MUSCLE & NERVE

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STIFF PERSONS SYNDROME

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Introduction: Stiff-person syndrome (SP) is an uncommon disorder characterized by progressive muscle stiffness, rigidity, and painful spasms involving the axial muscles and proximal portion of lower limbs, resulting in severely impaired ambulation. Phenomena may occur with dysatonomic manifestations

In some occasions it is associated with type 1 diabetes mellitus, or Myasthenia Gravis, showing a pathogenetic features in common.

The diagnosis is usually late, confirmed by the presence of continuous activity of motor unit in paraspinal muscles and in lower limbs in the EMG.

SP presents favorable response to symptomatic and immunomodulatory treatment.

Purpose: We described a patient with clinical and neurophysiological diagnosis of Stiff Person Syndrome detailing the evolution, their clinical characteristics and treatment.

Material and methods: A healthy 13-year-old boy, who started acutely with stiffness, rigidity, hypertonia, painful spams and dysautonomic phenomena.

Results: We found activity of continuous motor unit in EMG / neuromyotonia that affects the muscles of both proximal and distal limbs. Mild sensory and motor polyradiculopathy. Post-synaptic neuromuscular transmission compromise. CK 1900 IU/l. Antibodies antiperoxidase positive, high levels of TSH, and antibodies anti GAD positive in CSF, negative in blood. Antibodies antichannel of potassium negative in blood and CSF. Other studies were negative discarding paraneoplastic syndrome and other autoimmune diseases. Normal brain and spinal cord MRI.

We started treatment with intravenous pulses of gamma globulin and oral carbamazepine with good response.

Conclusions: The diagnosis of this rare autoimmune variant of the disease allowed the establishment of appropriate therapy.

Early recognition and initiation of treatment resulted in notorious improvement.

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CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY SECONDARY (CIDP) TO INFLAMMATORY BOWEL DISEASE (IBD) AND ASSOCIATED TO VITAMIN B12 DEFICIENCY

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Introduction: CIDP is characterized by symmetrical weakness in both proximal and distal muscles that progressively increases for more than two months. It may be associated to auto-immune and infectious conditions. We report a case in which the diagnosis of CIDP has led to a diagnosis of IBD, although the first hypothesis was Vitamin B12 deficiency.

Methods: Case report and literature review on the themes.

Case Description: Twelve-year-old boy was admitted with four-week weakness in the lower limbs without any sensitive alteration or pain. He had a history of chronic diarrhoea since age 4, associated with malnutrition and recurrent bronchospasm. His exam showed gait instability, lower limbs weakness and absence of deep tendon reflexes of both legs. After four weeks hospitalized, evolved with loss of gait and tactile sensitive alteration, worsening bronchospasm and pneumonia. He had Vitamin B12 deficiency, macrocytic anemia, elevated muscle enzymes, normal CSF and hyperintense signal of the conus medullaris on the MRI. The electroneuromyography showed motor-sensitive chronic demyelinating polyrradiculoneuropathy with axonal predominance. After hydroxycobalamin reposition, he had a slight improvement, but only got better after corticosteroid pulse therapy. On corticosteroid suspension, at the follow up, his diarrhoea returned and colonoscopy demonstrated colon stenosis suggesting IBD.

Conclusion/Discussion: CIDP is an underdiagnosed and potentially treatable disease. In our case, although we do not have a nerve biopsy, the clinical pattern and findings on electroneuromyography, besides the response to corticosteroid, can assure the diagnosis of CIDP, especially considering the association with IBD.

