric neurogeneticist and/or pediatric movement disorder specialist.

RESULTS: Patient evaluations and videos reviewed on multiple MCT8 deficiency patients revealed a common pattern of hypotonic quadriparesis with superimposed dystonia.

CONCLUSION/DISCUSSION: This multi-national pilot study has better characterized the motor impairments associated with MCT8 deficiency. It is anticipated that this information will allow physicians and therapists to focus their treatment efforts on the hyperkinetic elements of MCT8 deficiency rather than the conventionally suspected spasticity.

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TREATMENT OF TREMOR IN CEREBRAL PALSY WITH DEEP BRAIN STIMULATION

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Introduction: Deep brain stimulation (DBS) of the thalamic ventral intermediate nucleus (VIM) has been used to treat tremor in Parkinson Disease and Essential Tremor, but its use has not been described in the treatment of tremor related to cerebral palsy. We describe the use of DBS for treatment of medically refractory "rubral" tremor in a patient with ataxic cerebral palsy.

Case Description: A 16 year-old female with ataxic cerebral palsy and "rubral" tremor was treated with bilateral VIM DBS after modest benefit from propranolol, clonazepam, and primidone. Prior to DBS, she was unable to draw a spiral with her right hand. Tremor severity was assessed with the Fahn-Tolosa-Marin Tremor Rating Scale (TRS) with DBS off and on.

Results: Her "off" TRS score tremor score was 35/116 with moderate-severe handwriting and spiral drawing impairment. With DBS "on", TRS score was 27 and handwriting and spiral drawing improved slightly. Despite the numerical improvement, the TRS did not capture the full magnitude of her clinical benefit. After DBS, for the first time in her life, she was able to apply her own nail polish and make-up, carry a cup of hot coffee safely, and write more legibly in school. It also had a tremendous benefit for her confidence and hopefulness regarding her personal and career goals.

Conclusion: This case report illustrates the potential therapeutic benefit of VIM DBS to treat tremor associated with cerebral palsy. The TRS may be insensitive to meaningful functional improvement.