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ATYPICAL CLINICAL AND HISTOLOGICAL PRESENTATIONS IN PATIENTS WITH MUTATIONS ON THE RYR1 GENE

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The ryanodine receptor gene (**RYR1**) encodes the sarcoplasmic reticulum calcium release channel RYR1. **RYR1** mutations cause susceptibility to malignant hyperthermia (MH) and various congenital myopathy subtypes (congenital fiber-type disproportion, centronuclear myopathy, central core myopathy and multiminicore myopathy). Here we report clinical, pathologic and genetic features of three patients from two families with **RYR1** mutations and atypical clinical and histological phenotypes. The first case is an 18-year old female patient with stable and generalized muscle weakness, atrophy and skeletal deformities since the first months of life. Her muscle biopsy showed an intense dystrophic process without specific structural defects, and the initial diagnosis was congenital muscular dystrophy. She had the homozygous mutationc.122T>C. The second and third patients are two siblings with neonatal hypotonia, motor developmental delay, proximal weakness, calf hypertrophy, normal CK, no ophthalmoplegia and muscle biopsy compatible with centronuclear myopathy. They are compound heterozygous for two **RYR1** mutations: one frameshift deletion, c.6797-6_6798del, and a missense c.9892G>A. Our findings support the clinical and histological variability associated with mutations on **RYR1** gene, and confirm previous studies that indicate the need to investigate **RYR1** mutations in patients with congenital muscular dystrophies or centronuclear myopathies with no genetic confirmation.

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POLYSOMNOGRAPHIC ABNORMALITIES IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY

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Introduction: Duchenne Muscular Dystrophy (DMD) is a progressive neuromuscular disease of X-linked recessive inheritance caused by a defect in the dystrophin gene. Sleep-disordered breathing is common in these children, being ten times more prevalent than general population. The objective of this study is to analyze the polysomnographic characteristics in children with DMD.

Methods: Cross-sectional study of 29 patients with DMD who underwent a polysomnography between 2005-2011.

Results: Total of 29 patients, mean age 9 years (2-18). 20/29(69%) with preserved ambulation, of these, 19 being treated with corticosteroids (18 prednisone, 1 deflazacort). 9/29(31%) had lost gait (mean age 13 years), of these, 7 receiving corticosteroids (6 prednisone, 1 deflazacort). 3/29 were users of non-invasive ventilation (NIV). 13/29(45 %) had reduced sleep onset latency, 14/29(48%) decreased sleep efficiency, 8/29(27%) with sleep fragmentation. 13/29(44%) with increased superficial sleep and 11/29(37%) decreased REM sleep. Respiratory disturbance index (RDI) was increased in 18/29(62%), 6/18 showed obstructive apnoeas and hypopneas, 11/18 central apnoeas and 2/18 a mixed pattern. Of the 9 patients in wheelchair, 3 had an increased RDI. 2/29 with PCO_2>53 at the time of the exam. Snoring was present in 7/29(24%), of which two were associated with obstructive events. Mean baseline oxygen saturation was 97% and desaturation <90% present in 13/29(45%) patients.

Conclusions: The prevalence of sleep-disordered breathing in children with DMD is relevant. In this study the wheelchair-bound

patients didn't experience more respiratory events than patients who maintained ambulation. The number of patients receiving NIV was low.

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THE INVESTIGATION OF GENETICS, SERUM BIOCHEMISTRY AND PATHOLOGY IN DUCHENNE MUSCULAR DYSTROPHY

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Objective: To investigate the mutation of DMD gene in children, the dynamic of serum CK and transaminase and the correlation between the two enzymes of DMD, DMD muscle pathology, and the correlation with the age.

Methods: 177 boys with DMD were prospectively studied.

Results: (1) By MLPA assay, the deletion mutation was account for 71.17% of DMD and the duplication mutations was account for 10.43%, no gene mutations had been detected in 18.40%. Of 30 children whose gene mutations was not been found, 10 were found DMD gene point mutations with next-generation sequencing technology. (2) The CK, AST, ALT activity in infants with DMD were significantly increased more than the normal 10,7,5 times respectively, and were maintained until the age of 8, then gradually declined with age increasing. Enzymes showed a linear correlation between CK and AST (r=0.817, p<0.01) or CK and ALT (r=0.669, p<0.01) levels. (3) during the infant period of DMD, there were pathological changes, mainly as atrophy and hypertrophy of muscle fibers. At the late stage, there was little number of muscle fibers, fatty connective tissue proliferation. The elder the more serious.

Conclusion: (1) The deletion mutation of DMD gene is the main type of gene mutation. (2) Serum CK, AST, ALT level can reflect the DMD occurrences and progress, and provide an important basis for the early diagnosis of DMD. (3) The severity of clinical illness and pathological changes is associated with the course of the disease and the age of muscle biopsy.

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CLINICAL OF DIFFERENT PHENOTYPES WITH SPINAL MUSCULAR ATROPHY IN CHILDREN

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Objective: To compare the clinical characteristics of spinal muscular atrophy (SMA) in children within different phenotypes.

Methods: Totally 66 cases were enrolled in this study .The comparisons were performed in clinical manifestation, changes of electrophysiology, genetic test of homozygous deletions of survival motor neuron 1 gene (SMN1) and follow-up study. The statistics analysis was done by SPSS software for Windows.

Results: 63.3% of SMA1 patients were characterized by glossopharyngeal paralysis and 30.3% by paradoxical breathing, while the rate ratios were much higher in those with family history (80.0%, 50.0) of which the age onset were younger (1st month). For patients type II and type III, muscle fibrillation and deformity were more common and functional abilities of some patients might improved naturally. EMG in 73.3% of the patients suggests a motor neuron disease. There was no difference between 3 groups, age factor excluded. Approximately 90.5% of the patients in our cohort lacked both copies of SMN I. There was no significant difference in the deletion frequency or subtypes among the 3 groups.

Conclusions: Pulmonary cares are essential for SMA I with family histories in particular. Clinical manifestation of SMA II and SMA III are of great heterogeneity for which a period of follow-up is necessary and differential diagnosis should pay more attention. There is no significant difference in the deletion frequency among the subtypes in electrophysiology and gene tests.

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A CHILD WITH ANTI-GQ1B SYNDROME PRESENTING WITH COMPLETE OPHTHALMOPLEGIA AND UNILATERAL FACIAL PALSY.

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Introduction: The classical phenotype of Miller Fisher syndrome is characterized by ophthalmoplegia, ataxia and areflexia. However, less extensive forms have been described. We report a child with positive anti-GQ1b antibodies with unusual clinical findings.

Methods: The clinical records of a child with positive anti-GQ1b antibodies were reviewed and reported.

Results: a 14-years-old boy presented with headache, double vision and vomiting for 7 days. There was no history of fever, gait disturbances, altered sensorium or difficulty in swallowing. He had a viral respiratory infection 15 days prior to onset of symptoms. Examination revealed complete ophthalmoplegia, right lower-motor-neuron facial palsy, no limb weakness or cerebellar signs and normal fundus. CSF examination was normal with negative oligoclonal bands. MRI brain was normal. Electrophysiological studies done in second week of illness showed normal limb nerve conduction studies, low CMAP amplitude of right facial nerve, abnormal blink reflex and negative repetitive nerve stimulation test. The neostigmine challenge test was negative. Lyme disease serology was negative. Anti-GQ1b antibodies were positive with negative Anti-GM1, -GM2, -GM3, -GD1a, -GD1b, -GT1b antibodies. The child was managed conservatively. The child showed complete recovery after 3 months.

Conclusions: The patients with positive anti-GQ1b antibodies who do not demonstrate the full complement of the Miller Fisher syndrome triad have been reported previously. However, unilateral facial palsy has not been reported previously. This report further expands the phenotypic spectrum of anti-GQ1b syndrome.

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LIMB GIRDLE MYASTHENIA: AN UNCOMMON, TREATABLE CAUSE OF PROXIMAL MUSCLE WEAKNESS IN CHILDREN

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Introduction: Limb girdle myasthenia (LGM) is rare variant of myasthenia and differs from common forms in terms of presentation and distribution of weakness.

Presentation: *Child1*: 8 year-old previously healthy boy presented with severe abdominal pain aggravated on attempting to sit, stand/walk since 10 days without diurnal variation; difficulty in walking, getting up from floor since 4 days. Examination revealed hip girdle, truncal and neck flexor weakness and Gowers' sign. No facial, bulbar/extra-ocular weakness. Sinuous gait was noted when he started walking.

***Child2*:** 11 year, developmentally normal boy presented with severe pain in lower limbs and abdomen, difficulty in sitting, standing/walking since 15 days. He had 4-5 such episodes in past 2 years, improved spontaneously over 7-10 days with asymptomatic period of 3-4 months. Examination-similar to child1.

Management: Electrolytes, Thyroid profile, CPK, Urine-Porphobillinogen, ANA, Anti-AchR and Anti-Musk antibodies, CT chest were normal. NCS: Normal. Repetitive Nerve Stimulation Test: 60% & 90% decrement in nasalis, trapezius in child 1 and 2 respectively. Both showed dramatic improvement following IV Neostigmine, and continued on oral pyridostigmine.

Discussion: LGM is rare form of MG with prominent proximal muscle weakness, little/no ocular, bulbar or facial weakness. Most autoimmune LGM present after third decade and associated with thymoma. Episodic long-term fluctuations (precipitated by febrile illnesses) and absent diurnal fluctuations is characteristic. Cholinesterase inhibitors (pyridostigmine), corticosteroids (in acquired cases) are mainstay of therapy, with some cases requiring IV steroids, IVIG and/or immunomodulation**.**

Conclusion: LGM is rare treatable neuromuscular disorder. High index of suspicion helps in early diagnosis.

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COGNITIVE ASSESSMENT IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY

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Introduction: One third of children with Duchenne muscular dystrophy (DMD) have IQ (Intelligence Quotient) below 70, with a specific profile of cognitive affection and behavioural problems. There is paucity of reports from developing countries.

Objectives: Assessment of cognitive and behavioural impairment in DMD children and its predictors and relevance.

Materials and methods: Steroid naive DMD children were assessed by Malins Intelligence Scale (Indian adaptation of Weschlers intelligence scale) and also underwent MRI Brain with MRS and EEG.

Results and discussion: Twenty children fulfilling inclusion and exclusion were enrolled between March 2010 to February 2012. Mean age at enrolment was 9.52 years. The mean IQ was 78.3 (SD - 9.01) which is statistically significant compared to population mean ($p < 0.001$). Twenty percent had mild mental retardation, 50% had borderline intelligence and 30% had average intelligence (DSM IV criteria). Mean performance IQ was higher than verbal IQ. MRS showed reduced NAA in cerebellum which has not been described previously in literature. There is also increase in the incidence of cognitive impairment in children with deletion towards 3' end of the gene. There was more than expected association of behavioural problems, learning difficulties, sleep disorders and EEG abnormalities in children with DMD.

Conclusion: DMD children have high incidence of cognitive and behavioural impairment which is under recognized and has specific profile of affection. Cognitive assessment and therapy needs to be a part of integral approach in management. This area needs further research to elucidate the pathophysiology of the disease.

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UNUSUAL NEUROIMAGING FINDINGS IN TWO FAMILIES WITH GIANT AXONAL NEUROPATHY

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Introduction: Giant axonal neuropathy (GAN) is a severe autosomal recessive childhood disorder affecting both the peripheral and the central nervous system. The classical MRI brain findings include diffuse cerebral and cerebellar atrophy with white matter changes. We report the clinical, pathological and the unusual neuro-imaging findings in two families with GAN.

Methods: The clinical records of two families with affected children with GAN were reviewed and reported.

Results: In family-1, the index case presented at 8 years of age with global developmental delay, worsening gait abnormalities, poor school performance, no seizures with normal vision and hearing. Examination showed curly hair, spastic diplegia with absent reflexes. Fundus showed optic atrophy and retinitis-pigmentosa. His twin-sister had similar but milder phenotype. An elder sibling died at 16 years of age with similar illness. The MRI Brain showed T2/FLAIR hyperintensities in bilateral cerebellar dentate nuclei and internal-capsule In family-2, the index case presented at 8 years of age with clinical phenotype similar to the index case of family 1 except for hypotonia. The nerve conduction studies revealed sensori-motor axonal-neuropathy. The MRI-Brain showed T2/FLAIR hyperintensities in bilateral cerebellar dentate nuclei and globus pallidii. The diagnosis of GAN was based on the clinical phenotype, neuroimaging and the nerve biopsy. The nerve biopsy in both the cases revealed several giant-axons dispersed within the fascicles, variably labelled by NF-protein.

Conclusions: These cases further expand the neuroiamging spectrum seen in children with GAN.

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PSYCHOSOCIAL INTERVENTION PROGRAMME FOR FAMILIES OF PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY

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Introduction: Children with Duchenne Muscular Dystrophy and their families go through significant stress, which has been explored in previous research. Little effort has been made to report interventions targeting these issues. The present study aims to assess an intervention with families of children with DMD based on psychosocial typology of chronic genetic conditions.

Method: A pretest post test experimental design was used. Caregivers of 36 children with DMD attending neuromuscular disorders clinic of a tertiary referral center for Neurology were studied. All the children had the diagnosis of DMD confirmed by genetic analysis or absence of dystrophin staining on muscle Immunohistochemistry, were males, in the age range of 4 - 13 years. Caregivers included fathers (19), mothers (17) or both in age range of 27 – 43 yrs. All the parents received individualized intervention based on the psycho social conceptualization of chronic disease by Rolland (1987) targeting specific areas of psychosocial needs identified, caregiver knowledge & attitude, burden of care giving and coping.

Results: The programme was effective in increasing the Knowledge, attitude and wellbeing, and there was reduction in Family burden. There was shifting of coping strategies from emotional avoidant to active problem focused coping. The limitations of the study and its implications for practice, training and research will be discussed.

Conclusion: Parents will benefit from psychosocial intervention to help them develop coping skills to meet their children's needs and their own.

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ARRAY-CGH TECHNOLOGY IN THE DISCOVERY OF X-CHROMOSOME COPY NUMBER VARIANTS IN MALE PATIENTS WITH MENTAL RETARDATION SYNDROMES

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Introduction: Array comparative genomic hybridization (aCGH) is a high throughput and high resolution technique for the detection of chromosomal copy number variations (CNVs) in the form of deletions, amplifications and gains. Mental retardation is a genetically heterogeneous disorder and more than 90 genes for this disorder have been found on the X chromosome alone. CNVs of X-chromosome (X-CNVs) may be a significant cause of mental retardation in male patients. In this study, the application of this technology has enabled the detection of X-CNVs in males with mental retardation.

Methods: aCGH was performed on patients presented with non-syndromic mental retardation from Pantai Hospital Kuala Lumpur, Malaysia using the Oxford Gene Technology (OGT) microarray platform to identify CNVs and their location on the chromosome.

Results: Three patients have been identified to have CNVs at a specific region in chromosome X (Xq21.1). Of the three patients, two show a variant duplication resulting in an extra genetic material, while one has a deletion.

Discussion: The region of X-CNVs detected encompasses the MAGT1 (Magnesium transporter protein 1) gene. MAGT1 is involved in N-glycosylation through its association with N-oligosaccharyltransferase and in Mg2+ transport in epithelial cells. Based on a study conducted by Molinari et al. (2008), a mutation in the MAGT1 gene was identified in an Australian family where 4 out of 5 children suffered from mild to severe X-linked mental retardation.

Conclusion: This shows that aCGH technology can help in the identification of the clinically relevant X-CNVs in males with mental retardation syndromes.

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SÍNDROME SOMATOMORFO POSTERIOR A LA VACUNACIÓN CONTRA LA HEPATITIS B NOTIFICADO COMO ESAVI Y SU IMPACTO EN LA VACUNACIÓN CONTRA LA HEPATITIS B EN ICA, PERÚ

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Introducción: En el año 2007, el Ministerio de Salud organizó una campaña nacional de vacunación contra la hepatitis B con el objetivo de inmunizar a la mayor parte de la población. Reportamos la evaluación de un caso notificado como ESAVI y su impacto sanitario en un departamento del Perú.

Métodos: El sistema de vigilancia recibió la notificación de un Evento Supuestamente Atribuido a la Vacunación o Inmunización (ESAVI)

producido luego de la vacunación contra la hepatitis B. El caso fue evaluado por clínicos del Instituto Nacional de Salud del Niño (INSN) y por el Comité Nacional Asesor para la Clasificación de casos de ESAVI.

Descripción del caso: Adolescente mujer de 14 años que presenta cefalea intensa y agitación psicomotriz después de diez minutos de la vacunación. Es hospitalizada y diagnosticada de "encefalitis post vacunal", siendo luego transferida al INSN. La prensa informó del caso produciéndose alarma en la población. Las autoridades políticas suspendieron la campaña de vacunación. La paciente fue evaluada clínicamente y a través de exámenes de labratorio, resonancia magnética y electroencefalograma, con lo cual el Comité de ESAVI clasifica el caso como Sindrome Somatomorfo y como evento no relacionado a la vacuna.

Discusión: La evaluación integral identificó problemas psicológicos y sociales de fondo que incluían acoso sexual y maltrato. La vigilancia de ESAVI permitió el conocimiento del caso y la clasificación final. El error diagnóstico inicial como "necefalitis post-vacunal" ocasionó una inadecuada respuesta política y de la presenta, produciéndose un impacto sanitario negativo. Las coberturas de vacunación para hepatitis B y para otras vacunas cayeron a niveles críticos por más de un año en el departamento de Ica.

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POLIOMIELITISAGUDAPOSVACUNAL EN EL PERÚREPORTE DE CINCOCASOS Y ANÁLISIS DEL IMPACTO SOCIAL Y SANITARIO ENTRE LOS AÑOS 2009 Y 2011

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Introducción: La Vacuna de Poliovirus Oral (OPV en inglés) permitió la erradicación del virus salvaje de lasAméricas en 1994. Se sabeque la OPV puedeproducircasos de poliomielitisparalítica en tasasmenores a 1 casoporcadamillón de niñosvacunados.

Métodos: Reportamoscincocasos de poliomielitisparalíticaasociada y derivada de la vacunapresentados en el Perú entre los años 2009 y 2011.

Descripción de los casos: En dos de los casosreportados se aisló en heces un poliovirus con menos del 1% de variaciónrespecto al virus vacunal (casoasociado) y enuno de los casos se aisló un poliovirus con 2% de variación (casoderivado). Sin embargo, en los otros dos casos no se pudodeterminar el porcentaje de variaciónporhabersidodetectadostardíamente.

Discusión: La VigilanciaEpidemiológica de lasParálisisFláccidaspermitió la detección de los casosreportados. La identificación de un casoderivadosignificaqueexiste la probabilidad de otroscasos, por lo quelascoberturas de vacunación y vigilanciaepidemiológicadebenseroptimizadas. Se implementó la vacunación con OPV casa por casa y luego un "barrido" de vacunaciónnacional con OPV. La prensageneró un gran impacto social quellevó a modificar el esquema de vacunaciónintroduciendo la vacuna parenteral (IPV) y la entrega de unacompensacióneconómicapor parte del estado a lasfamilias.

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DOES ACUTE MOTOR AXONAL NEUROPATHY HURT?

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Guillain-Barré Syndrome is an acute immune-mediated polyradiculoneuropathy comprising a broad spectrum of clinical variants, which include the Acute Inflammatory Demyelinating Polyneuropathy, Miller Fisher Syndrome and the axonal loss variants, Acute Motor Axonal Neuropathy (AMAN) and Acute Motor and Sensory Axonal Neuropathy (AMSAN). AMAN is a rare pure motor axonal variant characterized by acute onset of distal weakness, hyporeflexia/areflexia and electrodiagnostic evidence of reduced compound muscle action potential amplitudes with normal motor conduction velocities and normal sensory nerve action potentials. Only a few cases are reported describing pain in AMAN, but this is not the norm. We describe the case of a 12-year-old girl diagnosed with AMAN after development of acute symmetric ascending weakness, painful dysesthesias without sensory deficits, generalized hyporeflexia, autonomic instability and electrodiagnostic evidence of pure motor axonal loss. Two courses of intravenous immunoglobulins were given with significant improvement of upper extremity weakness and mild improvement of painful dysesthesias after the second course. Lessening of pain occurred upon initiation of gabapentin that lasted for about 3 to 5 days, after which pain returned to initial intensity. Combined treatment with gabapentin, intravenous morphine, fentanyl patch, and physiotherapy was unsuccessful providing analgesia. This case presents the presence of pain in AMAN, which had been recently described in the literature. Further studies are needed to understand the pathophysiologic mechanisms of pain in AMAN to determine the best pain management options for this condition.

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RELIABILITY OF THE EK SCALE VERSUS HAND GRIP DYNAMOMETER TESTING IN NON-AMBULANT DUCHENNE MUSCULAR DYSTROPHY PATIENTS

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Background: Duchenne muscular dystrophy (DMD) is a progressive neuromuscular disorder where later disease progress is difficult to objectively measure. This study compared the Egen Klassification (EK) scale with grip strength testing in non-ambulant DMD patients.

Methods: This was a retrospective review of patients seen in the neuromuscular clinic at the National University Hospital from March 2011 to June 2013. EK scale was applied by trained physiotherapists. Strength testing with hand grip and pinch was performed with hand grip dynamometer in the dominant hand. EK scores and grip strength were correlated with age, functional stage, and lung function parameters.

Results: 19 non-ambulant DMD males aged 11 – 25 years (mean 17.4 years) were recruited. The mean EK score was 13.47 (range 4-27). EK score showed significant positive correlation with functional stage [r = 0.92 ($p<0.001$)] and age [r = 0.746 ($p<0.001$)]. EK showed significant negative correlation with FEV1 [r of -0.619 ($p<0.05$)] and FVC [r -0.497 ($p<0.05$)]. Mean grip strength was 3.92 kg (range of 1-19 kg). Hand grip showed negative correlation with patient's functional stage [r -0.456 ($p<0.05$)]. EK did not show any significant correlation with hand grip strength.

Conclusion: The EK scale was an easily used scale for the outpatient clinic setting with good correlation with lung function parameters as well as functional stage of the disease. Paradoxically this scale outperformed objective grip strength testing. Periodic assessment of non-ambulant patients should ideally comprise a combination of functional and objective strength testing especially in patients with limited motor abilities.

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TRIPLICATION OF PMP22 GENE REGION ASSOCIATED WITH CHARCOT-MARIE-TOOTH DISEASE-1A

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Hereditary peripheral neuropathies present a group of clinically and genetically heterogeneous entities. All known forms, including the various forms of Charcot-Marie-Tooth disease (CMT) are characterized as Mendelian traits and over 45 genes have been identified thus far. The mutational mechanism of the most common CMT type, CMT1A, is a 1.5 Mb chromosomal duplication at 17p12 that contains the gene PMP22. Deletion of PMP22 gene is associated with a different peripheral neuropathy, hereditary neuropathy with liability to pressure palsies (HNPP).

We report a case of nine years old boy seen in our out-patient department with CMT-1A whose microarray analysis revealed triplication in the region of PMP22 gene on the short arm of one chromosome 17. He presented with history of delayed walking at 4 years of age on the background of being floppy during infancy. Clinical examination revealed mild atrophy of calf muscles, mild hypotonia of lower limbs, pescavus, absent deep tendon reflexes and a high stepping gait. Nerve conduction revealed severe demyelination neuropathy. In the family history his father, paternal aunt and paternal grandfather had history

suggestive of lower motor neurone weakness but we did not have any further details.

Triplication of PMP22 region presenting with classical clinical and electrophysiological signs of CMT-1A has been described only once before to our knowledge. Whether it alters the long-term outcome of the condition is not known.

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INFANTILE AXONAL NEUROPATHY -A CASE SERIES

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Introduction: Early onset axonal neuropathies are a rare group of disorders associated with muscle weakness and early mortality. It is often difficult to elucidate an exact genetic mutation, largely due to a wide range of possible mutations. This poses ethical dilemma for professionals and families, in continuing (or withdrawing) intensive care due to scarce information regarding prognosis.

Methods: Review of case notes, investigations, clinical history and examination of patients.

Case Series: We present the clinical presentations, neurophysiological findings, and course of four infants with severe axonal neuropathy. All four infants presented within the first few weeks of life with poor feeding and or respiratory difficulties. Two needed assisted ventilation, one from birth and the other from 3 months of age, until the point they died. Neurophysiology showed purely motor axonal neuropathy in two infants and a sensorimotor axonal neuropathy in the other two. MRI of brain and spine were normal in three cases and showed cerebellar hypoplasia in one. Three of them died between 4 and 6 months and one survives with significant weakness at sixteen months. Extensive genetic investigations did not reveal any specific mutation. Nerve biopsy was performed in two of them; one showed giant axonal neuropathy and the other showed axonal neuropathy with no other diagnostic features.

Conclusion: This paper highlights the high rate of early mortality and the extremely low chance of finding a definitive cause in axonal neuropathies in infants. It also addresses the ethical issues that families and professionals face in managing such infants.

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PREPUBERTAL MYASTHENIA GRAVIS-IS INFECTION A TRIGGER OR THE CAUSE?

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Objective: Myasthenia gravis (MG) is an acquired autoimmune disorder of neuromuscular junction. Prepubertal children have a higher prevalence of ocular symptoms (OMG), lower frequency of acetylcholine receptor antibodies (AChR), and higher probability of achieving remission. We define presenting features and outcomes in our cohort.

Methods: We identified four prepubertal children with MG.

Results: Two males (M) and two females (F) with age of onset at 3(F1), 2.5(M1), 1.5(F2), and 10y (M2) were reviewed. All patients were tensilon test positive. They had electrophysiology, including the repetitive nerve stimulation (RNS) tests. All patients had antibodies sent against AChR and Muscle specific Kinase (MuSK). Neuroimaging was done in three who presented with acute symptoms.

Three children (F1, M1, F2) presented acutely following viral infections which included Chicken pox, diarrhoeal illness and herpetic stomatitis respectively. F1 presented with ptosis and bilateral divergent squint and was initially diagnosed with internuclearopthalmoplegia. Months later was diagnosed with OMG following positive tensilon. AChR antibodies are positive. Ptosis resolved with pyridostigmine but the ophthalmoplegia partially persists. F2 and M1 presented with ptosis and generalised weakness (M1). RNS showed a decrement in both. Both are antibody negative. Symptoms resolved with pyridostigmine in both.

M2 presented insidiously, is antibody positive and has a refractory course needing thymectomy, plasma exchange and rituximab.

Discussion: Could the relatively higher remission rates and lower levels of acetylcholine receptor antibodies seen in prepubertal MG be related to a post infectious autoimmune phenomenon? Or is there a genetic predisposition which is triggered by infection? A high index of suspicion is necessary to make an early diagnosis.